CURRICULUM VITAE

Bobby P.C. Koeleman

Education & Training:

2001-present
Associate Professor, University Medical Center Utrecht,
Department of Medical Genetics, Utrecht, The Netherlands.
Research lines: - Genetics of autoimmune disease
- Genetics of common epilepsies.

1999-2001
Postdoc, Leiden University Medical Center,
Department of Hematology and Bloodbank, Leiden, The Netherlands.
Subject: Identification of genetic risk factors for Type 1 Diabetes.

1997 - 1999
Postdoc, Oxford University (1998), Wellcome Trust Center of Human Genetics, and
Cambridge University, Medical Research Center (1999),
John Todd laboratory, Oxford and Cambridge, United Kingdom.
Subject: Identification of genetic risk factors for Type 1 Diabetes.

1991 - 1997
Doctorate University/College of Higher Education
Department of Thrombosis and Haematology,
Leiden University Medical Centre, The Netherlands.
Supervisor ('Promotor'): Prof Dr R. Bertina
Title of thesis: Genetic analysis of familial thrombophilia.

1986 - 1991
Undergraduate training in Biomedical Sciences. Leiden State University,
Faculty of Medicine, The Netherlands.
Internships were completed in four different research groups.

Grants:

- Dutch Diabetes Research Foundation (Diabetes Fonds Nederland, DFN) Network Grant. Title: An integrated approach from basic research to clinical intervention in type 1 diabetes. Period 2015-2019. I am one of two Expert Centers.
- EU-Fp7 program grant. Title: EPIPGX, Epilepsy Pharmacogenomics: delivering biomarkers for clinical use. Period: 2012-2015.
- Dutch Rheuma Research Foundation (Reumafonds Nederland). Title: Unravelling the molecular pathogenesis of Rheumatoid Arthritis without antibodies against citrullinated peptides using a Genome Wide Association Study. Period 2009-2013.
- Dutch Hart Foundation (Nederlandse Hart Stichting). Title: Gene discovery for venous thrombosis; from family-based linkage peaks to gene variant testing in population-based case-control studies. Period 2009-2012.
- Dutch Epilepsy Foundation (Nederlands Epilepsie Fonds). Title: Next generation sequencing for epilepsy disease gene discovery and improved DNA diagnostics. Period 2009-2012.
- Dutch Epilepsy Foundation (Nederlands Epilepsie Fonds). Title: Studying genetic, aetiological and clinical factors in treatment responses in the out-patient population-based epilepsy cohort (OPPEC). Period 2009-2013.

- Dutch Diabetes Research Foundation (Diabetes Fonds Nederland, DFN) Expert Center Grant. Title: An integrated approach from basic research to clinical intervention in type 1 diabetes. I am one of four Expert Centers.
- Co-applicant Dutch Rheuma Research Foundation with Dr Meyaard.
  Titel: Checks and balances in the immune system: genomic and functional analysis of inhibitory immune receptors in SLE. Period: 2007-2010.
- EU-Fp6 program grant. Title: Functional Genomics and Neurobiology of Epilepsy. A basis of new therapeutic strategies.
  Period: 2007-2011.
- NWO-VIDI grant ('Vernieuwingsimpuls'). Title: Genetics of common Epilepsies.
  Grant No: 917.66.315. Period: 2006-2010.
- Dutch Diabetes Research Foundation (Diabetes Fonds Nederland, DFN), International (JDRF), and The Netherlands Organization for Health Research and Development (ZonMW).
  Title: Functional genetics in Type 1 Diabetes. Grant No: 2001.10.004, I was coordinator of this network grant.
- National Epilepsy Funds (NEF). Title: The Dutch IGE-Genetics Study: Towards a Case-Control study of Idiopathic Generalised Epilepsy in the Dutch population. Grant No: 04-08. Period: 2004-2006
- Marie Curie Training and Mobility Grant. Title: Identification of genetic risk factors for Type 1 Diabetes. Period: 1997-1999.

Teaching:

Yearly:

- master course "Genetics and Genomics", University of Utrecht; I co-designed this course
- master course "Complex Genetics", University of Utrecht; I designed this course
- Bachelor course "Genes and Genomes", University of Utrecht.

