

1. Ahmadiharchegani F, Tobin RA, Degan C, Naveed A, Guglieri M, Jiménez-Requena A, de Vries SI, Szgyarto CAK, Spitali P, Tsonaka R, van der Burgt YEM, Diaz-Manera J, VBP15-004 investigators CINRG DNHS investigators, FOR-DMD investigators of the Muscle Study Group, Clemens PR, Hoffman EP, Dang UJ, Hathout Y (2025) Circulating protein biomarkers identified in two independent clinical trial cohorts of glucocorticoid-naive Duchenne muscular dystrophy patients. *SCI REP-UK* 15(1): 39997, doi: 10.1038/s41598-025-23758-6
2. Akkuratov EE, Sorrell F, Picton LD, Sousa VC, Paucar M, Jans D, Svensson LB, Lindskog M, Fritz N, Liebmann T, Sillar KT, Rosewich H, Svenningsson P, Brismar H, Miles GB, Aperia A (2025) ATP1A3 dysfunction causes motor hyperexcitability and afterhyperpolarization loss in a dystonia model. *BRAIN* 148(4): 1099-1105, doi: 10.1093/brain/awae373
3. Becker B, Cordts I, Becker J, Günther R, Baumann M, Bernert G, Eisenkölbl A, Fiedler B, Flotats-Bastardas M, Fieger M, Hagenacker T, Hahn A, Hobbiebrunken E, Bevo A, Jahnel J, Johannsen J, Kamm C, Koch JC, Köhler C, Kölbl H, Müller-Felber W, Neuwirth C, Plecko B, Stadler C, Smitka M, Von Moers A, Trollmann R, Weiler M, Ziegler A, Goldbach S, Probst-Schendzielorz K, Lochmüller H, Schara-Schmidt U, Walter MC, Kirschner J, Wirth B, Pechmann A, Deschauer M, with SMARtCARE study group (2025) Phenotypic intrafamilial variability of 5q-associated spinal muscular atrophy: A systematic multicentre sibling study. *J NEUROMUSCULAR DIS* 0: 22143602251370577, doi: 10.1177/22143602251370577
4. Becker M, Weiskorn J, Wiegand S, Röbl M, Meissner T, Schulz E, Golembowski S, Schwab KO, Meraner D, Holl RW (2025) Familial Hypercholesterolemia in Pediatric Patients With Type 1 Diabetes: Double Challenge for Diagnosis and Treatment. *DIABETES CARE* 48(12): 2119-2126, doi: 10.2337/dc25-1195
5. Böckmann I, Leifheit-Nestler M, Rehberg M, Spartà G, Evers K, Schlingmann KP, Kemper MJ, Richter-Unruh A, Hiort O, Grohmann-Held K, Derichs U, Freiberg C, Weitz M, Dunstheimer D, Schmid E, John-Kroegel U, Metzinger O, Heger S, Jorch N, Staude H, Patzer L, Wühl E, Zivicnjak M, Schnabel D, Haffner D, German Society for Pediatric Nephrology (GPN) and the German Society for Pediatric and Adolescent Endocrinology and Diabetology (DGPAED) (2025) Office Blood Pressure and Obesity in Children with X-Linked Hypophosphatemia. *CALCIFIED TISSUE INT* 116(1): 56, doi: 10.1007/s00223-025-01363-z
6. Boeckhaus J, Tönshoff B, Weber LT, Haffner D, Pape L, Latta K, Fehrenbach H, Lange-Sperandio B, Kettwig M, König S, John-Kroegel U, Gellermann J, Galiano M, Hafke A, Streit F, Gross O (2025) High adherence to angiotensin-converting enzyme inhibitor in children and adolescents with Alport syndrome: objective verification using liquid chromatography-mass spectrometry. *PEDIATR NEPHROL* NA: 1-10, doi: 10.1007/s00467-025-07053-0
