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Publications

Original Research Publications (166)

Wimmer K, Schamschula E, Wernstedt A, Traunfellner P, Amberger A, Zschocke J, Kroisel P, Chen Y, Callens T, Messiaen L (2020) AG-exclusion zone revisited: Lessons to learn from 91 intronic NF1 3' splice site mutations outside the canonical AG-dinucleotides. Hum Mutat 41:1145-1156.

Seidl-Philipp M, Schatz UA, Gasslitter I, Moosbrugger-Martinz V, Blunder S, Schossig AS, Zschocke J, Schmuth M, Gruber R (2020) Spectrum of ichthyoses in an Austrian ichthyosis cohort from 2004 to 2017. J Dtsch Dermatol Ges 18:17-25.

Schoner K, Witsch-Baumgartner M, Behunova J, Petrovic R, Bald R, Kircher SG, Ramaswamy A, Kluge B, Meyer-Wittkopf M, Schmitz R, Fritz B, Zschocke J, Laccone F, Rehder H (2020) Smith-Lemli-Opitz syndrome - Fetal phenotypes with special reference to the syndrome-specific internal malformation pattern. Birth Defects Res 112:175-185.

Perez-Valencia JA, Gallon R, Chen Y, Koch J, Keller M, Oberhuber K, Gomes A, Zschocke J, Burn J, Jackson MS, Santibanez-Koref M, Messiaen L, Wimmer K (2020) Constitutional mismatch repair deficiency is the diagnosis in 0.41% of pathogenic NF1/SPRED1 variant negative children suspected of sporadic neurofibromatosis type 1. Genet Med. In press

Oemer G, Koch J, Wohlfarter Y, Alam MT, Lackner K, Sailer S, Neumann L, Lindner HH, Watschinger K, Haltmeier M, Werner ER, Zschocke J, Keller MA (2020) Phospholipid Acyl Chain Diversity Controls the Tissue-Specific Assembly of Mitochondrial Cardiolipins. Cell Rep 30:4281-4291.e4284.

Koch J, Lackner K, Wohlfarter Y, Sailer S, Zschocke J, Werner ER, Watschinger K, Keller MA (2020) Unequivocal Mapping of Molecular Ether Lipid Species by LC-MS/MS in Plasmalogen-Deficient Mice. Anal Chem 92:11268-11276.

Kapferer-Seebacher I, Schnabl D, Zschocke J, Pope FM (2020) Dental Manifestations of Ehlers-Danlos Syndromes: A Systematic Review. Acta Derm Venereol 100:adv00092.

Hillert A, Anikster Y, Belanger-Quintana A, Burlina A, Burton BK, Carducci C, Chiesa AE, Christodoulou J, Đorđević M, Desviat LR, Eliyahu A, Evers RAF, Fajkusova L, Feillet F, Bonfim-Freitas PE, Giżewska M, Gundorova P, Karall D, Kneller K, Kutsev SI, Leuzzi V, Levy HL, Lichter-Konecki U, Muntau AC, Namour F, Oltarzewski M, Paras A, Perez B, Polak E, Polyakov AV, Porta F, Rohrbach M, Scholl-Bürgi S, Spécola N, Stojiljković M, Shen N, Santana-da Silva LC, Skouma A, van Spronsen F, Stoppioni V, Thöny B, Trefz FK, Vockley J, Yu Y, Zschocke J, Hoffmann GF, Garbade SF, Blau N (2020) The Genetic Landscape and Epidemiology of Phenylketonuria. Am J Hum Genet 107:234-250.

Weustenfeld M, Eidelpes R, Schmuth M, Rizzo WB, Zschocke J, Keller MA (2019) Genotype and phenotype variability in Sjögren-Larsson syndrome. Hum Mutat 40:177-186.

Peters V, Morath M, Mack M, Liesert M, Buckel W, Hoffmann GF, Vockley J, Ghisla S, Zschocke J (2019) Formation of 3-hydroxyglutaric acid in glutaric aciduria type I: in vitro participation of medium chain acyl-CoA dehydrogenase. JIMD Rep 47:30-34.

Lima Cunha D, Alakloby OM, Gruber R, Kakar N, Ahmad J, Alawbathani S, Plank R, Eckl K, Krabichler B, Altmüller J, Nürnberg P, Zschocke J, Borck G, Schmuth M, Alabdulkareem AS, Abdulaziz Alnutaifi K, Hennies HC (2019) Unknown mutations and genotype/phenotype correlations of autosomal recessive congenital ichthyosis in patients from Saudi Arabia and Pakistan. Mol Genet Genomic Med 7:e539.

Kapferer-Seebacher I, Waisfisz Q, Boesch S, Bronk M, van Tintelen P, Gizewski ER, Groebner R, Zschocke J, van der Knaap MS (2019) Periodontal Ehlers-Danlos syndrome is associated with leukoencephalopathy. Neurogenetics 20:1-8.

Huber A, Oemer G, Malanovic N, Lohner K, Kovács L, Salvenmoser W, Zschocke J, Keller MA, Marx F (2019) Membrane Sphingolipids Regulate the Fitness and Antifungal Protein Susceptibility of Neurospora crassa. Front Microbiol 10:605.

Hsieh TC, Mensah MA, Pantel JT, Aguilar D, Bar O, Bayat A, Becerra-Solano L, Bentzen HB, Biskup S, Borisov O, Braaten O, Ciaccio C, Coutelier M, Cremer K, Danyel M, Daschkey S, Eden HD, Devriendt K, Wilson S, Douzgou S, Đukić D, Ehmke N, Fauth C, Fischer-Zirnsak B, Fleischer N, Gabriel H, Graul-Neumann L, Gripp KW, Gurovich Y, Gusina A, Haddad N, Hajjir N, Hanani Y, Hertzberg J, Hoertnagel K, Howell J, Ivanovski I, Kaindl A, Kamphans T, Kamphausen S, Karimov C, Kathom H, Keryan A, Knaus A, Köhler S, Kornak U, Lavrov A, Leitheiser M, Lyon GJ, Mangold E, Reina PM, Carrascal AM, Mitter D, Herrador LM, Nadav G, Nöthen M, Orrico A, Ott CE, Park K, Peterlin B, Pölsler L, Raas-Rothschild A, Randolph L, Revencu N, Fagerberg CR, Robinson PN, Rosnev S, Rudnik S, Rudolf G, Schatz U, Schossig A, Schubach M, Shanoon O, Sheridan E, Smirin-Yosef P, Spielmann M, Suk EK, Sznajer Y, Thiel CT, Thiel G, Verloes A, Vrecar I, Wahl D, Weber I, Winter K, Wiśniewska M, Wollnik B, Yeung MW, Zhao M, Zhu N, Zschocke J, Mundlos S, Horn D, Krawitz PM (2019) PEDIA: prioritization of exome data by image analysis. Genet Med 21:2807-2814.

Gröbner R, Kapferer-Seebacher I, Amberger A, Redolfi R, Dalonneau F, Björck E, Milnes D, Bally I, Rossi V, Thielens N, Stoiber H, Gaboriaud C, Zschocke J (2019) C1R Mutations Trigger Constitutive Complement 1 Activation in Periodontal Ehlers-Danlos Syndrome. Front Immunol 10:2537.

