Clinical Genetics

Cat02 SCI/SSCI journal (WI-1)

The Pathogenesis of Ventral Idiopathic Herniation of the Spinal Cord: A Hypothesis Based on the Review of the Literature
Bartels, R. H. M. A., Brunner, H., Hosman, A., van Alfen, N. & Grotenhuis, J. A. 11 Sep 2017 In : Frontiers in Neurology. 8, 10 p., 476
Research output: Scientific - peer-review › Article

International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases
Boycott, K. M. , Rath, A. , Chong, J. X. , Hartley, T. , Alkuraya, F. S. , Baynam, G. , Brookes, A. J. , Brudno, M. , Carracedo, A. , den Dunnen, J. T. , Dyke, S. O. M. , Estivill, X. , Goldblatt, J. , Gonthier, C. , Groft, S. C. , Gut, I. , Hamosh, A. , Hieter, P. , Hoehn, S. , Hurles, M. E. & 20 others Kaufmann, P., Knoppers, B. M., Krischer, J. P., Macek, M., Matthijs, G., Olry, A., Parker, S., Paschall, J., Philippakis, A. A., Rehm, H. L., Robinson, P. N., Sham, P-C., Stefanov, R., Taruscio, D., Unni, D., Vanstone, M. R., Zhang, F., Brunner, H., Bamshad, M. J. & Lochmueller, H. 4 May 2017 In : American Journal of Human Genetics. 100, 5, p. 695-705 11 p.
Research output: Scientific - peer-review › Article

Heterozygous *HNRNPU* variants cause early onset epilepsy and severe intellectual disability
Bramswig, N. C. , Luedecke, H-J. , Hamdan, F. F. , Altmueller, J. , Beleggia, F. , Elcioglu, N. H. , Freyer, C. , Gerkes, E. H. , Demirkol, Y. K. , Knupp, K. G. , Kuechler, A. , Li, Y. , Lowenstein, D. H. , Michaud, J. L. , Park, K. , Stegmann, A. P. A. , Veenstra-Knol, H. E. , Wieland, T. , Wollnik, B. , Engels, H. & 3 others Strom, T. M., Kleefstra, T. & Wieczorek, D. Jul 2017 In : Human Genetics. 136, 7, p. 821-834 14 p.
Research output: Scientific - peer-review › Article

Expanding the clinical spectrum of recessive truncating mutations of KLHL7 to a Bohring-Opitz-like phenotype
Bruel, A-L. , Bigoni, S. , Kennedy, J. , Whiteford, M. , Buxton, C. , Parmeggiani, G. , Wherlock, M. , Woodward, G. , Greenslade, M. , Williams, M. , St-Onge, J. , Ferlini, A. , Garani, G. , Ballardini, E. , van Bon, B. W. , Acuna-Hidalgo, R. , Bohring, A. , Deleuze, J-F. , Boland, A. , Meyer, V. & 10 others Olaso, R., Ginglinger, E., Riviere, J-B., Brunner, H. G., Hoischen, A., Newbury-Ecob, R., Faivre, L., Thauvin-Robinet, C., Thevenon, J. & DDD Study Dec 2017 In : Journal of Medical Genetics. 54, 12, p. 830-835 6 p.
Research output: Scientific - peer-review › Article

Classic Galactosemia: Study on the Late Prenatal Development of GALT Specific Activity in a Sheep Model
Coelho, A. I., Bierau, J., Lindhout, M., Achten, J., Kramer, B. W. & Rubio-Gozalbo, M. E. Sep 2017 In : The Anatomical Record: advances in integrative anatomy and evolutionary biology. 300, 9, p. 1570-1575 6 p.
Research output: Scientific - peer-review › Article

Effects of preparatory and action planning instructions on situation specific and general fruit and snack intake
de Bruijn, G-J., Minh Hao Nguyen, Rhodes, R. E. & van Osch, L. 1 Jan 2017 In : Appetite. 108, p. 161-170 10 p.
Research output: Scientific - peer-review › Article

Estimates of live birth prevalence of children with Down syndrome in the period 1991-2015 in the Netherlands
de Graaf, G., Engelen, J. J. M., Gijsbers, A. C. J., Hochstenbach, R., Hoffer, M. J. V., Kooper, A. J. A., Sikkema-Raddatz, B., Srebniak, M. I., van der Kevie-Kersemaekers, A. M. F., van Zutven, L. J. C. M. & Voorhoeve, E. May 2017 In : Journal of Intellectual Disability Research. 61, 5, p. 461-470 10 p.
Research output: Scientific - peer-review › Article

The 6p25 deletion syndrome: An update on a rare neurocristopathy
de Vos, I. J. H. M., Stegmann, A. P. A., Webers, C. A. B. & Stumpel, C. T. R. M. 2017 In : Ophthalmic Genetics. 38, 2, p. 101-107 7 p.
Research output: Scientific - peer-review › Review article

Review of familial cerebral cavernous malformations and report of seven additional families
de Vos, I. J. H. M., Vreeburg, M., Koek, G. H. & van Steensel, M. A. M. Feb 2017 In : American Journal of Medical Genetics Part A. Part A 173A, 2, p. 338-351 14 p.
Research output: Scientific - peer-review › Review article

BRCA1 mutation carriers have a lower number of mature oocytes after ovarian stimulation for IVF/PGD
Derks-Smeets, I. A. P., van Tilborg, T. C., van Montfoort, A., Smits, L., Torrance, H. L., Meijer-Hoogeveen, M., Broekmans, F., Dreesen, J. C. F. M., Paulussen, A. D. C., Tjan-Heijnen, V. C. G., Homminga, I., van den Berg, M. M. J., Ausems, M. G. E. M., de Rycke, M., de Die-Smulders, C. E. M., Verpoest, W. & van Golde, R. Nov 2017 In : Journal of Assisted Reproduction and Genetics. 34, 11, p. 1475-1482 8 p.
Research output: Scientific - peer-review › Article

Uptake of prenatal diagnostic testing for retinoblastoma compared to other hereditary cancer syndromes in the Netherlands
Dommering, C. J., Henneman, L., van der Hout, A. H., Jonker, M. A., Tops, C. M. J., van den Ouweland, A. M. W., van der Luijt, R. B., Mensenkamp, A. R., Hogervorst, F. B. L., Redeker, E. J. W., de Die-Smulders, C. E. M., Moll, A. C. & Meijers-Heijboer, H. Apr 2017 In : Familial Cancer. 16, 2, p. 271-277 7 p.
Research output: Scientific - peer-review › Article

Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability
Dudding-Byth, T., Baxter, A., Holliday, E. G., Hackett, A., O'Donnell, S., White, S. M., Attia, J., Brunner, H., de Vries, B., Koolen, D., Kleefstra, T., Ratwatte, S., Riveros, C., Brain, S. & Lovell, B. C. 19 Dec 2017 In : BMC Biotechnology. 17, 9 p., 90
Research output: Scientific - peer-review › Article

Allelic Expression Imbalance Promoting a Mutant *PEX6* Allele Causes Zellweger Spectrum Disorder
Falkenberg, K. D., Braverman, N. E., Moser, A. B., Steinberg, S. J., Klouwer, F. C. C., Schluter, A., Ruiz, M., Pujol, A., Engvall, M., Naess, K., van Spronsen, F., Korver-Keularts, I., Rubio-Gozalbo, M. E., Ferdinandusse, S., Wanders, R. J. A. & Waterham, H. R. 7 Dec 2017 In : American Journal of Human Genetics. 101, 6, p. 965-976 12 p.
Research output: Scientific - peer-review › Article

TSC2 c.1864C > T Variant Aassociated with Mild Cases of Tuberous Sclerosis Complex
Farach, L. S., Gibson, W. T., Sparagana, S. P., Nellist, M., Stumpel, C. T. R. M., Hietala, M., Friedman, E., Pearson, D. A., Creighton, S. P., Wagemans, A., Segel, R., Ben-Shalom, E., Au, K. S. & Northrup, H. Mar 2017 In : American Journal of Medical Genetics Part A. 173, 3, p. 771-775 5 p.
Research output: Scientific - peer-review › Article

The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families
Fountain, M. D. , Aten, E. , Cho, M. T. , Juusola, J. , Walkiewicz, M. A. , Ray, J. W. , Xia, F. , Yang, Y. , Graham, B. H. , Bacino, C. A. , Potocki, L. , van Haeringen, A. , Ruivenkamp, C. A. L. , Mancias, P. , Northrup, H. , Kukolich, M. K. , Weiss, M. M. , van Ravenswaaij-Arts, C. M. A. , Mathijssen, I. B. , Levesque, S. & 19 others Meeks, N., Rosenfeld, J. A., Lemke, D., Hamosh, A., Lewis, S. K., Race, S., Stewart, L. L., Hay, B., Lewis, A. M., Guerreiro, R. L., Bras, J. T., Martins, M. P., Derksen-Lubsen, G., Peeters, E., Stumpel, C., Stegmann, S., Bok, L. A., Santen, G. W. E. & Schaaf, C. P. Jan 2017 In : Genetics in Medicine. 19, 1, p. 45-52 8 p.
Research output: Scientific - peer-review › Article

YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction
Gabriele, M. , Vulto-van Silfhout, A. T. , Germain, P-L. , Vitriolo, A. , Kumar, R. , Douglas, E. , Haan, E. , Kosaki, K. , Takenouchi, T. , Rauch, A. , Steindl, K. , Frengen, E. , Misceo, D. , Pedurupillay, C. R. J. , Stromme, P. , Rosenfeld, J. A. , Shao, Y. , Craigen, W. J. , Schaaf, C. P. , Rodriguez-Buritica, D. & 31 others Farach, L., Friedman, J., Thulin, P., McLean, S. D., Nugent, K. M., Morton, J., Nicholl, J., Andrieux, J., Stray-Pedersen, A., Chambon, P., Patrier, S., Lynch, S. A., Kjaergaard, S., Torring, P. M., Brasch-Andersen, C., Ronan, A., van Haeringen, A., Anderson, P. J., Powis, Z., Brunner, H. G., Pfundt, R., Schuurs-Hoeijmakers, J. H. M., van Bon, B. W. M., Lelieveld, S., Gilissen, C., Nillesen, W. M., Vissers, L. E. L. M., Gecz, J., Koolen, D. A., Testa, G. & de Vries, B. B. A. 1 Jun 2017 In : American Journal of Human Genetics. 100, 6, p. 907-925 19 p.
Research output: Scientific - peer-review › Article

Awareness and attitude regarding reproductive options of persons carrying a BRCA mutation and their partners
Gietel-Habets, J. J. G., de Die-Smulders, C. E. M., Derks-Smeets, I. A. P., Tibben, A., Tjan-Heijnen, V. C. G., van Golde, R., Gomez-Garcia, E., Kets, C. M. & van Osch, L. A. D. M. Mar 2017 In : Human Reproduction. 32, 3, p. 588-597 10 p.
Research output: Scientific - peer-review › Article

Diagnostic exome sequencing in 266 Dutch patients with visual impairment
Haer-Wigman, L. , van Zelst-Stams, W. A. G. , Pfundt, R. , van den Born, L. I. , Klaver, C. C. W. , Verheij, J. B. G. M. , Hoyng, C. B. , Breuning, M. H. , Boon, C. J. F. , Kievit, A. J. , Verhoeven, V. J. M. , Pott, J. W. R. , Sallevelt, S. C. E. H. , van Hagen, J. M. , Plomp, A. S. , Kroes, H. Y. , Lelieveld, S. H. , Hehir-Kwa, J. Y. , Castelein, S. , Nelen, M. & 5 others Scheffer, H., Lugtenberg, D., Cremers, F. P. M., Hoefsloot, L. & Yntema, H. G. May 2017 In : European Journal of Human Genetics. 25, 5, p. 591-599 9 p.
Research output: Scientific - peer-review › Article

Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3
Hamdi, Y. , Soucy, P. , Kuchenbaeker, K. B. , Pastinen, T. , Droit, A. , Lemacon, A. , Adlard, J. , Aittomaki, K. , Andrulis, I. L. , Arason, A. , Arnold, N. , Arun, B. K. , Azzollini, J. , Bane, A. , Barjhoux, L. , Barrowdale, D. , Benitez, J. , Berthet, P. , Blok, M. J. , Bobolis, K. & 148 others Bonadona, V., Bonanni, B., Bradbury, A. R., Brewer, C., Buecher, B., Buys, S. S., Caligo, M. A., Chiquette, J., Chung, W. K., Claes, K. B. M., Daly, M. B., Damiola, F., Davidson, R., De la Hoya, M., De Leeneer, K., Diez, O., Ding, Y. C., Dolcetti, R., Domchek, S. M., Dorfling, C. M., Eccles, D., Eeles, R., Einbeigi, Z., Ejlertsen, B., Engel, C., Evans, D. G., Feliubadalo, L., Foretova, L., Fostira, F., Foulkes, W. D., Fountzilas, G., Friedman, E., Frost, D., Ganschow, P., Ganz, P. A., Garber, J., Gayther, S. A., Gerdes, A-M., Glendon, G., Godwin, A. K., Goldgar, D. E., Greene, M. H., Gronwald, J., Hahnen, E., Hamann, U., Hansen, T. V. O., Hart, S., Hays, J. L., Hogervorst, F. B. L., Hulick, P. J., Imyanitov, E. N., Isaacs, C., Izatt, L., Jakubowska, A., James, P., Janavicius, R., Jensen, U. B., John, E. M., Joseph, V., Just, W., Kaczmarek, K., Karlan, B. Y., Kets, C. M., Kirk, J., Kriege, M., Laitman, Y., Laurent, M., Lazaro, C., Leslie, G., Lester, J., Lesueur, F., Liljegren, A., Loman, N., Loud, J. T., Manoukian, S., Mariani, M., Mazoyer, S., McGuffog, L., Meijers-Heijboer, H. E. J., Meindl, A., Miller, A., Montagna, M., Mulligan, A. M., Nathanson, K. L., Neuhausen, S. L., Nevanlinna, H., Nussbaum, R. L., Olah, E., Olopade, O. I., Ong, K., Oosterwijk, J. C., Osorio, A., Papi, L., Park, S. K., Pedersen, I. S., Peissel, B., Segura, P. P., Peterlongo, P., Phelan, C. M., Radice, P., Rantala, J., Rappaport-Fuerhauser, C., Rennert, G., Richardson, A., Robson, M., Rodriguez, G. C., Rookus, M. A., Schmutzler, R. K., Sevenet, N., Shah, P. D., Singer, C. F., Slavin, T. P., Snape, K., Sokolowska, J., Sonderstrup, I. M. H., Southey, M., Spurdle, A. B., Stadler, Z., Stoppa-Lyonnet, D., Sukiennicki, G., Sutter, C., Tan, Y., Tea, M-K., Teixeira, M. R., Teule, A., Teo, S-H., Terry, M. B., Thomassen, M., Tihomirova, L., Tischkowitz, M., Tognazzo, S., Toland, A. E., Tung, N., van den Ouweland, A. M. W., van der Luijt, R. B., van Engelen, K., van Rensburg, E. J., Varon-Mateeva, R., Wappenschmidt, B., Wijnen, J. T., Rebbeck, T., Chenevix-Trench, G., Offit, K., Couch, F. J., Nord, S., Easton, D. F., Antoniou, A. C. & Simard, J. Jan 2017 In : Breast Cancer Research and Treatment. 161, 1, p. 117-134 18 p.
Research output: Scientific - peer-review › Article

Novel *SLC25A32* mutation in a patient with a severe neuromuscular phenotype
Hellebrekers, D. M., Sallevelt, S. C. E. H., Theunissen, T. E. J., Hendrickx, A. T. M., Gottschalk, R. W., Hoeijmakers, J. G. J., Habets, D. D., Bierau, J., Schoonderwoerd, K. G. & Smeets, H. J. M. Jun 2017 In : European Journal of Human Genetics. 25, 7, p. 886-888 3 p.
Research output: Scientific - peer-review › Article

Novel genetic loci associated with hippocampal volume

Hibar, D. P. , Adams, H. H. H. , Jahanshad, N. , Chauhan, G. , Stein, J. L. , Hofer, E. , Renteria, M. E. , Bis, J. C. , Arias-Vasquez, A. , Ikram, M. K. , Desrivieres, S. , Vernooij, M. W. , Abramovic, L. , Alhusaini, S. , Amin, N. , Andersson, M. , Arfanakis, K. , Aribisala, B. S. , Armstrong, N. J. , Athanasiu, L. & 312 others Axelsson, T., Beecham, A. H., Beiser, A., Bernard, M., Blanton, S. H., Bohlken, M. M., Boks, M. P., Bralten, J., Brickman, A. M., Carmichael, O., Chakravarty, M. M., Chen, Q., Ching, C. R. K., Chouraki, V., Cuellar-Partida, G., Crivello, F., Den Braber, A., Nhat Trung Doan, Ehrlich, S., Giddaluru, S., Goldman, A. L., Gottesman, R. F., Grimm, O., Griswold, M. E., Guadalupe, T., Gutman, B. A., Hass, J., Haukvik, U. K., Hoehn, D., Holmes, A. J., Hoogman, M., Janowitz, D., Jia, T., Jorgensen, K. N., Karbalai, N., Kasperaviciute, D., Kim, S., Klein, M., Kraemer, B., Lee, P. H., Liewald, D. C. M., Lopez, L. M., Luciano, M., Macare, C., Marquand, A. F., Matarin, M., Mather, K. A., Mattheisen, M., McKay, D. R., Milaneschi, Y., Maniega, S. M., Nho, K., Nugent, A. C., Nyquist, P., Loohuis, L. M. O., Oosterlaan, J., Papmeyer, M., Pirpamer, L., Puetz, B., Ramasamy, A., Richards, J. S., Risacher, S. L., Roiz-Santianez, R., Rommelse, N., Ropele, S., Rose, E. J., Royle, N. A., Rundek, T., Saemann, P. G., Saremi, A., Satizabal, C. L., Schmaal, L., Schork, A. J., Shen, L., Shin, J., Shumskaya, E., Smith, A. V., Sprooten, E., Strike, L. T., Teumer, A., Tordesillas-Gutierrez, D., Toro, R., Trabzuni, D., Trompet, S., Vaidya, D., Van der Grond, J., Van der Lee, S. J., Van der Meer, D., Van Donkelaar, M. M. J., Van Eijk, K. R., Van Erp, T. G. M., Van Rooij, D., Walton, E., Westlye, L. T., Whelan, C. D., Windham, B. G., Winkler, A. M., Wittfeld, K., Woldehawariat, G., Wolf, C., Wolfers, T., Yanek, L. R., Yang, J., Zijdenbos, A., Zwiers, M. P., Agartz, I., Almasy, L., Ames, D., Amouyel, P., Andreassen, O. A., Arepalli, S., Assareh, A. A., Barral, S., Bastin, M. E., Becker, D. M., Becker, J. T., Bennett, D. A., Blangero, J., van Bokhoven, H., Boomsma, D. I., Brodaty, H., Brouwer, R. M., Brunner, H. G., Buckner, R. L., Buitelaar, J. K., Bulayeva, K. B., Cahn, W., Calhoun, V. D., Cannon, D. M., Cavalleri, G. L., Cheng, C-Y., Cichon, S., Cookson, M. R., Corvin, A., Crespo-Facorro, B., Curran, J. E., Czisch, M., Dale, A. M., Davies, G. E., De Craen, A. J. M., De Geus, E. J. C., De Jager, P. L., De Zubicaray, G. I., Deary, I. J., Debette, S., DeCarli, C., Delanty, N., Depondt, C., DeStefano, A., Dillman, A., Djurovic, S., Donohoe, G., Drevets, W. C., Duggirala, R., Dyer, T. D., Enzinger, C., Erk, S., Espeseth, T., Fedko, I. O., Fernandez, G., Ferrucci, L., Fisher, S. E., Fleischman, D. A., Ford, I., Fornage, M., Foroud, T. M., Fox, P. T., Francks, C., Fukunaga, M., Gibbs, J. R., Glahn, D. C., Gollub, R. L., Goring, H. H. H., Green, R. C., Gruber, O., Gudnason, V., Guelfi, S., Haberg, A. K., Hansell, N. K., Hardy, J., Hartman, C. A., Hashimoto, R., Hegenscheid, K., Heinz, A., Le Hellard, S., Hernandez, D. G., Heslenfeld, D. J., Ho, B-C., Hoekstra, P. J., Hoffmann, W., Hofman, A., Holsboer, F., Homuth, G., Hosten, N., Hottenga, J-J., Huentelman, M., Pol, H. E. H., Ikeda, M., Jack, C. R., Jenkinson, M., Johnson, R., Joensson, E. G., Jukema, J. W., Kahn, R. S., Kanai, R., Kloszewska, I., Knopman, D. S., Kochunov, P., Kwok, J. B., Lawrie, S. M., Lemaitre, H., Liu, X., Longo, D. L., Lopez, O. L., Lovestone, S., Martinez, O., Martinot, J-L., Mattay, V. S., McDonald, C., McIntosh, A. M., McMahon, F. J., McMahon, K. L., Mecocci, P., Melle, I., Meyer-Lindenberg, A., Mohnke, S., Montgomery, G. W., Morris, D. W., Mosley, T. H., Muhleisen, T. W., Mueller-Myhsok, B., Nalls, M. A., Nauck, M., Nichols, T. E., Niessen, W. J., Nothen, M. M., Nyberg, L., Ohi, K., Olvera, R. L., Ophoff, R. A., Pandolfo, M., Paus, T., Pausova, Z., Penninx, B. W. J. H., Pike, G. B., Potkin, S. G., Psaty, B. M., Reppermund, S., Rietschel, M., Roffman, J. L., Romanczuk-Seiferth, N., Rotter, J. I., Ryten, M., Sacco, R. L., Sachdev, P. S., Saykin, A. J., Schmidt, R., Schmidt, H., Schofield, P. R., Sigursson, S., Simmons, A., Singleton, A., Sisodiya, S. M., Smith, C., Smoller, J. W., Soininen, H., Steen, V. M., Stott, D. J., Sussmann, J. E., Thalamuthu, A., Toga, A. W., Traynor, B. J., Troncoso, J., Tsolaki, M., Tzourio, C., Uitterlinden, A. G., Hernandez, M. C. V., Van der Brug, M., van der Lugt, A., van der Wee, N. J. A., Van Haren, N. E. M., van't Ent, D., Van Tol, M-J., Vardarajan, B. N., Vellas, B., Veltman, D. J., Voelzke, H., Walter, H., Wardlaw, J. M., Wassink, T. H., Weale, M. E., Weinberger, D. R., Weiner, M. W., Wen, W., Westman, E., White, T., Wong, T. Y., Wright, C. B., Zielke, R. H., Zonderman, A. B., Martin, N. G., Van Duijn, C. M., Wright, M. J., Longstreth, W. T., Schumann, G., Grabe, H. J., Franke, B., Launer, L. J., Medland, S. E., Seshadri, S., Thompson, P. M. & Ikram, M. A. 18 Jan 2017 In : Nature Communications. 8, 12 p., 13624

Research output: Scientific - peer-review › Article

Food ingestion in an upright sitting position increases postprandial amino acid availability when compared with food ingestion in a lying down position

Holwerda, A. M., Lenaerts, K., Bierau, J., Wodzig, W. K. W. H. & van Loon, L. J. C. Jul 2017 In : Applied Physiology Nutrition and Metabolism-Physiologie appliquee nutrition et metabolisme. 42, 7, p. 738-743 6 p.

Research output: Scientific - peer-review › Article

Lamin A/C-Related Cardiac Disease Late Onset With a Variable and Mild Phenotype in a Large Cohort of Patients With the Lamin A/C p.(Arg331Gln) Founder Mutation
Hoorntje, E. T. , Bollen, I. A. , Barge-Schaapveld, D. Q. , van Tienen, F. H. , te Meerman, G. J. , Jansweijer, J. A. , van Essen, A. J. , Volders, P. G. , Constantinescu, A. A. , van den Akker, P. C. , van Spaendonck-Zwarts, K. Y. , Oldenburg, R. A. , Marcelis, C. L. , van der Smagt, J. J. , Hennekam, E. A. , Vink, A. , Bootsma, M. , Aten, E. , Wilde, A. A. , van den Wijngaard, A. & 5 others Broers, J. L., Jongbloed, J. D., van der Velden, J., van den Berg, M. P. & van Tintelen, J. P. Aug 2017 In : Circulation : Cardiovascular Genetics. 10, 4, 36 p., 001631
Research output: Scientific - peer-review › Article

Delusional and Psychotic Disorders in Juvenile Myotonic Dystrophy Type-1
Jacobs, D., Willekens, D., de Die-Smulders, C., Frijns, J-P. & Steyaert, J. Jun 2017 In : American Journal of Medical Genetics Part B-neuropsychiatric Genetics. 174, 4, p. 359-366 8 p.
Research output: Scientific - peer-review › Article

De Novo Truncating Mutations in the Last and Penultimate Exons of *PPM1D* Cause an Intellectual Disability Syndrome
Jansen, S. , Geuer, S. , Pfundt, R. , Brough, R. , Ghongane, P. , Herkert, J. C. , Marco, E. J. , Willemsen, M. H. , Kleefstra, T. , Hannibal, M. , Shieh, J. T. , Lynch, S. A. , Flinter, F. , FitzPatrick, D. R. , Gardham, A. , Bernhard, B. , Ragge, N. , Newbury-Ecob, R. , Bernier, R. , Kvarnung, M. & 10 others Magnusson, E. A. H., Wessels, M. W., van Slegtenhorst, M. A., Monaghan, K. G., de Vries, P., Veltman, J. A., Lord, C. J., Vissers, L. E. L. M., de Vries, B. B. A. & Deciphering Dev Disorders Study 6 Apr 2017 In : American Journal of Human Genetics. 100, 4, p. 650-658 9 p.
Research output: Scientific - peer-review › Article

Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification
Kamps, R., Brandao, R. D., van den Bosch, B. J., Paulussen, A. D. C., Xanthoulea, S., Blok, M. J. & Romano, A. Feb 2017 In : International Journal of Molecular Sciences. 18, 2, 57 p., 308
Research output: Scientific - peer-review › Review article

Network topology of NaV1.7 mutations in sodium channel-related painful disorders
Kapetis, D., Sassone, J., Yang, Y., Galbardi, B., Xenakis, M. N., Westra, R. L., Szklarczyk, R., Lindsey, P., Faber, C. G., Gerrits, M., Merkies, I. S. J., Dib-Hajj, S. D., Mantegazza, M., Waxman, S. G., PROPANE Study Grp, J.M. Smeets, H. & Lauria, G. 24 Feb 2017 In : BMC Systems Biology. 11, 16 p., 28
Research output: Scientific - peer-review › Article

From action planning and plan enactment to fruit consumption: moderated mediation effects
Kasten, S., van Osch, L., Eggers, S. M. & de Vries, H. 23 Oct 2017 In : BMC Public Health. 17, 11 p., 832
Research output: Scientific - peer-review › Article

Structural asymmetries of the human cerebellum in relation to cerebral cortical asymmetries and handedness
Kavaklioglu, T., Guadalupe, T., Zwiers, M., Marquand, A. F., Onnink, M., Shumskaya, E., Brunner, H., Fernandez, G., Fisher, S. E. & Francks, C. May 2017 In : Brain Structure & Function. 222, 4, p. 1611-1623 13 p.
Research output: Scientific - peer-review › Article

Novel IRF6 Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing
Khandelwal, K. D. , Ishorst, N. , Zhou, H. , Ludwig, K. U. , Venselaar, H. , Gilissen, C. , Thonissen, M. , van Rooij, I. A. L. M. , Dreesen, K. , Steehouwer, M. , van de Vorst, M. , Bloemen, M. , van Beusekom, E. , Roosenboom, J. , Borstlap, W. , Admiraal, R. , Dormaar, T. , Schoenaers, J. , Vander Poorten, V. , Hens, G. & 10 others Verdonck, A., Berge, S., Roeleveldt, N., Vriend, G., Devriendt, K., Brunner, H. G., Mangold, E., Hoischen, A., van Bokhoven, H. & Carels, C. E. L. Feb 2017 In : Journal of Dental Research. 96, 2, p. 179-185 7 p.
Research output: Scientific - peer-review › Article

CAD mutations and uridine-responsive epileptic encephalopathy
Koch, J. , Mayr, J. A. , Alhaddad, B. , Rauscher, C. , Bierau, J. , Kovacs-Nagy, R. , Coene, K. L. M. , Bader, I. , Holzhacker, M. , Prokisch, H. , Venselaar, H. , Wevers, R. A. , Distelmaier, F. , Polster, T. , Leiz, S. , Betzler, C. , Strom, T. M. , Sperl, W. , Meitinger, T. , Wortmann, S. B. & 1 others Haack, T. B. Feb 2017 In : Brain. 140, p. 279-286 8 p.
Research output: Scientific - peer-review › Article

Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder
Koemans, T. S., Kleefstra, T., Chubak, M. C., Stone, M. H., Reijnders, M. R. F., de Munnik, S., Willemsen, M. H., Fenckova, M., Stumpel, C. T. R. M., Bok, L. A., Saenz, M. S., Byerly, K. A., Baughn, L. B., Stegmann, A. P. A., Pfundt, R., Zhou, H., van Bokhoven, H., Schenck, A. & Kramer, J. M. Oct 2017 In : Plos Genetics. 13, 10, 24 p., 1006864
Research output: Scientific - peer-review › Article

Both basal and post-prandial muscle protein synthesis rates, following the ingestion of a leucine-enriched whey protein supplement, are not impaired in sarcopenic older males
Kramer, I. F., Verdijk, L. B., Hamer, H. M., Verlaan, S., Luiking, Y. C., Kouw, I. W. K., Senden, J. M., van Kranenburg, J., Gijsen, A. P., Bierau, J., Poeze, M. & van Loon, L. J. C. Oct 2017 In : Clinical Nutrition. 36, 5, p. 1440-1449 10 p.
Research output: Scientific - peer-review › Article

Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype
Lamers, I. J. C. , Reijnders, M. R. F. , Venselaar, H. , Kraus, A. , Jansen, S. , de Vries, B. B. A. , Houge, G. , Gradek, G. A. , Seo, J. , Choi, M. , Chae, J-H. , van der Burgt, I. , Pfundt, R. , Letteboer, S. J. F. , van Beersum, S. E. C. , Dusseljee, S. , Brunner, H. G. , Doherty, D. , Kleefstra, T. , Roepman, R. & 1 others DDD Study 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 824-832 9 p.
Research output: Scientific - peer-review › Article

Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes
Lelieveld, S. H., Wiel, L., Venselaar, H., Pfundt, R., Vriend, G., Veltman, J. A., Brunner, H. G., Vissers, L. E. L. M. & Gilissen, C. 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 478-484 7 p.
Research output: Scientific - peer-review › Article

***De Novo* Missense Mutations in *DHX30* Impair Global Translation and Cause a Neurodevelopmental Disorder**
Lessel, D. , Schob, C. , Kuery, S. , Reinders, M. R. F. , Harel, T. , Eldomery, M. K. , Coban-Akdemir, Z. , Denecke, J. , Edvardson, S. , Colin, E. , Stegmann, A. P. A. , Gerkes, E. H. , Tessarech, M. , Bonneau, D. , Barth, M. , Besnard, T. , Cogne, B. , Revah-Politi, A. , Strom, T. M. , Rosenfeld, J. A. & 24 others Yang, Y., Posey, J. E., Immken, L., Oundjian, N., Helbig, K. L., Meeks, N., Zegar, K., Morton, J., Schieving, J. H., Claasen, A., Huentelman, M., Narayanan, V., Ramsey, K., Brunner, H. G., Elpeleg, O., Mercier, S., Bezieau, S., Kubisch, C., Kleefstra, T., Kindler, S., Lupski, J. R., Kreienkamp, H-J., DDD study & C4RCD Res Grp 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 716-724 9 p.
Research output: Scientific - peer-review › Article

Palmitate-Induced Vacuolar-Type H(+)-ATPase Inhibition Feeds Forward Into Insulin Resistance and Contractile Dysfunction
Liu, Y., Steinbusch, L. K. M., Nabben, M., Kapsokalyvas, D., van Zandvoort, M., Schonleitner, P., Antoons, G., Simons, P. J., Coumans, W. A., Geomini, A., Chanda, D., Glatz, J. F. C., Neumann, D. & Luiken, J. J. F. P. 1 Jun 2017 In : Diabetes. 66, 6, p. 1521-1534 14 p.
Research output: Scientific - peer-review › Article

Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability
Marom, R. , Jain, M. , Burrage, L. C. , Song, I-W. , Graham, B. H. , Brown, C. W. , Stevens, S. J. C. , Stegmann, A. P. A. , Gunter, A. T. , Kaplan, J. D. , Gavrilova, R. H. , Shinawi, M. , Rosenfeld, J. A. , Bae, Y. , Tran, A. A. , Chen, Y. , Lu, J. T. , Gibbs, R. A. , Eng, C. , Yang, Y. & 4 others Rousseau, J., de Vries, B. B. A., Campeau, P. M. & Lee, B. Oct 2017 In : Human Mutation. 38, 10, p. 1365-1371 7 p.
Research output: Scientific - peer-review › Article

B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies
Maroofian, R., Riemersma, M., Jae, L. T., Zhianabed, N., Willemsen, M. H., Wissink-Lindhout, W. M., Willemsen, M. A., de Brouwer, A. P. M., Mehrjardi, M. Y. V., Ashrafi, M. R., Kusters, B., Kleefstra, T., Jamshidi, Y., Nasseri, M., Pfundt, R., Brummelkamp, T. R., Abbaszadegan, M. R., Lefeber, D. J. & van Bokhoven, H. 22 Dec 2017 In : Genome Medicine. 9, 11 p., 118
Research output: Scientific - peer-review › Article

COL6A5 variants in familial neuropathic chronic itch
Martinelli-Boneschi, F. , Colombi, M. , Castori, M. , Devigili, G. , Eleopra, R. , Malik, R. A. , Ritelli, M. , Zoppi, N. , Dordoni, C. , Sorosina, M. , Grammatico, P. , Fadavi, H. , Gerrits, M. M. , Almomani, R. , Faber, C. G. , Merkies, I. S. J. , Toniolo, D. , Cocca, M. , Doglioni, C. , Waxman, S. G. & 10 others Dib-Hajj, S. D., Taiana, M. M., Sassone, J., Lombardi, R., Cazzato, D., Zauli, A., Santoro, S., Marchi, M., Lauria, G. & INGI Network Mar 2017 In : Brain. 140, p. 555-567 13 p.
Research output: Scientific - peer-review › Article

Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia
Meyer, E. , Carss, K. J. , Rankin, J. , Nichols, J. M. E. , Grozeva, D. , Joseph, A. P. , Mencacci, N. E. , Papandreou, A. , Ng, J. , Barra, S. , Ngoh, A. , Ben-Pazi, H. , Willemsen, M. A. , Arkadir, D. , Barnicoat, A. , Bergman, H. , Bhate, S. , Boys, A. , Darin, N. , Foulds, N. & 53 others Gutowski, N., Hills, A., Houlden, H., Hurst, J. A., Israe, Z., Kaminska, M., Limousin, P., Lumsden, D., Mckee, S., Misra, S., Mohammed, S. S., Nakou, V., Nicolai, J., Nilsson, M., Pall, H., Peall, K. J., Peters, G. B., Prabhakar, P., Reuter, M. S., Rump, P., Sege, R., Sinnema, M., Smith, M., Turnpenny, P., White, S. M., Wieczorek, D., Wiethoff, S., Wilson, B. T., Winter, G., Wragg, C., Pope, S., Heales, S. J. H., Morrogh, D., Pittman, A., Carr, L. J., Perez-Duenas, B., Lin, J-P., Reis, A., Gahl, W. A., Toro, C., Bhatia, K. P., Wood, N. W., Kamsteeg, E-J., Chong, W. K., Gissen, P., Topf, M., Dale, R. C., Chubby, J. R., Raymond, F. L., Kurian, M. A., UK10K Consortium, Deciphering Dev Disorders Study & NIHR BioResource Rare Feb 2017 In : Nature Genetics. 49, 2, p. 223-237 15 p.
Research output: Scientific - peer-review › Article

Mendelian Disorders of Cornification Caused by Defects in Intracellular Calcium Pumps: Mutation Update and Database for Variants in ATP2A2 and ATP2C1 Associated with Darier Disease and Hailey-Hailey Disease
Nellen, R. G. L., Steijlen, P. M., van Steensel, M. A. M., Vreeburg, M., Frank, J., van Geel, M. & European Professional Contributors Apr 2017 In : Human Mutation. 38, 4, p. 343-356 14 p.
Research output: Scientific - peer-review › Article

Duplicated Enhancer Region Increases Expression of CTSB and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families
Ngcungcu, T. , Oti, M. , Sitek, J. C. , Haukanes, B. I. , Linghu, B. , Bruccoleri, R. , Stokowy, T. , Oakeley, E. J. , Yang, F. , Zhu, J. , Sultan, M. , Schalkwijk, J. , van Vlijmen-Willems, I. M. J. J. , von der Lippe, C. , Brunner, H. G. , Ersland, K. M. , Grayson, W. , Buechmann-Moller, S. , Sundnes, O. , Nirmala, N. & 9 others Morgan, T. M., van Bokhoven, H., Steen, V. M., Hull, P. R., Szustakowski, J., Staedtler, F., Zhou, H., Fiskerstrand, T. & Ramsay, M. 4 May 2017 In : American Journal of Human Genetics. 100, 5, p. 737-750 14 p.
Research output: Scientific - peer-review › Article

Novel pathogenic SLC25A46 splice-site mutation causes an optic atrophy spectrum disorder
Nguyen, M., Boesten, I., Hellebrekers, D. M. E. I., Mulder-den Hartog, N. M., de Coo, I. F. M., Smeets, H. J. M. & Gerards, M. Jan 2017 In : Clinical Genetics. 91, 1, p. 121-125 5 p.
Research output: Scientific - peer-review › Article

Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome
Oud, M. M. , Tuijnenburg, P. , Hempel, M. , van Vlies, N. , Ren, Z. , Ferdinandusse, S. , Jansen, M. H. , Santer, R. , Johannsen, J. , Bacchelli, C. , Alders, M. , Li, R. , Davies, R. , Dupuis, L. , Cale, C. M. , Wanders, R. J. A. , Pals, S. T. , Ocaka, L. , James, C. , Mueller, I. & 18 others Lehmberg, K., Strom, T., Engels, H., Williams, H. J., Beales, P., Roepmand, R., Dias, P., Brunner, H. G., Cobben, J-M., Hall, C., Hartley, T., Stabej, P. L. Q., Mendoza-Londono, R., Davies, E. G., de Sousa, S. B., Lesse, D., Arts, H. H. & Kuijpers, T. W. 2 Feb 2017 In : American Journal of Human Genetics. 100, 2, p. 281-296 16 p.
Research output: Scientific - peer-review › Article

NGS panel analysis in 24 ectopia lentis patients; a clinically relevant test with a high diagnostic yield
Overwater, E. , Floor, K. , van Beek, D. , de Boer, K. , van Dijk, T. , Hilhorst-Hofstee, Y. , Hoogeboom, A. J. M. , van Kaam, K. J. , van de Kamp, J. M. , Kempers, M. , Krapels, I. P. C. , Kroes, H. Y. , Loeys, B. , Salemink, S. , Stumpel, C. T. R. M. , Verhoeven, V. J. M. , Wijnands-van den Berg, E. , Cobben, J. M. , van Tintelen, J. P. , Weiss, M. M. & 2 others Houweling, A. C. & Maugeri, A. Sep 2017 In : European Journal of Medical Genetics. 60, 9, p. 465-473 9 p.
Research output: Scientific - peer-review › Article

Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders

Pfundt, R. , del Rosario, M. , Vissers, L. E. L. M. , Kwint, M. P. , Janssen, I. M. , de Leeuw, N. , Yntema, H. G. , Nelen, M. R. , Lugtenberg, D. , Kamsteeg, E-J. , Wieskamp, N. , Stegmann, A. P. A. , Stevens, S. J. C. , Rodenburg, R. J. T. , Simons, A. , Mensenkamp, A. R. , Rinne, T. , Gilissen, C. , Scheffer, H. , Veltman, J. A. & 1 others Hehir-Kwa, J. Y. Jun 2017 In : Genetics in Medicine. 19, 6, p. 667-675 9 p.

Research output: Scientific - peer-review › Article

Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer

Phelan, C. M. , Kuchenbaecker, K. B. , Tyrer, J. P. , Kar, S. P. , Lawrenson, K. , Winham, S. J. , Dennis, J. , Pirie, A. , Riggan, M. J. , Chornokur, G. , Earp, M. A. , Lyra, P. C. , Lee, J. M. , Coetzee, S. , Beesley, J. , McGuffog, L. , Soucy, P. , Dicks, E. , Lee, A. , Barrowdale, D. & 400 others Lecarpentier, J., Leslie, G., Aalfs, C. M., Aben, K. K. H., Adams, M., Adlard, J., Andrulis, I. L., Anton-Culver, H., Antonenkova, N., Aravantinos, G., Arnold, N., Arun, B. K., Arver, B., Azzollini, J., Balmana, J., Banerjee, S. N., Barjhoux, L., Barkardottir, R. B., Bean, Y., Beckmann, M. W., Beeghly-Fadiel, A., Benitez, J., Bermisheva, M., Bernardini, M. Q., Birrer, M. J., Bjorge, L., Black, A., Blankstein, K., Blok, M. J., Bodelon, C., Bogdanova, N., Bojesen, A., Bonanni, B., Borg, A., Bradbury, A. R., Brenton, J. D., Brewer, C., Brinton, L., Broberg, P., Brooks-Wilson, A., Bruinsma, F., Brunet, J., Buecher, B., Butzow, R., Buys, S. S., Caldes, T., Caligo, M. 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Research output: Scientific - peer-review › Article

The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies
Redin, C. , Brand, H. , Collins, R. L. , Kammin, T. , Mitchell, E. , Hodge, J. C. , Hanscom, C. , Pillalamarri, V. , Seabra, C. M. , Abbott, M-A. , Abdul-Rahman, O. A. , Aberg, E. , Adley, R. , Alcaraz-Estrada, S. L. , Alkuraya, F. S. , An, Y. , Anderson, M-A. , Antolik, C. , Anyane-Yeboa, K. , Atkin, J. F. & 129 others Bartell, T., Bernstein, J. A., Beyer, E., Blumenthal, I., Bongers, E. M. H. F., Brilstra, E. H., Brown, C. W., Bruggenwirth, H. T., Callewaert, B., Chiang, C., Corning, K., Cox, H., Cuppen, E., Currall, B. B., Cushing, T., David, D., Deardorff, M. A., Dheedene, A., D'Hooghe, M., de Vries, B. B. A., Earl, D. L., Ferguson, H. L., Fisher, H., FitzPatrick, D. R., Gerrol, P., Giachino, D., Glessner, J. T., Gliem, T., Grady, M., Graham, B. H., Griffis, C., Gripp, K. W., Gropman, A. L., Hanson-Kahn, A., Harris, D. J., Hayden, M. A., Hill, R., Hochstenbach, R., Hoffman, J. D., Hopkin, R. J., Hubshman, M. W., Innes, A. M., Irons, M., Irving, M., Jacobsen, J. C., Janssens, S., Jewett, T., Johnson, J. P., Jongmans, M. C., Kahler, S. G., Koolen, D. A., Korzelius, J., Kroisel, P. M., Lacassie, Y., Lawless, W., Lemyre, E., Leppig, K., Levin, A. V., Li, H., Li, H., Liao, E. C., Lim, C., Lose, E. J., Lucente, D., Macera, M. J., Manavalan, P., Mandrile, G., Marcelis, C. L., Margolin, L., Mason, T., Masser-Frye, D., McClellan, M. W., Mendoza, C. J. Z., Menten, B., Middelkamp, S., Mikami, L. R., Moe, E., Mohammed, S., Mononen, T., Mortenson, M. E., Moya, G., Nieuwint, A. W., Ordulu, Z., Parkash, S., Pauker, S. P., Pereira, S., Perrin, D., Phelan, K., Pina Aguilar, R. E., Poddighe, P. J., Pregno, G., Raskin, S., Reis, L., Rhead, W., Rita, D., Renkens, I., Roelens, F., Ruliera, J., Rump, P., Schilit, S. L. P., Shaheen, R., Sparkes, R., Spiegel, E., Stevens, B., Stone, M. R., Tagoe, J., Thakuria, J. V., van Bon, B. W., van de Kamp, J., van Der Burgt, I., van Essen, T., van Ravenswaaij-Arts, C. M., van Roosmalen, M. J., Vergult, S., Volker-Touw, C. M. L., Warburton, D. P., Waterman, M. J., Wiley, S., Wilson, A., Yerena-de Vega, M. D. L. C. A., Zori, R. T., Levy, B., Brunner, H. G., de Leeuw, N., Kloosterman, W. P., Thorland, E. C., Morton, C. C., Gusella, J. F. & Talkowski, M. E. Jan 2017 In : Nature Genetics. 49, 1, p. 36-45 10 p.
Research output: Scientific - peer-review › Article

Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability
Reijnders, M. R. F. , Kousi, M. , van Woerden, G. M. , Klein, M. , Bralten, J. , Mancini, G. M. S. , van Essen, T. , Proietti-Onori, M. , Smeets, E. E. J. , van Gastel, M. , Stegmann, A. P. A. , Stevens, S. J. C. , Lelieveld, S. H. , Gilissen, C. , Pfundt, R. , Tan, P. L. , Kleefstra, T. , Franke, B. , Elgersma, Y. , Katsanis, N. & 1 others Brunner, H. G. 20 Oct 2017 In : Nature Communications. 8, 12 p., 1052
Research output: Scientific - peer-review › Article

RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes
Reijnders, M. R. F. , Ansor, N. M. , Kousi, M. , Yue, W. W. , Tan, P. L. , Clarkson, K. , Clayton-Smith, J. , Corning, K. , Jones, J. R. , Lam, W. W. K. , Mancini, G. M. S. , Marcelis, C. , Mohammed, S. , Pfundt, R. , Roifman, M. , Cohn, R. , Chitayat, D. , Millard, T. H. , Katsanis, N. , Brunner, H. G. & 2 others Banka, S. & Deciphering Dev Disorders Study 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 466-477 12 p.
Research output: Scientific - peer-review › Article

Postzygotic mosaicism in basal cell naevus syndrome
Reinders, M. G. H. C., Boersma, H. J., Leter, E. M., Vreeburg, M., Paulussen, A. D. C., Arits, A. H. M. M., Roemen, G. M. J. M., Speel, E. J. M., Steijlen, P. M., van Geel, M. & Mosterd, K. Jul 2017 In : British Journal of Dermatology. 177, 1, p. 249-252 4 p.
Research output: Scientific - peer-review › Article

Propionic acidemia as a cause of adult-onset dilated cardiomyopathy
Riemersma, M., Hazebroek, M. R., Helderman-van den Enden, A. T. J. M., Salomons, G. S., Ferdinandusse, S., Brouwers, M. C. G. J., van der Ploeg, L., Heymans, S., Glatz, J. F. C., van den Wijngaard, A., Krapels, I. P. C., Bierau, J. & Brunner, H. G. Nov 2017 In : European Journal of Human Genetics. 25, 11, p. 1195-1201 7 p.
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
Sallevelt, S. C. E. H., de Koning, B., Szklarczyk, R., Paulussen, A. D. C., de Die-Smulders, C. E. M. & Smeets, H. J. M. May 2017 In : Genetics in Medicine. 19, 5, p. 583-592 10 p.
Research output: Scientific - peer-review › Article

De novo mtDNA point mutations are common and have a low recurrence risk
Sallevelt, S. C. E. H., de Die-Smulders, C. E. M., Hendrickx, A. T. M., Hellebrekers, D. M. E. I., de Coo, I. F. M., Alston, C. L., Knowles, C., Taylor, R. W., McFarland, R. & Smeets, H. J. M. Feb 2017 In : Journal of Medical Genetics. 54, 2, p. 114-124 11 p.
Research output: Scientific - peer-review › Article

PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Druesedau, M., Hellebrekers, D. M. E. I., Paulussen, A. D. C., Coonen, E., Van Golde, R. J. T., Geraedts, J. P. M., Gianaroli, L., Magli, M. C., Zeviani, M., Smeets, H. J. M. & de Die-Smulders, C. E. M. Mar 2017 In : Human Reproduction. 32, 3, p. 698-703 6 p.
Research output: Scientific - peer-review › Article

Associations of plasma uric acid and purine metabolites with blood pressure in children: the KOALA Birth Cohort Study
Scheepers, L. E. J. M., Boonen, A., Pijnenburg, W., Bierau, J., Staessen, J. A., Stehouwer, C. D. A., Thijs, C. & Arts, I. C. W. May 2017 In : Journal of Hypertension. 35, 5, p. 982-993 12 p.
Research output: Scientific - peer-review › Article

Growth Hormone Therapy in Children with Kabuki Syndrome: 1-year Treatment Results
Schott, D. A., Gerver, W. J. M. & Stumpel, C. T. R. M. 2017 In : Hormone Research in Paediatrics. 88, 3-4, p. 1-7 7 p.
Research output: Scientific - peer-review › Article

The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands
Seco, C. Z. , Wesdorp, M. , Feenstra, I. , Pfundt, R. , Hehir-Kwa, J. Y. , Lelieveld, S. H. , Castelein, S. , Gilissen, C. , de Wijs, I. J. , Admiraal, R. J. C. , Pennings, R. J. E. , Kunst, H. P. M. , van de Kamp, J. M. , Tamminga, S. , Houweling, A. C. , Plomp, A. S. , Maas, S. M. , Gans, P. A. M. D. K. , Kant, S. G. , de Geus, C. M. & 10 others Frints, S. G. M., Vanhoutte, E. K., van Dooren, M. F., van den Boogaard, M-J. H., Scheffer, H., Nelen, M., Kremer, H., Hoefsloot, L., Schraders, M. & Yntema, H. G. Feb 2017 In : European Journal of Human Genetics. 25, 3, p. 308-314 7 p.
Research output: Scientific - peer-review › Article

A homozygous FITM2 mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy
Seco, C. Z. , Castells-Nobau, A. , Joo, S. , Schraders, M. , Foo, J. N. , van der Voet, M. , Velan, S. S. , Nijhof, B. , Oostrik, J. , de Vrieze, E. , Katana, R. , Mansoor, A. , Huynen, M. , Szklarczyk, R. , Oti, M. , Tranebjaerg, L. , van Wijk, E. , Scheffer-de Gooyert, J. M. , Siddique, S. , Baets, J. & 10 others de Jonghe, P., Kazmi, S. A. R., Sadananthan, S. A., van de Warrenburg, B. P., Khor, C. C., Goepfert, M. C., Qamar, R., Schenck, A., Kremer, H. & Siddiqi, S. 1 Feb 2017 In : Disease Models & Mechanisms. 10, 2, p. 105-118 14 p.
Research output: Scientific - peer-review › Article

De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects
Slavotinek, A. , Risolino, M. , Losa, M. , Cho, M. T. , Monaghan, K. G. , Schneidman-Duhovny, D. , Parisotto, S. , Herkert, J. C. , Stegmann, A. P. A. , Miller, K. , Shur, N. , Chui, J. , Muller, E. , DeBrosse, S. , Szot, J. O. , Chapman, G. , Pachter, N. S. , Winlaw, D. S. , Mendelsohn, B. A. , Dalton, J. & 7 others Sarafoglou, K., Karachunski, P. I., Lewis, J. M., Pedro, H., Dunwoodie, S. L., Selleri, L. & Shieh, J. 15 Dec 2017 In : Human Molecular Genetics. 26, 24, p. 4849-4860 12 p.
Research output: Scientific - peer-review › Article

Heritability in a SCN5A-mutation founder population with increased female susceptibility to non-nocturnal ventricular tachyarrhythmia and sudden cardiac death
ter Bekke, R. M. A., Isaacs, A., Barysenka, A., Hoos, M. B., Jongbloed, J. D. H., Hoorntje, J. C. A., Patelski, A. S. M., Helderman-van den Enden, A. T. J. M., van den Wijngaard, A., Stoll, M. & Volders, P. G. A. Dec 2017 In : Heart Rhythm. 14, 12, p. 1873-1881 9 p.
Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

Women's Experience with Non-Invasive Prenatal Testing and Emotional Well-being and Satisfaction after Test-Results
van Schendel, R. V., Page-Christiaens, G. C. M. L., Beulen, L., Bilardo, C. M., de Boer, M. A., Coumans, A. B. C., Faas, B. H. W., van Langen, I. M., Lichtenbelt, K. D., van Maarle, M. C., Macville, M. V. E., Oepkes, D., Pajkrt, E., Henneman, L. & Dutch NIPT Consortium Dec 2017 In : Journal of Genetic Counseling. 26, 6, p. 1348-1356 9 p.
Research output: Scientific - peer-review › Article

A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology
Vissers, L. E. L. M., van Nimwegen, K. J. M., Schieving, J. H., Kamsteeg, E-J., Kleefstra, T., Yntema, H. G., Pfundt, R., van der Wilt, G. J., Krabbenborg, L., Brunner, H. G., van der Burg, S., Grutters, J., Veltman, J. A. & Willemsen, M. A. A. P. Sep 2017 In : Genetics in Medicine. 19, 9, p. 1055-1063 9 p.
Research output: Scientific - peer-review › Article

Quantification of Phenotype Information Aids the Identification of Novel Disease Genes
Vulto-van Silfhout, A. T., Gilissen, C., Goeman, J. J., Jansen, S., van Amen-Hellebrekers, C. J. M., van Bon, B. W. M., Koolen, D. A., Sistermans, E. A., Brunner, H. G., de Brouwer, A. P. M. & de Vries, B. B. A. May 2017 In : Human Mutation. 38, 5, p. 594-599 6 p.
Research output: Scientific - peer-review › Article

Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas
Weren, R. D. A., Mensenkamp, A. R., Simons, M., Eijkelenboom, A., Sie, A. S., Ouchene, H., van Asseldonk, M., Gomez-Garcia, E. B., Blok, M. J., de Hullu, J. A., Nelen, M. R., Hoischen, A., Bulten, J., Tops, B. B. J., Hoogerbrugge, N. & Ligtenberg, M. J. L. Feb 2017 In : Human Mutation. 38, 2, p. 226-235 10 p.
Research output: Scientific - peer-review › Article

The hypoxanthine-xanthine oxidase axis is not involved in the initial phase of clinical transplantation-related ischemia-reperfusion injury
Wijermars, L. G. M., Bakker, J. A., de Vries, D. K., van Noorden, C. J. F., Bierau, J., Kostidis, S., Mayboroda, O. A., Tsikas, D., Schaapherder, A. F. & Lindeman, J. H. N. 1 Mar 2017 In : American Journal of Physiology-Renal Physiology. 312, 3, p. F457-F464 8 p.
Research output: Scientific - peer-review › Article

The tumour suppressor CDKN2A/p16(INK4a) regulates adipogenesis and bone marrow-dependent development of perivascular adipose tissue
Wouters, K., Deleye, Y., Hannou, S. A., Vanhoutte, J., Marechal, X., Coisne, A., Tagzirt, M., Derudas, B., Bouchaert, E., Duhem, C., Vallez, E., Schalkwijk, C. G., Pattou, F., Montaigne, D., Staels, B. & Paumelle, R. Nov 2017 In : Diabetes & Vascular Disease Research. 14, 6, p. 516-524 9 p.
Research output: Scientific - peer-review › Article

Mutations in the Chromatin Regulator Gene *BRPF1* Cause Syndromic Intellectual Disability and Deficient Histone Acetylation
Yan, K. , Rousseau, J. , Littlejohn, R. O. , Kiss, C. , Lehman, A. , Rosenfeld, J. A. , Stumpel, C. T. R. , Stegmann, A. P. A. , Robak, L. , Scaglia, F. , Thi Tuyet Mai Nguyen, , Fu, H. , Ajeawung, N. F. , Camurri, M. V. , Li, L. , Gardham, A. , Panis, B. , Almannai, M. , Sacoto, M. J. G. , Baskin, B. & 16 others Ruivenkamp, C., Xia, F., Bi, W., Cho, M. T., Potjer, T. P., Santen, G. W. E., Parker, M. J., Canham, N., McKinnon, M., Potocki, L., MacKenzie, J. J., Roeder, E. R., Campeau, P. M., Yang, X-J., DDD Study & CAUSES Study 5 Jan 2017 In : American Journal of Human Genetics. 100, 1, p. 91-104 14 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

Homozygosity Mapping and Targeted Sanger Sequencing Identifies Three Novel CRB1 (Crumbs homologue 1) Mutations in Iranian Retinal Degeneration Families
Ghofrani, M., Yahyaei, M., Brunner, H. G., Cremers, F. P. M., Movasat, M., Imran Khan, M. & Keramatipour, M. Sep 2017 In : Iranian Biomedical Journal. 21, 5, p. 294-302 9 p.
Research output: Scientific - peer-review › Article

A novel mutation in L1CAM causes a mild form of L1 syndrome: a case report
Otter, M., Wevers, M., Pisters, M., Pfundt, R., Vos, Y., Nievelstein, R. J. & Stumpel, C. Aug 2017 In : Clinical Case Reports. 5, 8, p. 1213-1217 5 p.
Research output: Scientific - peer-review › Article

Nutrients required for phospholipid synthesis are lower in blood and cerebrospinal fluid in mild cognitive impairment and Alzheimer's disease dementia
van Wijk, N., Slot, R. E. R., Duits, F. H., Strik, M., Biesheuvel, E., Sijben, J. W. C., Blankenstein, M. A., Bierau, J., van der Flier, W. M., Scheltens, P. & Teunissen, C. E. 2017 In : Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring. 8, p. 139-146 8 p.
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 A: Editorial Material in wi-1 journal

The annual meeting 1988-2017
Brunner, H. Dec 2017 In : European Journal of Human Genetics. 25, p. S35-S36 2 p.
Research output: Scientific - peer-review › Editorial

Cat04 Editorial material (edit) - Cat04 C: Editorial Material in wn journal

Ichthyosis vulgaris met populatiespecifieke filaggrine (FLG) mutaties
Clabbers, J. M. K., Dodemont, S. R. P., van Geel, M. & Steijlen, P. M. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 581-584 4 p.
Research output: Scientific › Editorial

Een zeldzame oorzaak voor het basaalcelnaevussyndroom
Cosgun, B., Gijezen, L. M. C., Vreeburg, M., de Boer, M., van Geel, M. & Mosterd, K. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 561-564 4 p.
Research output: Scientific › Editorial

Palmoplantaire keratodermie type Nagashima
Ramakers, N. A. M., Vreeburg, M., van Geel, M. & Steijlen, P. M. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 578-580 3 p.
Research output: Scientific › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer

Milne, R. L. , Kuchenbaecker, K. B. , Michailidou, K. , Beesley, J. , Kar, S. , Lindstrom, S. , Hui, S. , Lemacon, A. , Soucy, P. , Dennis, J. , Jiang, X. , Rostamianfar, A. , Finucane, H. , Bolla, M. K. , McGuffog, L. , Wang, Q. , Aalfs, C. M. , Adams, M. , Adlard, J. , Agata, S. & 456 others Ahmed, S., Ahsan, H., Aittomaki, K., Al-Ejeh, F., Allen, J., Ambrosone, C. B., Amos, C. I., Andrulis, I. L., Anton-Culver, H., Antonenkova, N. N., Arndt, V., Arnold, N., Aronson, K. J., Auber, B., Auer, P. L., Ausems, M. G. E. M., Azzollini, J., Bacot, F., Balmana, J., Barile, M., Barjhoux, L., Barkardottir, R. B., Barrdahl, M., Barnes, D., Barrowdale, D., Baynes, C., Beckmann, M. W., Benitez, J., Bermisheva, M., Bernstein, L., Bignon, Y-J., Blazer, K. R., Blok, M. J., Blomqvist, C., Blot, W., Bobolis, K., Boeckx, B., Bogdanova, N. V., Bojesen, A., Bojesen, S. E., Bonanni, B., Borresen-Dale, A-L., Bozsik, A., Bradbury, A. R., Brand, J. 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Closas, M., Schmidt, M. K., Antoniou, A. C., Simard, J., ABCTB Investigators, EMBRACE, GEMO Study Collaborators, HEBON, kConFab AOCS Investigators & NBSC Collaborators Dec 2017 In : Nature Genetics. 49, 12, p. 1767-1778 12 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Biallelicframeshift mutation in RIN2 in a patient with intellectual disability and cataract, without RIN2 syndrome
van Amen-Hellebrekers, C. J. M., Jansen, S., Stegmann, A. P. A., Stevens, S. J. C., Pfundt, R. & de Vries, B. B. A. Dec 2017 In : American Journal of Medical Genetics Part A. 173, 12, p. 3238-3240 3 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Paroxysmal sensory (spinal) attacks without hyperexplexia in a patient with a variant in the GLRA1 gene
Zwarts, M. J., Willemsen, M. H., Kamsteeg, E. -J. & Schelhaas, H. J. 15 Jul 2017 In : Journal of the Neurological Sciences. 378, p. 175-176 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat06 Scientific national journal

Epilepsie en genetica
Rouhl, R., Schelhaas, H. J. & Willemsen, M. 2017 In : Neuron. 22, p. 22-27 6 p.
Research output: Scientific - peer-review › Article

Zeldzaam subtype van congenitale ichthyosis: Ichthyosis prematuriteitsyndroom (IPS)
van Oosterhout, M. M., van Geel, M., Stuurman, K. E., Pasmans, S. G. M. A., Steijlen, P. & Kuijpers, A. L. A. 1 Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 9, p. 495-498
Research output: Scientific › Article

Cat02 SCI/SSCI journal (WI-1)

The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands
Seco, C. Z. , Wesdorp, M. , Feenstra, I. , Pfundt, R. , Hehir-Kwa, J. Y. , Lelieveld, S. H. , Castelein, S. , Gilissen, C. , de Wijs, I. J. , Admiraal, R. J. C. , Pennings, R. J. E. , Kunst, H. P. M. , van de Kamp, J. M. , Tamminga, S. , Houweling, A. C. , Plomp, A. S. , Maas, S. M. , Gans, P. A. M. D. K. , Kant, S. G. , de Geus, C. M. & 10 others Frints, S. G. M., Vanhoutte, E. K., van Dooren, M. F., van den Boogaard, M-J. H., Scheffer, H., Nelen, M., Kremer, H., Hoefsloot, L., Schraders, M. & Yntema, H. G. Feb 2017 In : European Journal of Human Genetics. 25, 3, p. 308-314 7 p.
Research output: Scientific - peer-review › Article

Heterozygous *HNRNPU* variants cause early onset epilepsy and severe intellectual disability
Bramswig, N. C. , Luedecke, H-J. , Hamdan, F. F. , Altmueller, J. , Beleggia, F. , Elcioglu, N. H. , Freyer, C. , Gerkes, E. H. , Demirkol, Y. K. , Knupp, K. G. , Kuechler, A. , Li, Y. , Lowenstein, D. H. , Michaud, J. L. , Park, K. , Stegmann, A. P. A. , Veenstra-Knol, H. E. , Wieland, T. , Wollnik, B. , Engels, H. & 3 others Strom, T. M., Kleefstra, T. & Wieczorek, D. Jul 2017 In : Human Genetics. 136, 7, p. 821-834 14 p.
Research output: Scientific - peer-review › Article

Classic Galactosemia: Study on the Late Prenatal Development of GALT Specific Activity in a Sheep Model
Coelho, A. I., Bierau, J., Lindhout, M., Achten, J., Kramer, B. W. & Rubio-Gozalbo, M. E. Sep 2017 In : The Anatomical Record: advances in integrative anatomy and evolutionary biology. 300, 9, p. 1570-1575 6 p.
Research output: Scientific - peer-review › Article

Estimates of live birth prevalence of children with Down syndrome in the period 1991-2015 in the Netherlands
de Graaf, G., Engelen, J. J. M., Gijsbers, A. C. J., Hochstenbach, R., Hoffer, M. J. V., Kooper, A. J. A., Sikkema-Raddatz, B., Srebniak, M. I., van der Kevie-Kersemaekers, A. M. F., van Zutven, L. J. C. M. & Voorhoeve, E. May 2017 In : Journal of Intellectual Disability Research. 61, 5, p. 461-470 10 p.
Research output: Scientific - peer-review › Article

The 6p25 deletion syndrome: An update on a rare neurocristopathy
de Vos, I. J. H. M., Stegmann, A. P. A., Webers, C. A. B. & Stumpel, C. T. R. M. 2017 In : Ophthalmic Genetics. 38, 2, p. 101-107 7 p.
Research output: Scientific - peer-review › Review article

BRCA1 mutation carriers have a lower number of mature oocytes after ovarian stimulation for IVF/PGD
Derks-Smeets, I. A. P., van Tilborg, T. C., van Montfoort, A., Smits, L., Torrance, H. L., Meijer-Hoogeveen, M., Broekmans, F., Dreesen, J. C. F. M., Paulussen, A. D. C., Tjan-Heijnen, V. C. G., Homminga, I., van den Berg, M. M. J., Ausems, M. G. E. M., de Rycke, M., de Die-Smulders, C. E. M., Verpoest, W. & van Golde, R. Nov 2017 In : Journal of Assisted Reproduction and Genetics. 34, 11, p. 1475-1482 8 p.
Research output: Scientific - peer-review › Article

Allelic Expression Imbalance Promoting a Mutant *PEX6* Allele Causes Zellweger Spectrum Disorder
Falkenberg, K. D., Braverman, N. E., Moser, A. B., Steinberg, S. J., Klouwer, F. C. C., Schluter, A., Ruiz, M., Pujol, A., Engvall, M., Naess, K., van Spronsen, F., Korver-Keularts, I., Rubio-Gozalbo, M. E., Ferdinandusse, S., Wanders, R. J. A. & Waterham, H. R. 7 Dec 2017 In : American Journal of Human Genetics. 101, 6, p. 965-976 12 p.
Research output: Scientific - peer-review › Article

The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families
Fountain, M. D. , Aten, E. , Cho, M. T. , Juusola, J. , Walkiewicz, M. A. , Ray, J. W. , Xia, F. , Yang, Y. , Graham, B. H. , Bacino, C. A. , Potocki, L. , van Haeringen, A. , Ruivenkamp, C. A. L. , Mancias, P. , Northrup, H. , Kukolich, M. K. , Weiss, M. M. , van Ravenswaaij-Arts, C. M. A. , Mathijssen, I. B. , Levesque, S. & 19 others Meeks, N., Rosenfeld, J. A., Lemke, D., Hamosh, A., Lewis, S. K., Race, S., Stewart, L. L., Hay, B., Lewis, A. M., Guerreiro, R. L., Bras, J. T., Martins, M. P., Derksen-Lubsen, G., Peeters, E., Stumpel, C., Stegmann, S., Bok, L. A., Santen, G. W. E. & Schaaf, C. P. Jan 2017 In : Genetics in Medicine. 19, 1, p. 45-52 8 p.
Research output: Scientific - peer-review › Article

Association of breast cancer risk in BRCA1 and BRCA2 mutation carriers with genetic variants showing differential allelic expression: identification of a modifier of breast cancer risk at locus 11q22.3
Hamdi, Y. , Soucy, P. , Kuchenbaeker, K. B. , Pastinen, T. , Droit, A. , Lemacon, A. , Adlard, J. , Aittomaki, K. , Andrulis, I. L. , Arason, A. , Arnold, N. , Arun, B. K. , Azzollini, J. , Bane, A. , Barjhoux, L. , Barrowdale, D. , Benitez, J. , Berthet, P. , Blok, M. J. , Bobolis, K. & 148 others Bonadona, V., Bonanni, B., Bradbury, A. R., Brewer, C., Buecher, B., Buys, S. S., Caligo, M. A., Chiquette, J., Chung, W. K., Claes, K. B. M., Daly, M. B., Damiola, F., Davidson, R., De la Hoya, M., De Leeneer, K., Diez, O., Ding, Y. C., Dolcetti, R., Domchek, S. M., Dorfling, C. M., Eccles, D., Eeles, R., Einbeigi, Z., Ejlertsen, B., Engel, C., Evans, D. G., Feliubadalo, L., Foretova, L., Fostira, F., Foulkes, W. D., Fountzilas, G., Friedman, E., Frost, D., Ganschow, P., Ganz, P. A., Garber, J., Gayther, S. A., Gerdes, A-M., Glendon, G., Godwin, A. K., Goldgar, D. E., Greene, M. H., Gronwald, J., Hahnen, E., Hamann, U., Hansen, T. V. O., Hart, S., Hays, J. L., Hogervorst, F. B. L., Hulick, P. J., Imyanitov, E. N., Isaacs, C., Izatt, L., Jakubowska, A., James, P., Janavicius, R., Jensen, U. B., John, E. M., Joseph, V., Just, W., Kaczmarek, K., Karlan, B. Y., Kets, C. M., Kirk, J., Kriege, M., Laitman, Y., Laurent, M., Lazaro, C., Leslie, G., Lester, J., Lesueur, F., Liljegren, A., Loman, N., Loud, J. T., Manoukian, S., Mariani, M., Mazoyer, S., McGuffog, L., Meijers-Heijboer, H. E. J., Meindl, A., Miller, A., Montagna, M., Mulligan, A. M., Nathanson, K. L., Neuhausen, S. L., Nevanlinna, H., Nussbaum, R. L., Olah, E., Olopade, O. I., Ong, K., Oosterwijk, J. C., Osorio, A., Papi, L., Park, S. K., Pedersen, I. S., Peissel, B., Segura, P. P., Peterlongo, P., Phelan, C. M., Radice, P., Rantala, J., Rappaport-Fuerhauser, C., Rennert, G., Richardson, A., Robson, M., Rodriguez, G. C., Rookus, M. A., Schmutzler, R. K., Sevenet, N., Shah, P. D., Singer, C. F., Slavin, T. P., Snape, K., Sokolowska, J., Sonderstrup, I. M. H., Southey, M., Spurdle, A. B., Stadler, Z., Stoppa-Lyonnet, D., Sukiennicki, G., Sutter, C., Tan, Y., Tea, M-K., Teixeira, M. R., Teule, A., Teo, S-H., Terry, M. B., Thomassen, M., Tihomirova, L., Tischkowitz, M., Tognazzo, S., Toland, A. E., Tung, N., van den Ouweland, A. M. W., van der Luijt, R. B., van Engelen, K., van Rensburg, E. J., Varon-Mateeva, R., Wappenschmidt, B., Wijnen, J. T., Rebbeck, T., Chenevix-Trench, G., Offit, K., Couch, F. J., Nord, S., Easton, D. F., Antoniou, A. C. & Simard, J. Jan 2017 In : Breast Cancer Research and Treatment. 161, 1, p. 117-134 18 p.
Research output: Scientific - peer-review › Article