7. Boeckhaus J, Tönshoff B, Weber LT, Pape L, Latta K, Fehrenbach H, Lange-Sperandio B, Kettwig M, König S, John-Kroegel U, Gellermann J, Galiano M, Jami S, Pieper D, Dihazi GH, Hafke A, Kohl S, Liebau MC, König J, Haffner D, Gross O, Wallbach M (2025) Urinary Dickkopf-related protein 3 as a novel biomarker for kidney function decline in children with Alport syndrome. *PEDIATR NEPHROL* 40(7): 2205-2213, doi: 10.1007/s00467-025-06696-3
8. Bondarenko MS, Kuseyri Hübschmann O, Kulhánek J, Pons R, Pearson TS, Jeltsch K, Badnjarevic I, Wassenberg T, Horvath G, Stevanovic G, Kurian MA, Cortès-Saladelafont E, Roubertie A, Leuzzi V, Bertoldi M, Mastrangelo M, Assmann B, THD Guidelines Working Group, Garcia-Cazorla A, Opladen T (2025) Consensus Guideline for the Diagnosis and Treatment of Tyrosine Hydroxylase (TH) Deficiency. *J INHERIT METAB DIS* 48(6): e70106, doi: 10.1002/jimd.70106
9. Brieger LC, Przygodda S, Bohlen AV, Rehberg M, Konrad M, Schlingmann KP, Hiort O, Schmidt D, John-Kroegel U, Wuehl E, Kemper MJ, Derichs U, Patzer L, Albers N, Dunstheimer D, Heger S, Grohmann-Held K, Schroeder C, Jorch N, Schmid E, Staude H, Weitz M, Freiberg C, Huebner A, Heitmeyer-Pyper A, Sparta G, Partsch CJ, Marx M, Land C, Baus I, Wilkening F, Moeller K, Simic-Schleicher G, Empting S, Metzinger O, Wagner V, Holder M, Žebec MS, Schnabel D, Haffner D, Zivicnjak M, on behalf of, German Society for Pediatric Nephrology (GPN), German Society for Pediatric, Adolescent Endocrinology Diabetology (DGKED) (2025) Growth dynamics of transversal body dimensions and proportions, with related clinical determinants in children with X-linked hypophosphatemia treated with phosphate supplements and active vitamin D. *PEDIATR NEPHROL* 40(10): 3187-3200, doi: 10.1007/s00467-025-06841-y
10. Cordts I, Fuetterer C, Wachinger A, von Heynitz R, Kessler T, Freigang M, Quinten AL, Bjelica B, Brakemeier S, Hobbiebrunken E, Hagenacker T, Petri S, Koch JC, Hahn A, Lingor P, Deschauer M, Günther R, Weiler M, Haller B, Feneberg E (2025) Long-Term Dynamics of CSF and Serum Neurofilament Light Chain in Adult Patients With 5q Spinal Muscular Atrophy Treated With Nusinersen. *NEUROLOGY* 104(5): e213371, doi: 10.1212/WNL.0000000000213371

11. Debatin KM, Gärtner J, Klein C, Körner A, Mall MA, Muntau AC, van den Berg N (2025) The German Center for Child and Adolescent Health - A new structure for translational research in pediatrics shaping the health of children today and future generations. *MOL CELL PEDIATR* 12(1): 12, doi: 10.1186/s40348-025-00198-w
12. Dekker J, Schot R, Aldinger KA, Everman DB, Washington C, Jones JR, Sullivan JA, Spillmann RC, Shashi V, Vitobello A, Denommé-Pichon AS, Mosca-Boidron AL, Perrin L, Auvin S, Zaki MS, Gleeson JG, Meave N, Wallace C, Nambot S, Delanne J, Ruggiero SM, Helbig I, Fitzgerald MP, Leventer RJ, Grange DK, Argilli E, Sherr EH, Prakash S, Neilson DE, Nicita F, Sferra A, Bertini ES, Aiello C, Brockmann K, Kuranov AB, Kaulfuss S, Basit S, Alluqmani M, Almatrafi A, Friedman JM, Guimond C, Mohammed F, Sharma P, Goel D, Wirth T, Anheim M, Bahena P, Koparir A, Kolokotronis K, Vona B, Haaf T, Kunstmann