Gallon R, Mühlegger B, Wenzel SS, Sheth H, Hayes C, Aretz S, Dahan K, Foulkes W, Kratz CP, Ripperger T, Azizi AA, Baris Feldman H, Chong AL, Demirsoy U, Florkin B, Imschweiler T, Januszkiewicz-Lewandowska D, Lobitz S, Nathrath M, Pander HJ, Perez-Alonso V, Perne C, Ragab I, Rosenbaum T, Rueda D, Seidel MG, Suerink M, Taeubner J, Zimmermann SY, Zschocke J, Borthwick GM, Burn J, Jackson MS, Santibanez-Koref M, Wimmer K (2019) A sensitive and scalable microsatellite instability assay to diagnose constitutional mismatch repair deficiency by sequencing of peripheral blood leukocytes. Hum Mutat 40:649-655.

Bally I, Dalonneau F, Chouquet A, Gröbner R, Amberger A, Kapferer-Seebacher I, Stoiber H, Zschocke J, Thielens NM, Rossi V, Gaboriaud C (2019) Two Different Missense C1S Mutations, Associated to Periodontal Ehlers-Danlos Syndrome, Lead to Identical Molecular Outcomes. Front Immunol 10:2962.

Wolf NI, Zschocke J, Jakobs C, Rating D, Hoffmann GF (2018) ATAD1 encephalopathy and stiff baby syndrome: a recognizable clinical presentation. Brain 141:e49.

Weigand T, Singler B, Fleming T, Nawroth P, Klika KD, Thiel C, Baelde H, Garbade SF, Wagner AH, Hecker M, Yard BA, Amberger A, Zschocke J, Schmitt CP, Peters V (2018) Carnosine Catalyzes the Formation of the Oligo/Polymeric Products of Methylglyoxal. Cell Physiol Biochem 46:713-726.

Watschinger K, Keller MA, Golderer G, Coassin S, Zschocke J, Werner ER (2018) Biochemical Characterization of AGMO Variants Implicated in Relapses in Visceral Leishmaniasis. J Infect Dis 217:1846-1847.

Tesch VK, H IJ, Raicht A, Rueda D, Dominguez-Pinilla N, Allende LM, Colas C, Rosenbaum T, Ilencikova D, Baris HN, Nathrath MHM, Suerink M, Januszkiewicz-Lewandowska D, Ragab I, Azizi AA, Wenzel SS, Zschocke J, Schwinger W, Kloor M, Blattmann C, Brugieres L, van der Burg M, Wimmer K, Seidel MG (2018) No Overt Clinical Immunodeficiency Despite Immune Biological Abnormalities in Patients With Constitutional Mismatch Repair Deficiency. Front Immunol 9:1506.

Rinner A, Zschocke J, Schossig A, Gröbner R, Strobl H, Kapferer-Seebacher I (2018) High risk of peri-implant disease in periodontal Ehlers-Danlos Syndrome. A case series. Clin Oral Implants Res 29:1101-1106.

Oerum S, Roovers M, Rambo RP, Kopec J, Bailey HJ, Fitzpatrick F, Newman JA, Newman WG, Amberger A, Zschocke J, Droogmans L, Oppermann U, Yue WW (2018) Structural insight into the human mitochondrial tRNA purine N1-methyltransferase and ribonuclease P complexes. J Biol Chem 293:12862-12876.

Oemer G, Lackner K, Muigg K, Krumschnabel G, Watschinger K, Sailer S, Lindner H, Gnaiger E, Wortmann SB, Werner ER, Zschocke J, Keller MA (2018) Molecular structural diversity of mitochondrial cardiolipins. Proc Natl Acad Sci U S A 115:4158-4163.

Milev MP, Graziano C, Karall D, Kuper WFE, Al-Deri N, Cordelli DM, Haack TB, Danhauser K, Iuso A, Palombo F, Pippucci T, Prokisch H, Saint-Dic D, Seri M, Stanga D, Cenacchi G, van Gassen KLI, Zschocke J, Fauth C, Mayr JA, Sacher M, van Hasselt PM (2018) Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. J Med Genet 55:753-764.

Giunta C, Baumann M, Fauth C, Lindert U, Abdalla EM, Brady AF, Collins J, Dastgir J, Donkervoort S, Ghali N, Johnson DS, Kariminejad A, Koch J, Kraenzlin M, Lahiri N, Lozic B, Manzur AY, Morton JEV, Pilch J, Pollitt RC, Schreiber G, Shannon NL, Sobey G, Vandersteen A, van Dijk FS, Witsch-Baumgartner M, Zschocke J, Pope FM, Bönnemann CG, Rohrbach M (2018) A cohort of 17 patients with kyphoscoliotic Ehlers-Danlos syndrome caused by biallelic mutations in FKBP14: expansion of the clinical and mutational spectrum and description of the natural history. Genet Med 20:42-54.

Schossig A, Bloch-Zupan A, Lussi A, Wolf NI, Raskin S, Cohen M, Giuliano F, Jurgens J, Krabichler B, Koolen DA, de Macena Sobreira NL, Maurer E, Muller-Bolla M, Penzien J, Zschocke J, Kapferer-Seebacher I (2017) SLC13A5 is the second gene associated with Kohlschütter-Tönz syndrome. J Med Genet 54:54-62.

Povysil G, Tzika A, Vogt J, Haunschmid V, Messiaen L, Zschocke J, Klambauer G, Hochreiter S, Wimmer K (2017) panelcn.MOPS: Copy-number detection in targeted NGS panel data for clinical diagnostics. Hum Mutat 38:889-897.

Kapferer-Seebacher I, Lundberg P, Malfait F, Zschocke J (2017) Periodontal manifestations of Ehlers-Danlos syndromes: A systematic review. J Clin Periodontol 44:1088-1100.

Baumann M, Steichen-Gersdorf E, Krabichler B, Petersen BS, Weber U, Schmidt WM, Zschocke J, Müller T, Bittner RE, Janecke AR (2017) Homozygous SYNE1 mutation causes congenital onset of muscular weakness with distal arthrogryposis: a genotype-phenotype correlation. Eur J Hum Genet 25:262-266.

Vogt J, Wernstedt A, Ripperger T, Pabst B, Zschocke J, Kratz C, Wimmer K (2016) PMS2 inactivation by a complex rearrangement involving an HERV retroelement and the inverted 100-kb duplicon on 7p22.1. Eur J Hum Genet 24:1598-1604.

Staufner C, Haack TB, Köpke MG, Straub BK, Kölker S, Thiel C, Freisinger P, Baric I, McKiernan PJ, Dikow N, Harting I, Beisse F, Burgard P, Kotzaeridou U, Lenz D, Kühr J, Himbert U, Taylor RW, Distelmaier F, Vockley J, Ghaloul-Gonzalez L, Ozolek JA, Zschocke J, Kuster A, Dick A, Das AM, Wieland T, Terrile C, Strom TM, Meitinger T, Prokisch H, Hoffmann GF (2016) Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. J Inherit Metab Dis 39:3-16.

Pölsler L, Fiegl H, Wimmer K, Oberaigner W, Amberger A, Traunfellner P, Morscher RJ, Weber I, Fauth C, Wernstedt A, Sperner-Unterweger B, Oberguggenberger A, Hubalek M, Marth C, Zschocke J (2016) High prevalence of BRCA1 stop mutation c.4183C>T in the Tyrolean population: implications for genetic testing. Eur J Hum Genet 24:258-262.

Peters V, Kebbewar M, Janssen B, Hoffmann GF, Möller K, Wygoda S, Charbit M, Fernandes-Teixeira A, Jeck N, Zschocke J, Schmitt CP, Schäfer F, Wühl E (2016) CNDP1 genotype and renal survival in pediatric nephropathies. J Pediatr Endocrinol Metab 29:827-833.

Oberguggenberger A, Sztankay M, Morscher RJ, Sperner-Unterweger B, Weber I, Hubalek M, Kemmler G, Zschocke J, Martini C, Egle D, Dünser M, Gamper E, Meraner V (2016) Psychosocial outcomes and counselee satisfaction following genetic counseling for hereditary breast and ovarian cancer: A patient-reported outcome study. J Psychosom Res 89:39-45.