Novel *SLC25A32* mutation in a patient with a severe neuromuscular phenotype
Hellebrekers, D. M., Sallevelt, S. C. E. H., Theunissen, T. E. J., Hendrickx, A. T. M., Gottschalk, R. W., Hoeijmakers, J. G. J., Habets, D. D., Bierau, J., Schoonderwoerd, K. G. & Smeets, H. J. M. Jun 2017 In : European Journal of Human Genetics. 25, 7, p. 886-888 3 p.
Research output: Scientific - peer-review › Article

Food ingestion in an upright sitting position increases postprandial amino acid availability when compared with food ingestion in a lying down position
Holwerda, A. M., Lenaerts, K., Bierau, J., Wodzig, W. K. W. H. & van Loon, L. J. C. Jul 2017 In : Applied Physiology Nutrition and Metabolism-Physiologie appliquee nutrition et metabolisme. 42, 7, p. 738-743 6 p.
Research output: Scientific - peer-review › Article

Lamin A/C-Related Cardiac Disease Late Onset With a Variable and Mild Phenotype in a Large Cohort of Patients With the Lamin A/C p.(Arg331Gln) Founder Mutation
Hoorntje, E. T. , Bollen, I. A. , Barge-Schaapveld, D. Q. , van Tienen, F. H. , te Meerman, G. J. , Jansweijer, J. A. , van Essen, A. J. , Volders, P. G. , Constantinescu, A. A. , van den Akker, P. C. , van Spaendonck-Zwarts, K. Y. , Oldenburg, R. A. , Marcelis, C. L. , van der Smagt, J. J. , Hennekam, E. A. , Vink, A. , Bootsma, M. , Aten, E. , Wilde, A. A. , van den Wijngaard, A. & 5 others Broers, J. L., Jongbloed, J. D., van der Velden, J., van den Berg, M. P. & van Tintelen, J. P. Aug 2017 In : Circulation : Cardiovascular Genetics. 10, 4, 36 p., 001631
Research output: Scientific - peer-review › Article

Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification
Kamps, R., Brandao, R. D., van den Bosch, B. J., Paulussen, A. D. C., Xanthoulea, S., Blok, M. J. & Romano, A. Feb 2017 In : International Journal of Molecular Sciences. 18, 2, 57 p., 308
Research output: Scientific - peer-review › Review article

Network topology of NaV1.7 mutations in sodium channel-related painful disorders
Kapetis, D., Sassone, J., Yang, Y., Galbardi, B., Xenakis, M. N., Westra, R. L., Szklarczyk, R., Lindsey, P., Faber, C. G., Gerrits, M., Merkies, I. S. J., Dib-Hajj, S. D., Mantegazza, M., Waxman, S. G., PROPANE Study Grp, J.M. Smeets, H. & Lauria, G. 24 Feb 2017 In : BMC Systems Biology. 11, 16 p., 28
Research output: Scientific - peer-review › Article

CAD mutations and uridine-responsive epileptic encephalopathy
Koch, J. , Mayr, J. A. , Alhaddad, B. , Rauscher, C. , Bierau, J. , Kovacs-Nagy, R. , Coene, K. L. M. , Bader, I. , Holzhacker, M. , Prokisch, H. , Venselaar, H. , Wevers, R. A. , Distelmaier, F. , Polster, T. , Leiz, S. , Betzler, C. , Strom, T. M. , Sperl, W. , Meitinger, T. , Wortmann, S. B. & 1 others Haack, T. B. Feb 2017 In : Brain. 140, p. 279-286 8 p.
Research output: Scientific - peer-review › Article

Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder
Koemans, T. S., Kleefstra, T., Chubak, M. C., Stone, M. H., Reijnders, M. R. F., de Munnik, S., Willemsen, M. H., Fenckova, M., Stumpel, C. T. R. M., Bok, L. A., Saenz, M. S., Byerly, K. A., Baughn, L. B., Stegmann, A. P. A., Pfundt, R., Zhou, H., van Bokhoven, H., Schenck, A. & Kramer, J. M. Oct 2017 In : Plos Genetics. 13, 10, 24 p., 1006864
Research output: Scientific - peer-review › Article

Both basal and post-prandial muscle protein synthesis rates, following the ingestion of a leucine-enriched whey protein supplement, are not impaired in sarcopenic older males
Kramer, I. F., Verdijk, L. B., Hamer, H. M., Verlaan, S., Luiking, Y. C., Kouw, I. W. K., Senden, J. M., van Kranenburg, J., Gijsen, A. P., Bierau, J., Poeze, M. & van Loon, L. J. C. Oct 2017 In : Clinical Nutrition. 36, 5, p. 1440-1449 10 p.
Research output: Scientific - peer-review › Article

***De Novo* Missense Mutations in *DHX30* Impair Global Translation and Cause a Neurodevelopmental Disorder**
Lessel, D. , Schob, C. , Kuery, S. , Reinders, M. R. F. , Harel, T. , Eldomery, M. K. , Coban-Akdemir, Z. , Denecke, J. , Edvardson, S. , Colin, E. , Stegmann, A. P. A. , Gerkes, E. H. , Tessarech, M. , Bonneau, D. , Barth, M. , Besnard, T. , Cogne, B. , Revah-Politi, A. , Strom, T. M. , Rosenfeld, J. A. & 24 others Yang, Y., Posey, J. E., Immken, L., Oundjian, N., Helbig, K. L., Meeks, N., Zegar, K., Morton, J., Schieving, J. H., Claasen, A., Huentelman, M., Narayanan, V., Ramsey, K., Brunner, H. G., Elpeleg, O., Mercier, S., Bezieau, S., Kubisch, C., Kleefstra, T., Kindler, S., Lupski, J. R., Kreienkamp, H-J., DDD study & C4RCD Res Grp 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 716-724 9 p.
Research output: Scientific - peer-review › Article

Palmitate-Induced Vacuolar-Type H(+)-ATPase Inhibition Feeds Forward Into Insulin Resistance and Contractile Dysfunction
Liu, Y., Steinbusch, L. K. M., Nabben, M., Kapsokalyvas, D., van Zandvoort, M., Schonleitner, P., Antoons, G., Simons, P. J., Coumans, W. A., Geomini, A., Chanda, D., Glatz, J. F. C., Neumann, D. & Luiken, J. J. F. P. 1 Jun 2017 In : Diabetes. 66, 6, p. 1521-1534 14 p.
Research output: Scientific - peer-review › Article

Heterozygous variants in ACTL6A, encoding a component of the BAF complex, are associated with intellectual disability

Marom, R. , Jain, M. , Burrage, L. C. , Song, I-W. , Graham, B. H. , Brown, C. W. , Stevens, S. J. C. , Stegmann, A. P. A. , Gunter, A. T. , Kaplan, J. D. , Gavrilova, R. H. , Shinawi, M. , Rosenfeld, J. A. , Bae, Y. , Tran, A. A. , Chen, Y. , Lu, J. T. , Gibbs, R. A. , Eng, C. , Yang, Y. & 4 others Rousseau, J., de Vries, B. B. A., Campeau, P. M. & Lee, B. Oct 2017 In : Human Mutation. 38, 10, p. 1365-1371 7 p.

Research output: Scientific - peer-review › Article

COL6A5 variants in familial neuropathic chronic itch

Martinelli-Boneschi, F. , Colombi, M. , Castori, M. , Devigili, G. , Eleopra, R. , Malik, R. A. , Ritelli, M. , Zoppi, N. , Dordoni, C. , Sorosina, M. , Grammatico, P. , Fadavi, H. , Gerrits, M. M. , Almomani, R. , Faber, C. G. , Merkies, I. S. J. , Toniolo, D. , Cocca, M. , Doglioni, C. , Waxman, S. G. & 10 others Dib-Hajj, S. D., Taiana, M. M., Sassone, J., Lombardi, R., Cazzato, D., Zauli, A., Santoro, S., Marchi, M., Lauria, G. & INGI Network Mar 2017 In : Brain. 140, p. 555-567 13 p.

Research output: Scientific - peer-review › Article

Mendelian Disorders of Cornification Caused by Defects in Intracellular Calcium Pumps: Mutation Update and Database for Variants in ATP2A2 and ATP2C1 Associated with Darier Disease and Hailey-Hailey Disease

Nellen, R. G. L., Steijlen, P. M., van Steensel, M. A. M., Vreeburg, M., Frank, J., van Geel, M. & European Professional Contributors Apr 2017 In : Human Mutation. 38, 4, p. 343-356 14 p.

Research output: Scientific - peer-review › Article

Novel pathogenic SLC25A46 splice-site mutation causes an optic atrophy spectrum disorder

Nguyen, M., Boesten, I., Hellebrekers, D. M. E. I., Mulder-den Hartog, N. M., de Coo, I. F. M., Smeets, H. J. M. & Gerards, M. Jan 2017 In : Clinical Genetics. 91, 1, p. 121-125 5 p.

Research output: Scientific - peer-review › Article

Detection of clinically relevant copy-number variants by exome sequencing in a large cohort of genetic disorders

Pfundt, R. , del Rosario, M. , Vissers, L. E. L. M. , Kwint, M. P. , Janssen, I. M. , de Leeuw, N. , Yntema, H. G. , Nelen, M. R. , Lugtenberg, D. , Kamsteeg, E-J. , Wieskamp, N. , Stegmann, A. P. A. , Stevens, S. J. C. , Rodenburg, R. J. T. , Simons, A. , Mensenkamp, A. R. , Rinne, T. , Gilissen, C. , Scheffer, H. , Veltman, J. A. & 1 others Hehir-Kwa, J. Y. Jun 2017 In : Genetics in Medicine. 19, 6, p. 667-675 9 p.

Research output: Scientific - peer-review › Article

Identification of 12 new susceptibility loci for different histotypes of epithelial ovarian cancer

Phelan, C. M. , Kuchenbaecker, K. B. , Tyrer, J. P. , Kar, S. P. , Lawrenson, K. , Winham, S. J. , Dennis, J. , Pirie, A. , Riggan, M. J. , Chornokur, G. , Earp, M. A. , Lyra, P. C. , Lee, J. M. , Coetzee, S. , Beesley, J. , McGuffog, L. , Soucy, P. , Dicks, E. , Lee, A. , Barrowdale, D. & 400 others Lecarpentier, J., Leslie, G., Aalfs, C. M., Aben, K. K. H., Adams, M., Adlard, J., Andrulis, I. L., Anton-Culver, H., Antonenkova, N., Aravantinos, G., Arnold, N., Arun, B. K., Arver, B., Azzollini, J., Balmana, J., Banerjee, S. N., Barjhoux, L., Barkardottir, R. B., Bean, Y., Beckmann, M. W., Beeghly-Fadiel, A., Benitez, J., Bermisheva, M., Bernardini, M. Q., Birrer, M. J., Bjorge, L., Black, A., Blankstein, K., Blok, M. J., Bodelon, C., Bogdanova, N., Bojesen, A., Bonanni, B., Borg, A., Bradbury, A. R., Brenton, J. D., Brewer, C., Brinton, L., Broberg, P., Brooks-Wilson, A., Bruinsma, F., Brunet, J., Buecher, B., Butzow, R., Buys, S. S., Caldes, T., Caligo, M. 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Research output: Scientific - peer-review › Article

Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability
Reijnders, M. R. F. , Kousi, M. , van Woerden, G. M. , Klein, M. , Bralten, J. , Mancini, G. M. S. , van Essen, T. , Proietti-Onori, M. , Smeets, E. E. J. , van Gastel, M. , Stegmann, A. P. A. , Stevens, S. J. C. , Lelieveld, S. H. , Gilissen, C. , Pfundt, R. , Tan, P. L. , Kleefstra, T. , Franke, B. , Elgersma, Y. , Katsanis, N. & 1 others Brunner, H. G. 20 Oct 2017 In : Nature Communications. 8, 12 p., 1052
Research output: Scientific - peer-review › Article

Postzygotic mosaicism in basal cell naevus syndrome
Reinders, M. G. H. C., Boersma, H. J., Leter, E. M., Vreeburg, M., Paulussen, A. D. C., Arits, A. H. M. M., Roemen, G. M. J. M., Speel, E. J. M., Steijlen, P. M., van Geel, M. & Mosterd, K. Jul 2017 In : British Journal of Dermatology. 177, 1, p. 249-252 4 p.
Research output: Scientific - peer-review › Article

Propionic acidemia as a cause of adult-onset dilated cardiomyopathy
Riemersma, M., Hazebroek, M. R., Helderman-van den Enden, A. T. J. M., Salomons, G. S., Ferdinandusse, S., Brouwers, M. C. G. J., van der Ploeg, L., Heymans, S., Glatz, J. F. C., van den Wijngaard, A., Krapels, I. P. C., Bierau, J. & Brunner, H. G. Nov 2017 In : European Journal of Human Genetics. 25, 11, p. 1195-1201 7 p.
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
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Research output: Scientific - peer-review › Article

De novo mtDNA point mutations are common and have a low recurrence risk
Sallevelt, S. C. E. H., de Die-Smulders, C. E. M., Hendrickx, A. T. M., Hellebrekers, D. M. E. I., de Coo, I. F. M., Alston, C. L., Knowles, C., Taylor, R. W., McFarland, R. & Smeets, H. J. M. Feb 2017 In : Journal of Medical Genetics. 54, 2, p. 114-124 11 p.
Research output: Scientific - peer-review › Article

PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Druesedau, M., Hellebrekers, D. M. E. I., Paulussen, A. D. C., Coonen, E., Van Golde, R. J. T., Geraedts, J. P. M., Gianaroli, L., Magli, M. C., Zeviani, M., Smeets, H. J. M. & de Die-Smulders, C. E. M. Mar 2017 In : Human Reproduction. 32, 3, p. 698-703 6 p.
Research output: Scientific - peer-review › Article

Associations of plasma uric acid and purine metabolites with blood pressure in children: the KOALA Birth Cohort Study
Scheepers, L. E. J. M., Boonen, A., Pijnenburg, W., Bierau, J., Staessen, J. A., Stehouwer, C. D. A., Thijs, C. & Arts, I. C. W. May 2017 In : Journal of Hypertension. 35, 5, p. 982-993 12 p.
Research output: Scientific - peer-review › Article

A homozygous FITM2 mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy
Seco, C. Z. , Castells-Nobau, A. , Joo, S. , Schraders, M. , Foo, J. N. , van der Voet, M. , Velan, S. S. , Nijhof, B. , Oostrik, J. , de Vrieze, E. , Katana, R. , Mansoor, A. , Huynen, M. , Szklarczyk, R. , Oti, M. , Tranebjaerg, L. , van Wijk, E. , Scheffer-de Gooyert, J. M. , Siddique, S. , Baets, J. & 10 others de Jonghe, P., Kazmi, S. A. R., Sadananthan, S. A., van de Warrenburg, B. P., Khor, C. C., Goepfert, M. C., Qamar, R., Schenck, A., Kremer, H. & Siddiqi, S. 1 Feb 2017 In : Disease Models & Mechanisms. 10, 2, p. 105-118 14 p.
Research output: Scientific - peer-review › Article

De novo, deleterious sequence variants that alter the transcriptional activity of the homeoprotein PBX1 are associated with intellectual disability and pleiotropic developmental defects
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Research output: Scientific - peer-review › Article

Heritability in a SCN5A-mutation founder population with increased female susceptibility to non-nocturnal ventricular tachyarrhythmia and sudden cardiac death
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Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

Women's Experience with Non-Invasive Prenatal Testing and Emotional Well-being and Satisfaction after Test-Results
van Schendel, R. V., Page-Christiaens, G. C. M. L., Beulen, L., Bilardo, C. M., de Boer, M. A., Coumans, A. B. C., Faas, B. H. W., van Langen, I. M., Lichtenbelt, K. D., van Maarle, M. C., Macville, M. V. E., Oepkes, D., Pajkrt, E., Henneman, L. & Dutch NIPT Consortium Dec 2017 In : Journal of Genetic Counseling. 26, 6, p. 1348-1356 9 p.
Research output: Scientific - peer-review › Article

Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas
Weren, R. D. A., Mensenkamp, A. R., Simons, M., Eijkelenboom, A., Sie, A. S., Ouchene, H., van Asseldonk, M., Gomez-Garcia, E. B., Blok, M. J., de Hullu, J. A., Nelen, M. R., Hoischen, A., Bulten, J., Tops, B. B. J., Hoogerbrugge, N. & Ligtenberg, M. J. L. Feb 2017 In : Human Mutation. 38, 2, p. 226-235 10 p.
Research output: Scientific - peer-review › Article

The hypoxanthine-xanthine oxidase axis is not involved in the initial phase of clinical transplantation-related ischemia-reperfusion injury
Wijermars, L. G. M., Bakker, J. A., de Vries, D. K., van Noorden, C. J. F., Bierau, J., Kostidis, S., Mayboroda, O. A., Tsikas, D., Schaapherder, A. F. & Lindeman, J. H. N. 1 Mar 2017 In : American Journal of Physiology-Renal Physiology. 312, 3, p. F457-F464 8 p.
Research output: Scientific - peer-review › Article

Mutations in the Chromatin Regulator Gene *BRPF1* Cause Syndromic Intellectual Disability and Deficient Histone Acetylation
Yan, K. , Rousseau, J. , Littlejohn, R. O. , Kiss, C. , Lehman, A. , Rosenfeld, J. A. , Stumpel, C. T. R. , Stegmann, A. P. A. , Robak, L. , Scaglia, F. , Thi Tuyet Mai Nguyen, , Fu, H. , Ajeawung, N. F. , Camurri, M. V. , Li, L. , Gardham, A. , Panis, B. , Almannai, M. , Sacoto, M. J. G. , Baskin, B. & 16 others Ruivenkamp, C., Xia, F., Bi, W., Cho, M. T., Potjer, T. P., Santen, G. W. E., Parker, M. J., Canham, N., McKinnon, M., Potocki, L., MacKenzie, J. J., Roeder, E. R., Campeau, P. M., Yang, X-J., DDD Study & CAUSES Study 5 Jan 2017 In : American Journal of Human Genetics. 100, 1, p. 91-104 14 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

Nutrients required for phospholipid synthesis are lower in blood and cerebrospinal fluid in mild cognitive impairment and Alzheimer's disease dementia
van Wijk, N., Slot, R. E. R., Duits, F. H., Strik, M., Biesheuvel, E., Sijben, J. W. C., Blankenstein, M. A., Bierau, J., van der Flier, W. M., Scheltens, P. & Teunissen, C. E. 2017 In : Alzheimer's and Dementia: Diagnosis, Assessment and Disease Monitoring. 8, p. 139-146 8 p.
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 C: Editorial Material in wn journal

Ichthyosis vulgaris met populatiespecifieke filaggrine (FLG) mutaties
Clabbers, J. M. K., Dodemont, S. R. P., van Geel, M. & Steijlen, P. M. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 581-584 4 p.
Research output: Scientific › Editorial

Een zeldzame oorzaak voor het basaalcelnaevussyndroom
Cosgun, B., Gijezen, L. M. C., Vreeburg, M., de Boer, M., van Geel, M. & Mosterd, K. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 561-564 4 p.
Research output: Scientific › Editorial

Palmoplantaire keratodermie type Nagashima

Ramakers, N. A. M., Vreeburg, M., van Geel, M. & Steijlen, P. M. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 578-580 3 p.

Research output: Scientific › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Identification of ten variants associated with risk of estrogen-receptor-negative breast cancer

Milne, R. L. , Kuchenbaecker, K. B. , Michailidou, K. , Beesley, J. , Kar, S. , Lindstrom, S. , Hui, S. , Lemacon, A. , Soucy, P. , Dennis, J. , Jiang, X. , Rostamianfar, A. , Finucane, H. , Bolla, M. K. , McGuffog, L. , Wang, Q. , Aalfs, C. M. , Adams, M. , Adlard, J. , Agata, S. & 456 others Ahmed, S., Ahsan, H., Aittomaki, K., Al-Ejeh, F., Allen, J., Ambrosone, C. B., Amos, C. I., Andrulis, I. L., Anton-Culver, H., Antonenkova, N. N., Arndt, V., Arnold, N., Aronson, K. J., Auber, B., Auer, P. L., Ausems, M. G. E. M., Azzollini, J., Bacot, F., Balmana, J., Barile, M., Barjhoux, L., Barkardottir, R. B., Barrdahl, M., Barnes, D., Barrowdale, D., Baynes, C., Beckmann, M. W., Benitez, J., Bermisheva, M., Bernstein, L., Bignon, Y-J., Blazer, K. R., Blok, M. J., Blomqvist, C., Blot, W., Bobolis, K., Boeckx, B., Bogdanova, N. V., Bojesen, A., Bojesen, S. E., Bonanni, B., Borresen-Dale, A-L., Bozsik, A., Bradbury, A. R., Brand, J. 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Research output: Scientific - peer-review › Comment/Letter to the editor

Biallelicframeshift mutation in RIN2 in a patient with intellectual disability and cataract, without RIN2 syndrome
van Amen-Hellebrekers, C. J. M., Jansen, S., Stegmann, A. P. A., Stevens, S. J. C., Pfundt, R. & de Vries, B. B. A. Dec 2017 In : American Journal of Medical Genetics Part A. 173, 12, p. 3238-3240 3 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat06 Scientific national journal

Zeldzaam subtype van congenitale ichthyosis: Ichthyosis prematuriteitsyndroom (IPS)
van Oosterhout, M. M., van Geel, M., Stuurman, K. E., Pasmans, S. G. M. A., Steijlen, P. & Kuijpers, A. L. A. 1 Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 9, p. 495-498
Research output: Scientific › Article

Cat02 SCI/SSCI journal (WI-1)

Effects of preparatory and action planning instructions on situation specific and general fruit and snack intake
de Bruijn, G-J., Minh Hao Nguyen, Rhodes, R. E. & van Osch, L. 1 Jan 2017 In : Appetite. 108, p. 161-170 10 p.
Research output: Scientific - peer-review › Article

The 6p25 deletion syndrome: An update on a rare neurocristopathy
de Vos, I. J. H. M., Stegmann, A. P. A., Webers, C. A. B. & Stumpel, C. T. R. M. 2017 In : Ophthalmic Genetics. 38, 2, p. 101-107 7 p.
Research output: Scientific - peer-review › Review article

Review of familial cerebral cavernous malformations and report of seven additional families
de Vos, I. J. H. M., Vreeburg, M., Koek, G. H. & van Steensel, M. A. M. Feb 2017 In : American Journal of Medical Genetics Part A. Part A 173A, 2, p. 338-351 14 p.
Research output: Scientific - peer-review › Review article

BRCA1 mutation carriers have a lower number of mature oocytes after ovarian stimulation for IVF/PGD
Derks-Smeets, I. A. P., van Tilborg, T. C., van Montfoort, A., Smits, L., Torrance, H. L., Meijer-Hoogeveen, M., Broekmans, F., Dreesen, J. C. F. M., Paulussen, A. D. C., Tjan-Heijnen, V. C. G., Homminga, I., van den Berg, M. M. J., Ausems, M. G. E. M., de Rycke, M., de Die-Smulders, C. E. M., Verpoest, W. & van Golde, R. Nov 2017 In : Journal of Assisted Reproduction and Genetics. 34, 11, p. 1475-1482 8 p.
Research output: Scientific - peer-review › Article

Uptake of prenatal diagnostic testing for retinoblastoma compared to other hereditary cancer syndromes in the Netherlands
Dommering, C. J., Henneman, L., van der Hout, A. H., Jonker, M. A., Tops, C. M. J., van den Ouweland, A. M. W., van der Luijt, R. B., Mensenkamp, A. R., Hogervorst, F. B. L., Redeker, E. J. W., de Die-Smulders, C. E. M., Moll, A. C. & Meijers-Heijboer, H. Apr 2017 In : Familial Cancer. 16, 2, p. 271-277 7 p.
Research output: Scientific - peer-review › Article

TSC2 c.1864C > T Variant Aassociated with Mild Cases of Tuberous Sclerosis Complex
Farach, L. S., Gibson, W. T., Sparagana, S. P., Nellist, M., Stumpel, C. T. R. M., Hietala, M., Friedman, E., Pearson, D. A., Creighton, S. P., Wagemans, A., Segel, R., Ben-Shalom, E., Au, K. S. & Northrup, H. Mar 2017 In : American Journal of Medical Genetics Part A. 173, 3, p. 771-775 5 p.
Research output: Scientific - peer-review › Article

The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families
Fountain, M. D. , Aten, E. , Cho, M. T. , Juusola, J. , Walkiewicz, M. A. , Ray, J. W. , Xia, F. , Yang, Y. , Graham, B. H. , Bacino, C. A. , Potocki, L. , van Haeringen, A. , Ruivenkamp, C. A. L. , Mancias, P. , Northrup, H. , Kukolich, M. K. , Weiss, M. M. , van Ravenswaaij-Arts, C. M. A. , Mathijssen, I. B. , Levesque, S. & 19 others Meeks, N., Rosenfeld, J. A., Lemke, D., Hamosh, A., Lewis, S. K., Race, S., Stewart, L. L., Hay, B., Lewis, A. M., Guerreiro, R. L., Bras, J. T., Martins, M. P., Derksen-Lubsen, G., Peeters, E., Stumpel, C., Stegmann, S., Bok, L. A., Santen, G. W. E. & Schaaf, C. P. Jan 2017 In : Genetics in Medicine. 19, 1, p. 45-52 8 p.
Research output: Scientific - peer-review › Article

Awareness and attitude regarding reproductive options of persons carrying a BRCA mutation and their partners
Gietel-Habets, J. J. G., de Die-Smulders, C. E. M., Derks-Smeets, I. A. P., Tibben, A., Tjan-Heijnen, V. C. G., van Golde, R., Gomez-Garcia, E., Kets, C. M. & van Osch, L. A. D. M. Mar 2017 In : Human Reproduction. 32, 3, p. 588-597 10 p.
Research output: Scientific - peer-review › Article

Diagnostic exome sequencing in 266 Dutch patients with visual impairment
Haer-Wigman, L. , van Zelst-Stams, W. A. G. , Pfundt, R. , van den Born, L. I. , Klaver, C. C. W. , Verheij, J. B. G. M. , Hoyng, C. B. , Breuning, M. H. , Boon, C. J. F. , Kievit, A. J. , Verhoeven, V. J. M. , Pott, J. W. R. , Sallevelt, S. C. E. H. , van Hagen, J. M. , Plomp, A. S. , Kroes, H. Y. , Lelieveld, S. H. , Hehir-Kwa, J. Y. , Castelein, S. , Nelen, M. & 5 others Scheffer, H., Lugtenberg, D., Cremers, F. P. M., Hoefsloot, L. & Yntema, H. G. May 2017 In : European Journal of Human Genetics. 25, 5, p. 591-599 9 p.
Research output: Scientific - peer-review › Article

Novel *SLC25A32* mutation in a patient with a severe neuromuscular phenotype
Hellebrekers, D. M., Sallevelt, S. C. E. H., Theunissen, T. E. J., Hendrickx, A. T. M., Gottschalk, R. W., Hoeijmakers, J. G. J., Habets, D. D., Bierau, J., Schoonderwoerd, K. G. & Smeets, H. J. M. Jun 2017 In : European Journal of Human Genetics. 25, 7, p. 886-888 3 p.
Research output: Scientific - peer-review › Article

Delusional and Psychotic Disorders in Juvenile Myotonic Dystrophy Type-1
Jacobs, D., Willekens, D., de Die-Smulders, C., Frijns, J-P. & Steyaert, J. Jun 2017 In : American Journal of Medical Genetics Part B-neuropsychiatric Genetics. 174, 4, p. 359-366 8 p.
Research output: Scientific - peer-review › Article

De Novo Truncating Mutations in the Last and Penultimate Exons of *PPM1D* Cause an Intellectual Disability Syndrome
Jansen, S. , Geuer, S. , Pfundt, R. , Brough, R. , Ghongane, P. , Herkert, J. C. , Marco, E. J. , Willemsen, M. H. , Kleefstra, T. , Hannibal, M. , Shieh, J. T. , Lynch, S. A. , Flinter, F. , FitzPatrick, D. R. , Gardham, A. , Bernhard, B. , Ragge, N. , Newbury-Ecob, R. , Bernier, R. , Kvarnung, M. & 10 others Magnusson, E. A. H., Wessels, M. W., van Slegtenhorst, M. A., Monaghan, K. G., de Vries, P., Veltman, J. A., Lord, C. J., Vissers, L. E. L. M., de Vries, B. B. A. & Deciphering Dev Disorders Study 6 Apr 2017 In : American Journal of Human Genetics. 100, 4, p. 650-658 9 p.
Research output: Scientific - peer-review › Article

From action planning and plan enactment to fruit consumption: moderated mediation effects
Kasten, S., van Osch, L., Eggers, S. M. & de Vries, H. 23 Oct 2017 In : BMC Public Health. 17, 11 p., 832
Research output: Scientific - peer-review › Article

Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder
Koemans, T. S., Kleefstra, T., Chubak, M. C., Stone, M. H., Reijnders, M. R. F., de Munnik, S., Willemsen, M. H., Fenckova, M., Stumpel, C. T. R. M., Bok, L. A., Saenz, M. S., Byerly, K. A., Baughn, L. B., Stegmann, A. P. A., Pfundt, R., Zhou, H., van Bokhoven, H., Schenck, A. & Kramer, J. M. Oct 2017 In : Plos Genetics. 13, 10, 24 p., 1006864
Research output: Scientific - peer-review › Article

B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies
Maroofian, R., Riemersma, M., Jae, L. T., Zhianabed, N., Willemsen, M. H., Wissink-Lindhout, W. M., Willemsen, M. A., de Brouwer, A. P. M., Mehrjardi, M. Y. V., Ashrafi, M. R., Kusters, B., Kleefstra, T., Jamshidi, Y., Nasseri, M., Pfundt, R., Brummelkamp, T. R., Abbaszadegan, M. R., Lefeber, D. J. & van Bokhoven, H. 22 Dec 2017 In : Genome Medicine. 9, 11 p., 118
Research output: Scientific - peer-review › Article

Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia
Meyer, E. , Carss, K. J. , Rankin, J. , Nichols, J. M. E. , Grozeva, D. , Joseph, A. P. , Mencacci, N. E. , Papandreou, A. , Ng, J. , Barra, S. , Ngoh, A. , Ben-Pazi, H. , Willemsen, M. A. , Arkadir, D. , Barnicoat, A. , Bergman, H. , Bhate, S. , Boys, A. , Darin, N. , Foulds, N. & 53 others Gutowski, N., Hills, A., Houlden, H., Hurst, J. A., Israe, Z., Kaminska, M., Limousin, P., Lumsden, D., Mckee, S., Misra, S., Mohammed, S. S., Nakou, V., Nicolai, J., Nilsson, M., Pall, H., Peall, K. J., Peters, G. B., Prabhakar, P., Reuter, M. S., Rump, P., Sege, R., Sinnema, M., Smith, M., Turnpenny, P., White, S. M., Wieczorek, D., Wiethoff, S., Wilson, B. T., Winter, G., Wragg, C., Pope, S., Heales, S. J. H., Morrogh, D., Pittman, A., Carr, L. J., Perez-Duenas, B., Lin, J-P., Reis, A., Gahl, W. A., Toro, C., Bhatia, K. P., Wood, N. W., Kamsteeg, E-J., Chong, W. K., Gissen, P., Topf, M., Dale, R. C., Chubby, J. R., Raymond, F. L., Kurian, M. A., UK10K Consortium, Deciphering Dev Disorders Study & NIHR BioResource Rare Feb 2017 In : Nature Genetics. 49, 2, p. 223-237 15 p.
Research output: Scientific - peer-review › Article

Mendelian Disorders of Cornification Caused by Defects in Intracellular Calcium Pumps: Mutation Update and Database for Variants in ATP2A2 and ATP2C1 Associated with Darier Disease and Hailey-Hailey Disease
Nellen, R. G. L., Steijlen, P. M., van Steensel, M. A. M., Vreeburg, M., Frank, J., van Geel, M. & European Professional Contributors Apr 2017 In : Human Mutation. 38, 4, p. 343-356 14 p.
Research output: Scientific - peer-review › Article

NGS panel analysis in 24 ectopia lentis patients; a clinically relevant test with a high diagnostic yield
Overwater, E. , Floor, K. , van Beek, D. , de Boer, K. , van Dijk, T. , Hilhorst-Hofstee, Y. , Hoogeboom, A. J. M. , van Kaam, K. J. , van de Kamp, J. M. , Kempers, M. , Krapels, I. P. C. , Kroes, H. Y. , Loeys, B. , Salemink, S. , Stumpel, C. T. R. M. , Verhoeven, V. J. M. , Wijnands-van den Berg, E. , Cobben, J. M. , van Tintelen, J. P. , Weiss, M. M. & 2 others Houweling, A. C. & Maugeri, A. Sep 2017 In : European Journal of Medical Genetics. 60, 9, p. 465-473 9 p.
Research output: Scientific - peer-review › Article

Postzygotic mosaicism in basal cell naevus syndrome
Reinders, M. G. H. C., Boersma, H. J., Leter, E. M., Vreeburg, M., Paulussen, A. D. C., Arits, A. H. M. M., Roemen, G. M. J. M., Speel, E. J. M., Steijlen, P. M., van Geel, M. & Mosterd, K. Jul 2017 In : British Journal of Dermatology. 177, 1, p. 249-252 4 p.
Research output: Scientific - peer-review › Article