E, Maroofian R, Sczakiel HL, Boschann F, Misra-Isrie M, Louie RJ, Stoleran ES, Sanchez-Lara PA, Mergler S, Oegema R, Zarate YA, Kariminejad A, Tajsharghi H, Zeidler S, Kievit AJA, Bouman A, Cappuccio G, Brunetti-Pierri N, Stuurman KE, Swols DM, Tekin M, Upadia J, Martin DM, Craven D, Hiatt SM, van de Pol LA, D'Arco F, Margot H, Wilke M, Yousefi S, Barakat TS, van Veghel-Plandsoen MM, Aronica E, Anink J, Rogers SL, Slep KC, Doherty D, Dobyns WB, Mancini GMS (2025) A clinical and genotype-phenotype analysis of MACF1 variants. *AM J HUM GENET* 112(10): 2363-2380, doi: 10.1016/j.ajhg.2025.08.010
13. Dibaj P, Harun AA (2025) Follow-Up of Neurosarcoidosis With Longitudinally Extensive Myelitis: A Case Report and Review of the Literature. *Cureus* 17(8): e90238, doi: 10.7759/cureus.90238
14. Disse S, Ramantani G, Küpper H, Bock A, Korenke GC, Weidner B, Preisel M, Trollmann R, Wiemer-Kruel A, Wellmann S, Brockmann K, Schroeder S, Meyer S (2025) Sturge Weber syndrome in a multinational pediatric cohort: a systematic analysis of different types. *ORPHANET J RARE DIS* 20(1): 336, doi: 10.1186/s13023-025-03769-2
15. Disse SC, Küpper H, Bock A, Korenke GC, Ramantani G, Weidner B, Preisel M, Trollmann R, Wiemer-Kruel A, Brockmann K, Schroeder S, Meyer S (2025) The natural history of pediatric Sturge-Weber Syndrome: A multinational cross-sectional study. *EUR J PAEDIATR NEURO* 54: 200-209, doi: 10.1016/j.ejpn.2025.02.004
16. Driedger JH, Schröter J, PROTECT-Study Group, Syrbe S, Saffari A (2025) Long-term neuropsychologic outcome of pre-emptive mTOR inhibitor treatment in children with tuberous sclerosis complex (TSC) under 4 months of age (PROTECT), a two-arm, randomized, observer-blind, controlled phase IIb national multicentre clinical trial: study protocol. *ORPHANET J RARE DIS* 20(1): 2, doi: 10.1186/s13023-024-03495-1
17. Frosch M, Shimizu T, Wogram E, Amann L, Gruber L, Groisman AI, Fliegauf M, Schwabenland M, Chhatbar C, Zechel S, Rosewich H, Gärtner J, Quintana FJ, Buescher JM, Blank T, Binder H, Stadelmann C, Letzku JJ, Hopf C, Masuda T, Knobloch KP, Prinz M (2025) Microglia-neuron crosstalk through Hex-GM2-MGL2 maintains brain homeostasis. *NATURE* 646(8086): 913-924, doi: 10.1038/s41586-025-09477-y
18. García-Cazorla Á, Sevin C, Constante JR, Yazbeck E, Rosewich H, Jimenez S, Chia-Yi Chiang G, Rapalino O, Caruso P, Balentine D, Helmer KG, Bennett S, Emanuele M, Rodriguez-Pascual L, Pizcueta P, Pina G, Vilà A, Rovira M, Mantilla A, Meya U, Mistry A, Pascual M, Pascual S, Martinell M, Musolino PL, Mallack E (2025) Safety and efficacy of leriglitazone in childhood cerebral adrenoleukodystrophy (NEXUS): an interim analysis of an open-label, phase 2/3 trial. *ECLINICALMEDICINE* 84: 103265, doi: 10.1016/j.eclinm.2025.103265
19. Garnica-Agudelo D, Smith SDW, van de Velden D, Stier C, Brockmann K, Schroeder S, Neef NE, Focke NK (2025) Source reconstruction of clinical resting-state EEG reveals differences in power and functional connectivity in children with developmental dyslexia. *NEUROPSYCHOLOGIA* 219: 109289, doi: 10.1016/j.neuropsychologia.2025.109289