Kapferer-Seebacher I, Pepin M, Werner R, Aitman TJ, Nordgren A, Stoiber H, Thielens N, Gaboriaud C, Amberger A, Schossig A, Gruber R, Giunta C, Bamshad M, Björck E, Chen C, Chitayat D, Dorschner M, Schmitt-Egenolf M, Hale CJ, Hanna D, Hennies HC, Heiss-Kisielewsky I, Lindstrand A, Lundberg P, Mitchell AL, Nickerson DA, Reinstein E, Rohrbach M, Romani N, Schmuth M, Silver R, Taylan F, Vandersteen A, Vandrovcova J, Weerakkody R, Yang M, Pope FM, Byers PH, Zschocke J (2016) Periodontal Ehlers-Danlos Syndrome Is Caused by Mutations in C1R and C1S, which Encode Subcomponents C1r and C1s of Complement. Am J Hum Genet 99:1005-1014.

Fauth C, Steindl K, Toutain A, Farrell S, Witsch-Baumgartner M, Karall D, Joset P, Böhm S, Baumer A, Maier O, Zschocke J, Weksberg R, Marshall CR, Rauch A (2016) A recurrent germline mutation in the PIGA gene causes Simpson-Golabi-Behmel syndrome type 2. Am J Med Genet A 170a:392-402.

Degen C, Zschocke J, Toro P, Sattler C, Wahl HW, Schönknecht P, Schröder J (2016) The COMTp.Val158Met Polymorphism and Cognitive Performance in Adult Development, Healthy Aging and Mild Cognitive Impairment. Dement Geriatr Cogn Disord 41:27-34.

Burgard P, Mönch E, Zschocke J, Wendel U, Langenbeck U (2016) Development of Metabolic Phenotype in Phenylketonuria: Evaluation of the Blaskovics Protein Loading Test at 5 Years of Age. JIMD Rep 29:77-84.

Amberger A, Deutschmann AJ, Traunfellner P, Moser P, Feichtinger RG, Kofler B, Zschocke J (2016) 17β-Hydroxysteroid dehydrogenase type 10 predicts survival of patients with colorectal cancer and affects mitochondrial DNA content. Cancer Lett 374:149-155.

Wortmann SB, van Hasselt PM, Barić I, Burlina A, Darin N, Hörster F, Coker M, Ucar SK, Krumina Z, Naess K, Ngu LH, Pronicka E, Riordan G, Santer R, Wassmer E, Zschocke J, Schiff M, de Meirleir L, Alowain MA, Smeitink JA, Morava E, Kozicz T, Wevers RA, Wolf NI, Willemsen MA (2015) Eyes on MEGDEL: distinctive basal ganglia involvement in dystonia deafness syndrome. Neuropediatrics 46:98-103.

Pitt JJ, Peters H, Boneh A, Yaplito-Lee J, Wieser S, Hinderhofer K, Johnson D, Zschocke J (2015) Mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency: urinary organic acid profiles and expanded spectrum of mutations. J Inherit Metab Dis 38:459-466.

Pichler K, Scholl-Buergi S, Birnbacher R, Freilinger M, Straub S, Brunner J, Zschocke J, Bittner RE, Karall D (2015) A novel therapeutic approach for LPIN1 mutation-associated rhabdomyolysis--The Austrian experience. Muscle Nerve 52:437-439.

Peters V, Lanthaler B, Amberger A, Fleming T, Forsberg E, Hecker M, Wagner AH, Yue WW, Hoffmann GF, Nawroth P, Zschocke J, Schmitt CP (2015) Carnosine metabolism in diabetes is altered by reactive metabolites. Amino Acids 47:2367-2376.

Peters V, Klessens CQ, Baelde HJ, Singler B, Veraar KA, Zutinic A, Drozak J, Zschocke J, Schmitt CP, de Heer E (2015) Intrinsic carnosine metabolism in the human kidney. Amino Acids 47:2541-2550.

Moosbrugger-Martinz V, Jalili A, Schossig AS, Jahn-Bassler K, Zschocke J, Schmuth M, Stingl G, Eckl KM, Hennies HC, Gruber R (2015) Epidermal barrier abnormalities in exfoliative ichthyosis with a novel homozygous loss-of-function mutation in CSTA. Br J Dermatol 172:1628-1632.

Haack TB, Staufner C, Köpke MG, Straub BK, Kölker S, Thiel C, Freisinger P, Baric I, McKiernan PJ, Dikow N, Harting I, Beisse F, Burgard P, Kotzaeridou U, Kühr J, Himbert U, Taylor RW, Distelmaier F, Vockley J, Ghaloul-Gonzalez L, Zschocke J, Kremer LS, Graf E, Schwarzmayr T, Bader DM, Gagneur J, Wieland T, Terrile C, Strom TM, Meitinger T, Hoffmann GF, Prokisch H (2015) Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. Am J Hum Genet 97:163-169.

Fauth C, Rostasy K, Rath M, Gizewski E, Lederer AG, Sure U, Zschocke J, Felbor U (2015) Highly variable intrafamilial manifestations of a CCM3 mutation ranging from acute childhood cerebral haemorrhage to late-onset meningiomas. Clin Neurol Neurosurg 128:41-43.

Danecka MK, Woidy M, Zschocke J, Feillet F, Muntau AC, Gersting SW (2015) Mapping the functional landscape of frequent phenylalanine hydroxylase (PAH) genotypes promotes personalised medicine in phenylketonuria. J Med Genet 52:175-185.

van Karnebeek CD, Shevell M, Zschocke J, Moeschler JB, Stockler S (2014) The metabolic evaluation of the child with an intellectual developmental disorder: Diagnostic algorithm for identification of treatable causes and new digital resource. Mol Genet Metab.

Spreiz A, Haberlandt E, Baumann M, Baumgartner Sigl S, Fauth C, Gautsch K, Karall D, Janetschek C, Rostasy K, Scholl-Bürgi S, Zotter S, Utermann G, Zschocke J, Kotzot D (2014) Chromosomal microaberrations in patients with epilepsy, intellectual disability, and congenital anomalies. Clin Genet 86:361-366.

Peters V, Riedl E, Braunagel M, Höger S, Hauske S, Pfister F, Zschocke J, Lanthaler B, Benck U, Hammes HP, Krämer BK, Schmitt CP, Yard BA, Köppel H (2014) Carnosine treatment in combination with ACE inhibition in diabetic rats. Regul Pept 194-195:36-40.

Pabinger S, Dander A, Fischer M, Snajder R, Sperk M, Efremova M, Krabichler B, Speicher MR, Zschocke J, Trajanoski Z (2014) A survey of tools for variant analysis of next-generation genome sequencing data. Brief Bioinform 15:256-278.

Lanthaler B, Wieser S, Deutschmann A, Schossig A, Fauth C, Zschocke J, Witsch-Baumgartner M (2014) Genotype-based databases for variants causing rare diseases. Gene 550:136-140.

Hofer P, Fiegl H, Angerer J, Mueller-Holzner E, Chamson M, Klocker H, Steiner E, Hauffe H, Zschocke J, Goebel G (2014) A concept of a MIABIS based register of biosample collections at the Medical University of Innsbruck. Stud Health Technol Inform 205:293-297.

Deutschmann AJ, Amberger A, Zavadil C, Steinbeisser H, Mayr JA, Feichtinger RG, Oerum S, Yue WW, Zschocke J (2014) Mutation or knock-down of 17β-hydroxysteroid dehydrogenase type 10 cause loss of

MRPP1 and impaired processing of mitochondrial heavy strand transcripts. Hum Mol Genet 23:3618-3628.