Propionic acidemia as a cause of adult-onset dilated cardiomyopathy
Riemersma, M., Hazebroek, M. R., Helderman-van den Enden, A. T. J. M., Salomons, G. S., Ferdinandusse, S., Brouwers, M. C. G. J., van der Ploeg, L., Heymans, S., Glatz, J. F. C., van den Wijngaard, A., Krapels, I. P. C., Bierau, J. & Brunner, H. G. Nov 2017 In : European Journal of Human Genetics. 25, 11, p. 1195-1201 7 p.
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
Sallevelt, S. C. E. H., de Koning, B., Szklarczyk, R., Paulussen, A. D. C., de Die-Smulders, C. E. M. & Smeets, H. J. M. May 2017 In : Genetics in Medicine. 19, 5, p. 583-592 10 p.
Research output: Scientific - peer-review › Article

De novo mtDNA point mutations are common and have a low recurrence risk
Sallevelt, S. C. E. H., de Die-Smulders, C. E. M., Hendrickx, A. T. M., Hellebrekers, D. M. E. I., de Coo, I. F. M., Alston, C. L., Knowles, C., Taylor, R. W., McFarland, R. & Smeets, H. J. M. Feb 2017 In : Journal of Medical Genetics. 54, 2, p. 114-124 11 p.
Research output: Scientific - peer-review › Article

PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Druesedau, M., Hellebrekers, D. M. E. I., Paulussen, A. D. C., Coonen, E., Van Golde, R. J. T., Geraedts, J. P. M., Gianaroli, L., Magli, M. C., Zeviani, M., Smeets, H. J. M. & de Die-Smulders, C. E. M. Mar 2017 In : Human Reproduction. 32, 3, p. 698-703 6 p.
Research output: Scientific - peer-review › Article

Growth Hormone Therapy in Children with Kabuki Syndrome: 1-year Treatment Results
Schott, D. A., Gerver, W. J. M. & Stumpel, C. T. R. M. 2017 In : Hormone Research in Paediatrics. 88, 3-4, p. 1-7 7 p.
Research output: Scientific - peer-review › Article

The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands
Seco, C. Z. , Wesdorp, M. , Feenstra, I. , Pfundt, R. , Hehir-Kwa, J. Y. , Lelieveld, S. H. , Castelein, S. , Gilissen, C. , de Wijs, I. J. , Admiraal, R. J. C. , Pennings, R. J. E. , Kunst, H. P. M. , van de Kamp, J. M. , Tamminga, S. , Houweling, A. C. , Plomp, A. S. , Maas, S. M. , Gans, P. A. M. D. K. , Kant, S. G. , de Geus, C. M. & 10 others Frints, S. G. M., Vanhoutte, E. K., van Dooren, M. F., van den Boogaard, M-J. H., Scheffer, H., Nelen, M., Kremer, H., Hoefsloot, L., Schraders, M. & Yntema, H. G. Feb 2017 In : European Journal of Human Genetics. 25, 3, p. 308-314 7 p.
Research output: Scientific - peer-review › Article

Heritability in a SCN5A-mutation founder population with increased female susceptibility to non-nocturnal ventricular tachyarrhythmia and sudden cardiac death
ter Bekke, R. M. A., Isaacs, A., Barysenka, A., Hoos, M. B., Jongbloed, J. D. H., Hoorntje, J. C. A., Patelski, A. S. M., Helderman-van den Enden, A. T. J. M., van den Wijngaard, A., Stoll, M. & Volders, P. G. A. Dec 2017 In : Heart Rhythm. 14, 12, p. 1873-1881 9 p.
Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas
Weren, R. D. A., Mensenkamp, A. R., Simons, M., Eijkelenboom, A., Sie, A. S., Ouchene, H., van Asseldonk, M., Gomez-Garcia, E. B., Blok, M. J., de Hullu, J. A., Nelen, M. R., Hoischen, A., Bulten, J., Tops, B. B. J., Hoogerbrugge, N. & Ligtenberg, M. J. L. Feb 2017 In : Human Mutation. 38, 2, p. 226-235 10 p.
Research output: Scientific - peer-review › Article

The tumour suppressor CDKN2A/p16(INK4a) regulates adipogenesis and bone marrow-dependent development of perivascular adipose tissue
Wouters, K., Deleye, Y., Hannou, S. A., Vanhoutte, J., Marechal, X., Coisne, A., Tagzirt, M., Derudas, B., Bouchaert, E., Duhem, C., Vallez, E., Schalkwijk, C. G., Pattou, F., Montaigne, D., Staels, B. & Paumelle, R. Nov 2017 In : Diabetes & Vascular Disease Research. 14, 6, p. 516-524 9 p.
Research output: Scientific - peer-review › Article

Mutations in the Chromatin Regulator Gene *BRPF1* Cause Syndromic Intellectual Disability and Deficient Histone Acetylation
Yan, K. , Rousseau, J. , Littlejohn, R. O. , Kiss, C. , Lehman, A. , Rosenfeld, J. A. , Stumpel, C. T. R. , Stegmann, A. P. A. , Robak, L. , Scaglia, F. , Thi Tuyet Mai Nguyen, , Fu, H. , Ajeawung, N. F. , Camurri, M. V. , Li, L. , Gardham, A. , Panis, B. , Almannai, M. , Sacoto, M. J. G. , Baskin, B. & 16 others Ruivenkamp, C., Xia, F., Bi, W., Cho, M. T., Potjer, T. P., Santen, G. W. E., Parker, M. J., Canham, N., McKinnon, M., Potocki, L., MacKenzie, J. J., Roeder, E. R., Campeau, P. M., Yang, X-J., DDD Study & CAUSES Study 5 Jan 2017 In : American Journal of Human Genetics. 100, 1, p. 91-104 14 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

A novel mutation in L1CAM causes a mild form of L1 syndrome: a case report
Otter, M., Wevers, M., Pisters, M., Pfundt, R., Vos, Y., Nievelstein, R. J. & Stumpel, C. Aug 2017 In : Clinical Case Reports. 5, 8, p. 1213-1217 5 p.
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 C: Editorial Material in wn journal

Een zeldzame oorzaak voor het basaalcelnaevussyndroom
Cosgun, B., Gijezen, L. M. C., Vreeburg, M., de Boer, M., van Geel, M. & Mosterd, K. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 561-564 4 p.
Research output: Scientific › Editorial

Palmoplantaire keratodermie type Nagashima
Ramakers, N. A. M., Vreeburg, M., van Geel, M. & Steijlen, P. M. Oct 2017 In : Nederlands Tijdschrift voor Dermatologie en Venereologie. 27, 10, p. 578-580 3 p.
Research output: Scientific › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Paroxysmal sensory (spinal) attacks without hyperexplexia in a patient with a variant in the GLRA1 gene
Zwarts, M. J., Willemsen, M. H., Kamsteeg, E. -J. & Schelhaas, H. J. 15 Jul 2017 In : Journal of the Neurological Sciences. 378, p. 175-176 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat06 Scientific national journal

Epilepsie en genetica
Rouhl, R., Schelhaas, H. J. & Willemsen, M. 2017 In : Neuron. 22, p. 22-27 6 p.
Research output: Scientific - peer-review › Article

Complex Genetics

Cat02 SCI/SSCI journal (WI-1)

The effect of hypohydration on endothelial function in young healthy adults
Arnaoutis, G., Kavouras, S. A., Stratakis, N., Likka, M., Mitrakou, A., Papamichael, C., Sidossis, L. S. & Stamatelopoulos, K. Apr 2017 In : European Journal of Nutrition. 56, 3, p. 1211-1217 7 p.
Research output: Scientific - peer-review › Article

Telomere tracking from birth to adulthood and residential traffic exposure
Bijnens, E. M., Zeegers, M. P., Derom, C., Martens, D. S., Gielen, M., Hageman, G. J., Plusquin, M., Thiery, E., Vlietinck, R. & Nawrot, T. S. 21 Nov 2017 In : BMC Medicine. 15, 10 p., 205
Research output: Scientific - peer-review › Article

Blood pressure in young adulthood and residential greenness in the early-life environment of twins
Bijnens, E. M., Nawrot, T. S., Loos, R. J. F., Gielen, M., Vlietinck, R., Derom, C. & Zeegers, M. P. 5 Jun 2017 In : Environmental Health. 16, 8 p., 53
Research output: Scientific - peer-review › Article

Associations between maternal long-chain polyunsaturated fatty acid concentrations and child cognition at 7 years of age: The MEFAB birth cohort
Brouwer-Brolsma, E. M., van de Resta, O., Godschalk, R., Zeegers, M. P. A., Gielen, M. & de Groot, R. H. M. Nov 2017 In : Prostaglandins Leukotrienes and Essential Fatty Acids. 126, p. 92-97 6 p.
Research output: Scientific - peer-review › Article

Multiplex screening of 422 candidate serum biomarkers in bladder cancer patients identifies syndecan-1 and macrophage colony-stimulating factor 1 as prognostic indicators
Bryan, R. T., Gordon, N. S., Abbotts, B., Zeegers, M. P., Cheng, K. K., James, N. D. & Ward, D. G. Jun 2017 In : Translational Cancer Research. 6, p. S657-S665 9 p.
Research output: Scientific - peer-review › Article

Treatment of lumbar degenerative disc disease-associated radicular pain with culture-expanded autologous mesenchymal stem cells: a pilot study on safety and efficacy
Centeno, C., Markle, J., Dodson, E., Stemper, I., Williams, C. J., Hyzy, M., Ichim, T. & Freeman, M. 22 Sep 2017 In : Journal of Translational Medicine. 15, 12 p., 197
Research output: Scientific - peer-review › Article

Serotonin 5-HTTLPR Genotype Modulates Reactive Visual Scanning of Social and Non-social Affective Stimuli in Young Children
Christou, A. I., Wallis, Y., Bair, H., Zeegers, M. & McCleery, J. P. 23 Jun 2017 In : Frontiers in Behavioral Neuroscience. 11, 12 p., 118
Research output: Scientific - peer-review › Article

Interleukin-17-positive mast cells influence outcomes from BCG for patients with CIS: Data from a comprehensive characterisation of the immune microenvironment of urothelial bladder cancer
Dowell, A. C., Cobby, E., Wen, K., Devall, A. J., During, V., Anderson, J., James, N. D., Cheng, K. K., Zeegers, M. P., Bryan, R. T. & Taylor, G. S. 20 Sep 2017 In : PLoS One. 12, 9, 17 p., 0184841
Research output: Scientific - peer-review › Article

Scientific citations favor positive results: a systematic review and meta-analysis
Duyx, B., Urlings, M. J. E., Swaen, G. M. H., Bouter, L. M. & Zeegers, M. P. Aug 2017 In : Journal of Clinical Epidemiology. 88, p. 92-101 10 p.
Research output: Scientific - peer-review › Review article

UroMark-a urinary biomarker assay for the detection of bladder cancer
Feber, A. , Dhami, P. , Dong, L. , de Winter, P. , Tan, W. S. , Martinez-Fernandez, M. , Paul, D. S. , Hynes-Allen, A. , Rezaee, S. , Gurung, P. , Rodney, S. , Mehmood, A. , Villacampa, F. , de la Rosa, F. , Jameson, C. , Cheng, K. K. , Zeegers, M. P. , Bryan, R. T. , James, N. D. , Paramio, J. M. & 3 others Freeman, A., Beck, S. & Kelly, J. D. 31 Jan 2017 In : Clinical Epigenetics. 9, 10 p., 8
Research output: Scientific - peer-review › Article

A multistate population-based analysis of linked maternal and neonatal discharge records to identify risk factors for neonatal brachial plexus injury
Freeman, M. D., Goodyear, S. M. & Leith, W. M. Mar 2017 In : International Journal of Gynecology & Obstetrics. 136, 3, p. 331-336 6 p.
Research output: Scientific - peer-review › Article

Epidemiology and burden of systemic lupus erythematosus in a Southern European population: data from the community-based lupus registry of Crete, Greece
Gergianaki, I., Fanouriakis, A., Repa, A., Tzanakakis, M., Adamichou, C., Pompieri, A., Spirou, G., Bertsias, A., Kabouraki, E., Tzanakis, I., Chatzi, L., Sidiropoulos, P., Boumpas, D. T. & Bertsias, G. K. Dec 2017 In : Annals of the Rheumatic Diseases. 76, 12, p. 1992-2000 9 p.
Research output: Scientific - peer-review › Article

Improvements in the Long-Term Outcome of Crohn's Disease Over the Past Two Decades and the Relation to Changes in Medical Management: Results from the Population-Based IBDSL Cohort
Jeuring, S. F. G., van den Heuvel, T. R. A., Liu, L. Y. L., Zeegers, M. P., Hameeteman, W. H., Romberg-Camps, M. J. L., Oostenbrug, L. E., Masclee, A. A. M., Jonkers, D. M. A. E. & Pierik, M. J. Feb 2017 In : American Journal of Gastroenterology. 112, 2, p. 325-336 12 p.
Research output: Scientific - peer-review › Article

Network topology of NaV1.7 mutations in sodium channel-related painful disorders
Kapetis, D., Sassone, J., Yang, Y., Galbardi, B., Xenakis, M. N., Westra, R. L., Szklarczyk, R., Lindsey, P., Faber, C. G., Gerrits, M., Merkies, I. S. J., Dib-Hajj, S. D., Mantegazza, M., Waxman, S. G., PROPANE Study Grp, J.M. Smeets, H. & Lauria, G. 24 Feb 2017 In : BMC Systems Biology. 11, 16 p., 28
Research output: Scientific - peer-review › Article

Precision medicine in circadian rhythm sleep-wake disorders: current state and future perspectives
Keijzer, H., Snitselaar, M. A., Smits, M. G., Spruyt, K., Zee, P. C., Ehrhart, F. & Curfs, L. M. G. Mar 2017 In : Personalized Medicine. 14, 2, p. 171-182 12 p.
Research output: Scientific - peer-review › Review article

Can dim light melatonin onset be predicted by the timing of sleep in patients with possible circadian sleep-wake rhythm disorders?
Keijzer, H., Spruyt, K., Smits, M. G., de Geest, A. & Curfs, L. M. G. 2017 In : Biological Rhythm Research. 48, 4, p. 557-566 10 p.
Research output: Scientific - peer-review › Article

Incontinence and psychological symptoms in individuals with Mowat-Wilson Syndrome
Niemczyk, J., Einfeld, S., Mowat, D., Equit, M., Wagner, C., Curfs, L. & von Gontard, A. Mar 2017 In : Research in Developmental Disabilities. 62, p. 230-237 8 p.
Research output: Scientific - peer-review › Article

Incontinence in persons with Down Syndrome
Niemczyk, J., von Gontard, A., Equit, M., Medoff, D., Wagner, C. & Curfs, L. Aug 2017 In : Neurourology and Urodynamics. 36, 6, p. 1550-1556 7 p.
Research output: Scientific - peer-review › Article

Vitamin D insufficient levels during pregnancy and micronuclei frequency in peripheral blood T lymphocytes mothers and newborns (Rhea cohort, Crete)
O'Callaghan-Gordo, C., Kogevinas, M., Fthenou, E., Pedersen, M., Espinosa, A., Chalkiadaki, G., Daraki, V., Dermitzaki, E., Decordier, I., Georgiou, V., Merlo, D. F., Roumeliotaki, T., Loock, K. V., Kleinjans, J., Kirsch-Volders, M. & Chatzi, L. Aug 2017 In : Clinical Nutrition. 36, 4, p. 1029-1035 7 p.
Research output: Scientific - peer-review › Article

Associations of cord blood metabolites with perinatal characteristics, newborn anthropometry, and cord blood hormones in project viva
Perng, W., Rifas-Shiman, S. L., McCulloc, S., Chatzi, L., Mantzoros, C., Hivert, M-F. & Oken, E. Nov 2017 In : Metabolism-Clinical and Experimental. 76, p. 11-22 12 p.
Research output: Scientific - peer-review › Article

Physical activity and risk of prostate and bladder cancer in China: The South and East China case-control study on prostate and bladder cancer
Reulen, R. C., de Vogel, S., Zhong, W., Zhong, Z., Xie, L-P., Hu, Z., Deng, Y., Yang, K., Liang, Y., Zeng, X., Wong, Y. C., Tam, P-C., Hemelt, M. & Zeegers, M. P. 2 Jun 2017 In : PLoS One. 12, 6, 11 p., e0178613
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
Sallevelt, S. C. E. H., de Koning, B., Szklarczyk, R., Paulussen, A. D. C., de Die-Smulders, C. E. M. & Smeets, H. J. M. May 2017 In : Genetics in Medicine. 19, 5, p. 583-592 10 p.
Research output: Scientific - peer-review › Article

A homozygous FITM2 mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy
Seco, C. Z. , Castells-Nobau, A. , Joo, S. , Schraders, M. , Foo, J. N. , van der Voet, M. , Velan, S. S. , Nijhof, B. , Oostrik, J. , de Vrieze, E. , Katana, R. , Mansoor, A. , Huynen, M. , Szklarczyk, R. , Oti, M. , Tranebjaerg, L. , van Wijk, E. , Scheffer-de Gooyert, J. M. , Siddique, S. , Baets, J. & 10 others de Jonghe, P., Kazmi, S. A. R., Sadananthan, S. A., van de Warrenburg, B. P., Khor, C. C., Goepfert, M. C., Qamar, R., Schenck, A., Kremer, H. & Siddiqi, S. 1 Feb 2017 In : Disease Models & Mechanisms. 10, 2, p. 105-118 14 p.
Research output: Scientific - peer-review › Article

Polyunsaturated fatty acid levels at birth and child-to-adult growth: Results from the MEFAB cohort
Stratakis, N., Gielen, M., Margetaki, K., Godschalk, R. W., van der Wurff, I. S. M., Rouschop, S., Ibrahim, A., Antoniou, E., Chatzi, L., de Groot, R. H. M. & Zeegers, M. P. 2017 In : Prostaglandins Leukotrienes and Essential Fatty Acids. 126, p. 72-78 7 p.
Research output: Scientific - peer-review › Article

Fish and seafood consumption during pregnancy and the risk of asthma and allergic rhinitis in childhood: a pooled analysis of 18 European and US birth cohorts
Stratakis, N. , Roumeliotaki, T. , Oken, E. , Ballester, F. , Barros, H. , Basterrechea, M. , Cordier, S. , de Groot, R. , den Dekker, H. T. , Duijts, L. , Eggesbo, M. , Fantini, M. P. , Forastiere, F. , Gehring, U. , Gielen, M. , Gori, D. , Govarts, E. , Inskip, H. M. , Iszatt, N. , Jansen, M. & 22 others Kelleher, C., Mehegan, J., Molto-Puigmarti, C., Mommers, M., Oliveira, A., Olsen, S. F., Pele, F., Pizzi, C., Porta, D., Richiardi, L., Rifas-Shiman, S. L., Robinson, S. M., Schoeters, G., Strom, M., Sunyer, J., Thijs, C., Vrijheid, M., Vrijkotte, T. G. M., Wijga, A. H., Kogevinas, M., Zeegers, M. P. & Chatzi, L. Oct 2017 In : International Journal of Epidemiology. 46, 5, p. 1465-1477 13 p.
Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

A 20-Year Temporal Change Analysis in Incidence, Presenting Phenotype and Mortality, in the Dutch IBDSL Cohort-Can Diagnostic Factors Explain the Increase in IBD Incidence?
van den Heuvel, T. R. A., Jeuring, S. F. G., Zeegers, M. P., van Dongen, D. H. E., Wolters, A., Masclee, A. A. M., Hameeteman, W. H., Romberg-Camps, M. J. L., Oostenbrug, L. E., Pierik, M. J. & Jonkers, D. M. Oct 2017 In : Journal of Crohns & Colitis. 11, 10, p. 1169-1179 11 p.
Research output: Scientific - peer-review › Article

Cohort Profile: The Inflammatory Bowel Disease South Limburg Cohort (IBDSL)
van den Heuvel, T. R. A., Jonkers, D. M., Jeuring, S. F. G., Romberg-Camps, M. J. L., Oostenbrug, L. E., Zeegers, M. P., Masclee, A. A. & Pierik, M. J. Apr 2017 In : International Journal of Epidemiology. 46, 2, p. e7 9 p.
Research output: Scientific - peer-review › Article

A Review of Recruitment, Adherence and Drop-Out Rates in Omega-3 Polyunsaturated Fatty Acid Supplementation Trials in Children and Adolescents
van der Wurff, I. S. M., Meyer, B. J. & de Groot, R. H. M. May 2017 In : Nutrients. 9, 5, 32 p., 474
Research output: Scientific - peer-review › Review article

Do-Not-Attempt-Resuscitation orders for people with intellectual disabilities: dilemmas and uncertainties for ID physicians and trainees. The importance of the deliberation process
Wagemans, A. M. A., Lantman-de Valk, H. M. J. V. S., Proot, I. M., Bressers, A. M., Metsemakers, J., Tuffrey-Wijne, I., Groot, M. & Curfs, L. M. G. Mar 2017 In : Journal of Intellectual Disability Research. 61, 3, p. 245-254 10 p.
Research output: Scientific - peer-review › Article

Incontinence in persons with Angelman syndrome
Wagner, C., Niemczyk, J., Equit, M., Curfs, L. & von Gontard, A. Feb 2017 In : European Journal of Pediatrics. 176, 2, p. 225-232 8 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

The use of lumbar epidural injection of platelet lysate for treatment of radicular pain
Centeno, C., Markle, J., Dodson, E., Stemper, I., Hyzy, M., Williams, C. & Freeman, M. 25 Nov 2017 In : Journal of Experimental Orthopaedics. 4, 11 p., 38
Research output: Scientific - peer-review › Article

Selective citation in the literature on swimming in chlorinated water and childhood asthma: a network analysis
Duyx, B., Urlings, M. J. E., Swaen, G. M. H., Bouter, L. M. & Zeegers, M. P. 2017 In : Research integrity and peer review. 2, p. 17
Research output: Scientific - peer-review › Article

Drug Overdose Surveillance and Information Sharing Via a Public Database: The Role of the Medical Examiner/Coroner
Williams, K. E. & Freeman, M. Mar 2017 In : Academic Forensic Pathology. 7, 1, p. 60-72
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 A: Editorial Material in wi-1 journal

Cohort Profile: The Mother-Child Cohort in Crete, Greece (Rhea Study)
Chatzi, L., Leventakou, V., Vafeiadi, M., Koutra, K., Roumeliotaki, T., Chalkiadaki, G., Karachaliou, M., Daraki, V., Kyriklaki, A., Kampouri, M., Fthenou, E., Sarri, K., Vassilaki, M., Fasoulaki, M., Bitsios, P., Koutis, A., Stephanou, E. G. & Kogevinas, M. Oct 2017 In : International Journal of Epidemiology. 46, 5, p. 1392-1393k 13 p.
Research output: Scientific - peer-review › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Re: Lifestyle and bladder cancer prevention: no consistent evidence from cohort studies
Wesselius, A. & Zeegers, M. Nov 2017 In : European Journal of Epidemiology. 32, 11, p. 1037-1038 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat06 Scientific national journal

Veranderingen in de behandeling van ziekte van Crohn*: Minder ziekenhuisopnames, operaties en gebruik van glucocorticoïden
Jeuring, S., van den Heuvel, T., Zeegers, M., Haans, J. J., Romberg-Camps, M. J. L., Oostenbrug, L. E., Stassen, L., Masclee, A., Jonkers, D. & Pierik, M. 2017 In : Nederlands Tijdschrift voor Geneeskunde. 161, D1641
Research output: Scientific - peer-review › Article

Cat07 Book(contribution)/congrespapers - Cat07 Book contribution (BB)

Maternal fish intake during pregnancy and effects on the offspring
Chatzi, L. & Stratakis, N. Oct 2017 *Diet, Nutrition, and Fetal Programming.* Humana Press, p. 241-260
Research output: Scientific › Chapter

Genetic Factors and Alcohol Consumption
Weijenberg, M., Kok, G., Roozen, S. & Curfs, L. 1 Mar 2017 *Fetal Alcohol Spectrum Disorder: A knowledge synthesis.* Maastricht: Datawyse / Universitaire Pers Maastricht, p. 30-32 3 p.
Research output: Scientific › Chapter

Genetics & Cell Biology

Cat02 SCI/SSCI journal (WI-1)

Inhibition of CD40-TRAF6 interactions by the small molecule inhibitor 6877002 reduces neuroinflammation
Aarts, S. A. B. M., Seijkens, T. T. P., Kusters, P. J. H., van der Pol, S. M. A., Zarzycka, B., Heijnen, P. D. A. M., Beckers, L., den Toom, M., Gijbels, M. J. J., Boon, L., Weber, C., de Vries, H. E., Nicolaes, G. A. F., Dijkstra, C. D., Kooij, G. & Lutgens, E. 12 May 2017 In : Journal of Neuroinflammation. 14, 14 p., 105
Research output: Scientific - peer-review › Article

Diabetic db/db mice do not develop heart failure upon pressure overload: a longitudinal in vivo PET, MRI, and MRS study on cardiac metabolic, structural, and functional adaptations
Abdurrachim, D., Nabben, M., Hoerr, V., Kuhlmann, M. T., Bovenkamp, P., Ciapaite, J., Geraets, I. M. E., Coumans, W., Luiken, J. J. F. P., Glatz, J. F. C., Schaefers, M., Nicolay, K., Faber, C., Hermann, S. & Prompers, J. J. 1 Aug 2017 In : Cardiovascular Research. 113, 10, p. 1148-1160 13 p.
Research output: Scientific - peer-review › Article

The effect of hypohydration on endothelial function in young healthy adults
Arnaoutis, G., Kavouras, S. A., Stratakis, N., Likka, M., Mitrakou, A., Papamichael, C., Sidossis, L. S. & Stamatelopoulos, K. Apr 2017 In : European Journal of Nutrition. 56, 3, p. 1211-1217 7 p.
Research output: Scientific - peer-review › Article

Inhibition of atherogenesis by the COP9 signalosome subunit 5 in vivo
Asare, Y. , Ommer, M. , Azombo, F. A. , Alampour-Rajabi, S. , Sternkopf, M. , Sanati, M. , Gijbels, M. J. , Schmitz, C. , Sinitski, D. , Tilstam, P. V. , Lue, H. , Gessner, A. , Lange, D. , Schmid, J. A. , Weber, C. , Dichgans, M. , Jankowski, J. , Pardi, R. , de Winther, M. P. J. , Noels, H. & 1 others Bernhagen, J. 14 Mar 2017 In : Proceedings of the National Academy of Sciences of the United States of America. 114, 13, p. E2766-E2775 10 p.
Research output: Scientific - peer-review › Article

A Liver-Specific Long Noncoding RNA With a Role in Cell Viability Is Elevated in Human Nonalcoholic Steatohepatitis
Atanasovska, B., Rensen, S. S., van der Sijde, M. R., Marsman, G., Kumar, V., Jonkers, I., Withoff, S., Shiri-Sverdlov, R., Greve, J. W. M., Faber, K. N., Moshage, H., Wijmenga, C., van de Sluis, B., Hofker, M. H. & Fu, J. Sep 2017 In : Hepatology. 66, 3, p. 794-808 15 p.
Research output: Scientific - peer-review › Article

The Pathogenesis of Ventral Idiopathic Herniation of the Spinal Cord: A Hypothesis Based on the Review of the Literature
Bartels, R. H. M. A., Brunner, H., Hosman, A., van Alfen, N. & Grotenhuis, J. A. 11 Sep 2017 In : Frontiers in Neurology. 8, 10 p., 476
Research output: Scientific - peer-review › Article

Telomere tracking from birth to adulthood and residential traffic exposure
Bijnens, E. M., Zeegers, M. P., Derom, C., Martens, D. S., Gielen, M., Hageman, G. J., Plusquin, M., Thiery, E., Vlietinck, R. & Nawrot, T. S. 21 Nov 2017 In : BMC Medicine. 15, 10 p., 205
Research output: Scientific - peer-review › Article

Blood pressure in young adulthood and residential greenness in the early-life environment of twins
Bijnens, E. M., Nawrot, T. S., Loos, R. J. F., Gielen, M., Vlietinck, R., Derom, C. & Zeegers, M. P. 5 Jun 2017 In : Environmental Health. 16, 8 p., 53
Research output: Scientific - peer-review › Article

Functional mucous layer and healing of proximal colonic anastomoses in an experimental model
Bosmans, J. W. A. M., Jongen, A. C. H. M., Birchenough, G. M. H., Nystrom, E. E. L., Gijbels, M. J. J., Derikx, J. P. M., Bouvy, N. D. & Hansson, G. C. Apr 2017 In : British Journal of Surgery. 104, 5, p. 619-630 12 p.
Research output: Scientific - peer-review › Article

Comparison of three different application routes of butyrate to improve colonic anastomotic strength in rats
Bosmans, J. W. A. M., Jongen, A. C. H. M., Boonen, B. T. C., van Rijn, S., Scognamiglio, F., Stucchi, L., Gijbels, M. J. J., Marsich, E. & Bouvy, N. D. Mar 2017 In : International Journal of Colorectal Disease. 32, 3, p. 305-313 9 p.
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International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases
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Research output: Scientific - peer-review › Article

Combining traditional dietary assessment methods with novel metabolomics techniques: present efforts by the Food Biomarker Alliance
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Research output: Scientific - peer-review › Article

Associations between maternal long-chain polyunsaturated fatty acid concentrations and child cognition at 7 years of age: The MEFAB birth cohort
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Research output: Scientific - peer-review › Article

Expanding the clinical spectrum of recessive truncating mutations of KLHL7 to a Bohring-Opitz-like phenotype
Bruel, A-L. , Bigoni, S. , Kennedy, J. , Whiteford, M. , Buxton, C. , Parmeggiani, G. , Wherlock, M. , Woodward, G. , Greenslade, M. , Williams, M. , St-Onge, J. , Ferlini, A. , Garani, G. , Ballardini, E. , van Bon, B. W. , Acuna-Hidalgo, R. , Bohring, A. , Deleuze, J-F. , Boland, A. , Meyer, V. & 10 others Olaso, R., Ginglinger, E., Riviere, J-B., Brunner, H. G., Hoischen, A., Newbury-Ecob, R., Faivre, L., Thauvin-Robinet, C., Thevenon, J. & DDD Study Dec 2017 In : Journal of Medical Genetics. 54, 12, p. 830-835 6 p.
Research output: Scientific - peer-review › Article

Multiplex screening of 422 candidate serum biomarkers in bladder cancer patients identifies syndecan-1 and macrophage colony-stimulating factor 1 as prognostic indicators
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Research output: Scientific - peer-review › Article

Malondialdehyde Epitopes Are Sterile Mediators of Hepatic Inflammation in Hypercholesterolemic Mice
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Treatment of lumbar degenerative disc disease-associated radicular pain with culture-expanded autologous mesenchymal stem cells: a pilot study on safety and efficacy
Centeno, C., Markle, J., Dodson, E., Stemper, I., Williams, C. J., Hyzy, M., Ichim, T. & Freeman, M. 22 Sep 2017 In : Journal of Translational Medicine. 15, 12 p., 197
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2-Arachidonoylglycerol ameliorates inflammatory stress-induced insulin resistance in cardiomyocytes
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Research output: Scientific - peer-review › Article

Serotonin 5-HTTLPR Genotype Modulates Reactive Visual Scanning of Social and Non-social Affective Stimuli in Young Children
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Research output: Scientific - peer-review › Article

Electrochemical reduction of CerMet fuels for transmutation using surrogate CeO_2-Mo pellets
Claux, B., Soucek, P., Malmbeck, R., Rodrigues, A. & Glatz, J. -P. Aug 2017 In : Journal of Nuclear Materials. 491, p. 190-198 9 p.
Research output: Scientific - peer-review › Article

Estimates of live birth prevalence of children with Down syndrome in the period 1991-2015 in the Netherlands
de Graaf, G., Engelen, J. J. M., Gijsbers, A. C. J., Hochstenbach, R., Hoffer, M. J. V., Kooper, A. J. A., Sikkema-Raddatz, B., Srebniak, M. I., van der Kevie-Kersemaekers, A. M. F., van Zutven, L. J. C. M. & Voorhoeve, E. May 2017 In : Journal of Intellectual Disability Research. 61, 5, p. 461-470 10 p.
Research output: Scientific - peer-review › Article

Assessment of Chlamydia suis Infection in Pig Farmers
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Research output: Scientific - peer-review › Article

The 6p25 deletion syndrome: An update on a rare neurocristopathy
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Prevalence of Trichomonas vaginalis infection and protozoan load in South African women: a cross-sectional study
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Research output: Scientific - peer-review › Article

BRCA1 mutation carriers have a lower number of mature oocytes after ovarian stimulation for IVF/PGD
Derks-Smeets, I. A. P., van Tilborg, T. C., van Montfoort, A., Smits, L., Torrance, H. L., Meijer-Hoogeveen, M., Broekmans, F., Dreesen, J. C. F. M., Paulussen, A. D. C., Tjan-Heijnen, V. C. G., Homminga, I., van den Berg, M. M. J., Ausems, M. G. E. M., de Rycke, M., de Die-Smulders, C. E. M., Verpoest, W. & van Golde, R. Nov 2017 In : Journal of Assisted Reproduction and Genetics. 34, 11, p. 1475-1482 8 p.
Research output: Scientific - peer-review › Article

Uptake of prenatal diagnostic testing for retinoblastoma compared to other hereditary cancer syndromes in the Netherlands
Dommering, C. J., Henneman, L., van der Hout, A. H., Jonker, M. A., Tops, C. M. J., van den Ouweland, A. M. W., van der Luijt, R. B., Mensenkamp, A. R., Hogervorst, F. B. L., Redeker, E. J. W., de Die-Smulders, C. E. M., Moll, A. C. & Meijers-Heijboer, H. Apr 2017 In : Familial Cancer. 16, 2, p. 271-277 7 p.
Research output: Scientific - peer-review › Article

Interleukin-17-positive mast cells influence outcomes from BCG for patients with CIS: Data from a comprehensive characterisation of the immune microenvironment of urothelial bladder cancer
Dowell, A. C., Cobby, E., Wen, K., Devall, A. J., During, V., Anderson, J., James, N. D., Cheng, K. K., Zeegers, M. P., Bryan, R. T. & Taylor, G. S. 20 Sep 2017 In : PLoS One. 12, 9, 17 p., 0184841
Research output: Scientific - peer-review › Article

Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability
Dudding-Byth, T., Baxter, A., Holliday, E. G., Hackett, A., O'Donnell, S., White, S. M., Attia, J., Brunner, H., de Vries, B., Koolen, D., Kleefstra, T., Ratwatte, S., Riveros, C., Brain, S. & Lovell, B. C. 19 Dec 2017 In : BMC Biotechnology. 17, 9 p., 90
Research output: Scientific - peer-review › Article