20. Garnica-Agudelo D, Smith SDW, van de Velden D, Weise D, Brockmann K, Focke NK (2025) Increase in EEG functional connectivity and power during wakefulness in self-limited epilepsy with centrotemporal spikes. *CLIN NEUROPHYSIOL* 171: 107-123, doi: 10.1016/j.clinph.2024.12.028
21. Gauß KF, Krätzner R, Budde K, Hafke A, Henning AK, Fenzlaff M, Friedrich N, Nauck M, Petersmann A (2025) Comprehensive utilization of NMR spectra—evaluation of NMR-based quantification of amino acids for research and patient care. *J LAB MED* -: -
22. Gavazzi F, Yu E, Tashnim Z, Woidill S, Sevagamoorthy A, Arnold K, Ammann-Schnell L, Groeschel S, Krägeloh-Mann I, Breitling V, Schlotawa L, Ahrens-Nicklas R, Adang LA (2025) Exploration Into Lived Experiences of Multiple Sulfatase Deficiency-Affected Individuals and Their Families. *J CHILD NEUROL* 40(10): 852-861, doi: 10.1177/08830738251339848

23. Groeschel S, [Rosewich H](#) (2025) Seizures in childhood cerebral adrenoleukodystrophy: New insights and remaining knowledge gaps. *DEV MED CHILD NEUROL* 67(11): 1381-1382, doi: 10.1111/dmcn.16352
24. Hirn E, [Huppke B](#), Wilken B, Kiehnopf M, Huppke P (2025) Rett Syndrome: Specific MECP2 Variants are Associated With Elevated Serum Neurofilament Light Chain. *PEDIATR NEUROL* 172: 1-7, doi: 10.1016/j.pediatrneurol.2025.07.016
25. Hummel-Abmeier H, Naxer S, Kadas EM, Zimmermann H, Knaack B, Huppke P, Kowallick A, [Meier K](#), Brandt AU, Paul F, Schittkowski M, Oertel FC, [Gärtner J](#) (2025) The Inner Nuclear Layer in Pediatric Multiple Sclerosis. *NEUROL-NEUROIMMUNOL* 12(3): e200387, doi: 10.1212/NXI.000000000200387
26. [Huppke B](#), Vries Hd, Blaschek A, Huppke P (2025) Severe Disease Activation after Fingolimod Discontinuation in a Pediatric Multiple Sclerosis Patient: A Case Report and Literature Review. *NEUROPEDIATRICS* 56(2): 147-150, doi: 10.1055/a-2496-5294
27. Kokaly N, Guerreiro H, Bredow J, Dreha-Kulaczewski S, [Ohlenbusch A](#), Köhler W, Reinhardt T, Schön G, Volk AE, Sigel H, Bley A (2025) Description of the Hamburg Alexander Leukodystrophy Cohort-Insights into Practical Classification and the Care Situation. *J CLIN MED* 14(19): 1, doi: 10.3390/jcm14196918
28. Laue T, Junge N, [Leiskau C](#), Mutschler F, Ohlendorf J, Baumann U (2025) Effectiveness of hepatitis A immunization after pediatric liver transplantation: A retrospective observational analysis. *AM J TRANSPLANT* 25(5): 1086-1097, doi: 10.1016/j.ajt.2024.12.009
29. Laugwitz L, Buchert R, Olgún P, Estiar MA, Atanasova M, Jr WM, Enssle J, Marsden B, Avilés J, González-Gutiérrez A, Candia N, Fabiano M, Morlot S, Peralta S, Groh A, Schillinger C, Kuehn C, Sofan L, Sturm M, Bender B, Tomaselli PJ, Diebold U, Mueller AJ, Spranger S, Fuchs M, Freua F, Melo US, Mattas L, Ashtiani S, Suchowersky O, Groeschel S, Rouleau GA, Yosovich K, Michelson M, Leibovitz Z, Bilal M, Uctepe E, Yesilyurt A, Ozdogan O, Celik T, Krägeloh-Mann I, Riess O, [Rosewich H](#), Umair M, Lev D, Zuchner S, Schweizer U, Lynch DS, Gan-Or Z, Haack TB (2025) EEFSEC deficiency: A selenopathy with early-onset neurodegeneration. *AM J HUM GENET* 112(1): 168-180, doi: 10.1016/j.ajhg.2024.12.001