Balci S, Zschocke J, Kotzot D, Ergün MA, Spreiz A (2014) Formation of a familial ring chromosome 18 investigated by SNP-array analysis. Am J Med Genet A 164a:1854-1856.

Baguet A, Everaert I, Yard B, Peters V, Zschocke J, Zutinic A, de Heer E, Podgorski T, Domaszewska K, Derave W (2014) Does low serum carnosinase activity favour high-intensity exercise capacity? J Appl Physiol (1985).

Tucci A, Kara E, Schossig A, Wolf NI, Plagnol V, Fawcett K, Paisan-Ruiz C, Moore M, Hernandez D, Musumeci S, Tennison M, Hennekam R, Palmeri S, Malandrini A, Raskin S, Donnai D, Hennig C, Tzschach A, Hordijk R, Bast T, Wimmer K, Lo CN, Shorvon S, Mefford H, Eichler EE, Hall R, Hayes I, Hardy J, Singleton A, Zschocke J, Houlden H (2013) Kohlschutter-Tonz syndrome: mutations in ROGDI and evidence of genetic heterogeneity. Hum Mutat 34:296-300.

Szklarczyk R, Wanschers BF, Nijtmans LG, Rodenburg RJ, Zschocke J, Dikow N, van den Brand MA, Hendriks-Franssen MG, Gilissen C, Veltman JA, Nooteboom M, Koopman WJ, Willems PH, Smeitink JA, Huynen MA, van den Heuvel LP (2013) A mutation in the FAM36A gene, the human ortholog of COX20, impairs cytochrome c oxidase assembly and is associated with ataxia and muscle hypotonia. Hum Mol Genet 22:656-667.

Sterl E, Paul K, Paschke E, Zschocke J, Brunner-Krainz M, Windisch E, Konstantopoulou V, Moslinger D, Karall D, Scholl-Burgi S, Sperl W, Lagler F, Plecko B (2013) Prevalence of tetrahydrobiopterine (BH4)-responsive alleles among Austrian patients with PAH deficiency: comprehensive results from molecular analysis in 147 patients. J Inherit Metab Dis 36:7-13.

Spreiz A, Haberlandt E, Baumann M, Baumgartner Sigl S, Fauth C, Gautsch K, Karall D, Janetschek C, Rostasy K, Scholl-Burgi S, Zotter S, Utermann G, Zschocke J, Kotzot D (2013) Chromosomal microaberrations in patients with epilepsy, intellectual disability, and congenital anomalies. Clin Genet.

Spreiz A, Guilherme RS, Castellan C, Green A, Rittinger O, Wellek B, Utermann B, Erdel M, Fauth C, Haberlandt E, Kim CA, Kulikowski LD, Meloni VA, Utermann G, Zschocke J, Melaragno MI, Kotzot D (2013) Single-nucleotide polymorphism array-based characterization of ring chromosome 18. J Pediatr 163:1174-1178 e1173.

Pabinger S, Dander A, Fischer M, Snajder R, Sperk M, Efremova M, Krabichler B, Speicher MR, Zschocke J, Trajanoski Z (2013) A survey of tools for variant analysis of next-generation genome sequencing data. Brief Bioinform.

Muller T, Mizumoto S, Suresh I, Komatsu Y, Vodopiutz J, Dundar M, Straub V, Lingenhel A, Melmer A, Lechner S, Zschocke J, Sugahara K, Janecke AR (2013) Loss of dermatan sulfate epimerase (DSE) function results in musculocontractural Ehlers-Danlos syndrome. Hum Mol Genet 22:3761-3772.

Lanthaler B, Steichen-Gersdorf E, Kollerits B, Zschocke J, Witsch-Baumgartner M (2013) Maternal ABCA1 genotype is associated with severity of Smith-Lemli-Opitz syndrome and with viability of patients homozygous for null mutations. Eur J Hum Genet 21:286-293.

Houten SM, Te Brinke H, Denis S, Ruiter JP, Knegt AC, de Klerk JB, Augoustides-Savvopoulou P, Haberle J, Baumgartner MR, Coskun T, Zschocke J, Sass JO, Poll-The BT, Wanders RJ, Duran M (2013) Genetic basis of hyperlysinemia. Orphanet J Rare Dis 8:57.

Fruhmesser A, Vogt PH, Zimmer J, Witsch-Baumgartner M, Fauth C, Zschocke J, Pinggera GM, Kotzot D (2013) Single nucleotide polymorphism array analysis in men with idiopathic azoospermia or oligoasthenozoospermia syndrome. Fertil Steril 100:81-87.

Wernstedt A, Valtorta E, Armelao F, Togni R, Girlando S, Baudis M, Heinimann K, Messiaen L, Staehli N, Zschocke J, Marra G, Wimmer K (2012) Improved multiplex ligation-dependent probe amplification analysis identifies a deleterious PMS2 allele generated by recombination with crossover between PMS2 and PMS2CL. Genes Chromosomes Cancer 51:819-831.

Schossig A, Wolf NI, Fischer C, Fischer M, Stocker G, Pabinger S, Dander A, Steiner B, Tonz O, Kotzot D, Haberlandt E, Amberger A, Burwinkel B, Wimmer K, Fauth C, Grond-Ginsbach C, Koch MJ, Deichmann A, von Kalle C, Bartram CR, Kohlschutter A, Trajanoski Z, Zschocke J (2012) Mutations in ROGDI Cause Kohlschutter-Tonz Syndrome. Am J Hum Genet 90:701-707.

Scholl-Burgi S, Sass JO, Zschocke J, Karall D (2012) Amino acid metabolism in patients with propionic acidaemia. J Inherit Metab Dis 35:65-70.

Peters V, Schmitt CP, Zschocke J, Gross ML, Brismar K, Forsberg E (2012) Carnosine treatment largely prevents alterations of renal carnosine metabolism in diabetic mice. Amino Acids 42:2411-2416.

Krabichler B, Rostasy K, Baumann M, Karall D, Scholl-Burgi S, Schwarzer C, Gautsch K, Spreiz A, Kotzot D, Zschocke J, Fauth C, Haberlandt E (2012) Novel mutation in potassium channel related gene KCTD7 and progressive myoclonic epilepsy. Ann Hum Genet 76:326-331.

Kalb S, Caglayan AO, Degerliyurt A, Schmid S, Ceylaner S, Hatipoglu N, Hinderhofer K, Rehder H, Kurtoglu S, Ceylaner G, Zschocke J, Witsch-Baumgartner M (2012) High frequency of p.Thr93Met in Smith-Lemli-Opitz syndrome patients in Turkey. Clin Genet 81:598-601.

Hockner M, Spreiz A, Fruhmesser A, Tzschach A, Dufke A, Rittinger O, Kalscheuer V, Singer S, Erdel M, Fauth C, Grossmann V, Utermann G, Zschocke J, Kotzot D (2012) Parental origin of de novo cyto-genetically balanced reciprocal non-Robertsonian translocations. Cytogenet Genome Res 136:242-245.

Fischer M, Snajder R, Pabinger S, Dander A, Schossig A, Zschocke J, Trajanoski Z, Stocker G (2012) SIMPLEX: cloud-enabled pipeline for the comprehensive analysis of exome sequencing data. PLoS One 7:e41948.

Danhauser K, Sauer SW, Haack TB, Wieland T, Staufner C, Graf E, Zschocke J, Strom TM, Traub T, Okun JG, Meitinger T, Hoffmann GF, Prokisch H, Kolker S (2012) DHTKD1 mutations cause 2-aminoadipic and 2-oxoadipic aciduria. Am J Hum Genet 91:1082-1087.