Regularly:

- course "Complex Genetics" "Vervolgopleiding Klinische Genetica"
- course on epilepsy genetics, Int League Against Epilepsy meetings.
- invited master class epilepsy genetics

Committees and Management:

- 2014- .. : External Scientific Advisory Board member, PreciseAds consortium
- 2010- … : Member Scientific Board Diabetes Foundation Netherlands (DFN).
- 2007-2010: Member teaching commission, Department Medical Genetics Utrecht.
- 2007-2009: Interim head section Complex Genetics group, Department Medical Genetics Utrecht.
- 2007-2009**:** Member management team, Department Medical Genetics Utrecht.

List of publications, 166 publications, H-index 36.

1. Remarkable Phenytoin Sensitivity in 4 Children with SCN8A-related Epilepsy: A Molecular Neuropharmacological Approach. Boerma RS, Braun KP, van de Broek MP, van Berkestijn FM, Swinkels ME, Hagebeuk EO, Lindhout D, van Kempen M, Boon M, Nicolai J, de Kovel CG, Brilstra EH, Koeleman BP. Neurotherapeutics. 2015 Epub
2. Complex SCN8A DNA-abnormalities in an individual with therapy resistant absence epilepsy. Berghuis B, de Kovel CG, van Iterson L, Lamberts RJ, Sander JW, Lindhout D, Koeleman BP. Epilepsy Res. 2015 ;115:141-4.
3. Vitamin D receptor polymorphisms and growth until adulthood after very premature birth.
4. Finken MJ, Schrevel M, Houwing-Duistermaat JJ, Kharagjitsingh AV, Dekker FW, Koeleman BP, Roep BO, Wit JM. J Bone Miner Metab. 2015. Epub .