30. Paciello LM, Quante M, [Rosewich H](#), Shellhaas RA (2025) The role of sleep in neonatal neurocritical care and the influence on long-term outcome. *SEMIN PERINATOL* 49(8): 152127, doi: 10.1016/j.semperi.2025.152127
31. Planas-Serra L, Rodríguez-Ruiz M, Anderson EN, Rodríguez-Palmero A, Vélez-Santamaria V, Schlüter A, Verdura E, Gereñu G, Jiménez-Zúñiga A, Iñáñez A, Casas J, Bech JJ, De La Torre C, Martínez JJ, Ruiz M, Fourcade S, Iascone M, Tenconi R, [Meier K](#), [Diegmann S](#), Lee RHC, Beland B, Mir A, Darvish H, Chung W, Karimiani EG, Leal SM, Schrauwen I, Öhman S, Järvelä I, Granvik J, Reinson K, Kurvinen E, Öunap K, Schwan A, Platzer K, Kalayci T, Sharifi S, Korenke GC, Houlden H, Maroofian R, López de Munáin A, Casasnovas C, Pandey UB, Pujol A (2025) Bi-allelic variants in the ribosomal protein RPS6KC1 cause a complex neurodevelopmental disorder. *AM J HUM GENET* 112(11): 2643-2664, doi: 10.1016/j.ajhg.2025.09.015
32. Safavi-Abbasi S, Venezia E, Sughrue M, [Dibaj P](#) (2025) Effectiveness of mindfulness meditation on the quality of life, pain intensity, mobility and physical function in adults with chronic low back pain: a systematic review and metaanalysis. *DISCOV PSYCHOL* 5 - 183: 1
33. Stoldt S, Maass F, Weber M, Dennerlein S, Ilgen P, [Gärtner J](#), Canfes A, Schweighofer SV, Jans DC, Rehling P, Jakobs S (2025) Super-resolution microscopy of mitochondrial mRNAs. *NAT COMMUN* 16(1): 6391, doi: 10.1038/s41467-025-61577-5
34. Tricoli L, Sase S, Hacker JL, Pham V, Chappell M, Breda L, Hurwitz SN, Tanaka N, Castruccio Castracani C, Guerra A, Hou Z, [Schlotawa L](#), Radhakrishnan K, Hogenauer M, Roche A, Everett J, Bushman F, Kurre P, Ahrens-Nicklas R, Adang LA, Vanderver AL, Rivella S (2025) Effective gene therapy for metachromatic leukodystrophy achieved with minimal lentiviral genomic integrations. *MOL THER-NUCL ACIDS* 36(1): 102464, doi: 10.1016/j.omtn.2025.102464
35. van der Ham M, Hoytema van Konijnenburg E, van Rossum W, Gerrits J, van Hasselt P, Prinsen H, Jans J, [Schlotawa L](#), Laugwitz L, de Sain-van der Velden M (2025) Profiling and semi-quantitation of urine sulfatides by UHPLC-Orbitrap-HRMS. *ANAL CHIM ACTA* 1350: 343824, doi: 10.1016/j.aca.2025.343824
36. von Steinbuechel N, Zeldovich M, Bockhop F, Krenz U, Timmermann D, Buchheim A, Koerte IK, Bonfert MV, Berweck S, Kieslich M, [Brockmann K](#), Roediger M, Greving S, Neu A, Wartemann U, Suss J, Auer C, Muehlan H, Cunitz K (2025) Quality of Life after Brain Injury in children aged six and seven years (QOLIBRI-KIDDY) - development and scale analysis of the first disease-specific self-report instrument for young children after traumatic brain injury. *J PATIENT-REP OUTCOM* 9(1): 53, doi: 10.1186/s41687-025-00890-5