Scientific citations favor positive results: a systematic review and meta-analysis
Duyx, B., Urlings, M. J. E., Swaen, G. M. H., Bouter, L. M. & Zeegers, M. P. Aug 2017 In : Journal of Clinical Epidemiology. 88, p. 92-101 10 p.
Research output: Scientific - peer-review › Review article

TSC2 c.1864C > T Variant Aassociated with Mild Cases of Tuberous Sclerosis Complex
Farach, L. S., Gibson, W. T., Sparagana, S. P., Nellist, M., Stumpel, C. T. R. M., Hietala, M., Friedman, E., Pearson, D. A., Creighton, S. P., Wagemans, A., Segel, R., Ben-Shalom, E., Au, K. S. & Northrup, H. Mar 2017 In : American Journal of Medical Genetics Part A. 173, 3, p. 771-775 5 p.
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UroMark-a urinary biomarker assay for the detection of bladder cancer
Feber, A. , Dhami, P. , Dong, L. , de Winter, P. , Tan, W. S. , Martinez-Fernandez, M. , Paul, D. S. , Hynes-Allen, A. , Rezaee, S. , Gurung, P. , Rodney, S. , Mehmood, A. , Villacampa, F. , de la Rosa, F. , Jameson, C. , Cheng, K. K. , Zeegers, M. P. , Bryan, R. T. , James, N. D. , Paramio, J. M. & 3 others Freeman, A., Beck, S. & Kelly, J. D. 31 Jan 2017 In : Clinical Epigenetics. 9, 10 p., 8
Research output: Scientific - peer-review › Article

The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families
Fountain, M. D. , Aten, E. , Cho, M. T. , Juusola, J. , Walkiewicz, M. A. , Ray, J. W. , Xia, F. , Yang, Y. , Graham, B. H. , Bacino, C. A. , Potocki, L. , van Haeringen, A. , Ruivenkamp, C. A. L. , Mancias, P. , Northrup, H. , Kukolich, M. K. , Weiss, M. M. , van Ravenswaaij-Arts, C. M. A. , Mathijssen, I. B. , Levesque, S. & 19 others Meeks, N., Rosenfeld, J. A., Lemke, D., Hamosh, A., Lewis, S. K., Race, S., Stewart, L. L., Hay, B., Lewis, A. M., Guerreiro, R. L., Bras, J. T., Martins, M. P., Derksen-Lubsen, G., Peeters, E., Stumpel, C., Stegmann, S., Bok, L. A., Santen, G. W. E. & Schaaf, C. P. Jan 2017 In : Genetics in Medicine. 19, 1, p. 45-52 8 p.
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A multistate population-based analysis of linked maternal and neonatal discharge records to identify risk factors for neonatal brachial plexus injury
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Research output: Scientific - peer-review › Article

YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction
Gabriele, M. , Vulto-van Silfhout, A. T. , Germain, P-L. , Vitriolo, A. , Kumar, R. , Douglas, E. , Haan, E. , Kosaki, K. , Takenouchi, T. , Rauch, A. , Steindl, K. , Frengen, E. , Misceo, D. , Pedurupillay, C. R. J. , Stromme, P. , Rosenfeld, J. A. , Shao, Y. , Craigen, W. J. , Schaaf, C. P. , Rodriguez-Buritica, D. & 31 others Farach, L., Friedman, J., Thulin, P., McLean, S. D., Nugent, K. M., Morton, J., Nicholl, J., Andrieux, J., Stray-Pedersen, A., Chambon, P., Patrier, S., Lynch, S. A., Kjaergaard, S., Torring, P. M., Brasch-Andersen, C., Ronan, A., van Haeringen, A., Anderson, P. J., Powis, Z., Brunner, H. G., Pfundt, R., Schuurs-Hoeijmakers, J. H. M., van Bon, B. W. M., Lelieveld, S., Gilissen, C., Nillesen, W. M., Vissers, L. E. L. M., Gecz, J., Koolen, D. A., Testa, G. & de Vries, B. B. A. 1 Jun 2017 In : American Journal of Human Genetics. 100, 6, p. 907-925 19 p.
Research output: Scientific - peer-review › Article

A two dimensional electromechanical model of a cardiomyocyte to assess intra-cellular regional mechanical heterogeneities
Garcia-Canadilla, P., Rodriguez, J. F., Palazzi, M. J., Gonzalez-Tendero, A., Schonleitner, P., Balicevic, V., Loncaric, S., Luiken, J. J. F. P., Ceresa, M., Camara, O., Antoons, G., Crispi, F., Gratacos, E. & Bijnens, B. 24 Aug 2017 In : PLoS One. 12, 8, 20 p., 0182915
Research output: Scientific - peer-review › Article

Epidemiology and burden of systemic lupus erythematosus in a Southern European population: data from the community-based lupus registry of Crete, Greece
Gergianaki, I., Fanouriakis, A., Repa, A., Tzanakakis, M., Adamichou, C., Pompieri, A., Spirou, G., Bertsias, A., Kabouraki, E., Tzanakis, I., Chatzi, L., Sidiropoulos, P., Boumpas, D. T. & Bertsias, G. K. Dec 2017 In : Annals of the Rheumatic Diseases. 76, 12, p. 1992-2000 9 p.
Research output: Scientific - peer-review › Article

Awareness and attitude regarding reproductive options of persons carrying a BRCA mutation and their partners
Gietel-Habets, J. J. G., de Die-Smulders, C. E. M., Derks-Smeets, I. A. P., Tibben, A., Tjan-Heijnen, V. C. G., van Golde, R., Gomez-Garcia, E., Kets, C. M. & van Osch, L. A. D. M. Mar 2017 In : Human Reproduction. 32, 3, p. 588-597 10 p.
Research output: Scientific - peer-review › Article

From fat to FAT (CD36/SR-B2): Understanding the regulation of cellular fatty acid uptake
Glatz, J. F. C. & Luiken, J. J. F. P. May 2017 In : Biochimie. 136, p. 21-26 6 p.
Research output: Scientific - peer-review › Article

Proposed guidelines to evaluate scientific validity and evidence for genotype-based dietary advice
Grimaldi, K. A. , van Ommen, B. , Ordovas, J. M. , Parnell, L. D. , Mathers, J. C. , Bendik, I. , Brennan, L. , Celis-Morales, C. , Cirillo, E. , Daniel, H. , de Kok, B. , El-Sohemy, A. , Fairweather-Tait, S. J. , Fallaize, R. , Fenech, M. , Ferguson, L. R. , Gibney, E. R. , Gibney, M. , Gjelstad, I. M. F. , Kaput, J. & 16 others Karlsen, A. S., Kolossa, S., Lovegrove, J., Macready, A. L., Marsaux, C. F. M., Martinez, J. A., Milagro, F., Navas-Carretero, S., Roche, H. M., Saris, W. H. M., Traczyk, I., van Kranen, H., Verschuren, L., Virgili, F., Weber, P. & Bouwman, J. 15 Dec 2017 In : Genes and nutrition. 12, 12 p., 35
Research output: Scientific - peer-review › Review article

Comprehensive global genome dynamics of Chlamydia trachomatis show ancient diversification followed by contemporary mixing and recent lineage expansion
Hadfield, J. , Harris, S. R. , Seth-Smith, H. M. B. , Parmar, S. , Andersson, P. , Giffard, P. M. , Schachter, J. , Moncada, J. , Ellison, L. , Gallo Vaulet, M. L. , Rodriguez Fermepin, M. , Radebe, F. , Mendoza, S. , Ouburg, S. , Morre, S. A. , Sachse, K. , Puolakkainen, M. , Korhonen, S. J. , Sonnex, C. , Wiggins, R. & 18 others Jalal, H., Brunelli, T., Casprini, P., Pitt, R., Ison, C., Savicheva, A., Shipitsyna, E., Hadad, R., Kari, L., Burton, M. J., Mabey, D., Solomon, A. W., Lewis, D., Marsh, P., Unemo, M., Clarke, I. N., Parkhill, J. & Thomson, N. R. Jul 2017 In : Genome Research. 27, 7, p. 1220-1229 10 p.
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Protocol for intraoperative assessment of the human cerebrovascular glycocalyx
Haeren, R. H. L., Vink, H., Staals, J., van Zandvoort, M. A. M. J., Dings, J., van Overbeeke, J. J., Hoogland, G., Rijkers, K. & Schijns, O. E. M. G. Jan 2017 In : BMJ Open. 7, 1, 7 p., 013954
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Diagnostic exome sequencing in 266 Dutch patients with visual impairment
Haer-Wigman, L. , van Zelst-Stams, W. A. G. , Pfundt, R. , van den Born, L. I. , Klaver, C. C. W. , Verheij, J. B. G. M. , Hoyng, C. B. , Breuning, M. H. , Boon, C. J. F. , Kievit, A. J. , Verhoeven, V. J. M. , Pott, J. W. R. , Sallevelt, S. C. E. H. , van Hagen, J. M. , Plomp, A. S. , Kroes, H. Y. , Lelieveld, S. H. , Hehir-Kwa, J. Y. , Castelein, S. , Nelen, M. & 5 others
Scheffer, H., Lugtenberg, D., Cremers, F. P. M., Hoefsloot, L. & Yntema, H. G. May 2017 In : European Journal of Human Genetics. 25, 5, p. 591-599 9 p.
Research output: Scientific - peer-review › Article

Novel *SLC25A32* mutation in a patient with a severe neuromuscular phenotype
Hellebrekers, D. M., Sallevelt, S. C. E. H., Theunissen, T. E. J., Hendrickx, A. T. M., Gottschalk, R. W., Hoeijmakers, J. G. J., Habets, D. D., Bierau, J., Schoonderwoerd, K. G. & Smeets, H. J. M. Jun 2017 In : European Journal of Human Genetics. 25, 7, p. 886-888 3 p.
Research output: Scientific - peer-review › Article

Novel genetic loci associated with hippocampal volume

Hibar, D. P. , Adams, H. H. H. , Jahanshad, N. , Chauhan, G. , Stein, J. L. , Hofer, E. , Renteria, M. E. , Bis, J. C. , Arias-Vasquez, A. , Ikram, M. K. , Desrivieres, S. , Vernooij, M. W. , Abramovic, L. , Alhusaini, S. , Amin, N. , Andersson, M. , Arfanakis, K. , Aribisala, B. S. , Armstrong, N. J. , Athanasiu, L. & 312 others Axelsson, T., Beecham, A. H., Beiser, A., Bernard, M., Blanton, S. H., Bohlken, M. M., Boks, M. P., Bralten, J., Brickman, A. M., Carmichael, O., Chakravarty, M. M., Chen, Q., Ching, C. R. K., Chouraki, V., Cuellar-Partida, G., Crivello, F., Den Braber, A., Nhat Trung Doan, Ehrlich, S., Giddaluru, S., Goldman, A. L., Gottesman, R. F., Grimm, O., Griswold, M. E., Guadalupe, T., Gutman, B. A., Hass, J., Haukvik, U. K., Hoehn, D., Holmes, A. J., Hoogman, M., Janowitz, D., Jia, T., Jorgensen, K. N., Karbalai, N., Kasperaviciute, D., Kim, S., Klein, M., Kraemer, B., Lee, P. H., Liewald, D. C. M., Lopez, L. M., Luciano, M., Macare, C., Marquand, A. F., Matarin, M., Mather, K. A., Mattheisen, M., McKay, D. R., Milaneschi, Y., Maniega, S. M., Nho, K., Nugent, A. C., Nyquist, P., Loohuis, L. M. O., Oosterlaan, J., Papmeyer, M., Pirpamer, L., Puetz, B., Ramasamy, A., Richards, J. S., Risacher, S. L., Roiz-Santianez, R., Rommelse, N., Ropele, S., Rose, E. J., Royle, N. A., Rundek, T., Saemann, P. G., Saremi, A., Satizabal, C. L., Schmaal, L., Schork, A. J., Shen, L., Shin, J., Shumskaya, E., Smith, A. V., Sprooten, E., Strike, L. T., Teumer, A., Tordesillas-Gutierrez, D., Toro, R., Trabzuni, D., Trompet, S., Vaidya, D., Van der Grond, J., Van der Lee, S. J., Van der Meer, D., Van Donkelaar, M. M. J., Van Eijk, K. R., Van Erp, T. G. M., Van Rooij, D., Walton, E., Westlye, L. T., Whelan, C. D., Windham, B. G., Winkler, A. M., Wittfeld, K., Woldehawariat, G., Wolf, C., Wolfers, T., Yanek, L. R., Yang, J., Zijdenbos, A., Zwiers, M. P., Agartz, I., Almasy, L., Ames, D., Amouyel, P., Andreassen, O. A., Arepalli, S., Assareh, A. A., Barral, S., Bastin, M. E., Becker, D. M., Becker, J. T., Bennett, D. A., Blangero, J., van Bokhoven, H., Boomsma, D. I., Brodaty, H., Brouwer, R. M., Brunner, H. G., Buckner, R. L., Buitelaar, J. K., Bulayeva, K. B., Cahn, W., Calhoun, V. D., Cannon, D. M., Cavalleri, G. L., Cheng, C-Y., Cichon, S., Cookson, M. R., Corvin, A., Crespo-Facorro, B., Curran, J. E., Czisch, M., Dale, A. M., Davies, G. E., De Craen, A. J. M., De Geus, E. J. C., De Jager, P. L., De Zubicaray, G. I., Deary, I. J., Debette, S., DeCarli, C., Delanty, N., Depondt, C., DeStefano, A., Dillman, A., Djurovic, S., Donohoe, G., Drevets, W. C., Duggirala, R., Dyer, T. D., Enzinger, C., Erk, S., Espeseth, T., Fedko, I. O., Fernandez, G., Ferrucci, L., Fisher, S. E., Fleischman, D. A., Ford, I., Fornage, M., Foroud, T. M., Fox, P. T., Francks, C., Fukunaga, M., Gibbs, J. R., Glahn, D. C., Gollub, R. L., Goring, H. H. H., Green, R. C., Gruber, O., Gudnason, V., Guelfi, S., Haberg, A. K., Hansell, N. K., Hardy, J., Hartman, C. A., Hashimoto, R., Hegenscheid, K., Heinz, A., Le Hellard, S., Hernandez, D. G., Heslenfeld, D. J., Ho, B-C., Hoekstra, P. J., Hoffmann, W., Hofman, A., Holsboer, F., Homuth, G., Hosten, N., Hottenga, J-J., Huentelman, M., Pol, H. E. H., Ikeda, M., Jack, C. R., Jenkinson, M., Johnson, R., Joensson, E. G., Jukema, J. W., Kahn, R. S., Kanai, R., Kloszewska, I., Knopman, D. S., Kochunov, P., Kwok, J. B., Lawrie, S. M., Lemaitre, H., Liu, X., Longo, D. L., Lopez, O. L., Lovestone, S., Martinez, O., Martinot, J-L., Mattay, V. S., McDonald, C., McIntosh, A. M., McMahon, F. J., McMahon, K. L., Mecocci, P., Melle, I., Meyer-Lindenberg, A., Mohnke, S., Montgomery, G. W., Morris, D. W., Mosley, T. H., Muhleisen, T. W., Mueller-Myhsok, B., Nalls, M. A., Nauck, M., Nichols, T. E., Niessen, W. J., Nothen, M. M., Nyberg, L., Ohi, K., Olvera, R. L., Ophoff, R. A., Pandolfo, M., Paus, T., Pausova, Z., Penninx, B. W. J. H., Pike, G. B., Potkin, S. G., Psaty, B. M., Reppermund, S., Rietschel, M., Roffman, J. L., Romanczuk-Seiferth, N., Rotter, J. I., Ryten, M., Sacco, R. L., Sachdev, P. S., Saykin, A. J., Schmidt, R., Schmidt, H., Schofield, P. R., Sigursson, S., Simmons, A., Singleton, A., Sisodiya, S. M., Smith, C., Smoller, J. W., Soininen, H., Steen, V. M., Stott, D. J., Sussmann, J. E., Thalamuthu, A., Toga, A. W., Traynor, B. J., Troncoso, J., Tsolaki, M., Tzourio, C., Uitterlinden, A. G., Hernandez, M. C. V., Van der Brug, M., van der Lugt, A., van der Wee, N. J. A., Van Haren, N. E. M., van't Ent, D., Van Tol, M-J., Vardarajan, B. N., Vellas, B., Veltman, D. J., Voelzke, H., Walter, H., Wardlaw, J. M., Wassink, T. H., Weale, M. E., Weinberger, D. R., Weiner, M. W., Wen, W., Westman, E., White, T., Wong, T. Y., Wright, C. B., Zielke, R. H., Zonderman, A. B., Martin, N. G., Van Duijn, C. M., Wright, M. J., Longstreth, W. T., Schumann, G., Grabe, H. J., Franke, B., Launer, L. J., Medland, S. E., Seshadri, S., Thompson, P. M. & Ikram, M. A. 18 Jan 2017 In : Nature Communications. 8, 12 p., 13624

Research output: Scientific - peer-review › Article

Correlated evolutionary rates across genomic compartments in Annonaceae

Hoekstra, P. H., Wieringa, J. J., Smets, E., Brandão, R. D., Lopes, J. D. C., Erkens, R. H. J. & Chatrou, L. W. Sep 2017 In : Molecular Phylogenetics and Evolution. 114, p. 63-72 10 p.

Research output: Scientific - peer-review › Article

The Netherlands Chlamydia cohort study (NECCST) protocol to assess the risk of late complications following Chlamydia trachomatis infection in women
Hoenderboom, B. M., van Oeffelen, A. A. M., van Benthem, B. H. B., van Bergen, J. E. A. M., Dukers-Muijrers, N. H. T. M., Gotz, H. M., Hoebe, C. J. P. A., Hogewoning, A. A., van der Klis, F. R. M., van Baarle, D., Land, J. A., van der Sande, M. A. B., van Veen, M. G., de Vries, F., Morre, S. A. & van den Broek, I. V. F. 11 Apr 2017 In : BMC Infectious Diseases. 17, 9 p., 264
Research output: Scientific - peer-review › Article

Lamin A/C-Related Cardiac Disease Late Onset With a Variable and Mild Phenotype in a Large Cohort of Patients With the Lamin A/C p.(Arg331Gln) Founder Mutation
Hoorntje, E. T. , Bollen, I. A. , Barge-Schaapveld, D. Q. , van Tienen, F. H. , te Meerman, G. J. , Jansweijer, J. A. , van Essen, A. J. , Volders, P. G. , Constantinescu, A. A. , van den Akker, P. C. , van Spaendonck-Zwarts, K. Y. , Oldenburg, R. A. , Marcelis, C. L. , van der Smagt, J. J. , Hennekam, E. A. , Vink, A. , Bootsma, M. , Aten, E. , Wilde, A. A. , van den Wijngaard, A. & 5 others Broers, J. L., Jongbloed, J. D., van der Velden, J., van den Berg, M. P. & van Tintelen, J. P. Aug 2017 In : Circulation : Cardiovascular Genetics. 10, 4, 36 p., 001631
Research output: Scientific - peer-review › Article

Frequency of chest pain in primary care, diagnostic tests performed and final diagnoses
Hoorweg, B. B. N., Willemsen, R. T. A., Cleef, L. E., Boogaerts, T., Buntinx, F., Glatz, J. F. C. & Dinant, G. J. Nov 2017 In : Heart. 103, 21, p. 1727-1732 6 p.
Research output: Scientific - peer-review › Article

Blood-derived macrophages prone to accumulate lysosomal lipids trigger oxLDL-dependent murine hepatic inflammation
Houben, T., Oligschlaeger, Y., Bitorina, A. V., Hendrikx, T., Walenbergh, S. M. A., Lenders, M-H., Gijbels, M. J. J., Verheyen, F., Luetjohann, D., Hofker, M. H., Binder, C. J. & Shiri-Sverdlov, R. 2 Oct 2017 In : Scientific Reports. 7, 9 p., 12550
Research output: Scientific - peer-review › Article

Cathepsin D regulates lipid metabolism in murine steatohepatitis
Houben, T., Oligschlaeger, Y., Hendrikx, T., Bitorina, A. V., Walenbergh, S. M. A., van Gorp, P. J., Gijbels, M. J. J., Friedrichs, S., Plat, J., Schaap, F. G., Luetjohann, D., Hofker, M. H. & Shiri-Sverdlov, R. 14 Jun 2017 In : Scientific Reports. 7, 10 p., 3494
Research output: Scientific - peer-review › Article

Oxidized LDL at the crossroads of immunity in non-alcoholic steatohepatitis
Houben, T., Brandsma, E., Walenbergh, S. M. A., Hofker, M. H. & Shiri-Sverdlov, R. Apr 2017 In : Biochimica et Biophysica Acta-Molecular and Cell Biology of Lipids. 1862, 4, p. 416-429 14 p.
Research output: Scientific - peer-review › Review article

Delusional and Psychotic Disorders in Juvenile Myotonic Dystrophy Type-1
Jacobs, D., Willekens, D., de Die-Smulders, C., Frijns, J-P. & Steyaert, J. Jun 2017 In : American Journal of Medical Genetics Part B-neuropsychiatric Genetics. 174, 4, p. 359-366 8 p.
Research output: Scientific - peer-review › Article

International differences in the evaluation of conditions for newborn bloodspot screening: a review of scientific literature and policy documents
Jansen, M. E., Metternick-Jones, S. C. & Lister, K. J. Jan 2017 In : European Journal of Human Genetics. 25, 1, p. 10-16 7 p.
Research output: Scientific - peer-review › Review article

Modulation of the gut microbiota impacts nonalcoholic fatty liver disease: a potential role for bile acids
Janssen, A. W. F., Houben, T., Katiraei, S., Dijk, W., Boutens, L., van der Bolt, N., Wang, Z., Brown, J. M., Hazen, S. L., Mandard, S., Shiri-Sverdlov, R., Kuipers, F., van Dijk, K. W., Vervoort, J., Stienstra, R., Hooiveld, G. J. E. J. & Kersten, S. Jul 2017 In : Journal of Lipid Research. 58, 7, p. 1399-1416 18 p.
Research output: Scientific - peer-review › Article

Improvements in the Long-Term Outcome of Crohn's Disease Over the Past Two Decades and the Relation to Changes in Medical Management: Results from the Population-Based IBDSL Cohort
Jeuring, S. F. G., van den Heuvel, T. R. A., Liu, L. Y. L., Zeegers, M. P., Hameeteman, W. H., Romberg-Camps, M. J. L., Oostenbrug, L. E., Masclee, A. A. M., Jonkers, D. M. A. E. & Pierik, M. J. Feb 2017 In : American Journal of Gastroenterology. 112, 2, p. 325-336 12 p.
Research output: Scientific - peer-review › Article

Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification
Kamps, R., Brandao, R. D., van den Bosch, B. J., Paulussen, A. D. C., Xanthoulea, S., Blok, M. J. & Romano, A. Feb 2017 In : International Journal of Molecular Sciences. 18, 2, 57 p., 308
Research output: Scientific - peer-review › Review article

Network topology of NaV1.7 mutations in sodium channel-related painful disorders
Kapetis, D., Sassone, J., Yang, Y., Galbardi, B., Xenakis, M. N., Westra, R. L., Szklarczyk, R., Lindsey, P., Faber, C. G., Gerrits, M., Merkies, I. S. J., Dib-Hajj, S. D., Mantegazza, M., Waxman, S. G., PROPANE Study Grp, J.M. Smeets, H. & Lauria, G. 24 Feb 2017 In : BMC Systems Biology. 11, 16 p., 28
Research output: Scientific - peer-review › Article

Investigating the race for the surface and skin integration in clinically retrieved abutments with two-photon microscopy
Kapsokalyvas, D., van Hoof, M., Wigren, S., Chimhanda, T., Kuijpers, H. J., Ramaekers, F. C. S., Stokroos, R. J. & van Zandvoort, M. A. M. J. 1 Nov 2017 In : Colloids and Surfaces B-Biointerfaces. 159, p. 97-107 11 p.
Research output: Scientific - peer-review › Article

Structural asymmetries of the human cerebellum in relation to cerebral cortical asymmetries and handedness
Kavaklioglu, T., Guadalupe, T., Zwiers, M., Marquand, A. F., Onnink, M., Shumskaya, E., Brunner, H., Fernandez, G., Fisher, S. E. & Francks, C. May 2017 In : Brain Structure & Function. 222, 4, p. 1611-1623 13 p.
Research output: Scientific - peer-review › Article

Precision medicine in circadian rhythm sleep-wake disorders: current state and future perspectives
Keijzer, H., Snitselaar, M. A., Smits, M. G., Spruyt, K., Zee, P. C., Ehrhart, F. & Curfs, L. M. G. Mar 2017 In : Personalized Medicine. 14, 2, p. 171-182 12 p.
Research output: Scientific - peer-review › Review article

Can dim light melatonin onset be predicted by the timing of sleep in patients with possible circadian sleep-wake rhythm disorders?
Keijzer, H., Spruyt, K., Smits, M. G., de Geest, A. & Curfs, L. M. G. 2017 In : Biological Rhythm Research. 48, 4, p. 557-566 10 p.
Research output: Scientific - peer-review › Article

Novel IRF6 Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing
Khandelwal, K. D. , Ishorst, N. , Zhou, H. , Ludwig, K. U. , Venselaar, H. , Gilissen, C. , Thonissen, M. , van Rooij, I. A. L. M. , Dreesen, K. , Steehouwer, M. , van de Vorst, M. , Bloemen, M. , van Beusekom, E. , Roosenboom, J. , Borstlap, W. , Admiraal, R. , Dormaar, T. , Schoenaers, J. , Vander Poorten, V. , Hens, G. & 10 others Verdonck, A., Berge, S., Roeleveldt, N., Vriend, G., Devriendt, K., Brunner, H. G., Mangold, E., Hoischen, A., van Bokhoven, H. & Carels, C. E. L. Feb 2017 In : Journal of Dental Research. 96, 2, p. 179-185 7 p.
Research output: Scientific - peer-review › Article

Molecular characterization, prevalence and clinical relevance of Phodopus sungorus papillomavirus type 1 (PsuPV1) naturally infecting Siberian hamsters (&ITPhodopus sungorus&IT)
Kocjan, B. J., Hosnjak, L., Racnik, J., Zadravec, M., Bakovnik, N., Cigler, B., Ummelen, M., Hopman, A. H. N., Gale, N., Svara, T., Gombac, M. & Poljak, M. Nov 2017 In : Journal of General Virology. 98, 11, p. 2799-2809 11 p.
Research output: Scientific - peer-review › Article

PBCA-based polymeric microbubbles for molecular imaging and drug delivery
Koczera, P., Appold, L., Shi, Y., Liu, M., Dasgupta, A., Pathak, V., Ojha, T., Fokong, S., Wu, Z., van Zandvoort, M., Iranzo, O., Kuehne, A. J. C., Pich, A., Kiessling, F. & Lammers, T. 10 Aug 2017 In : Journal of Controlled Release. 259, p. 128-135 8 p.
Research output: Scientific - peer-review › Article

Parenteral nutrition dysregulates bile salt homeostasis in a rat model of parenteral nutrition-associated liver disease
Koelfat, K. V. K., Schaap, F. G., Hodin, C. M. J. M., Visschers, R. G. J., Svavarsson, B. I., Lenicek, M., Shiri-Sverdlov, R., Lenaerts, K. & Damink, S. W. M. O. Oct 2017 In : Clinical Nutrition. 36, 5, p. 1403-1410 8 p.
Research output: Scientific - peer-review › Article

Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder
Koemans, T. S., Kleefstra, T., Chubak, M. C., Stone, M. H., Reijnders, M. R. F., de Munnik, S., Willemsen, M. H., Fenckova, M., Stumpel, C. T. R. M., Bok, L. A., Saenz, M. S., Byerly, K. A., Baughn, L. B., Stegmann, A. P. A., Pfundt, R., Zhou, H., van Bokhoven, H., Schenck, A. & Kramer, J. M. Oct 2017 In : Plos Genetics. 13, 10, 24 p., 1006864
Research output: Scientific - peer-review › Article

Investigations of Glucocorticoid Action in GN
Kuppe, C., van Roeyen, C., Leuchtle, K., Kabgani, N., Vogt, M., Van Zandvoort, M., Smeets, B., Floege, J., Groene, H-J. & Moellert, M. J. May 2017 In : Journal of the American Society of Nephrology. 28, 5, p. 1408-1420 13 p.
Research output: Scientific - peer-review › Article

Constitutive CD40 Signaling in Dendritic Cells Limits Atherosclerosis by Provoking Inflammatory Bowel Disease and Ensuing Cholesterol Malabsorption
Kusters, P., Seijkens, T., Buerger, C., Legein, B., Winkels, H., Gijbels, M., Barthels, C., Bennett, R., Beckers, L., Atzler, D., Biessen, E., Brocker, T., Weber, C., Gerdes, N. & Lutgenst, E. Dec 2017 In : American Journal of Pathology. 187, 12, p. 2912-2919 8 p.
Research output: Scientific - peer-review › Article

Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype
Lamers, I. J. C. , Reijnders, M. R. F. , Venselaar, H. , Kraus, A. , Jansen, S. , de Vries, B. B. A. , Houge, G. , Gradek, G. A. , Seo, J. , Choi, M. , Chae, J-H. , van der Burgt, I. , Pfundt, R. , Letteboer, S. J. F. , van Beersum, S. E. C. , Dusseljee, S. , Brunner, H. G. , Doherty, D. , Kleefstra, T. , Roepman, R. & 1 others DDD Study 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 824-832 9 p.
Research output: Scientific - peer-review › Article

Risk of eating disorders in a non-western setting: an exploratory study in Khartoum state, Sudan
Lau, C. C. L. & Ambrosino, E. Dec 2017 In : Eating and Weight Disorders-Studies on Anorexia Bulimia and Obesity. 22, 4, p. 649-656 8 p.
Research output: Scientific - peer-review › Article

Amiloride-sensitive cation channel 2 genotype affects the response to a carbon dioxide panic challenge
Leibold, N. K., van den Hove, D. L. A., Viechtbauer, W., Kenis, G., Goossens, L., Lange, I., Knuts, I., Smeets, H. J., Myin-Germeys, I., Steinbusch, H. W. M. & Schruers, K. R. J. Oct 2017 In : Journal of Psychopharmacology. 31, 10, p. 1294-1301 8 p.
Research output: Scientific - peer-review › Article

Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes
Lelieveld, S. H., Wiel, L., Venselaar, H., Pfundt, R., Vriend, G., Veltman, J. A., Brunner, H. G., Vissers, L. E. L. M. & Gilissen, C. 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 478-484 7 p.
Research output: Scientific - peer-review › Article

***De Novo* Missense Mutations in *DHX30* Impair Global Translation and Cause a Neurodevelopmental Disorder**
Lessel, D. , Schob, C. , Kuery, S. , Reinders, M. R. F. , Harel, T. , Eldomery, M. K. , Coban-Akdemir, Z. , Denecke, J. , Edvardson, S. , Colin, E. , Stegmann, A. P. A. , Gerkes, E. H. , Tessarech, M. , Bonneau, D. , Barth, M. , Besnard, T. , Cogne, B. , Revah-Politi, A. , Strom, T. M. , Rosenfeld, J. A. & 24 others Yang, Y., Posey, J. E., Immken, L., Oundjian, N., Helbig, K. L., Meeks, N., Zegar, K., Morton, J., Schieving, J. H., Claasen, A., Huentelman, M., Narayanan, V., Ramsey, K., Brunner, H. G., Elpeleg, O., Mercier, S., Bezieau, S., Kubisch, C., Kleefstra, T., Kindler, S., Lupski, J. R., Kreienkamp, H-J., DDD study & C4RCD Res Grp 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 716-724 9 p.
Research output: Scientific - peer-review › Article

Palmitate-Induced Vacuolar-Type H(+)-ATPase Inhibition Feeds Forward Into Insulin Resistance and Contractile Dysfunction
Liu, Y., Steinbusch, L. K. M., Nabben, M., Kapsokalyvas, D., van Zandvoort, M., Schonleitner, P., Antoons, G., Simons, P. J., Coumans, W. A., Geomini, A., Chanda, D., Glatz, J. F. C., Neumann, D. & Luiken, J. J. F. P. 1 Jun 2017 In : Diabetes. 66, 6, p. 1521-1534 14 p.
Research output: Scientific - peer-review › Article

The attitudes of Dutch fertility specialists towards the addition of genetic testing in screening of tubal factor infertility
Malogajski, J., Jansen, M. E., Ouburg, S., Ambrosino, E., Terwee, C. B. & Morre, S. A. Jun 2017 In : Sexual & Reproductive Healthcare. 12, p. 123-127 5 p.
Research output: Scientific - peer-review › Article

Inhibition of sarcolemmal FAT/CD36 by sulfo-N-succinimidyl oleate rapidly corrects metabolism and restores function in the diabetic heart following hypoxia/reoxygenation
Mansor, L. S., Fialho, M. D. L. S., Yea, G., Coumans, W. A., West, J. A., Kerr, M., Carr, C. A., Luiken, J. J. F. P., Glatz, J. F. C., Evans, R. D., Griffin, J. L., Tyler, D. J., Clarke, K. & Heather, L. C. 1 Jun 2017 In : Cardiovascular Research. 113, 7, p. 737-748 12 p.
Research output: Scientific - peer-review › Article

B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies
Maroofian, R., Riemersma, M., Jae, L. T., Zhianabed, N., Willemsen, M. H., Wissink-Lindhout, W. M., Willemsen, M. A., de Brouwer, A. P. M., Mehrjardi, M. Y. V., Ashrafi, M. R., Kusters, B., Kleefstra, T., Jamshidi, Y., Nasseri, M., Pfundt, R., Brummelkamp, T. R., Abbaszadegan, M. R., Lefeber, D. J. & van Bokhoven, H. 22 Dec 2017 In : Genome Medicine. 9, 11 p., 118
Research output: Scientific - peer-review › Article

Cervical Carcinogenesis and Immune Response Gene Polymorphisms: A Review
Mehta, A. M., Mooij, M., Brankovic, I., Ouburg, S., Morre, S. A. & Jordanova, E. S. 2017 In : Journal of Immunology Research. 2017, 12 p., 8913860
Research output: Scientific - peer-review › Review article

MicroRNAs in glaucoma and neurodegenerative diseases
Molasy, M., Walczak, A., Szaflik, J., Szaflik, J. P. & Majsterek, I. Jan 2017 In : Journal of Human Genetics. 62, 1, p. 105-112 8 p.
Research output: Scientific - peer-review › Review article

A new leptin-mediated mechanism for stimulating fatty acid oxidation: a pivotal role for sarcolemmal FAT/CD36
Momken, I., Chabowski, A., Dirkx, E., Nabben, M., Jain, S. S., McFarlan, J. T., Glatz, J. F. C., Luiken, J. J. F. P. & Bonen, A. 1 Jan 2017 In : Biochemical Journal. 474, 1, p. 149-162 14 p.
Research output: Scientific - peer-review › Article