5. Effect of vaccinations on seizure risk and disease course in Dravet syndrome. Verbeek NE, van der Maas NA, Sonsma AC, Ippel E, Vermeer-de Bondt PE, Hagebeuk E, Jansen FE, Geesink HH, Braun KP, de Louw A, Augustijn PB, Neuteboom RF, Schieving JH, Stroink H, Vermeulen RJ, Nicolai J, Brouwer OF, van Kempen M, de Kovel CG, Kemmeren JM, Koeleman BP, Knoers NV, Lindhout D, Gunning WB, Brilstra EH. Neurology. 2015; Epub
6. Genetic Factors for the Severity of ACPA-negative Rheumatoid Arthritis in 2 Cohorts of Early Disease: A Genome-wide Study. de Rooy DP, Tsonaka R, Andersson ML, Forslind K, Zhernakova A, Frank-Bertoncelj M, de Kovel CG, Koeleman BP, van der Heijde DM, Huizinga TW, Toes RE, Houwing-Duistermaat JJ, Ospelt C, Svensson B, van der Helm-van Mil AH. J Rheumatol. 2015 ;42(8):1383-91.
7. Clinical and genetic analysis of a family with two rare reflex epilepsies. Kasteleijn-Nolst Trenité DG, Volkers L, Strengman E, Schippers HM, Perquin W, de Haan GJ, Gkountidi AO, Slot RV, de Graaf SF, Jocic-Jakubi B, Capovilla G, Covanis A, Parisi P, Veggiotti P, Brinciotti M, Incorpora G, Piccioli M, Cantonetti L, Berkovic SF, Scheffer IE, Brilstra EH, Sonsma AC, Bader AJ, de Kovel CG, Koeleman BP. Seizure. 2015 ;29:90-6.
8. Burden analysis of rare microdeletions suggests a strong impact of neurodevelopmental genes in genetic generalised epilepsies. Lal D, Ruppert AK, Trucks H, Schulz H, de Kovel CG, Kasteleijn-Nolst Trenité D, Sonsma AC, Koeleman BP, Lindhout D, Weber YG, Lerche H, Kapser C, Schankin CJ, Kunz WS, Surges R, Elger CE, Gaus V, Schmitz B, Helbig I, Muhle H, Stephani U, Klein KM, Rosenow F, Neubauer BA, Reinthaler EM, Zimprich F, Feucht M, Møller RS, Hjalgrim H, De Jonghe P, Suls A, Lieb W, Franke A, Strauch K, Gieger C, Schurmann C, Schminke U, Nürnberg P; EPICURE Consortium, Sander T. PLoS Genet. 2015;11(5):e1005226.
9. A large-scale genetic analysis reveals a strong contribution of the HLA class II region to giant cell arteritis susceptibility. Carmona FD, Mackie SL, Martín JE, Taylor JC, Vaglio A, Eyre S, Bossini-Castillo L, Castañeda S, Cid MC, Hernández-Rodríguez J, Prieto-González S, Solans R, Ramentol-Sintas M, González-Escribano MF, Ortiz-Fernández