Baumann M, Giunta C, Krabichler B, Ruschendorf F, Zoppi N, Colombi M, Bittner RE, Quijano-Roy S, Muntoni F, Cirak S, Schreiber G, Zou Y, Hu Y, Romero NB, Carlier RY, Amberger A, Deutschmann A, Straub V, Rohrbach M, Steinmann B, Rostasy K, Karall D, Bonnemann CG, Zschocke J, Fauth C (2012) Mutations in FKBP14 cause a variant of Ehlers-Danlos syndrome with progressive kyphoscoliosis, myopathy, and hearing loss. Am J Hum Genet 90:201-216.

Peters V, Jansen EE, Jakobs C, Riedl E, Janssen B, Yard BA, Wedel J, Hoffmann GF, Zschocke J, Gotthardt D, Fischer C, Koppel H (2011) Anserine inhibits carnosine degradation but in human serum carnosinase (CN1) is not correlated with histidine dipeptide concentration. Clin Chim Acta 412:263-267.

Mayr JA, Zimmermann FA, Fauth C, Bergheim C, Meierhofer D, Radmayr D, Zschocke J, Koch J, Sperl W (2011) Lipoic acid synthetase deficiency causes neonatal-onset epilepsy, defective mitochondrial energy metabolism, and glycine elevation. Am J Hum Genet 89:792-797.

Jungraithmayr TC, Hofer K, Cochat P, Chernin G, Cortina G, Fargue S, Grimm P, Knueppel T, Kowarsch A, Neuhaus T, Pagel P, Pfeiffer KP, Schafer F, Schonermarck U, Seeman T, Toenshoff B, Weber S, Winn MP, Zschocke J, Zimmerhackl LB (2011) Screening for NPHS2 mutations may help predict FSGS recurrence after transplantation. J Am Soc Nephrol 22:579-585.

Al-Dirbashi OY, Kolker S, Ng D, Fisher L, Rupar T, Lepage N, Rashed MS, Santa T, Goodman SI, Geraghty MT, Zschocke J, Christensen E, Hoffmann GF, Chakraborty P (2011) Diagnosis of glutaric aciduria type 1 by measuring 3-hydroxyglutaric acid in dried urine spots by liquid chromatography tandem mass spectrometry. J Inherit Metab Dis 34:173-180.

Shafqat N, Turnbull A, Zschocke J, Oppermann U, Yue WW (2010) Crystal structures of human HMG-CoA synthase isoforms provide insights into inherited ketogenesis disorders and inhibitor design. J Mol Biol 398:497-506.

Rauschenberger K, Scholer K, Sass JO, Sauer S, Djuric Z, Rumig C, Wolf NI, Okun JG, Kolker S, Schwarz H, Fischer C, Grziwa B, Runz H, Numann A, Shafqat N, Kavanagh KL, Hammerling G, Wanders RJ, Shield JP, Wendel U, Stern D, Nawroth P, Hoffmann GF, Bartram CR, Arnold B, Bierhaus A, Oppermann U, Steinbeisser H, Zschocke J (2010) A non-enzymatic function of 17beta-hydroxysteroid dehydrogenase type 10 is required for mitochondrial integrity and cell survival. EMBO Mol Med 2:51-62.

Peters V, Kebbewar M, Jansen EW, Jakobs C, Riedl E, Koeppel H, Frey D, Adelmann K, Klingbeil K, Mack M, Hoffmann GF, Janssen B, Zschocke J, Yard BA (2010) Relevance of allosteric conformations and homocarnosine concentration on carnosinase activity. Amino Acids 38:1607-1615.

Holiner I, Simma B, Reiter A, Sass JO, Zschocke J, Huemer M (2010) Compliance to clinical guidelines determines outcome in glutaric aciduria type I in the era of newborn screening. Klin Padiatr 222:35-37.

Heringer J, Boy SP, Ensenauer R, Assmann B, Zschocke J, Harting I, Lucke T, Maier EM, Muhlhausen C, Haege G, Hoffmann GF, Burgard P, Kolker S (2010) Use of guidelines improves the neurological outcome in glutaric aciduria type I. Ann Neurol 68:743-752.

Grossmann V, Hockner M, Karmous-Benailly H, Liang D, Puttinger R, Quadrelli R, Rothlisberger B, Huber A, Wu L, Spreiz A, Fauth C, Erdel M, Zschocke J, Utermann G, Kotzot D (2010) Parental origin of apparently balanced de novo complex chromosomal rearrangements investigated by microdissection, whole genome amplification, and microsatellite-mediated haplotype analysis. Clin Genet 78:548-553.

Ganster C, Wernstedt A, Kehrer-Sawatzki H, Messiaen L, Schmidt K, Rahner N, Heinimann K, Fonatsch C, Zschocke J, Wimmer K (2010) Functional PMS2 hybrid alleles containing a pseudogene-specific missense variant trace back to a single ancient intrachromosomal recombination event. Hum Mutat 31:552-560.

Gan-Schreier H, Kebbewar M, Fang-Hoffmann J, Wilrich J, Abdoh G, Ben-Omran T, Shahbek N, Bener A, Al Rifai H, Al Khal AL, Lindner M, Zschocke J, Hoffmann GF (2010) Newborn population screening for classic homocystinuria by determination of total homocysteine from Guthrie cards. J Pediatr 156:427-432.

Zschocke J, Kebbewar M, Gan-Schreier H, Fischer C, Fang-Hoffmann J, Wilrich J, Abdoh G, Ben-Omran T, Shahbek N, Lindner M, Al Rifai H, Al Khal AL, Hoffmann GF (2009) Molecular neonatal screening for homocystinuria in the Qatari population. Hum Mutat 30:1021-1022.

Spiekerkoetter U, Lindner M, Santer R, Grotzke M, Baumgartner MR, Boehles H, Das A, Haase C, Hennermann JB, Karall D, de Klerk H, Knerr I, Koch HG, Plecko B, Roschinger W, Schwab KO, Scheible D, Wijburg FA, Zschocke J, Mayatepek E, Wendel U (2009) Management and outcome in 75 individuals with long-chain fatty acid oxidation defects: results from a workshop. J Inherit Metab Dis 32:488-497.

Soufi M, Kurt B, Schweer H, Sattler AM, Klaus G, Zschocke J, Schaefer JR (2009) Genetics and kinetics of familial hypercholesterolemia, with the special focus on FH-(Marburg) p.W556R. Atheroscler Suppl 10:5-11.

Langenbeck U, Burgard P, Wendel U, Lindner M, Zschocke J, German Collaborative Study on Phenylketonuria H (2009) Metabolic phenotypes of phenylketonuria. Kinetic and molecular evaluation of the Blaskovics protein loading test. J Inherit Metab Dis 32:506-513.

Harting I, Seitz A, Rating D, Sartor K, Zschocke J, Janssen B, Ebinger F, Wolf NI (2009) Abnormal myelination in Angelman syndrome. Eur J Paediatr Neurol 13:271-276.

Harting I, Neumaier-Probst E, Seitz A, Maier EM, Assmann B, Baric I, Troncoso M, Muhlhausen C, Zschocke J, Boy NP, Hoffmann GF, Garbade SF, Kolker S (2009) Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I. Brain 132:1764-1782.

Zurfluh MR, Zschocke J, Lindner M, Feillet F, Chery C, Burlina A, Stevens RC, Thony B, Blau N (2008) Molecular genetics of tetrahydrobiopterin-responsive phenylalanine hydroxylase deficiency. Hum Mutat 29:167-175.