Dietary nitrate does not reduce oxygen cost of exercise or improve muscle mitochondrial function in patients with mitochondrial myopathy
Nabben, M., Schmitz, J. P. J., Ciapaite, J., le Clercq, C. M. P., van Riel, N. A., Haak, H. R., Nicolay, K., de Coo, I. F. M., Smeets, H., Praet, S. F., van Loon, L. J. & Prompers, J. J. 1 May 2017 In : American Journal of Physiology-regulatory Integrative and Comparative Physiology. 312, 5, p. R689-R701 13 p.
Research output: Scientific - peer-review › Article

Duplicated Enhancer Region Increases Expression of CTSB and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families
Ngcungcu, T. , Oti, M. , Sitek, J. C. , Haukanes, B. I. , Linghu, B. , Bruccoleri, R. , Stokowy, T. , Oakeley, E. J. , Yang, F. , Zhu, J. , Sultan, M. , Schalkwijk, J. , van Vlijmen-Willems, I. M. J. J. , von der Lippe, C. , Brunner, H. G. , Ersland, K. M. , Grayson, W. , Buechmann-Moller, S. , Sundnes, O. , Nirmala, N. & 9 others Morgan, T. M., van Bokhoven, H., Steen, V. M., Hull, P. R., Szustakowski, J., Staedtler, F., Zhou, H., Fiskerstrand, T. & Ramsay, M. 4 May 2017 In : American Journal of Human Genetics. 100, 5, p. 737-750 14 p.
Research output: Scientific - peer-review › Article

Novel pathogenic SLC25A46 splice-site mutation causes an optic atrophy spectrum disorder
Nguyen, M., Boesten, I., Hellebrekers, D. M. E. I., Mulder-den Hartog, N. M., de Coo, I. F. M., Smeets, H. J. M. & Gerards, M. Jan 2017 In : Clinical Genetics. 91, 1, p. 121-125 5 p.
Research output: Scientific - peer-review › Article

Incontinence and psychological symptoms in individuals with Mowat-Wilson Syndrome
Niemczyk, J., Einfeld, S., Mowat, D., Equit, M., Wagner, C., Curfs, L. & von Gontard, A. Mar 2017 In : Research in Developmental Disabilities. 62, p. 230-237 8 p.
Research output: Scientific - peer-review › Article

Incontinence in persons with Down Syndrome
Niemczyk, J., von Gontard, A., Equit, M., Medoff, D., Wagner, C. & Curfs, L. Aug 2017 In : Neurourology and Urodynamics. 36, 6, p. 1550-1556 7 p.
Research output: Scientific - peer-review › Article

Vitamin D insufficient levels during pregnancy and micronuclei frequency in peripheral blood T lymphocytes mothers and newborns (Rhea cohort, Crete)
O'Callaghan-Gordo, C., Kogevinas, M., Fthenou, E., Pedersen, M., Espinosa, A., Chalkiadaki, G., Daraki, V., Dermitzaki, E., Decordier, I., Georgiou, V., Merlo, D. F., Roumeliotaki, T., Loock, K. V., Kleinjans, J., Kirsch-Volders, M. & Chatzi, L. Aug 2017 In : Clinical Nutrition. 36, 4, p. 1029-1035 7 p.
Research output: Scientific - peer-review › Article

Folic acid reduces doxorubicin-induced cardiomyopathy by modulating endothelial nitric oxide synthase
Octavia, Y., Kararigas, G., de Boer, M., Chrifi, I., Kietadisorn, R., Swinnen, M., Duimel, H., Verheyen, F. K., Brandt, M. M., Fliegner, D., Cheng, C., Janssens, S., Duncker, D. J. & Moens, A. L. Dec 2017 In : Journal of Cellular and Molecular Medicine. 21, 12, p. 3277-3287 11 p.
Research output: Scientific - peer-review › Article

Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia
Oud, M. S., Ramos, L., O'Bryan, M. K., McLachlan, R. I., Okutman, O., Viville, S., de Vries, P. F., Smeets, D. F. C. M., Lugtenberg, D., Hehir-Kwa, J. Y., Gilissen, C., van de Vorst, M., Vissers, L. E. L. M., Hoischen, A., Meijerink, A. M., Fleischer, K., Veltman, J. A. & Noordam, M. J. Nov 2017 In : Human Mutation. 38, 11, p. 1592-1605 14 p.
Research output: Scientific - peer-review › Article

Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome
Oud, M. M. , Tuijnenburg, P. , Hempel, M. , van Vlies, N. , Ren, Z. , Ferdinandusse, S. , Jansen, M. H. , Santer, R. , Johannsen, J. , Bacchelli, C. , Alders, M. , Li, R. , Davies, R. , Dupuis, L. , Cale, C. M. , Wanders, R. J. A. , Pals, S. T. , Ocaka, L. , James, C. , Mueller, I. & 18 others Lehmberg, K., Strom, T., Engels, H., Williams, H. J., Beales, P., Roepmand, R., Dias, P., Brunner, H. G., Cobben, J-M., Hall, C., Hartley, T., Stabej, P. L. Q., Mendoza-Londono, R., Davies, E. G., de Sousa, S. B., Lesse, D., Arts, H. H. & Kuijpers, T. W. 2 Feb 2017 In : American Journal of Human Genetics. 100, 2, p. 281-296 16 p.
Research output: Scientific - peer-review › Article

Silencing of Anticoagulant Protein C Evokes Low-Incident but Spontaneous Atherothrombosis in Apolipoprotein E-Deficient Mice-Brief Report
Ouweneel, A. B., Heestermans, M., Verwilligen, R. A. F., Gijbels, M. J. J., Reitsma, P. H., Van Eck, M. & van Vlijmen, B. J. M. May 2017 In : Arteriosclerosis Thrombosis and Vascular Biology. 37, 5, p. 782-785 4 p.
Research output: Scientific - peer-review › Article

NGS panel analysis in 24 ectopia lentis patients; a clinically relevant test with a high diagnostic yield
Overwater, E. , Floor, K. , van Beek, D. , de Boer, K. , van Dijk, T. , Hilhorst-Hofstee, Y. , Hoogeboom, A. J. M. , van Kaam, K. J. , van de Kamp, J. M. , Kempers, M. , Krapels, I. P. C. , Kroes, H. Y. , Loeys, B. , Salemink, S. , Stumpel, C. T. R. M. , Verhoeven, V. J. M. , Wijnands-van den Berg, E. , Cobben, J. M. , van Tintelen, J. P. , Weiss, M. M. & 2 others Houweling, A. C. & Maugeri, A. Sep 2017 In : European Journal of Medical Genetics. 60, 9, p. 465-473 9 p.
Research output: Scientific - peer-review › Article

Associations of cord blood metabolites with perinatal characteristics, newborn anthropometry, and cord blood hormones in project viva
Perng, W., Rifas-Shiman, S. L., McCulloc, S., Chatzi, L., Mantzoros, C., Hivert, M-F. & Oken, E. Nov 2017 In : Metabolism-Clinical and Experimental. 76, p. 11-22 12 p.
Research output: Scientific - peer-review › Article

Adventitial lymphatic capillary expansion impacts on plaque T cell accumulation in atherosclerosis
Rademakers, T., van der Vorst, E. P. C., Daissormont, I. T. M. N., Otten, J. J. T., Theodorou, K., Theelen, T. L., Gijbels, M., Anisimov, A., Nurmi, H., Lindeman, J. H. N., Schober, A., Heeneman, S., Alitalo, K. & Biessen, E. A. L. 28 Mar 2017 In : Scientific Reports. 7, 10 p., 45263
Research output: Scientific - peer-review › Article

The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies
Redin, C. , Brand, H. , Collins, R. L. , Kammin, T. , Mitchell, E. , Hodge, J. C. , Hanscom, C. , Pillalamarri, V. , Seabra, C. M. , Abbott, M-A. , Abdul-Rahman, O. A. , Aberg, E. , Adley, R. , Alcaraz-Estrada, S. L. , Alkuraya, F. S. , An, Y. , Anderson, M-A. , Antolik, C. , Anyane-Yeboa, K. , Atkin, J. F. & 129 others Bartell, T., Bernstein, J. A., Beyer, E., Blumenthal, I., Bongers, E. M. H. F., Brilstra, E. H., Brown, C. W., Bruggenwirth, H. T., Callewaert, B., Chiang, C., Corning, K., Cox, H., Cuppen, E., Currall, B. B., Cushing, T., David, D., Deardorff, M. A., Dheedene, A., D'Hooghe, M., de Vries, B. B. A., Earl, D. L., Ferguson, H. L., Fisher, H., FitzPatrick, D. R., Gerrol, P., Giachino, D., Glessner, J. T., Gliem, T., Grady, M., Graham, B. H., Griffis, C., Gripp, K. W., Gropman, A. L., Hanson-Kahn, A., Harris, D. J., Hayden, M. A., Hill, R., Hochstenbach, R., Hoffman, J. D., Hopkin, R. J., Hubshman, M. W., Innes, A. M., Irons, M., Irving, M., Jacobsen, J. C., Janssens, S., Jewett, T., Johnson, J. P., Jongmans, M. C., Kahler, S. G., Koolen, D. A., Korzelius, J., Kroisel, P. M., Lacassie, Y., Lawless, W., Lemyre, E., Leppig, K., Levin, A. V., Li, H., Li, H., Liao, E. C., Lim, C., Lose, E. J., Lucente, D., Macera, M. J., Manavalan, P., Mandrile, G., Marcelis, C. L., Margolin, L., Mason, T., Masser-Frye, D., McClellan, M. W., Mendoza, C. J. Z., Menten, B., Middelkamp, S., Mikami, L. R., Moe, E., Mohammed, S., Mononen, T., Mortenson, M. E., Moya, G., Nieuwint, A. W., Ordulu, Z., Parkash, S., Pauker, S. P., Pereira, S., Perrin, D., Phelan, K., Pina Aguilar, R. E., Poddighe, P. J., Pregno, G., Raskin, S., Reis, L., Rhead, W., Rita, D., Renkens, I., Roelens, F., Ruliera, J., Rump, P., Schilit, S. L. P., Shaheen, R., Sparkes, R., Spiegel, E., Stevens, B., Stone, M. R., Tagoe, J., Thakuria, J. V., van Bon, B. W., van de Kamp, J., van Der Burgt, I., van Essen, T., van Ravenswaaij-Arts, C. M., van Roosmalen, M. J., Vergult, S., Volker-Touw, C. M. L., Warburton, D. P., Waterman, M. J., Wiley, S., Wilson, A., Yerena-de Vega, M. D. L. C. A., Zori, R. T., Levy, B., Brunner, H. G., de Leeuw, N., Kloosterman, W. P., Thorland, E. C., Morton, C. C., Gusella, J. F. & Talkowski, M. E. Jan 2017 In : Nature Genetics. 49, 1, p. 36-45 10 p.
Research output: Scientific - peer-review › Article

Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability
Reijnders, M. R. F. , Kousi, M. , van Woerden, G. M. , Klein, M. , Bralten, J. , Mancini, G. M. S. , van Essen, T. , Proietti-Onori, M. , Smeets, E. E. J. , van Gastel, M. , Stegmann, A. P. A. , Stevens, S. J. C. , Lelieveld, S. H. , Gilissen, C. , Pfundt, R. , Tan, P. L. , Kleefstra, T. , Franke, B. , Elgersma, Y. , Katsanis, N. & 1 others Brunner, H. G. 20 Oct 2017 In : Nature Communications. 8, 12 p., 1052
Research output: Scientific - peer-review › Article

RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes
Reijnders, M. R. F. , Ansor, N. M. , Kousi, M. , Yue, W. W. , Tan, P. L. , Clarkson, K. , Clayton-Smith, J. , Corning, K. , Jones, J. R. , Lam, W. W. K. , Mancini, G. M. S. , Marcelis, C. , Mohammed, S. , Pfundt, R. , Roifman, M. , Cohn, R. , Chitayat, D. , Millard, T. H. , Katsanis, N. , Brunner, H. G. & 2 others Banka, S. & Deciphering Dev Disorders Study 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 466-477 12 p.
Research output: Scientific - peer-review › Article

Low-Dose Lipopolysaccharide Causes Biliary Injury by Blood Biliary Barrier Impairment in a Rat Hepatic Ischemia/Reperfusion Model
Reiling, J., Bridle, K. R., Gijbels, M., Schaap, F. G., Jaskowski, L., Santrampurwala, N., Britton, L. J., Campbell, C. M., Damink, S. W. M. O., Crawford, D. H. G., Dejong, C. H. C. & Fawcett, J. Feb 2017 In : Liver Transplantation. 23, 2, p. 194-206 13 p.
Research output: Scientific - peer-review › Article

Physical activity and risk of prostate and bladder cancer in China: The South and East China case-control study on prostate and bladder cancer
Reulen, R. C., de Vogel, S., Zhong, W., Zhong, Z., Xie, L-P., Hu, Z., Deng, Y., Yang, K., Liang, Y., Zeng, X., Wong, Y. C., Tam, P-C., Hemelt, M. & Zeegers, M. P. 2 Jun 2017 In : PLoS One. 12, 6, 11 p., e0178613
Research output: Scientific - peer-review › Article

Propionic acidemia as a cause of adult-onset dilated cardiomyopathy
Riemersma, M., Hazebroek, M. R., Helderman-van den Enden, A. T. J. M., Salomons, G. S., Ferdinandusse, S., Brouwers, M. C. G. J., van der Ploeg, L., Heymans, S., Glatz, J. F. C., van den Wijngaard, A., Krapels, I. P. C., Bierau, J. & Brunner, H. G. Nov 2017 In : European Journal of Human Genetics. 25, 11, p. 1195-1201 7 p.
Research output: Scientific - peer-review › Article

Small heterodimer partner (SHP) contributes to insulin resistance in cardiomyocytes
Rodriguez-Calvo, R., Chanda, D., Oligschlaeger, Y., Miglianico, M., Coumans, W. A., Barroso, E., Tajes, M., Luiken, J. J. F. P., Glatz, J. F. C., Vazquez-Carrera, M. & Neumann, D. May 2017 In : Biochimica et Biophysica Acta-Molecular and Cell Biology of Lipids. 1862, 5, p. 541-551 11 p.
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
Sallevelt, S. C. E. H., de Koning, B., Szklarczyk, R., Paulussen, A. D. C., de Die-Smulders, C. E. M. & Smeets, H. J. M. May 2017 In : Genetics in Medicine. 19, 5, p. 583-592 10 p.
Research output: Scientific - peer-review › Article

De novo mtDNA point mutations are common and have a low recurrence risk
Sallevelt, S. C. E. H., de Die-Smulders, C. E. M., Hendrickx, A. T. M., Hellebrekers, D. M. E. I., de Coo, I. F. M., Alston, C. L., Knowles, C., Taylor, R. W., McFarland, R. & Smeets, H. J. M. Feb 2017 In : Journal of Medical Genetics. 54, 2, p. 114-124 11 p.
Research output: Scientific - peer-review › Article

PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Druesedau, M., Hellebrekers, D. M. E. I., Paulussen, A. D. C., Coonen, E., Van Golde, R. J. T., Geraedts, J. P. M., Gianaroli, L., Magli, M. C., Zeviani, M., Smeets, H. J. M. & de Die-Smulders, C. E. M. Mar 2017 In : Human Reproduction. 32, 3, p. 698-703 6 p.
Research output: Scientific - peer-review › Article

Growth Hormone Therapy in Children with Kabuki Syndrome: 1-year Treatment Results
Schott, D. A., Gerver, W. J. M. & Stumpel, C. T. R. M. 2017 In : Hormone Research in Paediatrics. 88, 3-4, p. 1-7 7 p.
Research output: Scientific - peer-review › Article

Computer-assisted three-dimensional tracking of sensory innervation in the murine bladder mucosa with two-photon microscopy
Schueth, A., Spronck, B., van Zandvoort, M. A. M. J. & van Koeveringe, G. A. Nov 2017 In : Journal of Chemical Neuroanatomy. 85, p. 43-49 7 p.
Research output: Scientific - peer-review › Article

The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands

Seco, C. Z. , Wesdorp, M. , Feenstra, I. , Pfundt, R. , Hehir-Kwa, J. Y. , Lelieveld, S. H. , Castelein, S. , Gilissen, C. , de Wijs, I. J. , Admiraal, R. J. C. , Pennings, R. J. E. , Kunst, H. P. M. , van de Kamp, J. M. , Tamminga, S. , Houweling, A. C. , Plomp, A. S. , Maas, S. M. , Gans, P. A. M. D. K. , Kant, S. G. , de Geus, C. M. & 10 others Frints, S. G. M., Vanhoutte, E. K., van Dooren, M. F., van den Boogaard, M-J. H., Scheffer, H., Nelen, M., Kremer, H., Hoefsloot, L., Schraders, M. & Yntema, H. G. Feb 2017 In : European Journal of Human Genetics. 25, 3, p. 308-314 7 p.

Research output: Scientific - peer-review › Article

A homozygous FITM2 mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy

Seco, C. Z. , Castells-Nobau, A. , Joo, S. , Schraders, M. , Foo, J. N. , van der Voet, M. , Velan, S. S. , Nijhof, B. , Oostrik, J. , de Vrieze, E. , Katana, R. , Mansoor, A. , Huynen, M. , Szklarczyk, R. , Oti, M. , Tranebjaerg, L. , van Wijk, E. , Scheffer-de Gooyert, J. M. , Siddique, S. , Baets, J. & 10 others de Jonghe, P., Kazmi, S. A. R., Sadananthan, S. A., van de Warrenburg, B. P., Khor, C. C., Goepfert, M. C., Qamar, R., Schenck, A., Kremer, H. & Siddiqi, S. 1 Feb 2017 In : Disease Models & Mechanisms. 10, 2, p. 105-118 14 p.

Research output: Scientific - peer-review › Article

Prevalence of genital Chlamydia trachomatis infections in Russia: systematic literature review and multicenter study

Smelov, V., Thomas, P., Ouburg, S. & Morre, S. A. Oct 2017 In : Pathogens and Disease. 75, 7, 9 p., 081

Research output: Scientific - peer-review › Review article

Chlamydia trachomatis Strain Types Have Diversified Regionally and Globally with Evidence for Recombination across Geographic Divides

Smelov, V., Vrbanac, A., van Ess, E. F., Noz, M. P., Wan, R., Eklund, C., Morgan, T., Shrier, L. A., Sanders, B., Dillner, J., de Vries, H. J. C., Morre, S. A. & Dean, D. 13 Nov 2017 In : Frontiers in microbiology. 8, 14 p., 2195

Research output: Scientific - peer-review › Article

Sexuality and individual support plans for people with intellectual disabilities

Stoffelen, J. M. T., Herps, M. A., Buntinx, W. H. E., Schaafsma, D., Kok, G. & Curfs, L. M. G. 11 Oct 2017 In : Journal of Intellectual Disability Research. 61, 12, p. 1117-1129

Research output: Scientific - peer-review › Article

Polyunsaturated fatty acid levels at birth and child-to-adult growth: Results from the MEFAB cohort

Stratakis, N., Gielen, M., Margetaki, K., Godschalk, R. W., van der Wurff, I. S. M., Rouschop, S., Ibrahim, A., Antoniou, E., Chatzi, L., de Groot, R. H. M. & Zeegers, M. P. 2017 In : Prostaglandins Leukotrienes and Essential Fatty Acids. 126, p. 72-78 7 p.

Research output: Scientific - peer-review › Article

Fish and seafood consumption during pregnancy and the risk of asthma and allergic rhinitis in childhood: a pooled analysis of 18 European and US birth cohorts
Stratakis, N. , Roumeliotaki, T. , Oken, E. , Ballester, F. , Barros, H. , Basterrechea, M. , Cordier, S. , de Groot, R. , den Dekker, H. T. , Duijts, L. , Eggesbo, M. , Fantini, M. P. , Forastiere, F. , Gehring, U. , Gielen, M. , Gori, D. , Govarts, E. , Inskip, H. M. , Iszatt, N. , Jansen, M. & 22 others Kelleher, C., Mehegan, J., Molto-Puigmarti, C., Mommers, M., Oliveira, A., Olsen, S. F., Pele, F., Pizzi, C., Porta, D., Richiardi, L., Rifas-Shiman, S. L., Robinson, S. M., Schoeters, G., Strom, M., Sunyer, J., Thijs, C., Vrijheid, M., Vrijkotte, T. G. M., Wijga, A. H., Kogevinas, M., Zeegers, M. P. & Chatzi, L. Oct 2017 In : International Journal of Epidemiology. 46, 5, p. 1465-1477 13 p.
Research output: Scientific - peer-review › Article

Limited additive value of the Ki-67 proliferative index on patient survival in World Health Organization-classified pulmonary carcinoids
Swarts, D. R. A., Rudelius, M., Claessen, S. M. H., Cleutjens, J. P., Seidl, S., Volante, M., Ramaekers, F. C. S. & Speel, E. J. M. Feb 2017 In : Histopathology. 70, 3, p. 412-422 11 p.
Research output: Scientific - peer-review › Article

Early screening for Chlamydia trachomatis in young women for primary prevention of pelvic inflammatory disease (i-Predict): study protocol for a randomised controlled trial
Tamarelle, J., Thiebaut, A. C. M., Sabin, B., Bebear, C., Judlin, P., Fauconnier, A., Rahib, D., Meaude-Roufai, L., Ravel, J., Morre, S. A., de Barbeyrac, B., Delarocque-Astagneau, E. & I-Predict Study Grp 13 Nov 2017 In : Trials. 18, 11 p., 534
Research output: Scientific - peer-review › Article

Cellular strain avoidance is mediated by a functional actin cap - observations in an Lmna-deficient cell model
Tamiello, C., Halder, M., Kamps, M. A. F., Baaijens, F. P. T., Broers, J. L. V. & Bouten, C. V. C. 15 Feb 2017 In : Journal of Cell Science. 130, 4, p. 779-790 12 p.
Research output: Scientific - peer-review › Article

Whole body and hematopoietic ADAM8 deficiency does not influence advanced atherosclerotic lesion development, despite its association with human plaque progression
Theodorou, K., van der Vorst, E. P. C., Gijbels, M. J., Wolfs, I. M. J., Jeurissen, M., Theelen, T. L., Sluimer, J. C., Wijnands, E., Cleutjens, J. P., Li, Y., Jansen, Y., Weber, C., Ludwig, A., Bentzon, J. F., Bartsch, J. W., Biessen, E. A. L. & Donners, M. M. P. C. 15 Sep 2017 In : Scientific Reports. 7, 11 p., 11670
Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

Burden of Chlamydia trachomatis in India: a systematic literature review
Thomas, P., Spaargaren, J., Kant, R., Lawrence, R., Dayal, A., Lal, J. A. & Morre, S. A. Jul 2017 In : Pathogens and Disease. 75, 5, 10 p., 055
Research output: Scientific - peer-review › Review article

NDRG4, an early detection marker for colorectal cancer, is specifically expressed in enteric neurons
Vaes, N., Lentjes, M. H. F. M., Gijbels, M. J., Rademakers, G., Daenen, K. L., Boesmans, W., Wouters, K. A. D., Geuzens, A., Qu, X., Steinbusch, H. P. J., Rutten, B. P. F., Baldwin, S. H., Sharkey, K. A., Hofstra, R. M. W., van Engeland, M., Vanden Berghe, P. & Melotte, V. Sep 2017 In : Neurogastroenterology and Motility. 29, 9, 10 p., 13095
Research output: Scientific - peer-review › Article

Impact of tissue adhesives on the prevention of anastomotic leakage of colonic anastomoses: an in vivo study
Vakalopoulos, K. A., Bosmans, J. W. A. M., van Barneveld, K. W. Y., Vogels, R. R. M., Boersema, G. S. A., Wu, Z., Gijbels, M. J. J., Jeekel, J., Kleinrensink, G., Bouvy, N. D. & Lange, J. F. Jul 2017 In : International Journal of Colorectal Disease. 32, 7, p. 961-965 5 p.
Research output: Scientific - peer-review › Article

A 20-Year Temporal Change Analysis in Incidence, Presenting Phenotype and Mortality, in the Dutch IBDSL Cohort-Can Diagnostic Factors Explain the Increase in IBD Incidence?
van den Heuvel, T. R. A., Jeuring, S. F. G., Zeegers, M. P., van Dongen, D. H. E., Wolters, A., Masclee, A. A. M., Hameeteman, W. H., Romberg-Camps, M. J. L., Oostenbrug, L. E., Pierik, M. J. & Jonkers, D. M. Oct 2017 In : Journal of Crohns & Colitis. 11, 10, p. 1169-1179 11 p.
Research output: Scientific - peer-review › Article

Cohort Profile: The Inflammatory Bowel Disease South Limburg Cohort (IBDSL)
van den Heuvel, T. R. A., Jonkers, D. M., Jeuring, S. F. G., Romberg-Camps, M. J. L., Oostenbrug, L. E., Zeegers, M. P., Masclee, A. A. & Pierik, M. J. Apr 2017 In : International Journal of Epidemiology. 46, 2, p. e7 9 p.
Research output: Scientific - peer-review › Article

High-Density Lipoproteins Exert Pro-inflammatory Effects on Macrophages via Passive Cholesterol Depletion and PKC-NF-kappa B/STAT1-IRF1 Signaling
van der Vorst, E. P. C. , Theodorou, K. , Wu, Y. , Hoeksema, M. A. , Goossens, P. , Bursill, C. A. , Aliyev, T. , Huitema, L. F. A. , Tas, S. W. , Wolfs, I. M. J. , Kuijpers, M. J. E. , Gijbels, M. J. , Schalkwijk, C. G. , Koonen, D. P. Y. , Abdollahi-Roodsaz, S. , McDaniels, K. , Wang, C-C. , Leitges, M. , Lawrence, T. , Plat, J. & 6 others Van Eck, M., Rye, K-A., Touqui, L., de Winther, M. P. J., Biessen, E. A. L. & Donners, M. M. P. C. 10 Jan 2017 In : Cell Metabolism. 25, 1, p. 197-207 11 p.
Research output: Scientific - peer-review › Article

A Review of Recruitment, Adherence and Drop-Out Rates in Omega-3 Polyunsaturated Fatty Acid Supplementation Trials in Children and Adolescents
van der Wurff, I. S. M., Meyer, B. J. & de Groot, R. H. M. May 2017 In : Nutrients. 9, 5, 32 p., 474
Research output: Scientific - peer-review › Review article

Performance of the multitarget Mikrogen Chlamydia trachomatis IgG ELISA in the prediction of tubal factor infertility (TFI) in subfertile women: comparison with the Medac MOMP IgG ELISA plus
van Ess, E. F., Ouburg, S., Spaargaren, J., Land, J. A. & Morre, S. A. Oct 2017 In : Pathogens and Disease. 75, 7, 7 p., 067
Research output: Scientific - peer-review › Article

Lmna knockout mouse embryonic fibroblasts are less contractile than their wild-type counterparts
van Loosdregt, I. A. E. W., Kamps, M. A. F., Oomens, C. W. J., Loerakker, S., Broers, J. L. V. & Bouten, C. V. C. 1 Aug 2017 In : Integrative Biology. 9, 8, p. 709-721 13 p.
Research output: Scientific - peer-review › Article

A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology
Vissers, L. E. L. M., van Nimwegen, K. J. M., Schieving, J. H., Kamsteeg, E-J., Kleefstra, T., Yntema, H. G., Pfundt, R., van der Wilt, G. J., Krabbenborg, L., Brunner, H. G., van der Burg, S., Grutters, J., Veltman, J. A. & Willemsen, M. A. A. P. Sep 2017 In : Genetics in Medicine. 19, 9, p. 1055-1063 9 p.
Research output: Scientific - peer-review › Article

Quantification of Phenotype Information Aids the Identification of Novel Disease Genes
Vulto-van Silfhout, A. T., Gilissen, C., Goeman, J. J., Jansen, S., van Amen-Hellebrekers, C. J. M., van Bon, B. W. M., Koolen, D. A., Sistermans, E. A., Brunner, H. G., de Brouwer, A. P. M. & de Vries, B. B. A. May 2017 In : Human Mutation. 38, 5, p. 594-599 6 p.
Research output: Scientific - peer-review › Article

Do-Not-Attempt-Resuscitation orders for people with intellectual disabilities: dilemmas and uncertainties for ID physicians and trainees. The importance of the deliberation process
Wagemans, A. M. A., Lantman-de Valk, H. M. J. V. S., Proot, I. M., Bressers, A. M., Metsemakers, J., Tuffrey-Wijne, I., Groot, M. & Curfs, L. M. G. Mar 2017 In : Journal of Intellectual Disability Research. 61, 3, p. 245-254 10 p.
Research output: Scientific - peer-review › Article

Incontinence in persons with Angelman syndrome
Wagner, C., Niemczyk, J., Equit, M., Curfs, L. & von Gontard, A. Feb 2017 In : European Journal of Pediatrics. 176, 2, p. 225-232 8 p.
Research output: Scientific - peer-review › Article

Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas
Weren, R. D. A., Mensenkamp, A. R., Simons, M., Eijkelenboom, A., Sie, A. S., Ouchene, H., van Asseldonk, M., Gomez-Garcia, E. B., Blok, M. J., de Hullu, J. A., Nelen, M. R., Hoischen, A., Bulten, J., Tops, B. B. J., Hoogerbrugge, N. & Ligtenberg, M. J. L. Feb 2017 In : Human Mutation. 38, 2, p. 226-235 10 p.
Research output: Scientific - peer-review › Article

Multi-photon microscopy in cardiovascular research
Wu, Z., Rademakers, T., Kiessling, F., Vogt, M., Westein, E., Weber, C., Megens, R. T. A. & van Zandvoort, M. 1 Nov 2017 In : Methods. 130, p. 79-89 11 p.
Research output: Scientific - peer-review › Article

Mutations in the Chromatin Regulator Gene *BRPF1* Cause Syndromic Intellectual Disability and Deficient Histone Acetylation
Yan, K. , Rousseau, J. , Littlejohn, R. O. , Kiss, C. , Lehman, A. , Rosenfeld, J. A. , Stumpel, C. T. R. , Stegmann, A. P. A. , Robak, L. , Scaglia, F. , Thi Tuyet Mai Nguyen, , Fu, H. , Ajeawung, N. F. , Camurri, M. V. , Li, L. , Gardham, A. , Panis, B. , Almannai, M. , Sacoto, M. J. G. , Baskin, B. & 16 others Ruivenkamp, C., Xia, F., Bi, W., Cho, M. T., Potjer, T. P., Santen, G. W. E., Parker, M. J., Canham, N., McKinnon, M., Potocki, L., MacKenzie, J. J., Roeder, E. R., Campeau, P. M., Yang, X-J., DDD Study & CAUSES Study 5 Jan 2017 In : American Journal of Human Genetics. 100, 1, p. 91-104 14 p.
Research output: Scientific - peer-review › Article

Nilotinib Enhances Tumor Angiogenesis and Counteracts VEGFR2 Blockade in an Orthotopic Breast Cancer Xenograft Model with Desmoplastic Response
Zafarnia, S., Bzyl-Ibach, J., Spivak, I., Li, Y., Koletnik, S., Doleschel, D., Rix, A., Pochon, S., Tardy, I., Koyadan, S., van Zandvoort, M., Palmowski, M., Kiessling, F. & Lederle, W. Nov 2017 In : Neoplasia (Online). 19, 11, p. 896-907 12 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

The use of lumbar epidural injection of platelet lysate for treatment of radicular pain
Centeno, C., Markle, J., Dodson, E., Stemper, I., Hyzy, M., Williams, C. & Freeman, M. 25 Nov 2017 In : Journal of Experimental Orthopaedics. 4, 11 p., 38
Research output: Scientific - peer-review › Article

Selective citation in the literature on swimming in chlorinated water and childhood asthma: a network analysis
Duyx, B., Urlings, M. J. E., Swaen, G. M. H., Bouter, L. M. & Zeegers, M. P. 2017 In : Research integrity and peer review. 2, p. 17
Research output: Scientific - peer-review › Article

Homozygosity Mapping and Targeted Sanger Sequencing Identifies Three Novel CRB1 (Crumbs homologue 1) Mutations in Iranian Retinal Degeneration Families
Ghofrani, M., Yahyaei, M., Brunner, H. G., Cremers, F. P. M., Movasat, M., Imran Khan, M. & Keramatipour, M. Sep 2017 In : Iranian Biomedical Journal. 21, 5, p. 294-302 9 p.
Research output: Scientific - peer-review › Article

Policy Making in Newborn Screening Needs a Structured and Transparent Approach
Jansen, M. E., Lister, K. J., van Kranen, H. J. & Cornel, M. C. 21 Mar 2017 In : Frontiers in Public Health. 5, 6 p., 53
Research output: Scientific - peer-review › Review article

A novel mutation in L1CAM causes a mild form of L1 syndrome: a case report
Otter, M., Wevers, M., Pisters, M., Pfundt, R., Vos, Y., Nievelstein, R. J. & Stumpel, C. Aug 2017 In : Clinical Case Reports. 5, 8, p. 1213-1217 5 p.
Research output: Scientific - peer-review › Article

PRC1 Prevents Replication Stress during Chondrogenic Transit Amplification
Spaapen, F., Eijssen, L., Adriaens, M., Welting, T., Prickaerts, P., Salvaing, J., Dahlmans, V. E. H., Surtel, D., Kruitz, F., Kuijer, R., Takihara, Y., Marks, H., Stunnenberg, H., Wouters, B., Vidal, M. & Voncken, J. Dec 2017 In : Epigenomes. 1, 3, 31 p., 22
Research output: Scientific - peer-review › Article

Drug Overdose Surveillance and Information Sharing Via a Public Database: The Role of the Medical Examiner/Coroner
Williams, K. E. & Freeman, M. Mar 2017 In : Academic Forensic Pathology. 7, 1, p. 60-72
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 A: Editorial Material in wi-1 journal

The annual meeting 1988-2017
Brunner, H. Dec 2017 In : European Journal of Human Genetics. 25, p. S35-S36 2 p.
Research output: Scientific - peer-review › Editorial

Cohort Profile: The Mother-Child Cohort in Crete, Greece (Rhea Study)
Chatzi, L., Leventakou, V., Vafeiadi, M., Koutra, K., Roumeliotaki, T., Chalkiadaki, G., Karachaliou, M., Daraki, V., Kyriklaki, A., Kampouri, M., Fthenou, E., Sarri, K., Vassilaki, M., Fasoulaki, M., Bitsios, P., Koutis, A., Stephanou, E. G. & Kogevinas, M. Oct 2017 In : International Journal of Epidemiology. 46, 5, p. 1392-1393k 13 p.
Research output: Scientific - peer-review › Editorial