L, Morado IC, Narváez J, Miranda-Filloy JA; Spanish GCA Group, Beretta L, Lunardi C, Cimmino MA, Gianfreda D, Santilli D, Ramirez GA, Soriano A, Muratore F, Pazzola G, Addimanda O, Wijmenga C, Witte T, Schirmer JH, Moosig F, Schönau V, Franke A, Palm Ø, Molberg Ø, Diamantopoulos AP, Carette S, Cuthbertson D, Forbess LJ, Hoffman GS, Khalidi NA, Koening CL, Langford CA, McAlear CA, Moreland L, Monach PA, Pagnoux C, Seo P, Spiera R, Sreih AG, Warrington KJ, Ytterberg SR, Gregersen PK, Pease CT, Gough A, Green M, Hordon L, Jarrett S, Watts R, Levy S, Patel Y, Kamath S, Dasgupta B, Worthington J, Koeleman BP, de Bakker PI, Barrett JH, Salvarani C, Merkel PA, González-Gay MA, Morgan AW, Martín J. Am J Hum Genet. 2015 Apr 2;96(4):565-80. doi: 10.1016/j.ajhg.2015.02.009.
10. CHD2 variants are a risk factor for photosensitivity in epilepsy. Galizia EC, Myers CT, Leu C, de Kovel CG, Afrikanova T, Cordero-Maldonado ML, Martins TG, Jacmin M, Drury S, Krishna Chinthapalli V, Muhle H, Pendziwiat M, Sander T, Ruppert AK, Møller RS, Thiele H, Krause R, Schubert J, Lehesjoki AE, Nürnberg P, Lerche H; EuroEPINOMICS CoGIE Consortium, Palotie A, Coppola A, Striano S, Gaudio LD, Boustred C, Schneider AL, Lench N, Jocic-Jakubi B, Covanis A, Capovilla G, Veggiotti P, Piccioli M, Parisi P, Cantonetti L, Sadleir LG, Mullen SA, Berkovic SF, Stephani U, Helbig I, Crawford AD, Esguerra CV, Kasteleijn-Nolst Trenité DG, Koeleman BP, Mefford HC, Scheffer IE, Sisodiya SM. Brain. 2015 ;138(Pt 5):1198-207.
11. De novo loss- or gain-of-function mutations in KCNA2 cause epileptic encephalopathy. Syrbe S, Hedrich UB, Riesch E, Djémié T, Müller S, Møller RS, Maher B, Hernandez-Hernandez L, Synofzik M, Caglayan HS, Arslan M, Serratosa JM, Nothnagel M, May P, Krause R, Löffler H, Detert K, Dorn T, Vogt H, Krämer G, Schöls L, Mullis PE, Linnankivi T, Lehesjoki AE, Sterbova K, Craiu DC, Hoffman-Zacharska D, Korff CM, Weber YG, Steinlin M, Gallati S, Bertsche A, Bernhard MK, Merkenschlager A, Kiess W;

EuroEPINOMICS RES, Gonzalez M, Züchner S, Palotie A, Suls A, De Jonghe P, Helbig I, Biskup S, Wolff M, Maljevic S, Schüle R, Sisodiya SM, Weckhuysen S, Lerche H, Lemke JR. Nat Genet. 2015;47(4):393-9.