Zschocke J, Aulehla-Scholz C, Patton S (2008) Quality of diagnostic mutation analyses for phenylketonuria. J Inherit Metab Dis 31:697-702.

Sass JO, Ensenauer R, Roschinger W, Reich H, Steuerwald U, Schirrmacher O, Engel K, Haberle J, Andresen BS, Megarbane A, Lehnert W, Zschocke J (2008) 2-Methylbutyryl-coenzyme A dehydrogenase deficiency: functional and molecular studies on a defect in isoleucine catabolism. Mol Genet Metab 93:30-35.

Runz H, Dolle D, Schlitter AM, Zschocke J (2008) NPC-db, a Niemann-Pick type C disease gene variation database. Hum Mutat 29:345-350.

Wolf NI, Harting I, Innes AM, Patzer S, Zeitler P, Schneider A, Wolff A, Baier K, Zschocke J, Ebinger F, Boltshauser E, Rating D (2007) Ataxia, delayed dentition and hypomyelination: a novel leukoencephalopathy. Neuropediatrics 38:64-70.

Kolker S, Garbade SF, Boy N, Maier EM, Meissner T, Muhlhausen C, Hennermann JB, Lucke T, Haberle J, Baumkotter J, Haller W, Muller E, Zschocke J, Burgard P, Hoffmann GF (2007) Decline of acute encephalopathic crises in children with glutaryl-CoA dehydrogenase deficiency identified by newborn screening in Germany. Pediatr Res 62:357-363.

Hoffmann GF, Schmitt B, Windfuhr M, Wagner N, Strehl H, Bagci S, Franz AR, Mills PB, Clayton PT, Baumgartner MR, Steinmann B, Bast T, Wolf NI, Zschocke J (2007) Pyridoxal 5'-phosphate may be curative in early-onset epileptic encephalopathy. J Inherit Metab Dis 30:96-99.

Haas D, Morgenthaler J, Lacbawan F, Long B, Runz H, Garbade SF, Zschocke J, Kelley RI, Okun JG, Hoffmann GF, Muenke M (2007) Abnormal sterol metabolism in holoprosencephaly: studies in cultured lymphoblasts. J Med Genet 44:298-305.

Haas D, Garbade SF, Vohwinkel C, Muschol N, Trefz FK, Penzien JM, Zschocke J, Hoffmann GF, Burgard P (2007) Effects of cholesterol and simvastatin treatment in patients with Smith-Lemli-Opitz syndrome (SLOS). J Inherit Metab Dis 30:375-387.

Elsaid MF, Bener A, Lindner M, Alzyoud M, Shahbek N, Abdelrahman MO, Abdoh G, Bessisso MS, Zschocke J, Hoffmann GF (2007) Are heterocygotes for classical homocystinuria at risk of vitamin B12 and folic acid deficiency? Mol Genet Metab 92:100-103.

Dikow N, Nygren AO, Schouten JP, Hartmann C, Kramer N, Janssen B, Zschocke J (2007) Quantification of the methylation status of the PWS/AS imprinted region: comparison of two approaches based on bisulfite sequencing and methylation-sensitive MLPA. Mol Cell Probes 21:208-215.

Birk Moller L, Nygren AO, Scott P, Hougaard P, Bieber Nielsen J, Hartmann C, Guttler F, Tyfield L, Zschocke J (2007) Low proportion of whole exon deletions causing phenylketonuria in Denmark and Germany. Hum Mutat 28:207.

Zschocke J, Nebel A, Wicks K, Peters V, El Mokhtari NE, Krawczak M, van der Woude F, Janssen B, Schreiber S (2006) Allelic variation in the CNDP1 gene and its lack of association with longevity and coronary heart disease. Mech Ageing Dev 127:817-820.

Mack M, Schniegler-Mattox U, Peters V, Hoffmann GF, Liesert M, Buckel W, Zschocke J (2006) Biochemical characterization of human 3-methylglutaconyl-CoA hydratase and its role in leucine metabolism. FEBS J 273:2012-2022.

Mack M, Liesert M, Zschocke J, Peters V, Linder D, Buckel W (2006) 3-Methylglutaconyl-CoA hydratase from Acinetobacter sp. Arch Microbiol 185:297-306.

Kolker S, Garbade SF, Greenberg CR, Leonard JV, Saudubray JM, Ribes A, Kalkanoglu HS, Lund AM, Merinero B, Wajner M, Troncoso M, Williams M, Walter JH, Campistol J, Marti-Herrero M, Caswill M, Burlina AB, Lagler F, Maier EM, Schwahn B, Tokatli A, Dursun A, Coskun T, Chalmers RA, Koeller DM, Zschocke J, Christensen E, Burgard P, Hoffmann GF (2006) Natural history, outcome, and treatment efficacy in children and adults with glutaryl-CoA dehydrogenase deficiency. Pediatr Res 59:840-847.

El-Said MF, Badii R, Bessisso MS, Shahbek N, El-Ali MG, El-Marikhie M, El-Zyoid M, Salem MS, Bener A, Hoffmann GF, Zschocke J (2006) A common mutation in the CBS gene explains a high incidence of homocystinuria in the Qatari population. Hum Mutat 27:719.

Chalmers RA, Bain MD, Zschocke J (2006) Riboflavin-responsive glutaryl CoA dehydrogenase deficiency. Mol Genet Metab 88:29-37.

Wolf NI, Harting I, Boltshauser E, Wiegand G, Koch MJ, Schmitt-Mechelke T, Martin E, Zschocke J, Uhlenberg B, Hoffmann GF, Weber L, Ebinger F, Rating D (2005) Leukoencephalopathy with ataxia, hypodontia, and hypomyelination. Neurology 64:1461-1464.

Witsch-Baumgartner M, Clayton P, Clusellas N, Haas D, Kelley RI, Krajewska-Walasek M, Lechner S, Rossi M, Zschocke J, Utermann G (2005) Identification of 14 novel mutations in DHCR7 causing the Smith-Lemli-Opitz syndrome and delineation of the DHCR7 mutational spectra in Spain and Italy. Hum Mutat 25:412.

Schulze-Bergkamen A, Okun JG, Spiekerkotter U, Lindner M, Haas D, Kohlmuller D, Mayatepek E, Schulze-Bergkamen H, Greenberg CR, Zschocke J, Hoffmann GF, Kolker S (2005) Quantitative acylcarnitine profiling in peripheral blood mononuclear cells using in vitro loading with palmitic and 2-oxoadipic acids: biochemical confirmation of fatty acid oxidation and organic acid disorders. Pediatr Res 58:873-880.

Mills PB, Surtees RA, Champion MP, Beesley CE, Dalton N, Scambler PJ, Heales SJ, Briddon A, Scheimberg I, Hoffmann GF, Zschocke J, Clayton PT (2005) Neonatal epileptic encephalopathy caused by mutations in the PNPO gene encoding pyridox(am)ine 5'-phosphate oxidase. Hum Mol Genet 14:1077-1086.

Kulkens S, Harting I, Sauer S, Zschocke J, Hoffmann GF, Gruber S, Bodamer OA, Kolker S (2005) Late-onset neurologic disease in glutaryl-CoA dehydrogenase deficiency. Neurology 64:2142-2144.

Janssen B, Hohenadel D, Brinkkoetter P, Peters V, Rind N, Fischer C, Rychlik I, Cerna M, Romzova M, de Heer E, Baelde H, Bakker SJ, Zirie M, Rondeau E, Mathieson P, Saleem MA, Meyer J, Koppel H, Sauerhoefer S, Bartram CR, Nawroth P, Hammes HP, Yard BA, Zschocke J, van der Woude FJ (2005) Carnosine as a protective factor in diabetic nephropathy: association with a leucine repeat of the carnosinase gene CNDP1. Diabetes 54:2320-2327.