37. von Steinbuechel N, Zeldovich M, Holloway I, Mayer AC, Rojczyk P, Krenz U, Koerte IK, Bonfert MV, Berweck S, Kieslich M, Brockmann K, Roediger M, Lendt M, Staebler M, Auer C, Neu A, Kaiser A, Driemeyer J, Wartemann U, Pinggera D, Thomé C, Schoen V, Geiger P, Suss J, Buchheim A, Muehlan H, Cunitz K (2025) Quality of life after brain injury in children and adolescents questionnaire - validation of the proxy version (QOLIBRI-KID/ADO-Proxy). BRAIN INJURY 39(9): 742-757, doi: 10.1080/02699052.2025.2481435
38. Weiß C, Vill K, Baumann M, Bernert G, Blaschek A, Eisenkölbl A, Flotats-Bastardas M, Friese J, Ganter C, Goldhahn K, Hahn A, von der Hagen M, Hartmann H, Hasselmann O, Horber V, Husain RA, Illsinger S, Jacquier D, Johannsen J, Köhler C, Kölbl H, Kolodzig M, Klein A, Pechmann A, von Moers A, Müller-Felber W, Rauscher C, Schara-Schmidt U, Schreiber G, Schwartz O, Sproß J, Stettner GM, Stoltenburg C, Stumpe E, Trollmann R, Wiegand G, Wilichowski E, Kirschner J, Ziegler A, collaborators and members of the INTEGRATE ATMP consortium (2025) Delphi consensus on gene therapy of spinal muscular atrophy with onasemnogene abeparvovec in Germany, Austria and Switzerland-part I-systematic literature review and existing evidence. J NEUROMUSCULAR DIS 1: 22143602251387019, doi: 10.1177/22143602251387019
39. Wendland K, Irsfeld M, Schreiber K, Ternka K, Stadelmann C, Nessler S, Gärtner J, Kettwig M (2025) Unlocking microglia pyroptosis in a model of type I interferon-driven neuroinflammation: lessons from Rnaset2^{-/-} mice. CELL DEATH DIS 17(1): 138, doi: 10.1038/s41419-025-08350-0
40. Wilpert NM, Hewitt AL, Pons R, Henke MT, Dell'Orco A, Bauer M, Grolik C, Menz S, Wahle M, Zink A, Prigione A, Reinauer C, Lange C, Furth C, Brockmann K, Jung-Klawitter S, Christ S, Kaindl AM, Tietze A, Krude H, Opladen T, Schuelke M (2025) Patients with Allan-Herndon-Dudley Syndrome (MCT8 Deficiency) Display Symptoms of Parkinsonism in Childhood and Respond to Levodopa/Carbidopa Treatment. MOVEMENT DISORD 40(5): 938-949, doi: 10.1002/mds.30152
41. Wong KM, Maroofian R, Meier K, Diegmann S, Tkemaladze T, Alvi JR, Tasharrofi B, Efthymiou S, Munchau A, Korenke GC, Almontashiri N, Eyaid W, Kashgari A, Alotaibi M, Gärtner J, Huppke B, Asadollahi M, Chikvinidze G, Keramatipour M, Sultan T, Thiele H, Nürnberg P, Gräler MH, Houlden H, Huppke P (2025) Biallelic ELOVL1 Variants Are Linked to Hypomyelinating Leukodystrophy, Movement Disorder, and Ichthyosis. MOVEMENT DISORD 40(9): 1836-1850, doi: 10.1002/mds.30258
42. Zeldovich M, Krol L, Koerte IK, Cunitz K, Kieslich M, Henrich M, Brockmann K, Buchheim A, Lendt M, Auer C, Neu A, Driemeyer J, Wartemann U, Thomé C, Pinggera D, Berweck S, Bonfert MV, Suss J, Muehlan H, Steinbuechel Nv (2025) Correction: A short scale to measure health-related quality of life after traumatic brain injury in children and adolescents (QOLIBRI-OS-KID/ADO): psychometric properties and German reference values. QUAL LIFE RES 34(1): 287-288, doi: 10.1007/s11136-024-03845-3