Healthy children without fear: Reproductive options for patients or couples carrying inherited diseases
Geraedts, J. May 2017 In : Embo Reports. 18, 5, p. 666-669 4 p.
Research output: Scientific - peer-review › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Immediate and prolonged-release melatonin in children with neurodevelopmental disabilities. Author reply to Prof. Zisapel
Bruni, O., Alonso-Alconada, D., Besag, F., Biran, V., Braam, W., Cortese, S., Moavero, R., Parisi, P., Smits, M., Van der Heijden, K. & Curatolo, P. Mar 2017 In : European Journal of Paediatric Neurology. 21, 2, p. 420-421 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Thirty years of Dutch embryo selection
Geraedts, J. 30 Nov 2017 In : Nature. 551, 7682, p. 565-565 1 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Preliminary stop of the TOPical Imiquimod treatment of high-grade Cervical intraepithelial neoplasia (TOPIC) trial
Koeneman, M. M., Kruse, A. J., Kooreman, L. F. S., zur Hausen, A., Hopman, A. H. N., Sep, S. J. S., Van Gorp, T., Slangen, B. F. M., van Beekhuizen, H. J., van de Sande, A. J. M., Gerestein, C. G., Nijman, H. W. & Kruitwagen, R. F. P. M. 7 Feb 2017 In : BMC Cancer. 17, 2 p., 110
Research output: Scientific - peer-review › Comment/Letter to the editor

Concern regarding the alleged spread of hypervirulent lymphogranuloma venereum Chlamydia trachomatis strain in Europe
Seth-Smith, H. M., Galan, J. C., Goldenberger, D., Lewis, D. A., Peuchant, O., Bebear, C., de Barbeyrac, B., Benard, A., Carter, I., Kok, J., Bruisten, S. M., Versteeg, B., Morre, S. A., Thomson, N. R., Egli, A. & de Vries, H. J. 13 Apr 2017 In : Eurosurveillance. 22, 15, p. 1-2 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Re: Lifestyle and bladder cancer prevention: no consistent evidence from cohort studies
Wesselius, A. & Zeegers, M. Nov 2017 In : European Journal of Epidemiology. 32, 11, p. 1037-1038 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat06 Scientific national journal

Veranderingen in de behandeling van ziekte van Crohn*: Minder ziekenhuisopnames, operaties en gebruik van glucocorticoïden
Jeuring, S., van den Heuvel, T., Zeegers, M., Haans, J. J., Romberg-Camps, M. J. L., Oostenbrug, L. E., Stassen, L., Masclee, A., Jonkers, D. & Pierik, M. 2017 In : Nederlands Tijdschrift voor Geneeskunde. 161, D1641
Research output: Scientific - peer-review › Article

Cat07 Book(contribution)/congrespapers - Cat07 Book contribution (BB)

Optical Imaging
Alves, F., Bode, J., Cimalla, P., Hilger, I., Hofmann, M., Jaedicke, V., Koch, E., Licha, K., Rademakers, T., Razansky, D. & Van Zandvoort, M. A. M. J. 23 May 2017 *Small Animal Imaging: Basics and Practical Guide.* Kiessling, F., Pichler, B. J. & Hauff, P. (eds.). 2 ed. Springer International Publishing, p. 403-490 98 p.
Research output: Scientific › Chapter

Maternal fish intake during pregnancy and effects on the offspring
Chatzi, L. & Stratakis, N. Oct 2017 *Diet, Nutrition, and Fetal Programming.* Humana Press, p. 241-260
Research output: Scientific › Chapter

Development of the Uterine Cervix and Its Implications for the Pathogenesis of Cervical Cancer
Hopman, A. H. N. & Ramaekers, F. C. S. 10 Sep 2017 *Pathology of the Cervix.* Herrington, C. S. (ed.). 1 ed. Springer International Publishing, Vol. 3, p. 1-20 20 p.
Research output: Scientific › Chapter

Intravital imaging in small animals
Rademakers, T. & van Zandvoort, M. 2017 *Small Animal Imaging: Basics and Practical Guide (second edition).* Kiessling, F., Pichler, B. J. & hauff, P. (eds.). Springer Publishers, p. 403-490
Research output: Scientific › Chapter

Genetic Factors and Alcohol Consumption
Weijenberg, M., Kok, G., Roozen, S. & Curfs, L. 1 Mar 2017 *Fetal Alcohol Spectrum Disorder: A knowledge synthesis.* Maastricht: Datawyse / Universitaire Pers Maastricht, p. 30-32 3 p.
Research output: Scientific › Chapter

Institute for Public Health Genomics

Cat02 SCI/SSCI journal (WI-1)

Assessment of Chlamydia suis Infection in Pig Farmers
De Puysseleyr, L., De Puysseleyr, K., Braeckman, L., Morre, S. A., Cox, E. & Vanrompay, D. Jun 2017 In : Transboundary and Emerging Diseases. 64, 3, p. 826-833 8 p.
Research output: Scientific - peer-review › Article

Prevalence of Trichomonas vaginalis infection and protozoan load in South African women: a cross-sectional study
de Waaij, D. J., Dubbink, J. H., Ouburg, S., Peters, R. P. H. & Morre, S. A. Oct 2017 In : BMJ Open. 7, 10, 6 p., 016959
Research output: Scientific - peer-review › Article

Proposed guidelines to evaluate scientific validity and evidence for genotype-based dietary advice
Grimaldi, K. A. , van Ommen, B. , Ordovas, J. M. , Parnell, L. D. , Mathers, J. C. , Bendik, I. , Brennan, L. , Celis-Morales, C. , Cirillo, E. , Daniel, H. , de Kok, B. , El-Sohemy, A. , Fairweather-Tait, S. J. , Fallaize, R. , Fenech, M. , Ferguson, L. R. , Gibney, E. R. , Gibney, M. , Gjelstad, I. M. F. , Kaput, J. & 16 others Karlsen, A. S., Kolossa, S., Lovegrove, J., Macready, A. L., Marsaux, C. F. M., Martinez, J. A., Milagro, F., Navas-Carretero, S., Roche, H. M., Saris, W. H. M., Traczyk, I., van Kranen, H., Verschuren, L., Virgili, F., Weber, P. & Bouwman, J. 15 Dec 2017 In : Genes and nutrition. 12, 12 p., 35
Research output: Scientific - peer-review › Review article

Comprehensive global genome dynamics of Chlamydia trachomatis show ancient diversification followed by contemporary mixing and recent lineage expansion
Hadfield, J. , Harris, S. R. , Seth-Smith, H. M. B. , Parmar, S. , Andersson, P. , Giffard, P. M. , Schachter, J. , Moncada, J. , Ellison, L. , Gallo Vaulet, M. L. , Rodriguez Fermepin, M. , Radebe, F. , Mendoza, S. , Ouburg, S. , Morre, S. A. , Sachse, K. , Puolakkainen, M. , Korhonen, S. J. , Sonnex, C. , Wiggins, R. & 18 others Jalal, H., Brunelli, T., Casprini, P., Pitt, R., Ison, C., Savicheva, A., Shipitsyna, E., Hadad, R., Kari, L., Burton, M. J., Mabey, D., Solomon, A. W., Lewis, D., Marsh, P., Unemo, M., Clarke, I. N., Parkhill, J. & Thomson, N. R. Jul 2017 In : Genome Research. 27, 7, p. 1220-1229 10 p.
Research output: Scientific - peer-review › Article

The Netherlands Chlamydia cohort study (NECCST) protocol to assess the risk of late complications following Chlamydia trachomatis infection in women
Hoenderboom, B. M., van Oeffelen, A. A. M., van Benthem, B. H. B., van Bergen, J. E. A. M., Dukers-Muijrers, N. H. T. M., Gotz, H. M., Hoebe, C. J. P. A., Hogewoning, A. A., van der Klis, F. R. M., van Baarle, D., Land, J. A., van der Sande, M. A. B., van Veen, M. G., de Vries, F., Morre, S. A. & van den Broek, I. V. F. 11 Apr 2017 In : BMC Infectious Diseases. 17, 9 p., 264
Research output: Scientific - peer-review › Article

International differences in the evaluation of conditions for newborn bloodspot screening: a review of scientific literature and policy documents
Jansen, M. E., Metternick-Jones, S. C. & Lister, K. J. Jan 2017 In : European Journal of Human Genetics. 25, 1, p. 10-16 7 p.
Research output: Scientific - peer-review › Review article

Risk of eating disorders in a non-western setting: an exploratory study in Khartoum state, Sudan
Lau, C. C. L. & Ambrosino, E. Dec 2017 In : Eating and Weight Disorders-Studies on Anorexia Bulimia and Obesity. 22, 4, p. 649-656 8 p.
Research output: Scientific - peer-review › Article

The attitudes of Dutch fertility specialists towards the addition of genetic testing in screening of tubal factor infertility
Malogajski, J., Jansen, M. E., Ouburg, S., Ambrosino, E., Terwee, C. B. & Morre, S. A. Jun 2017 In : Sexual & Reproductive Healthcare. 12, p. 123-127 5 p.
Research output: Scientific - peer-review › Article

Cervical Carcinogenesis and Immune Response Gene Polymorphisms: A Review
Mehta, A. M., Mooij, M., Brankovic, I., Ouburg, S., Morre, S. A. & Jordanova, E. S. 2017 In : Journal of Immunology Research. 2017, 12 p., 8913860
Research output: Scientific - peer-review › Review article

Prevalence of genital Chlamydia trachomatis infections in Russia: systematic literature review and multicenter study
Smelov, V., Thomas, P., Ouburg, S. & Morre, S. A. Oct 2017 In : Pathogens and Disease. 75, 7, 9 p., 081
Research output: Scientific - peer-review › Review article

Chlamydia trachomatis Strain Types Have Diversified Regionally and Globally with Evidence for Recombination across Geographic Divides
Smelov, V., Vrbanac, A., van Ess, E. F., Noz, M. P., Wan, R., Eklund, C., Morgan, T., Shrier, L. A., Sanders, B., Dillner, J., de Vries, H. J. C., Morre, S. A. & Dean, D. 13 Nov 2017 In : Frontiers in microbiology. 8, 14 p., 2195
Research output: Scientific - peer-review › Article

Early screening for Chlamydia trachomatis in young women for primary prevention of pelvic inflammatory disease (i-Predict): study protocol for a randomised controlled trial
Tamarelle, J., Thiebaut, A. C. M., Sabin, B., Bebear, C., Judlin, P., Fauconnier, A., Rahib, D., Meaude-Roufai, L., Ravel, J., Morre, S. A., de Barbeyrac, B., Delarocque-Astagneau, E. & I-Predict Study Grp 13 Nov 2017 In : Trials. 18, 11 p., 534
Research output: Scientific - peer-review › Article

Burden of Chlamydia trachomatis in India: a systematic literature review
Thomas, P., Spaargaren, J., Kant, R., Lawrence, R., Dayal, A., Lal, J. A. & Morre, S. A. Jul 2017 In : Pathogens and Disease. 75, 5, 10 p., 055
Research output: Scientific - peer-review › Review article

Performance of the multitarget Mikrogen Chlamydia trachomatis IgG ELISA in the prediction of tubal factor infertility (TFI) in subfertile women: comparison with the Medac MOMP IgG ELISA plus
van Ess, E. F., Ouburg, S., Spaargaren, J., Land, J. A. & Morre, S. A. Oct 2017 In : Pathogens and Disease. 75, 7, 7 p., 067
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

Policy Making in Newborn Screening Needs a Structured and Transparent Approach
Jansen, M. E., Lister, K. J., van Kranen, H. J. & Cornel, M. C. 21 Mar 2017 In : Frontiers in Public Health. 5, 6 p., 53
Research output: Scientific - peer-review › Review article

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Concern regarding the alleged spread of hypervirulent lymphogranuloma venereum Chlamydia trachomatis strain in Europe
Seth-Smith, H. M., Galan, J. C., Goldenberger, D., Lewis, D. A., Peuchant, O., Bebear, C., de Barbeyrac, B., Benard, A., Carter, I., Kok, J., Bruisten, S. M., Versteeg, B., Morre, S. A., Thomson, N. R., Egli, A. & de Vries, H. J. 13 Apr 2017 In : Eurosurveillance. 22, 15, p. 1-2 2 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Clinical Genetics

Cat02 SCI/SSCI journal (WI-1)

The Pathogenesis of Ventral Idiopathic Herniation of the Spinal Cord: A Hypothesis Based on the Review of the Literature
Bartels, R. H. M. A., Brunner, H., Hosman, A., van Alfen, N. & Grotenhuis, J. A. 11 Sep 2017 In : Frontiers in Neurology. 8, 10 p., 476
Research output: Scientific - peer-review › Article

International Cooperation to Enable the Diagnosis of All Rare Genetic Diseases
Boycott, K. M. , Rath, A. , Chong, J. X. , Hartley, T. , Alkuraya, F. S. , Baynam, G. , Brookes, A. J. , Brudno, M. , Carracedo, A. , den Dunnen, J. T. , Dyke, S. O. M. , Estivill, X. , Goldblatt, J. , Gonthier, C. , Groft, S. C. , Gut, I. , Hamosh, A. , Hieter, P. , Hoehn, S. , Hurles, M. E. & 20 others Kaufmann, P., Knoppers, B. M., Krischer, J. P., Macek, M., Matthijs, G., Olry, A., Parker, S., Paschall, J., Philippakis, A. A., Rehm, H. L., Robinson, P. N., Sham, P-C., Stefanov, R., Taruscio, D., Unni, D., Vanstone, M. R., Zhang, F., Brunner, H., Bamshad, M. J. & Lochmueller, H. 4 May 2017 In : American Journal of Human Genetics. 100, 5, p. 695-705 11 p.
Research output: Scientific - peer-review › Article

Expanding the clinical spectrum of recessive truncating mutations of KLHL7 to a Bohring-Opitz-like phenotype
Bruel, A-L. , Bigoni, S. , Kennedy, J. , Whiteford, M. , Buxton, C. , Parmeggiani, G. , Wherlock, M. , Woodward, G. , Greenslade, M. , Williams, M. , St-Onge, J. , Ferlini, A. , Garani, G. , Ballardini, E. , van Bon, B. W. , Acuna-Hidalgo, R. , Bohring, A. , Deleuze, J-F. , Boland, A. , Meyer, V. & 10 others Olaso, R., Ginglinger, E., Riviere, J-B., Brunner, H. G., Hoischen, A., Newbury-Ecob, R., Faivre, L., Thauvin-Robinet, C., Thevenon, J. & DDD Study Dec 2017 In : Journal of Medical Genetics. 54, 12, p. 830-835 6 p.
Research output: Scientific - peer-review › Article

Estimates of live birth prevalence of children with Down syndrome in the period 1991-2015 in the Netherlands
de Graaf, G., Engelen, J. J. M., Gijsbers, A. C. J., Hochstenbach, R., Hoffer, M. J. V., Kooper, A. J. A., Sikkema-Raddatz, B., Srebniak, M. I., van der Kevie-Kersemaekers, A. M. F., van Zutven, L. J. C. M. & Voorhoeve, E. May 2017 In : Journal of Intellectual Disability Research. 61, 5, p. 461-470 10 p.
Research output: Scientific - peer-review › Article

The 6p25 deletion syndrome: An update on a rare neurocristopathy
de Vos, I. J. H. M., Stegmann, A. P. A., Webers, C. A. B. & Stumpel, C. T. R. M. 2017 In : Ophthalmic Genetics. 38, 2, p. 101-107 7 p.
Research output: Scientific - peer-review › Review article

BRCA1 mutation carriers have a lower number of mature oocytes after ovarian stimulation for IVF/PGD
Derks-Smeets, I. A. P., van Tilborg, T. C., van Montfoort, A., Smits, L., Torrance, H. L., Meijer-Hoogeveen, M., Broekmans, F., Dreesen, J. C. F. M., Paulussen, A. D. C., Tjan-Heijnen, V. C. G., Homminga, I., van den Berg, M. M. J., Ausems, M. G. E. M., de Rycke, M., de Die-Smulders, C. E. M., Verpoest, W. & van Golde, R. Nov 2017 In : Journal of Assisted Reproduction and Genetics. 34, 11, p. 1475-1482 8 p.
Research output: Scientific - peer-review › Article

Uptake of prenatal diagnostic testing for retinoblastoma compared to other hereditary cancer syndromes in the Netherlands
Dommering, C. J., Henneman, L., van der Hout, A. H., Jonker, M. A., Tops, C. M. J., van den Ouweland, A. M. W., van der Luijt, R. B., Mensenkamp, A. R., Hogervorst, F. B. L., Redeker, E. J. W., de Die-Smulders, C. E. M., Moll, A. C. & Meijers-Heijboer, H. Apr 2017 In : Familial Cancer. 16, 2, p. 271-277 7 p.
Research output: Scientific - peer-review › Article

Computer face-matching technology using two-dimensional photographs accurately matches the facial gestalt of unrelated individuals with the same syndromic form of intellectual disability
Dudding-Byth, T., Baxter, A., Holliday, E. G., Hackett, A., O'Donnell, S., White, S. M., Attia, J., Brunner, H., de Vries, B., Koolen, D., Kleefstra, T., Ratwatte, S., Riveros, C., Brain, S. & Lovell, B. C. 19 Dec 2017 In : BMC Biotechnology. 17, 9 p., 90
Research output: Scientific - peer-review › Article

TSC2 c.1864C > T Variant Aassociated with Mild Cases of Tuberous Sclerosis Complex
Farach, L. S., Gibson, W. T., Sparagana, S. P., Nellist, M., Stumpel, C. T. R. M., Hietala, M., Friedman, E., Pearson, D. A., Creighton, S. P., Wagemans, A., Segel, R., Ben-Shalom, E., Au, K. S. & Northrup, H. Mar 2017 In : American Journal of Medical Genetics Part A. 173, 3, p. 771-775 5 p.
Research output: Scientific - peer-review › Article

The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families
Fountain, M. D. , Aten, E. , Cho, M. T. , Juusola, J. , Walkiewicz, M. A. , Ray, J. W. , Xia, F. , Yang, Y. , Graham, B. H. , Bacino, C. A. , Potocki, L. , van Haeringen, A. , Ruivenkamp, C. A. L. , Mancias, P. , Northrup, H. , Kukolich, M. K. , Weiss, M. M. , van Ravenswaaij-Arts, C. M. A. , Mathijssen, I. B. , Levesque, S. & 19 others Meeks, N., Rosenfeld, J. A., Lemke, D., Hamosh, A., Lewis, S. K., Race, S., Stewart, L. L., Hay, B., Lewis, A. M., Guerreiro, R. L., Bras, J. T., Martins, M. P., Derksen-Lubsen, G., Peeters, E., Stumpel, C., Stegmann, S., Bok, L. A., Santen, G. W. E. & Schaaf, C. P. Jan 2017 In : Genetics in Medicine. 19, 1, p. 45-52 8 p.
Research output: Scientific - peer-review › Article

YY1 Haploinsufficiency Causes an Intellectual Disability Syndrome Featuring Transcriptional and Chromatin Dysfunction
Gabriele, M. , Vulto-van Silfhout, A. T. , Germain, P-L. , Vitriolo, A. , Kumar, R. , Douglas, E. , Haan, E. , Kosaki, K. , Takenouchi, T. , Rauch, A. , Steindl, K. , Frengen, E. , Misceo, D. , Pedurupillay, C. R. J. , Stromme, P. , Rosenfeld, J. A. , Shao, Y. , Craigen, W. J. , Schaaf, C. P. , Rodriguez-Buritica, D. & 31 others Farach, L., Friedman, J., Thulin, P., McLean, S. D., Nugent, K. M., Morton, J., Nicholl, J., Andrieux, J., Stray-Pedersen, A., Chambon, P., Patrier, S., Lynch, S. A., Kjaergaard, S., Torring, P. M., Brasch-Andersen, C., Ronan, A., van Haeringen, A., Anderson, P. J., Powis, Z., Brunner, H. G., Pfundt, R., Schuurs-Hoeijmakers, J. H. M., van Bon, B. W. M., Lelieveld, S., Gilissen, C., Nillesen, W. M., Vissers, L. E. L. M., Gecz, J., Koolen, D. A., Testa, G. & de Vries, B. B. A. 1 Jun 2017 In : American Journal of Human Genetics. 100, 6, p. 907-925 19 p.
Research output: Scientific - peer-review › Article

Awareness and attitude regarding reproductive options of persons carrying a BRCA mutation and their partners
Gietel-Habets, J. J. G., de Die-Smulders, C. E. M., Derks-Smeets, I. A. P., Tibben, A., Tjan-Heijnen, V. C. G., van Golde, R., Gomez-Garcia, E., Kets, C. M. & van Osch, L. A. D. M. Mar 2017 In : Human Reproduction. 32, 3, p. 588-597 10 p.
Research output: Scientific - peer-review › Article

Novel *SLC25A32* mutation in a patient with a severe neuromuscular phenotype
Hellebrekers, D. M., Sallevelt, S. C. E. H., Theunissen, T. E. J., Hendrickx, A. T. M., Gottschalk, R. W., Hoeijmakers, J. G. J., Habets, D. D., Bierau, J., Schoonderwoerd, K. G. & Smeets, H. J. M. Jun 2017 In : European Journal of Human Genetics. 25, 7, p. 886-888 3 p.
Research output: Scientific - peer-review › Article

Novel genetic loci associated with hippocampal volume

Hibar, D. P. , Adams, H. H. H. , Jahanshad, N. , Chauhan, G. , Stein, J. L. , Hofer, E. , Renteria, M. E. , Bis, J. C. , Arias-Vasquez, A. , Ikram, M. K. , Desrivieres, S. , Vernooij, M. W. , Abramovic, L. , Alhusaini, S. , Amin, N. , Andersson, M. , Arfanakis, K. , Aribisala, B. S. , Armstrong, N. J. , Athanasiu, L. & 312 others Axelsson, T., Beecham, A. H., Beiser, A., Bernard, M., Blanton, S. H., Bohlken, M. M., Boks, M. P., Bralten, J., Brickman, A. M., Carmichael, O., Chakravarty, M. M., Chen, Q., Ching, C. R. K., Chouraki, V., Cuellar-Partida, G., Crivello, F., Den Braber, A., Nhat Trung Doan, Ehrlich, S., Giddaluru, S., Goldman, A. L., Gottesman, R. F., Grimm, O., Griswold, M. E., Guadalupe, T., Gutman, B. A., Hass, J., Haukvik, U. K., Hoehn, D., Holmes, A. J., Hoogman, M., Janowitz, D., Jia, T., Jorgensen, K. N., Karbalai, N., Kasperaviciute, D., Kim, S., Klein, M., Kraemer, B., Lee, P. H., Liewald, D. C. M., Lopez, L. M., Luciano, M., Macare, C., Marquand, A. F., Matarin, M., Mather, K. A., Mattheisen, M., McKay, D. R., Milaneschi, Y., Maniega, S. M., Nho, K., Nugent, A. C., Nyquist, P., Loohuis, L. M. O., Oosterlaan, J., Papmeyer, M., Pirpamer, L., Puetz, B., Ramasamy, A., Richards, J. S., Risacher, S. L., Roiz-Santianez, R., Rommelse, N., Ropele, S., Rose, E. J., Royle, N. A., Rundek, T., Saemann, P. G., Saremi, A., Satizabal, C. L., Schmaal, L., Schork, A. J., Shen, L., Shin, J., Shumskaya, E., Smith, A. V., Sprooten, E., Strike, L. T., Teumer, A., Tordesillas-Gutierrez, D., Toro, R., Trabzuni, D., Trompet, S., Vaidya, D., Van der Grond, J., Van der Lee, S. J., Van der Meer, D., Van Donkelaar, M. M. J., Van Eijk, K. R., Van Erp, T. G. M., Van Rooij, D., Walton, E., Westlye, L. T., Whelan, C. D., Windham, B. G., Winkler, A. M., Wittfeld, K., Woldehawariat, G., Wolf, C., Wolfers, T., Yanek, L. R., Yang, J., Zijdenbos, A., Zwiers, M. P., Agartz, I., Almasy, L., Ames, D., Amouyel, P., Andreassen, O. A., Arepalli, S., Assareh, A. A., Barral, S., Bastin, M. E., Becker, D. M., Becker, J. T., Bennett, D. A., Blangero, J., van Bokhoven, H., Boomsma, D. I., Brodaty, H., Brouwer, R. M., Brunner, H. G., Buckner, R. L., Buitelaar, J. K., Bulayeva, K. B., Cahn, W., Calhoun, V. D., Cannon, D. M., Cavalleri, G. L., Cheng, C-Y., Cichon, S., Cookson, M. R., Corvin, A., Crespo-Facorro, B., Curran, J. E., Czisch, M., Dale, A. M., Davies, G. E., De Craen, A. J. M., De Geus, E. J. C., De Jager, P. L., De Zubicaray, G. I., Deary, I. J., Debette, S., DeCarli, C., Delanty, N., Depondt, C., DeStefano, A., Dillman, A., Djurovic, S., Donohoe, G., Drevets, W. C., Duggirala, R., Dyer, T. D., Enzinger, C., Erk, S., Espeseth, T., Fedko, I. O., Fernandez, G., Ferrucci, L., Fisher, S. E., Fleischman, D. A., Ford, I., Fornage, M., Foroud, T. M., Fox, P. T., Francks, C., Fukunaga, M., Gibbs, J. R., Glahn, D. C., Gollub, R. L., Goring, H. H. H., Green, R. C., Gruber, O., Gudnason, V., Guelfi, S., Haberg, A. K., Hansell, N. K., Hardy, J., Hartman, C. A., Hashimoto, R., Hegenscheid, K., Heinz, A., Le Hellard, S., Hernandez, D. G., Heslenfeld, D. J., Ho, B-C., Hoekstra, P. J., Hoffmann, W., Hofman, A., Holsboer, F., Homuth, G., Hosten, N., Hottenga, J-J., Huentelman, M., Pol, H. E. H., Ikeda, M., Jack, C. R., Jenkinson, M., Johnson, R., Joensson, E. G., Jukema, J. W., Kahn, R. S., Kanai, R., Kloszewska, I., Knopman, D. S., Kochunov, P., Kwok, J. B., Lawrie, S. M., Lemaitre, H., Liu, X., Longo, D. L., Lopez, O. L., Lovestone, S., Martinez, O., Martinot, J-L., Mattay, V. S., McDonald, C., McIntosh, A. M., McMahon, F. J., McMahon, K. L., Mecocci, P., Melle, I., Meyer-Lindenberg, A., Mohnke, S., Montgomery, G. W., Morris, D. W., Mosley, T. H., Muhleisen, T. W., Mueller-Myhsok, B., Nalls, M. A., Nauck, M., Nichols, T. E., Niessen, W. J., Nothen, M. M., Nyberg, L., Ohi, K., Olvera, R. L., Ophoff, R. A., Pandolfo, M., Paus, T., Pausova, Z., Penninx, B. W. J. H., Pike, G. B., Potkin, S. G., Psaty, B. M., Reppermund, S., Rietschel, M., Roffman, J. L., Romanczuk-Seiferth, N., Rotter, J. I., Ryten, M., Sacco, R. L., Sachdev, P. S., Saykin, A. J., Schmidt, R., Schmidt, H., Schofield, P. R., Sigursson, S., Simmons, A., Singleton, A., Sisodiya, S. M., Smith, C., Smoller, J. W., Soininen, H., Steen, V. M., Stott, D. J., Sussmann, J. E., Thalamuthu, A., Toga, A. W., Traynor, B. J., Troncoso, J., Tsolaki, M., Tzourio, C., Uitterlinden, A. G., Hernandez, M. C. V., Van der Brug, M., van der Lugt, A., van der Wee, N. J. A., Van Haren, N. E. M., van't Ent, D., Van Tol, M-J., Vardarajan, B. N., Vellas, B., Veltman, D. J., Voelzke, H., Walter, H., Wardlaw, J. M., Wassink, T. H., Weale, M. E., Weinberger, D. R., Weiner, M. W., Wen, W., Westman, E., White, T., Wong, T. Y., Wright, C. B., Zielke, R. H., Zonderman, A. B., Martin, N. G., Van Duijn, C. M., Wright, M. J., Longstreth, W. T., Schumann, G., Grabe, H. J., Franke, B., Launer, L. J., Medland, S. E., Seshadri, S., Thompson, P. M. & Ikram, M. A. 18 Jan 2017 In : Nature Communications. 8, 12 p., 13624

Research output: Scientific - peer-review › Article

Correlated evolutionary rates across genomic compartments in Annonaceae

Hoekstra, P. H., Wieringa, J. J., Smets, E., Brandão, R. D., Lopes, J. D. C., Erkens, R. H. J. & Chatrou, L. W. Sep 2017 In : Molecular Phylogenetics and Evolution. 114, p. 63-72 10 p.

Research output: Scientific - peer-review › Article

Delusional and Psychotic Disorders in Juvenile Myotonic Dystrophy Type-1
Jacobs, D., Willekens, D., de Die-Smulders, C., Frijns, J-P. & Steyaert, J. Jun 2017 In : American Journal of Medical Genetics Part B-neuropsychiatric Genetics. 174, 4, p. 359-366 8 p.
Research output: Scientific - peer-review › Article

Next-Generation Sequencing in Oncology: Genetic Diagnosis, Risk Prediction and Cancer Classification
Kamps, R., Brandao, R. D., van den Bosch, B. J., Paulussen, A. D. C., Xanthoulea, S., Blok, M. J. & Romano, A. Feb 2017 In : International Journal of Molecular Sciences. 18, 2, 57 p., 308
Research output: Scientific - peer-review › Review article

Network topology of NaV1.7 mutations in sodium channel-related painful disorders
Kapetis, D., Sassone, J., Yang, Y., Galbardi, B., Xenakis, M. N., Westra, R. L., Szklarczyk, R., Lindsey, P., Faber, C. G., Gerrits, M., Merkies, I. S. J., Dib-Hajj, S. D., Mantegazza, M., Waxman, S. G., PROPANE Study Grp, J.M. Smeets, H. & Lauria, G. 24 Feb 2017 In : BMC Systems Biology. 11, 16 p., 28
Research output: Scientific - peer-review › Article

Structural asymmetries of the human cerebellum in relation to cerebral cortical asymmetries and handedness
Kavaklioglu, T., Guadalupe, T., Zwiers, M., Marquand, A. F., Onnink, M., Shumskaya, E., Brunner, H., Fernandez, G., Fisher, S. E. & Francks, C. May 2017 In : Brain Structure & Function. 222, 4, p. 1611-1623 13 p.
Research output: Scientific - peer-review › Article

Novel IRF6 Mutations Detected in Orofacial Cleft Patients by Targeted Massively Parallel Sequencing
Khandelwal, K. D. , Ishorst, N. , Zhou, H. , Ludwig, K. U. , Venselaar, H. , Gilissen, C. , Thonissen, M. , van Rooij, I. A. L. M. , Dreesen, K. , Steehouwer, M. , van de Vorst, M. , Bloemen, M. , van Beusekom, E. , Roosenboom, J. , Borstlap, W. , Admiraal, R. , Dormaar, T. , Schoenaers, J. , Vander Poorten, V. , Hens, G. & 10 others Verdonck, A., Berge, S., Roeleveldt, N., Vriend, G., Devriendt, K., Brunner, H. G., Mangold, E., Hoischen, A., van Bokhoven, H. & Carels, C. E. L. Feb 2017 In : Journal of Dental Research. 96, 2, p. 179-185 7 p.
Research output: Scientific - peer-review › Article

Functional convergence of histone methyltransferases EHMT1 and KMT2C involved in intellectual disability and autism spectrum disorder
Koemans, T. S., Kleefstra, T., Chubak, M. C., Stone, M. H., Reijnders, M. R. F., de Munnik, S., Willemsen, M. H., Fenckova, M., Stumpel, C. T. R. M., Bok, L. A., Saenz, M. S., Byerly, K. A., Baughn, L. B., Stegmann, A. P. A., Pfundt, R., Zhou, H., van Bokhoven, H., Schenck, A. & Kramer, J. M. Oct 2017 In : Plos Genetics. 13, 10, 24 p., 1006864
Research output: Scientific - peer-review › Article

Recurrent De Novo Mutations Disturbing the GTP/GDP Binding Pocket of RAB11B Cause Intellectual Disability and a Distinctive Brain Phenotype
Lamers, I. J. C. , Reijnders, M. R. F. , Venselaar, H. , Kraus, A. , Jansen, S. , de Vries, B. B. A. , Houge, G. , Gradek, G. A. , Seo, J. , Choi, M. , Chae, J-H. , van der Burgt, I. , Pfundt, R. , Letteboer, S. J. F. , van Beersum, S. E. C. , Dusseljee, S. , Brunner, H. G. , Doherty, D. , Kleefstra, T. , Roepman, R. & 1 others DDD Study 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 824-832 9 p.
Research output: Scientific - peer-review › Article

Amiloride-sensitive cation channel 2 genotype affects the response to a carbon dioxide panic challenge
Leibold, N. K., van den Hove, D. L. A., Viechtbauer, W., Kenis, G., Goossens, L., Lange, I., Knuts, I., Smeets, H. J., Myin-Germeys, I., Steinbusch, H. W. M. & Schruers, K. R. J. Oct 2017 In : Journal of Psychopharmacology. 31, 10, p. 1294-1301 8 p.
Research output: Scientific - peer-review › Article

Spatial Clustering of de Novo Missense Mutations Identifies Candidate Neurodevelopmental Disorder-Associated Genes
Lelieveld, S. H., Wiel, L., Venselaar, H., Pfundt, R., Vriend, G., Veltman, J. A., Brunner, H. G., Vissers, L. E. L. M. & Gilissen, C. 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 478-484 7 p.
Research output: Scientific - peer-review › Article

***De Novo* Missense Mutations in *DHX30* Impair Global Translation and Cause a Neurodevelopmental Disorder**
Lessel, D. , Schob, C. , Kuery, S. , Reinders, M. R. F. , Harel, T. , Eldomery, M. K. , Coban-Akdemir, Z. , Denecke, J. , Edvardson, S. , Colin, E. , Stegmann, A. P. A. , Gerkes, E. H. , Tessarech, M. , Bonneau, D. , Barth, M. , Besnard, T. , Cogne, B. , Revah-Politi, A. , Strom, T. M. , Rosenfeld, J. A. & 24 others Yang, Y., Posey, J. E., Immken, L., Oundjian, N., Helbig, K. L., Meeks, N., Zegar, K., Morton, J., Schieving, J. H., Claasen, A., Huentelman, M., Narayanan, V., Ramsey, K., Brunner, H. G., Elpeleg, O., Mercier, S., Bezieau, S., Kubisch, C., Kleefstra, T., Kindler, S., Lupski, J. R., Kreienkamp, H-J., DDD study & C4RCD Res Grp 2 Nov 2017 In : American Journal of Human Genetics. 101, 5, p. 716-724 9 p.
Research output: Scientific - peer-review › Article

B3GALNT2 mutations associated with non-syndromic autosomal recessive intellectual disability reveal a lack of genotype-phenotype associations in the muscular dystrophy-dystroglycanopathies
Maroofian, R., Riemersma, M., Jae, L. T., Zhianabed, N., Willemsen, M. H., Wissink-Lindhout, W. M., Willemsen, M. A., de Brouwer, A. P. M., Mehrjardi, M. Y. V., Ashrafi, M. R., Kusters, B., Kleefstra, T., Jamshidi, Y., Nasseri, M., Pfundt, R., Brummelkamp, T. R., Abbaszadegan, M. R., Lefeber, D. J. & van Bokhoven, H. 22 Dec 2017 In : Genome Medicine. 9, 11 p., 118
Research output: Scientific - peer-review › Article

MicroRNAs in glaucoma and neurodegenerative diseases
Molasy, M., Walczak, A., Szaflik, J., Szaflik, J. P. & Majsterek, I. Jan 2017 In : Journal of Human Genetics. 62, 1, p. 105-112 8 p.
Research output: Scientific - peer-review › Review article

Dietary nitrate does not reduce oxygen cost of exercise or improve muscle mitochondrial function in patients with mitochondrial myopathy
Nabben, M., Schmitz, J. P. J., Ciapaite, J., le Clercq, C. M. P., van Riel, N. A., Haak, H. R., Nicolay, K., de Coo, I. F. M., Smeets, H., Praet, S. F., van Loon, L. J. & Prompers, J. J. 1 May 2017 In : American Journal of Physiology-regulatory Integrative and Comparative Physiology. 312, 5, p. R689-R701 13 p.
Research output: Scientific - peer-review › Article

Duplicated Enhancer Region Increases Expression of CTSB and Segregates with Keratolytic Winter Erythema in South African and Norwegian Families

Ngcungcu, T. , Oti, M. , Sitek, J. C. , Haukanes, B. I. , Linghu, B. , Bruccoleri, R. , Stokowy, T. , Oakeley, E. J. , Yang, F. , Zhu, J. , Sultan, M. , Schalkwijk, J. , van Vlijmen-Willems, I. M. J. J. , von der Lippe, C. , Brunner, H. G. , Ersland, K. M. , Grayson, W. , Buechmann-Moller, S. , Sundnes, O. , Nirmala, N. & 9 others Morgan, T. M., van Bokhoven, H., Steen, V. M., Hull, P. R., Szustakowski, J., Staedtler, F., Zhou, H., Fiskerstrand, T. & Ramsay, M. 4 May 2017 In : American Journal of Human Genetics. 100, 5, p. 737-750 14 p.