Janssen B, Hartmann C, Scholz V, Jauch A, Zschocke J (2005) MLPA analysis for the detection of deletions, duplications and complex rearrangements in the dystrophin gene: potential and pitfalls. Neurogenetics 6:29-35.

Stiasny-Kolster K, Moller JC, Zschocke J, Bandmann O, Cassel W, Oertel WH, Hoffmann GF (2004) Normal dopaminergic and serotonergic metabolites in cerebrospinal fluid and blood of restless legs syndrome patients. Mov Disord 19:192-196.

Saunders-Pullman R, Blau N, Hyland K, Zschocke J, Nygaard T, Raymond D, Shanker V, Mohrmann K, Arnold L, Tabbal S, deLeon D, Ford B, Brin M, Chouinard S, Ozelius L, Klein C, Bressman SB (2004) Phenylalanine loading as a diagnostic test for DRD: interpreting the utility of the test. Mol Genet Metab 83:207-212.

Mayatepek E, Okun JG, Meissner T, Assmann B, Hammond J, Zschocke J, Lehmann WD (2004) Synthesis and metabolism of leukotrienes in gamma-glutamyl transpeptidase deficiency. J Lipid Res 45:900-904.

Mayatepek E, Flock B, Zschocke J (2004) Benzydamine metabolism in vivo is impaired in patients with deficiency of flavin-containing monooxygenase 3. Pharmacogenetics 14:775-777.

Hartmann C, John AL, Klaes R, Hofmann W, Bielen R, Koehler R, Janssen B, Bartram CR, Arnold N, Zschocke J (2004) Large BRCA1 gene deletions are found in 3% of German high-risk breast cancer families. Hum Mutat 24:534.

Christensen E, Ribes A, Merinero B, Zschocke J (2004) Correlation of genotype and phenotype in glutaryl-CoA dehydrogenase deficiency. J Inherit Metab Dis 27:861-868.

Burlina AP, Zara G, Hoffmann GF, Zschocke J, Burlina AB (2004) Management of movement disorders in glutaryl-CoA dehydrogenase deficiency: anticholinergic drugs and botulinum toxin as additional therapeutic options. J Inherit Metab Dis 27:911-915.

Zschocke J, Preusse A, Sarnavka V, Fumic K, Mardesic D, Hoffmann GF, Baric I (2003) The molecular basis of phenylalanine hydroxylase deficiency in Croatia. Hum Mutat 21:399.

Zschocke J (2003) Phenylketonuria mutations in Europe. Hum Mutat 21:345-356.

Tighe O, Dunican D, O'Neill C, Bertorelle G, Beattie D, Graham C, Zschocke J, Cali F, Romano V, Hrabincova E, Kozak L, Nechyporenko M, Livshits L, Guldberg P, Jurkowska M, Zekanowski C, Perez B, Desviat LR, Ugarte M, Kucinskas V, Knappskog P, Treacy E, Naughten E, Tyfield L, Byck S, Scriver CR, Mayne PD, Croke DT (2003) Genetic diversity within the R408W phenylketonuria mutation lineages in Europe. Hum Mutat 21:387-393.

Ofman R, Ruiter JP, Feenstra M, Duran M, Poll-The BT, Zschocke J, Ensenauer R, Lehnert W, Sass JO, Sperl W, Wanders RJ (2003) 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency is caused by mutations in the HADH2 gene. Am J Hum Genet 72:1300-1307.

Ly TB, Peters V, Gibson KM, Liesert M, Buckel W, Wilcken B, Carpenter K, Ensenauer R, Hoffmann GF, Mack M, Zschocke J (2003) Mutations in the AUH gene cause 3-methylglutaconic aciduria type I. Hum Mutat 21:401-407.

Lindner M, Steinfeld R, Burgard P, Schulze A, Mayatepek E, Zschocke J (2003) Tetrahydrobiopterin sensitivity in German patients with mild phenylalanine hydroxylase deficiency. Hum Mutat 21:400.

Krawczak M, Zschocke J (2003) A role for overdominant selection in phenylketonuria? Evidence from molecular data. Hum Mutat 21:394-397.

Kolker S, Hoffmann GF, Schor DS, Feyh P, Wagner L, Jeffrey I, Pourfarzam M, Okun JG, Zschocke J, Baric I, Bain MD, Jakobs C, Chalmers RA (2003) Glutaryl-CoA dehydrogenase deficiency: region-specific analysis of organic acids and acylcarnitines in post mortem brain predicts vulnerability of the putamen. Neuropediatrics 34:253-260.

Bayatti N, Zschocke J, Behl C (2003) Brain region-specific neuroprotective action and signaling of corticotropin-releasing hormone in primary neurons. Endocrinology 144:4051-4060.

Bandmann O, Goertz M, Zschocke J, Deuschl G, Jost W, Hefter H, Muller U, Zofel P, Hoffmann G, Oertel W (2003) The phenylalanine loading test in the differential diagnosis of dystonia. Neurology 60:700-702.

Steinfeld R, Kohlschutter A, Zschocke J, Lindner M, Ullrich K, Lukacs Z (2002) Tetrahydrobiopterin monotherapy for phenylketonuria patients with common mild mutations. Eur J Pediatr 161:403-405.

Niklowitz P, Menke T, Wiesel T, Mayatepek E, Zschocke J, Okun JG, Andler W (2002) Coenzyme Q10 in plasma and erythrocytes: comparison of antioxidant levels in healthy probands after oral supplementation and in patients suffering from sickle cell anemia. Clin Chim Acta 326:155-161.

Neu IS, Metzger G, Zschocke J, Zelezny R, Mayatepek E (2002) Leukotrienes in patients with clinically active multiple sclerosis. Acta Neurol Scand 105:63-66.

Zschocke J, Schulze A, Lindner M, Fiesel S, Olgemoller K, Hoffmann GF, Penzien J, Ruiter JP, Wanders RJ, Mayatepek E (2001) Molecular and functional characterisation of mild MCAD deficiency. Hum Genet 108:404-408.

Zschocke J, Hoffmann GF (2001) Cycle sequencing of polymerase chain reaction-amplified genomic DNA with dye-labeled universal primers. Methods Mol Biol 167:113-117.

Weglage J, Pietsch M, Feldmann R, Koch HG, Zschocke J, Hoffmann G, Muntau-Heger A, Denecke J, Guldberg P, Guttler F, Moller H, Wendel U, Ullrich K, Harms E (2001) Normal clinical outcome in untreated subjects with mild hyperphenylalaninemia. Pediatr Res 49:532-536.

Rupp A, Kreis R, Zschocke J, Slotboom J, Boesch C, Rating D, Pietz J (2001) Variability of blood-brain ratios of phenylalanine in typical patients with phenylketonuria. J Cereb Blood Flow Metab 21:276-284.

Lindner M, Haas D, Mayatepek E, Zschocke J, Burgard P (2001) Tetrahydrobiopterin responsiveness in phenylketonuria differs between patients with the same genotype. Mol Genet Metab 73:104-106.

Langenbeck U, Zschocke J, Wendel U, Honig V (2001) Modelling the phenylalanine blood level response during treatment of phenylketonuria. J Inherit Metab Dis 24:805-814.

Aledo R, Zschocke J, Pie J, Mir C, Fiesel S, Mayatepek E, Hoffmann GF, Casals N, Hegardt FG (2001) Genetic basis of mitochondrial HMG-CoA synthase deficiency. Hum Genet 109:19-23.