12. Identification of Srp9 as a febrile seizure susceptibility gene. Hessel EV, de Wit M, Wolterink-Donselaar IG, Karst H, de Graaff E, van Lith HA, de Bruijn E, de Sonnaville S, Verbeek NE, Lindhout D, de Kovel CG, Koeleman BP, van Kempen M, Brilstra E, Cuppen E, Loos M, Spijker SS, Kan AA, Baars SE, van Rijen PC, Gosselaar PH, Groot Koerkamp MJ, Holstege FC, van Duijn C, Vergeer J, Moll HA, Taubøll E, Heuser K, Ramakers GM, Pasterkamp RJ, van Nieuwenhuizen O, Hoogenraad CC, Kas MJ, de Graan PN. Ann Clin Transl Neurol. 2014 Apr;1(4):239-50. doi: 10.1002/acn3.48.
13. The phenotypic spectrum of SCN8A encephalopathy. Larsen J, Carvill GL, Gardella E, Kluger G, Schmiedel G, Barisic N, Depienne C, Brilstra E, Mang Y, Nielsen JE, Kirkpatrick M, Goudie D, Goldman R, Jähn JA, Jepsen B, Gill D, Döcker M, Biskup S, McMahon JM, Koeleman B, Harris M, Braun K, de Kovel CG, Marini C, Specchio N, Djémié T, Weckhuysen S, Tommerup N, Troncoso M, Troncoso L, Bevot A, Wolff M, Hjalgrim H, Guerrini R, Scheffer IE, Mefford HC, Møller RS; EuroEPINOMICS RES Consortium CRP. Neurology. 2015 Feb 3;84(5):480-9.
14. Recessive loss-of-function mutations in AP4S1 cause mild fever-sensitive seizures, developmental delay and spastic paraplegia through loss of AP-4 complex assembly. Hardies K, May P, Djémié T, Tarta-Arsene O, Deconinck T, Craiu D; AR working group of the EuroEPINOMICS RES Consortium, Helbig I, Suls A, Balling R, Weckhuysen S, De Jonghe P, Hirst J. Hum Mol Genet. 2015 ;24(8):2218-27.
15. Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes. Schubert J, Siekierska A, Langlois M, May P, Huneau C, Becker F, Muhle H, Suls A, Lemke JR, de Kovel CG, Thiele H, Konrad K, Kawalia A, Toliat MR, Sander T, Rüschendorf F, Caliebe A, Nagel I, Kohl B, Kecskés A, Jacmin M, Hardies K, Weckhuysen S, Riesch E, Dorn T, Brilstra EH, Baulac S, Møller RS, Hjalgrim H, Koeleman BP; EuroEPINOMICS RES Consortium, Jurkat-Rott K, Lehman-Horn F, Roach JC, Glusman G, Hood L, Galas DJ, Martin B, de Witte PA, Biskup S, De Jonghe P, Helbig I, Balling R, Nürnberg P, Crawford AD, Esguerra CV, Weber YG, Lerche H. Nat Genet. 2014;46(12):1327-32.
16. De novo mutations in synaptic transmission genes including DNM1 cause epileptic encephalopathies. EuroEPINOMICS-RES Consortium; Epilepsy Phenome/Genome Project; Epi4K Consortium. Am J Hum Genet. 2014 ;95(4):360-70.
17. Characterization of a de novo SCN8A mutation in a patient with epileptic encephalopathy. de Kovel CG, Meisler MH, Brilstra EH, van Berkestijn FM, van 't Slot R, van Lieshout S, Nijman IJ, O'Brien JE, Hammer MF, Estacion M, Waxman SG, Dib-Hajj SD, Koeleman BP. Epilepsy Res. 2014 ;108(9):1511-8.
18. Identification of IL12RB1 as a novel systemic sclerosis susceptibility locus. López-Isac E, Bossini-Castillo L, Guerra SG, Denton C, Fonseca C, Assassi S, Zhou X, Mayes MD, Simeón CP, Ortego-Centeno N, Castellví I, Carreira P; Spanish Scleroderma Group, Gorlova O, Beretta L, Santaniello A, Lunardi C, Hesselstrand R, Nordin A, Riemekasten G, Witte T, Hunzelmann N, Kreuter A, Distler JH, Voskuyl AE, de Vries-Bouwstra J, Koeleman BP, Herrick A, Worthington J, Radstake TR, Martin J. Arthritis Rheumatol. 2014 ;66(12):3521-3.
19. Genetic determinants of common epilepsies: a meta-analysis of genome-wide association studies. International League Against Epilepsy Consortium on Complex Epilepsies. Electronic address: epilepsy-austin@unimelb.edu.au. Lancet Neurol. 2014 ;13(9):893-903.
20. Evaluation of genetic risk loci for intracranial aneurysms in sporadic arteriovenous malformations of the brain. Kremer PH, Koeleman BP, Pawlikowska L, Weinsheimer S, Bendjilali N, Sidney S, Zaroff JG, Rinkel GJ, van den Berg LH, Ruigrok YM, de Kort GA, Veldink JH, Kim H, Klijn CJ. J Neurol Neurosurg Psychiatry. 2015 ;86(5):524-9.