Research output: Scientific - peer-review › Article

Novel pathogenic SLC25A46 splice-site mutation causes an optic atrophy spectrum disorder

Nguyen, M., Boesten, I., Hellebrekers, D. M. E. I., Mulder-den Hartog, N. M., de Coo, I. F. M., Smeets, H. J. M. & Gerards, M. Jan 2017 In : Clinical Genetics. 91, 1, p. 121-125 5 p.

Research output: Scientific - peer-review › Article

Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia

Oud, M. S., Ramos, L., O'Bryan, M. K., McLachlan, R. I., Okutman, O., Viville, S., de Vries, P. F., Smeets, D. F. C. M., Lugtenberg, D., Hehir-Kwa, J. Y., Gilissen, C., van de Vorst, M., Vissers, L. E. L. M., Hoischen, A., Meijerink, A. M., Fleischer, K., Veltman, J. A. & Noordam, M. J. Nov 2017 In : Human Mutation. 38, 11, p. 1592-1605 14 p.

Research output: Scientific - peer-review › Article

Mutations in EXTL3 Cause Neuro-immuno-skeletal Dysplasia Syndrome

Oud, M. M. , Tuijnenburg, P. , Hempel, M. , van Vlies, N. , Ren, Z. , Ferdinandusse, S. , Jansen, M. H. , Santer, R. , Johannsen, J. , Bacchelli, C. , Alders, M. , Li, R. , Davies, R. , Dupuis, L. , Cale, C. M. , Wanders, R. J. A. , Pals, S. T. , Ocaka, L. , James, C. , Mueller, I. & 18 others Lehmberg, K., Strom, T., Engels, H., Williams, H. J., Beales, P., Roepmand, R., Dias, P., Brunner, H. G., Cobben, J-M., Hall, C., Hartley, T., Stabej, P. L. Q., Mendoza-Londono, R., Davies, E. G., de Sousa, S. B., Lesse, D., Arts, H. H. & Kuijpers, T. W. 2 Feb 2017 In : American Journal of Human Genetics. 100, 2, p. 281-296 16 p.

Research output: Scientific - peer-review › Article

NGS panel analysis in 24 ectopia lentis patients; a clinically relevant test with a high diagnostic yield

Overwater, E. , Floor, K. , van Beek, D. , de Boer, K. , van Dijk, T. , Hilhorst-Hofstee, Y. , Hoogeboom, A. J. M. , van Kaam, K. J. , van de Kamp, J. M. , Kempers, M. , Krapels, I. P. C. , Kroes, H. Y. , Loeys, B. , Salemink, S. , Stumpel, C. T. R. M. , Verhoeven, V. J. M. , Wijnands-van den Berg, E. , Cobben, J. M. , van Tintelen, J. P. , Weiss, M. M. & 2 others Houweling, A. C. & Maugeri, A. Sep 2017 In : European Journal of Medical Genetics. 60, 9, p. 465-473 9 p.

Research output: Scientific - peer-review › Article

The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies
Redin, C. , Brand, H. , Collins, R. L. , Kammin, T. , Mitchell, E. , Hodge, J. C. , Hanscom, C. , Pillalamarri, V. , Seabra, C. M. , Abbott, M-A. , Abdul-Rahman, O. A. , Aberg, E. , Adley, R. , Alcaraz-Estrada, S. L. , Alkuraya, F. S. , An, Y. , Anderson, M-A. , Antolik, C. , Anyane-Yeboa, K. , Atkin, J. F. & 129 others Bartell, T., Bernstein, J. A., Beyer, E., Blumenthal, I., Bongers, E. M. H. F., Brilstra, E. H., Brown, C. W., Bruggenwirth, H. T., Callewaert, B., Chiang, C., Corning, K., Cox, H., Cuppen, E., Currall, B. B., Cushing, T., David, D., Deardorff, M. A., Dheedene, A., D'Hooghe, M., de Vries, B. B. A., Earl, D. L., Ferguson, H. L., Fisher, H., FitzPatrick, D. R., Gerrol, P., Giachino, D., Glessner, J. T., Gliem, T., Grady, M., Graham, B. H., Griffis, C., Gripp, K. W., Gropman, A. L., Hanson-Kahn, A., Harris, D. J., Hayden, M. A., Hill, R., Hochstenbach, R., Hoffman, J. D., Hopkin, R. J., Hubshman, M. W., Innes, A. M., Irons, M., Irving, M., Jacobsen, J. C., Janssens, S., Jewett, T., Johnson, J. P., Jongmans, M. C., Kahler, S. G., Koolen, D. A., Korzelius, J., Kroisel, P. M., Lacassie, Y., Lawless, W., Lemyre, E., Leppig, K., Levin, A. V., Li, H., Li, H., Liao, E. C., Lim, C., Lose, E. J., Lucente, D., Macera, M. J., Manavalan, P., Mandrile, G., Marcelis, C. L., Margolin, L., Mason, T., Masser-Frye, D., McClellan, M. W., Mendoza, C. J. Z., Menten, B., Middelkamp, S., Mikami, L. R., Moe, E., Mohammed, S., Mononen, T., Mortenson, M. E., Moya, G., Nieuwint, A. W., Ordulu, Z., Parkash, S., Pauker, S. P., Pereira, S., Perrin, D., Phelan, K., Pina Aguilar, R. E., Poddighe, P. J., Pregno, G., Raskin, S., Reis, L., Rhead, W., Rita, D., Renkens, I., Roelens, F., Ruliera, J., Rump, P., Schilit, S. L. P., Shaheen, R., Sparkes, R., Spiegel, E., Stevens, B., Stone, M. R., Tagoe, J., Thakuria, J. V., van Bon, B. W., van de Kamp, J., van Der Burgt, I., van Essen, T., van Ravenswaaij-Arts, C. M., van Roosmalen, M. J., Vergult, S., Volker-Touw, C. M. L., Warburton, D. P., Waterman, M. J., Wiley, S., Wilson, A., Yerena-de Vega, M. D. L. C. A., Zori, R. T., Levy, B., Brunner, H. G., de Leeuw, N., Kloosterman, W. P., Thorland, E. C., Morton, C. C., Gusella, J. F. & Talkowski, M. E. Jan 2017 In : Nature Genetics. 49, 1, p. 36-45 10 p.
Research output: Scientific - peer-review › Article

Variation in a range of mTOR-related genes associates with intracranial volume and intellectual disability
Reijnders, M. R. F. , Kousi, M. , van Woerden, G. M. , Klein, M. , Bralten, J. , Mancini, G. M. S. , van Essen, T. , Proietti-Onori, M. , Smeets, E. E. J. , van Gastel, M. , Stegmann, A. P. A. , Stevens, S. J. C. , Lelieveld, S. H. , Gilissen, C. , Pfundt, R. , Tan, P. L. , Kleefstra, T. , Franke, B. , Elgersma, Y. , Katsanis, N. & 1 others Brunner, H. G. 20 Oct 2017 In : Nature Communications. 8, 12 p., 1052
Research output: Scientific - peer-review › Article

RAC1 Missense Mutations in Developmental Disorders with Diverse Phenotypes
Reijnders, M. R. F. , Ansor, N. M. , Kousi, M. , Yue, W. W. , Tan, P. L. , Clarkson, K. , Clayton-Smith, J. , Corning, K. , Jones, J. R. , Lam, W. W. K. , Mancini, G. M. S. , Marcelis, C. , Mohammed, S. , Pfundt, R. , Roifman, M. , Cohn, R. , Chitayat, D. , Millard, T. H. , Katsanis, N. , Brunner, H. G. & 2 others Banka, S. & Deciphering Dev Disorders Study 7 Sep 2017 In : American Journal of Human Genetics. 101, 3, p. 466-477 12 p.
Research output: Scientific - peer-review › Article

Propionic acidemia as a cause of adult-onset dilated cardiomyopathy
Riemersma, M., Hazebroek, M. R., Helderman-van den Enden, A. T. J. M., Salomons, G. S., Ferdinandusse, S., Brouwers, M. C. G. J., van der Ploeg, L., Heymans, S., Glatz, J. F. C., van den Wijngaard, A., Krapels, I. P. C., Bierau, J. & Brunner, H. G. Nov 2017 In : European Journal of Human Genetics. 25, 11, p. 1195-1201 7 p.
Research output: Scientific - peer-review › Article

Preimplantation genetic diagnosis for mitochondrial DNA mutations: analysis of one blastomere suffices
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Coonen, E., Paulussen, A. D. C., Hellebrekers, D. M. E. I., de Die-Smulders, C. E. M., Smeets, H. J. M. & Lindsey, P. Oct 2017 In : Journal of Medical Genetics. 54, 10, p. 693-697 5 p.
Research output: Scientific - peer-review › Article

A comprehensive strategy for exome-based preconception carrier screening
Sallevelt, S. C. E. H., de Koning, B., Szklarczyk, R., Paulussen, A. D. C., de Die-Smulders, C. E. M. & Smeets, H. J. M. May 2017 In : Genetics in Medicine. 19, 5, p. 583-592 10 p.
Research output: Scientific - peer-review › Article

De novo mtDNA point mutations are common and have a low recurrence risk
Sallevelt, S. C. E. H., de Die-Smulders, C. E. M., Hendrickx, A. T. M., Hellebrekers, D. M. E. I., de Coo, I. F. M., Alston, C. L., Knowles, C., Taylor, R. W., McFarland, R. & Smeets, H. J. M. Feb 2017 In : Journal of Medical Genetics. 54, 2, p. 114-124 11 p.
Research output: Scientific - peer-review › Article

PGD for the m.14487 T>C mitochondrial DNA mutation resulted in the birth of a healthy boy
Sallevelt, S. C. E. H., Dreesen, J. C. F. M., Druesedau, M., Hellebrekers, D. M. E. I., Paulussen, A. D. C., Coonen, E., Van Golde, R. J. T., Geraedts, J. P. M., Gianaroli, L., Magli, M. C., Zeviani, M., Smeets, H. J. M. & de Die-Smulders, C. E. M. Mar 2017 In : Human Reproduction. 32, 3, p. 698-703 6 p.
Research output: Scientific - peer-review › Article

Growth Hormone Therapy in Children with Kabuki Syndrome: 1-year Treatment Results
Schott, D. A., Gerver, W. J. M. & Stumpel, C. T. R. M. 2017 In : Hormone Research in Paediatrics. 88, 3-4, p. 1-7 7 p.
Research output: Scientific - peer-review › Article

The diagnostic yield of whole-exome sequencing targeting a gene panel for hearing impairment in The Netherlands
Seco, C. Z. , Wesdorp, M. , Feenstra, I. , Pfundt, R. , Hehir-Kwa, J. Y. , Lelieveld, S. H. , Castelein, S. , Gilissen, C. , de Wijs, I. J. , Admiraal, R. J. C. , Pennings, R. J. E. , Kunst, H. P. M. , van de Kamp, J. M. , Tamminga, S. , Houweling, A. C. , Plomp, A. S. , Maas, S. M. , Gans, P. A. M. D. K. , Kant, S. G. , de Geus, C. M. & 10 others Frints, S. G. M., Vanhoutte, E. K., van Dooren, M. F., van den Boogaard, M-J. H., Scheffer, H., Nelen, M., Kremer, H., Hoefsloot, L., Schraders, M. & Yntema, H. G. Feb 2017 In : European Journal of Human Genetics. 25, 3, p. 308-314 7 p.
Research output: Scientific - peer-review › Article

A homozygous FITM2 mutation causes a deafness-dystonia syndrome with motor regression and signs of ichthyosis and sensory neuropathy
Seco, C. Z. , Castells-Nobau, A. , Joo, S. , Schraders, M. , Foo, J. N. , van der Voet, M. , Velan, S. S. , Nijhof, B. , Oostrik, J. , de Vrieze, E. , Katana, R. , Mansoor, A. , Huynen, M. , Szklarczyk, R. , Oti, M. , Tranebjaerg, L. , van Wijk, E. , Scheffer-de Gooyert, J. M. , Siddique, S. , Baets, J. & 10 others de Jonghe, P., Kazmi, S. A. R., Sadananthan, S. A., van de Warrenburg, B. P., Khor, C. C., Goepfert, M. C., Qamar, R., Schenck, A., Kremer, H. & Siddiqi, S. 1 Feb 2017 In : Disease Models & Mechanisms. 10, 2, p. 105-118 14 p.
Research output: Scientific - peer-review › Article

Selection and Characterization of Palmitic Acid Responsive Patients with an OXPHOS Complex I Defect
Theunissen, T. E. J., Gerards, M., Hellebrekers, D. M. E. I., van Tienen, F. H., Kamps, R., Sallevelt, S. C. E. H., Hartog, E. N. M. M. -D., Scholte, H. R., Verdijk, R. M., Schoonderwoerd, K., de Coo, I. F. M., Szklarczyk, R. & Smeets, H. J. M. 18 Oct 2017 In : Frontiers in molecular neuroscience. 10, 12 p., 336
Research output: Scientific - peer-review › Article

Rapid Resolution of Blended or Composite Multigenic Disease in Infants by Whole-Exome Sequencing
Theunissen, T. E. J., Sallevelt, S. C. E. H., Hellebrekers, D. M. E. I., de Koning, B., Hendrickx, A. T. M., van den Bosch, B. J. C., Kamps, R., Schoonderwoerd, K., Szklarczyk, R., Hartog, E. N. M. M-D., de Coo, I. F. M. & Smeets, H. J. M. Mar 2017 In : The Journal of Pediatrics. 182, p. 371-374 4 p.
Research output: Scientific - peer-review › Article

A clinical utility study of exome sequencing versus conventional genetic testing in pediatric neurology
Vissers, L. E. L. M., van Nimwegen, K. J. M., Schieving, J. H., Kamsteeg, E-J., Kleefstra, T., Yntema, H. G., Pfundt, R., van der Wilt, G. J., Krabbenborg, L., Brunner, H. G., van der Burg, S., Grutters, J., Veltman, J. A. & Willemsen, M. A. A. P. Sep 2017 In : Genetics in Medicine. 19, 9, p. 1055-1063 9 p.
Research output: Scientific - peer-review › Article

Quantification of Phenotype Information Aids the Identification of Novel Disease Genes
Vulto-van Silfhout, A. T., Gilissen, C., Goeman, J. J., Jansen, S., van Amen-Hellebrekers, C. J. M., van Bon, B. W. M., Koolen, D. A., Sistermans, E. A., Brunner, H. G., de Brouwer, A. P. M. & de Vries, B. B. A. May 2017 In : Human Mutation. 38, 5, p. 594-599 6 p.
Research output: Scientific - peer-review › Article

Novel BRCA1 and BRCA2 Tumor Test as Basis for Treatment Decisions and Referral for Genetic Counselling of Patients with Ovarian Carcinomas
Weren, R. D. A., Mensenkamp, A. R., Simons, M., Eijkelenboom, A., Sie, A. S., Ouchene, H., van Asseldonk, M., Gomez-Garcia, E. B., Blok, M. J., de Hullu, J. A., Nelen, M. R., Hoischen, A., Bulten, J., Tops, B. B. J., Hoogerbrugge, N. & Ligtenberg, M. J. L. Feb 2017 In : Human Mutation. 38, 2, p. 226-235 10 p.
Research output: Scientific - peer-review › Article

Mutations in the Chromatin Regulator Gene *BRPF1* Cause Syndromic Intellectual Disability and Deficient Histone Acetylation
Yan, K. , Rousseau, J. , Littlejohn, R. O. , Kiss, C. , Lehman, A. , Rosenfeld, J. A. , Stumpel, C. T. R. , Stegmann, A. P. A. , Robak, L. , Scaglia, F. , Thi Tuyet Mai Nguyen, , Fu, H. , Ajeawung, N. F. , Camurri, M. V. , Li, L. , Gardham, A. , Panis, B. , Almannai, M. , Sacoto, M. J. G. , Baskin, B. & 16 others Ruivenkamp, C., Xia, F., Bi, W., Cho, M. T., Potjer, T. P., Santen, G. W. E., Parker, M. J., Canham, N., McKinnon, M., Potocki, L., MacKenzie, J. J., Roeder, E. R., Campeau, P. M., Yang, X-J., DDD Study & CAUSES Study 5 Jan 2017 In : American Journal of Human Genetics. 100, 1, p. 91-104 14 p.
Research output: Scientific - peer-review › Article

Cat03 Non SCI/SSCI journal (WI-2)

Homozygosity Mapping and Targeted Sanger Sequencing Identifies Three Novel CRB1 (Crumbs homologue 1) Mutations in Iranian Retinal Degeneration Families
Ghofrani, M., Yahyaei, M., Brunner, H. G., Cremers, F. P. M., Movasat, M., Imran Khan, M. & Keramatipour, M. Sep 2017 In : Iranian Biomedical Journal. 21, 5, p. 294-302 9 p.
Research output: Scientific - peer-review › Article

A novel mutation in L1CAM causes a mild form of L1 syndrome: a case report
Otter, M., Wevers, M., Pisters, M., Pfundt, R., Vos, Y., Nievelstein, R. J. & Stumpel, C. Aug 2017 In : Clinical Case Reports. 5, 8, p. 1213-1217 5 p.
Research output: Scientific - peer-review › Article

Cat04 Editorial material (edit) - Cat04 A: Editorial Material in wi-1 journal

The annual meeting 1988-2017
Brunner, H. Dec 2017 In : European Journal of Human Genetics. 25, p. S35-S36 2 p.
Research output: Scientific - peer-review › Editorial

Healthy children without fear: Reproductive options for patients or couples carrying inherited diseases
Geraedts, J. May 2017 In : Embo Reports. 18, 5, p. 666-669 4 p.
Research output: Scientific - peer-review › Editorial

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Thirty years of Dutch embryo selection
Geraedts, J. 30 Nov 2017 In : Nature. 551, 7682, p. 565-565 1 p.
Research output: Scientific - peer-review › Comment/Letter to the editor

Molecular Cell Biology

Cat02 SCI/SSCI journal (WI-1)

Protocol for intraoperative assessment of the human cerebrovascular glycocalyx
Haeren, R. H. L., Vink, H., Staals, J., van Zandvoort, M. A. M. J., Dings, J., van Overbeeke, J. J., Hoogland, G., Rijkers, K. & Schijns, O. E. M. G. Jan 2017 In : BMJ Open. 7, 1, 7 p., 013954
Research output: Scientific - peer-review › Article

Lamin A/C-Related Cardiac Disease Late Onset With a Variable and Mild Phenotype in a Large Cohort of Patients With the Lamin A/C p.(Arg331Gln) Founder Mutation
Hoorntje, E. T. , Bollen, I. A. , Barge-Schaapveld, D. Q. , van Tienen, F. H. , te Meerman, G. J. , Jansweijer, J. A. , van Essen, A. J. , Volders, P. G. , Constantinescu, A. A. , van den Akker, P. C. , van Spaendonck-Zwarts, K. Y. , Oldenburg, R. A. , Marcelis, C. L. , van der Smagt, J. J. , Hennekam, E. A. , Vink, A. , Bootsma, M. , Aten, E. , Wilde, A. A. , van den Wijngaard, A. & 5 others Broers, J. L., Jongbloed, J. D., van der Velden, J., van den Berg, M. P. & van Tintelen, J. P. Aug 2017 In : Circulation : Cardiovascular Genetics. 10, 4, 36 p., 001631
Research output: Scientific - peer-review › Article

Investigating the race for the surface and skin integration in clinically retrieved abutments with two-photon microscopy
Kapsokalyvas, D., van Hoof, M., Wigren, S., Chimhanda, T., Kuijpers, H. J., Ramaekers, F. C. S., Stokroos, R. J. & van Zandvoort, M. A. M. J. 1 Nov 2017 In : Colloids and Surfaces B-Biointerfaces. 159, p. 97-107 11 p.
Research output: Scientific - peer-review › Article

Molecular characterization, prevalence and clinical relevance of Phodopus sungorus papillomavirus type 1 (PsuPV1) naturally infecting Siberian hamsters (&ITPhodopus sungorus&IT)
Kocjan, B. J., Hosnjak, L., Racnik, J., Zadravec, M., Bakovnik, N., Cigler, B., Ummelen, M., Hopman, A. H. N., Gale, N., Svara, T., Gombac, M. & Poljak, M. Nov 2017 In : Journal of General Virology. 98, 11, p. 2799-2809 11 p.
Research output: Scientific - peer-review › Article

PBCA-based polymeric microbubbles for molecular imaging and drug delivery
Koczera, P., Appold, L., Shi, Y., Liu, M., Dasgupta, A., Pathak, V., Ojha, T., Fokong, S., Wu, Z., van Zandvoort, M., Iranzo, O., Kuehne, A. J. C., Pich, A., Kiessling, F. & Lammers, T. 10 Aug 2017 In : Journal of Controlled Release. 259, p. 128-135 8 p.
Research output: Scientific - peer-review › Article

Investigations of Glucocorticoid Action in GN
Kuppe, C., van Roeyen, C., Leuchtle, K., Kabgani, N., Vogt, M., Van Zandvoort, M., Smeets, B., Floege, J., Groene, H-J. & Moellert, M. J. May 2017 In : Journal of the American Society of Nephrology. 28, 5, p. 1408-1420 13 p.
Research output: Scientific - peer-review › Article

Palmitate-Induced Vacuolar-Type H(+)-ATPase Inhibition Feeds Forward Into Insulin Resistance and Contractile Dysfunction
Liu, Y., Steinbusch, L. K. M., Nabben, M., Kapsokalyvas, D., van Zandvoort, M., Schonleitner, P., Antoons, G., Simons, P. J., Coumans, W. A., Geomini, A., Chanda, D., Glatz, J. F. C., Neumann, D. & Luiken, J. J. F. P. 1 Jun 2017 In : Diabetes. 66, 6, p. 1521-1534 14 p.
Research output: Scientific - peer-review › Article

Folic acid reduces doxorubicin-induced cardiomyopathy by modulating endothelial nitric oxide synthase
Octavia, Y., Kararigas, G., de Boer, M., Chrifi, I., Kietadisorn, R., Swinnen, M., Duimel, H., Verheyen, F. K., Brandt, M. M., Fliegner, D., Cheng, C., Janssens, S., Duncker, D. J. & Moens, A. L. Dec 2017 In : Journal of Cellular and Molecular Medicine. 21, 12, p. 3277-3287 11 p.
Research output: Scientific - peer-review › Article

Computer-assisted three-dimensional tracking of sensory innervation in the murine bladder mucosa with two-photon microscopy
Schueth, A., Spronck, B., van Zandvoort, M. A. M. J. & van Koeveringe, G. A. Nov 2017 In : Journal of Chemical Neuroanatomy. 85, p. 43-49 7 p.
Research output: Scientific - peer-review › Article

Limited additive value of the Ki-67 proliferative index on patient survival in World Health Organization-classified pulmonary carcinoids
Swarts, D. R. A., Rudelius, M., Claessen, S. M. H., Cleutjens, J. P., Seidl, S., Volante, M., Ramaekers, F. C. S. & Speel, E. J. M. Feb 2017 In : Histopathology. 70, 3, p. 412-422 11 p.
Research output: Scientific - peer-review › Article

Cellular strain avoidance is mediated by a functional actin cap - observations in an Lmna-deficient cell model
Tamiello, C., Halder, M., Kamps, M. A. F., Baaijens, F. P. T., Broers, J. L. V. & Bouten, C. V. C. 15 Feb 2017 In : Journal of Cell Science. 130, 4, p. 779-790 12 p.
Research output: Scientific - peer-review › Article

Lmna knockout mouse embryonic fibroblasts are less contractile than their wild-type counterparts
van Loosdregt, I. A. E. W., Kamps, M. A. F., Oomens, C. W. J., Loerakker, S., Broers, J. L. V. & Bouten, C. V. C. 1 Aug 2017 In : Integrative Biology. 9, 8, p. 709-721 13 p.
Research output: Scientific - peer-review › Article

Multi-photon microscopy in cardiovascular research
Wu, Z., Rademakers, T., Kiessling, F., Vogt, M., Westein, E., Weber, C., Megens, R. T. A. & van Zandvoort, M. 1 Nov 2017 In : Methods. 130, p. 79-89 11 p.
Research output: Scientific - peer-review › Article

Nilotinib Enhances Tumor Angiogenesis and Counteracts VEGFR2 Blockade in an Orthotopic Breast Cancer Xenograft Model with Desmoplastic Response
Zafarnia, S., Bzyl-Ibach, J., Spivak, I., Li, Y., Koletnik, S., Doleschel, D., Rix, A., Pochon, S., Tardy, I., Koyadan, S., van Zandvoort, M., Palmowski, M., Kiessling, F. & Lederle, W. Nov 2017 In : Neoplasia (Online). 19, 11, p. 896-907 12 p.
Research output: Scientific - peer-review › Article

Cat05 Letter to the editor - Cat05 A: Letter to the editor in wi-1 journal

Preliminary stop of the TOPical Imiquimod treatment of high-grade Cervical intraepithelial neoplasia (TOPIC) trial
Koeneman, M. M., Kruse, A. J., Kooreman, L. F. S., zur Hausen, A., Hopman, A. H. N., Sep, S. J. S., Van Gorp, T., Slangen, B. F. M., van Beekhuizen, H. J., van de Sande, A. J. M., Gerestein, C. G., Nijman, H. W. & Kruitwagen, R. F. P. M. 7 Feb 2017 In : BMC Cancer. 17, 2 p., 110
Research output: Scientific - peer-review › Comment/Letter to the editor

Cat07 Book(contribution)/congrespapers - Cat07 Book contribution (BB)

Optical Imaging
Alves, F., Bode, J., Cimalla, P., Hilger, I., Hofmann, M., Jaedicke, V., Koch, E., Licha, K., Rademakers, T., Razansky, D. & Van Zandvoort, M. A. M. J. 23 May 2017 *Small Animal Imaging: Basics and Practical Guide.* Kiessling, F., Pichler, B. J. & Hauff, P. (eds.). 2 ed. Springer International Publishing, p. 403-490 98 p.
Research output: Scientific › Chapter

Development of the Uterine Cervix and Its Implications for the Pathogenesis of Cervical Cancer
Hopman, A. H. N. & Ramaekers, F. C. S. 10 Sep 2017 *Pathology of the Cervix.* Herrington, C. S. (ed.). 1 ed. Springer International Publishing, Vol. 3, p. 1-20 20 p.
Research output: Scientific › Chapter

Intravital imaging in small animals
Rademakers, T. & van Zandvoort, M. 2017 *Small Animal Imaging: Basics and Practical Guide (second edition).* Kiessling, F., Pichler, B. J. & hauff, P. (eds.). Springer Publishers, p. 403-490
Research output: Scientific › Chapter

Molecular Genetics

Cat02 SCI/SSCI journal (WI-1)

Inhibition of CD40-TRAF6 interactions by the small molecule inhibitor 6877002 reduces neuroinflammation
Aarts, S. A. B. M., Seijkens, T. T. P., Kusters, P. J. H., van der Pol, S. M. A., Zarzycka, B., Heijnen, P. D. A. M., Beckers, L., den Toom, M., Gijbels, M. J. J., Boon, L., Weber, C., de Vries, H. E., Nicolaes, G. A. F., Dijkstra, C. D., Kooij, G. & Lutgens, E. 12 May 2017 In : Journal of Neuroinflammation. 14, 14 p., 105
Research output: Scientific - peer-review › Article

Diabetic db/db mice do not develop heart failure upon pressure overload: a longitudinal in vivo PET, MRI, and MRS study on cardiac metabolic, structural, and functional adaptations
Abdurrachim, D., Nabben, M., Hoerr, V., Kuhlmann, M. T., Bovenkamp, P., Ciapaite, J., Geraets, I. M. E., Coumans, W., Luiken, J. J. F. P., Glatz, J. F. C., Schaefers, M., Nicolay, K., Faber, C., Hermann, S. & Prompers, J. J. 1 Aug 2017 In : Cardiovascular Research. 113, 10, p. 1148-1160 13 p.
Research output: Scientific - peer-review › Article

Inhibition of atherogenesis by the COP9 signalosome subunit 5 in vivo
Asare, Y. , Ommer, M. , Azombo, F. A. , Alampour-Rajabi, S. , Sternkopf, M. , Sanati, M. , Gijbels, M. J. , Schmitz, C. , Sinitski, D. , Tilstam, P. V. , Lue, H. , Gessner, A. , Lange, D. , Schmid, J. A. , Weber, C. , Dichgans, M. , Jankowski, J. , Pardi, R. , de Winther, M. P. J. , Noels, H. & 1 others Bernhagen, J. 14 Mar 2017 In : Proceedings of the National Academy of Sciences of the United States of America. 114, 13, p. E2766-E2775 10 p.
Research output: Scientific - peer-review › Article

A Liver-Specific Long Noncoding RNA With a Role in Cell Viability Is Elevated in Human Nonalcoholic Steatohepatitis
Atanasovska, B., Rensen, S. S., van der Sijde, M. R., Marsman, G., Kumar, V., Jonkers, I., Withoff, S., Shiri-Sverdlov, R., Greve, J. W. M., Faber, K. N., Moshage, H., Wijmenga, C., van de Sluis, B., Hofker, M. H. & Fu, J. Sep 2017 In : Hepatology. 66, 3, p. 794-808 15 p.
Research output: Scientific - peer-review › Article

Functional mucous layer and healing of proximal colonic anastomoses in an experimental model
Bosmans, J. W. A. M., Jongen, A. C. H. M., Birchenough, G. M. H., Nystrom, E. E. L., Gijbels, M. J. J., Derikx, J. P. M., Bouvy, N. D. & Hansson, G. C. Apr 2017 In : British Journal of Surgery. 104, 5, p. 619-630 12 p.
Research output: Scientific - peer-review › Article

Comparison of three different application routes of butyrate to improve colonic anastomotic strength in rats
Bosmans, J. W. A. M., Jongen, A. C. H. M., Boonen, B. T. C., van Rijn, S., Scognamiglio, F., Stucchi, L., Gijbels, M. J. J., Marsich, E. & Bouvy, N. D. Mar 2017 In : International Journal of Colorectal Disease. 32, 3, p. 305-313 9 p.
Research output: Scientific - peer-review › Article

Malondialdehyde Epitopes Are Sterile Mediators of Hepatic Inflammation in Hypercholesterolemic Mice
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Electrochemical reduction of CerMet fuels for transmutation using surrogate CeO_2-Mo pellets
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Modulation of the gut microbiota impacts nonalcoholic fatty liver disease: a potential role for bile acids
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Constitutive CD40 Signaling in Dendritic Cells Limits Atherosclerosis by Provoking Inflammatory Bowel Disease and Ensuing Cholesterol Malabsorption
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Palmitate-Induced Vacuolar-Type H(+)-ATPase Inhibition Feeds Forward Into Insulin Resistance and Contractile Dysfunction
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Inhibition of sarcolemmal FAT/CD36 by sulfo-N-succinimidyl oleate rapidly corrects metabolism and restores function in the diabetic heart following hypoxia/reoxygenation
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Dietary nitrate does not reduce oxygen cost of exercise or improve muscle mitochondrial function in patients with mitochondrial myopathy
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Research output: Scientific - peer-review › Article

Whole body and hematopoietic ADAM8 deficiency does not influence advanced atherosclerotic lesion development, despite its association with human plaque progression
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NDRG4, an early detection marker for colorectal cancer, is specifically expressed in enteric neurons
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Impact of tissue adhesives on the prevention of anastomotic leakage of colonic anastomoses: an in vivo study
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High-Density Lipoproteins Exert Pro-inflammatory Effects on Macrophages via Passive Cholesterol Depletion and PKC-NF-kappa B/STAT1-IRF1 Signaling
van der Vorst, E. P. C. , Theodorou, K. , Wu, Y. , Hoeksema, M. A. , Goossens, P. , Bursill, C. A. , Aliyev, T. , Huitema, L. F. A. , Tas, S. W. , Wolfs, I. M. J. , Kuijpers, M. J. E. , Gijbels, M. J. , Schalkwijk, C. G. , Koonen, D. P. Y. , Abdollahi-Roodsaz, S. , McDaniels, K. , Wang, C-C. , Leitges, M. , Lawrence, T. , Plat, J. & 6 others Van Eck, M., Rye, K-A., Touqui, L., de Winther, M. P. J., Biessen, E. A. L. & Donners, M. M. P. C. 10 Jan 2017 In : Cell Metabolism. 25, 1, p. 197-207 11 p.
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