21. A genome-wide association study identifies a functional ERAP2 haplotype associated with birdshot chorioretinopathy. Kuiper JJ, Van Setten J, Ripke S, Van 'T Slot R, Mulder F, Missotten T, Baarsma GS, Francioli LC, Pulit SL, De Kovel CG, Ten Dam-Van Loon N, Den Hollander AI, Huis in het Veld P, Hoyng CB, Cordero-Coma M, Martín J, Llorenç V, Arya B, Thomas D, Bakker SC, Ophoff RA, Rothova A, De Bakker PI, Mutis T, Koeleman BP. Hum Mol Genet. 2014 ;23(22):6081-7.
22. 16p11.2 600 kb Duplications confer risk for typical and atypical Rolandic epilepsy. Reinthaler EM, Lal D, Lebon S, Hildebrand MS, Dahl HH, Regan BM, Feucht M, Steinböck H, Neophytou B, Ronen GM, Roche L, Gruber-Sedlmayr U, Geldner J, Haberlandt E, Hoffmann P, Herms S, Gieger C, Waldenberger M, Franke A, Wittig M, Schoch S, Becker AJ, Hahn A, Männik K, Toliat MR, Winterer G; 16p11.2 European Consortium, Lerche H, Nürnberg P, Mefford H, Scheffer IE, Berkovic SF, Beckmann JS; EPICURE Consortium; EuroEPINOMICS Consortium, Sander T, Jacquemont S, Reymond A, Zimprich F, Neubauer BA. Hum Mol Genet. 2014 ;23(22):6069-80.
23. A genetic variant in osteoprotegerin is associated with progression of joint destruction in rheumatoid arthritis. Knevel R, de Rooy DP, Saxne T, Lindqvist E, Leijsma MK, Daha NA, Koeleman BP, Tsonaka R, Houwing-Duistermaat JJ, Schonkeren JJ, Toes RE, Huizinga TW, Brouwer E, Wilson AG, van der Helm-van Mil AH. Arthritis Res Ther. 2014;16(3):R108.
24. De novo mutations in HCN1 cause early infantile epileptic encephalopathy. Nava C, Dalle C, Rastetter A, Striano P, de Kovel CG, Nabbout R, Cancès C, Ville D, Brilstra EH, Gobbi G, Raffo E, Bouteiller D, Marie Y, Trouillard O, Robbiano A, Keren B, Agher D, Roze E, Lesage S, Nicolas A, Brice A, Baulac M, Vogt C, El Hajj N, Schneider E, Suls A, Weckhuysen S, Gormley P, Lehesjoki AE, De Jonghe P, Helbig I, Baulac S, Zara F, Koeleman BP; EuroEPINOMICS RES Consortium, Haaf T, LeGuern E, Depienne C. Nat Genet. 2014;46(6):640-5.
25. Using ancestry-informative markers to identify fine structure across 15 populations of European origin. Huckins LM, Boraska V, Franklin CS, Floyd JA, Southam L; GCAN; WTCCC3, Sullivan PF, Bulik CM, Collier DA, Tyler-Smith C, Zeggini E, Tachmazidou I; GCAN; WTCCC3. Eur J Hum Genet. 2014;22(10):1190-200.
26. A genome-wide association study of rheumatoid arthritis without antibodies against citrullinated peptides. Bossini-Castillo L, de Kovel C, Kallberg H, van 't Slot R, Italiaander A, Coenen M, Tak PP, Posthumus MD, Wijmenga C, Huizinga T, van der Helm-van Mil AH, Stoeken-Rijsbergen G, Rodriguez-Rodriguez L, Balsa A, González-Álvaro I, González-Gay MÁ, Gómez-Vaquero C, Franke B; LifeLines Cohort Study, Vermeulen S, van der Horst-Bruinsma Ie, Dijkmans BA, Wolbink GJ, Ophoff RA, Maehlen MT, van Riel P, Merriman M, Klareskog L, Lie BA, Merriman T, Crusius JB, Brouwer E, Martin J, de Vries N, Toes R, Padyukov L, Koeleman BP. Ann Rheum Dis. 2015 ;74(3):e15.
27. A genome-wide association study of anorexia nervosa. Boraska V, Franklin CS, Floyd JA, Thornton LM, Huckins LM, Southam L, .., Koeleman BP, .., Ritchie GR, Barrett JC; Wellcome Trust Case Control Consortium 3, Estivill X, Hinney A, Sullivan PF, Collier DA, Zeggini E, Bulik CM. Mol Psychiatry. 2014 ;19(10):1085-94.
28. A genome-wide association study follow-up suggests a possible role for PPARG in systemic sclerosis susceptibility. López-Isac E, Bossini-Castillo L, Simeon CP, Egurbide MV, Alegre-Sancho JJ, Callejas JL, Roman-Ivorra JA, Freire M, Beretta L, Santaniello A, Airó P, Lunardi C, Hunzelmann N, Riemekasten G, Witte T, Kreuter A, Distler JH, Schuerwegh AJ, Vonk MC, Voskuyl AE, Shiels PG, van Laar JM, Fonseca C, Denton C, Herrick A, Worthington J, Assassi S, Koeleman BP, Mayes MD, Radstake TR, Martin J; Spanish Scleroderma Group. Arthritis Res Ther. 2014 ;16(1):R6.
29. Immunochip analysis identifies multiple susceptibility loci for systemic sclerosis. Mayes MD, Bossini-Castillo L, Gorlova O, Martin JE, Zhou X, Chen WV, Assassi S, Ying J, Tan FK, Arnett FC, Reveille JD, Guerra S, Teruel M, Carmona FD, Gregersen PK, Lee AT, López-Isac E, Ochoa E, Carreira P, Simeón CP, Castellví I, González-Gay MÁ; Spanish

Scleroderma Group, Zhernakova A, Padyukov L, Alarcón-Riquelme M, Wijmenga C, Brown M, Beretta L, Riemekasten G, Witte T, Hunzelmann N, Kreuter A, Distler JH, Voskuyl AE, Schuerwegh AJ, Hesselstrand R, Nordin A, Airó P, Lunardi C, Shiels P, van Laar JM, Herrick A, Worthington J, Denton C, Wigley FM, Hummers LK, Varga J, Hinchcliff ME, Baron M, Hudson M, Pope JE, Furst DE, Khanna D, Phillips K, Schiopu E, Segal BM, Molitor JA, Silver RM, Steen VD, Simms RW, Lafyatis RA, Fessler BJ, Frech TM, Alkassab F, Docherty P, Kaminska E, Khalidi N, Jones HN, Markland J, Robinson D, Broen J, Radstake TR, Fonseca C, Koeleman BP, Martin J. Am J Hum Genet. 2014 ;94(1):47-61.

30. Do inhibitory immune receptors play a role in the etiology of autoimmune disease? Olde Nordkamp MJ, Koeleman BP, Meyaard L. Clin Immunol. 2014 ;150(1):31-42.
31. Polymorphisms in ACVRL1 and endoglin genes are not associated with sporadic and HHT-related brain AVMs in Dutch patients. Boshuisen K, Brundel M, de Kovel CG, Letteboer TG, Rinkel GJ, Westermann CJ, Kim H, Pawlikowska L, Koeleman BP, Klijn CJ. Transl Stroke Res. 2013 ;4(3):375-8.
32. Structural genomic variation in childhood epilepsies with complex phenotypes. Helbig I, Swinkels ME, Aten E, Caliebe A, van 't Slot R, Boor R, von Spiczak S, Muhle H, Jähn JA, van Binsbergen E, van Nieuwenhuizen O, Jansen FE, Braun KP, de Haan GJ, Tommerup N, Stephani U, Hjalgrim H, Poot M, Lindhout D, Brilstra EH, Møller RS, Koeleman BP. Eur J Hum Genet. 2014 ;22(7):896-901.
33. Febrile temperatures unmask biophysical defects in Nav1.1 epilepsy mutations supportive of seizure initiation. Volkers L, Kahlig KM, Das JH, van Kempen MJ, Lindhout D, Koeleman BP, Rook MB. J Gen Physiol. 2013 ;142(6):641-53.
34. De novo loss-of-function mutations in CHD2 cause a fever-sensitive myoclonic epileptic encephalopathy sharing features with Dravet syndrome. Suls A, Jaehn JA, Kecskés A, Weber Y, Weckhuysen S, Craiu DC, Siekierska A, Djémié T, Afrikanova T, Gormley P, von Spiczak S, Kluger G, Iliescu CM, Talvik T, Talvik I, Meral C, Caglayan HS, Giraldez BG, Serratosa J, Lemke JR, Hoffman-Zacharska D, Szczepanik E, Barisic N, Komarek V, Hjalgrim H, Møller RS, Linnankivi T, Dimova P, Striano P, Zara F, Marini C, Guerrini R, Depienne C, Baulac S, Kuhlenbäumer G, Crawford AD, Lehesjoki AE, de Witte PA, Palotie A, Lerche H, Esguerra CV, De Jonghe P, Helbig I; EuroEPINOMICS RES Consortium. Am J Hum Genet. 2013;93(5):967-75.
35. A genome-wide investigation of copy number variation in patients with sporadic brain arteriovenous malformation. Bendjilali N, Kim H, Weinsheimer S, Guo DE, Kwok PY, Zaroff JG, Sidney S, Lawton MT, McCulloch CE, Koeleman BP, Klijn CJ, Young WL, Pawlikowska L. PLoS One. 2013;8(10):e71434.
36. Epilepsy, hippocampal sclerosis and febrile seizures linked by common genetic variation around SCN1A. Kasperaviciute D, Catarino CB, Matarin M, Leu C, Novy J, Tostevin A, Leal B, Hessel EV, Hallmann K, Hildebrand MS, Dahl HH, Ryten M, Trabzuni D, Ramasamy A, Alhusaini S, Doherty CP, Dorn T, Hansen J, Krämer G, Steinhoff BJ, Zumsteg D, Duncan S, Kälviäinen RK, Eriksson KJ, Kantanen AM, Pandolfo M, Gruber-Sedlmayr U, Schlachter K, Reinthaler EM, Stogmann E, Zimprich F, Théâtre E, Smith C, O'Brien TJ, Meng Tan K, Petrovski S, Robbiano A, Paravidino R, Zara F, Striano P, Sperling MR, Buono RJ, Hakonarson H, Chaves J, Costa PP, Silva BM, da Silva AM, de Graan PN, Koeleman BP, Becker A, Schoch S, von Lehe M, Reif PS, Rosenow F, Becker F, Weber Y, Lerche H, Rössler K, Buchfelder M, Hamer HM, Kobow K, Coras R, Blumcke I, Scheffer IE, Berkovic SF, Weale ME; UK Brain Expression Consortium, Delanty N, Depondt C, Cavalleri GL, Kunz WS, Sisodiya SM. Brain. 2013;136(Pt 10):3140-50.
37. Consensus on diagnosis and management of JME: From founder's observations to current trends. Kasteleijn-Nolst Trenité DG, Schmitz B, Janz D, Delgado-Escueta AV, Thomas P, Hirsch E, Lerche H, Camfield C, Baykan B, Feucht M, Martínez-Juárez IE, Duron RM, Medina MT, Rubboli G, Jerney J, Hermann B, Yacubian E, Koutroumanidis M, Stephani U, Salas-Puig J, Reed RC, Woermann F, Wandschneider B, Bureau M, Gambardella A, Koepp

MJ, Gelisse P, Gurses C, Crespel A, Nguyen-Michel VH, Ferlazzo E, Grisar T, Helbig I, Koeleman BP, Striano P, Trimble M, Buono R, Cossette P, Represa A, Dravet C, Serafini A, Berglund IS, Sisodiya SM, Yamakawa K, Genton P. Epilepsy Behav. 2013;28 Suppl 1:S87-90.

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