Zschocke J, Ruiter JP, Brand J, Lindner M, Hoffmann GF, Wanders RJ, Mayatepek E (2000) Progressive infantile neurodegeneration caused by 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency: a novel inborn error of branched-chain fatty acid and isoleucine metabolism. Pediatr Res 48:852-855.

Zschocke J, Quak E, Guldberg P, Hoffmann GF (2000) Mutation analysis in glutaric aciduria type I. J Med Genet 37:177-181.

Zschocke J, Mayatepek E (2000) Biochemical and molecular studies in mild flavin monooxygenase 3 deficiency. J Inherit Metab Dis 23:378-382.

Greeves LG, Patterson CC, Carson DJ, Thom R, Wolfenden MC, Zschocke J, Graham CA, Nevin NC, Trimble ER (2000) Effect of genotype on changes in intelligence quotient after dietary relaxation in phenylketonuria and hyperphenylalaninaemia. Arch Dis Child 82:216-221.

Zschocke J, Kohlmueller D, Quak E, Meissner T, Hoffmann GF, Mayatepek E (1999) Mild trimethylaminuria caused by common variants in FMO3 gene. Lancet 354:834-835.

Zschocke J, Hoffmann GF (1999) Phenylketonuria mutations in Germany. Hum Genet 104:390-398.

Shin YS, Zschocke J, Das AM, Podskarbi T (1999) Molecular and biochemical basis for variants and deficiency forms of galactose-1-phosphate uridyltransferase. J Inherit Metab Dis 22:327-329.

Liesert M, Zschocke J, Hoffmann GF, Muhlhauser N, Buckel W (1999) Biochemistry of glutaric aciduria type I: activities of in vitro expressed wild-type and mutant cDNA encoding human glutaryl-CoA dehydrogenase. J Inherit Metab Dis 22:256-258.

Weglage J, Moller HE, Wiedermann D, Cipcic-Schmidt S, Zschocke J, Ullrich K (1998) In vivo NMR spectroscopy in patients with phenylketonuria: clinical significance of interindividual differences in brain phenylalanine concentrations. J Inherit Metab Dis 21:81-82.

Guldberg P, Rey F, Zschocke J, Romano V, Francois B, Michiels L, Ullrich K, Hoffmann GF, Burgard P, Schmidt H, Meli C, Riva E, Dianzani I, Ponzone A, Rey J, Guttler F (1998) A European multicenter study of phenylalanine hydroxylase deficiency: classification of 105 mutations and a general system for genotype-based prediction of metabolic phenotype. Am J Hum Genet 63:71-79.

Zschocke J, Mallory JP, Eiken HG, Nevin NC (1997) Phenylketonuria and the peoples of Northern Ireland. Hum Genet 100:189-194.

Guldberg P, Zschocke J, Dagbjartsson A, Henriksen KF, Guttler F (1997) A molecular survey of phenylketonuria in Iceland: identification of a founding mutation and evidence of predominant Norse settlement. Eur J Hum Genet 5:376-381.

Zschocke J, Graham CA, Carson DJ, Nevin NC (1995) Phenylketonuria mutation analysis in Northern Ireland: a rapid stepwise approach. Am J Hum Genet 57:1311-1317.

Zschocke J, Graham CA (1995) A fluorescent multiplex ARMS method for rapid mutation analysis. Mol Cell Probes 9:447-451.

Zschocke J, Carson DJ (1995) Transient neonatal hyperphenylalaninaemia is not related to mutations at the phenylalanine hydroxylase gene. J Inherit Metab Dis 18:369-370.

Zschocke J, Graham CA, Stewart FJ, Carson DJ, Nevin NC (1994) Non-phenylketonuria hyperphenylalaninaemia in Northern Ireland: frequent mutation allows screening and early diagnosis. Hum Mutat 4:114-118.

Zschocke J, Graham CA, Stewart FJ, Carson DJ, Nevin NC (1994) Automated sequencing detects all mutations in Northern Irish patients with phenylketonuria and mild hyperphenylalaninaemia. Acta Paediatr Suppl 407:37-38.

Zschocke J, Graham CA, McKnight JJ, Nevin NC (1994) The STR system in the human phenylalanine hydroxylase gene: true fragment length obtained with fluorescent labelled PCR primers. Acta Paediatr Suppl 407:41-42.

Guidelines (11)

Toth B, Baston-Büst DM, Behre HM, Bielfeld A, Bohlmann M, Bühling K, Dittrich R, Goeckenjan M, Hancke K, Kliesch S, Köhn FM, Krüssel J, Kuon R, Liebenthron J, Nawroth F, Nordhoff V, Pinggera GM, Rogenhofer N, Rudnik-Schöneborn S, Schuppe HC, Schüring A, Seifert-Klauss V, Strowitzki T, Tüttelmann F, Vomstein K, Wildt L, Wischmann T, Wunder D, Zschocke J (2019) Diagnosis and Therapy Before Assisted Reproductive Treatments. Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Register Number 015-085, February 2019) - Part 1, Basic Assessment of the Woman. Geburtshilfe Frauenheilkd 79:1278-1292.

Toth B, Baston-Büst DM, Behre HM, Bielfeld A, Bohlmann M, Bühling K, Dittrich R, Goeckenjan M, Hancke K, Kliesch S, Köhn FM, Krüssel J, Kuon R, Liebenthron J, Nawroth F, Nordhoff V, Pinggera GM, Rogenhofer N, Rudnik-Schöneborn S, Schuppe HC, Schüring A, Seifert-Klauss V, Strowitzki T, Tüttelmann F, Vomstein K, Wildt L, Wischmann T, Wunder D, Zschocke J (2019) Diagnosis and Treatment Before Assisted Reproductive Treatments. Guideline of the DGGG, OEGGG and SGGG (S2k Level, AWMF Register Number 015-085, February 2019) - Part 2, Hemostaseology, Andrology, Genetics and History of Malignant Disease. Geburtshilfe Frauenheilkd 79:1293-1308.

Toth B, Würfel W, Bohlmann M, Zschocke J, Rudnik-Schöneborn S, Nawroth F, Schleußner E, Rogenhofer N, Wischmann T, von Wolff M, Hancke K, von Otte S, Kuon R, Feil K, Tempfer C (2018) Recurrent Miscarriage: Diagnostic and Therapeutic Procedures. Guideline of the DGGG, OEGGG and SGGG (S2k-Level, AWMF Registry Number 015/050). Geburtshilfe Frauenheilkd 78:364-381.

Zeimet AG, Mori H, Petru E, Polterauer S, Reinthaller A, Schauer C, Scholl-Firon T, Singer C, Wimmer K, Zschocke J, Marth C (2017) AGO Austria recommendation on screening and diagnosis of Lynch syndrome (LS). Arch Gynecol Obstet 296:123-127.

Malfait F, Francomano C, Byers P, Belmont J, Berglund B, Black J, Bloom L, Bowen JM, Brady AF, Burrows NP, Castori M, Cohen H, Colombi M, Demirdas S, De Backer J, De Paepe A, Fournel-Gigleux S, Frank M, Ghali N, Giunta C, Grahame R, Hakim A, Jeunemaitre X, Johnson D, Juul-Kristensen B, Kapferer-Seebacher I, Kazkaz H, Kosho T, Lavallee ME, Levy H, Mendoza-Londono R, Pepin M, Pope FM, Reinstein E, Robert L, Rohrbach M, Sanders L, Sobey GJ, Van Damme T, Vandersteen A, van Mourik C, Voermans N, Wheeldon N, Zschocke J, Tinkle B (2017) The 2017 international classification of the Ehlers-Danlos syndromes. Am J Med Genet C Semin Med Genet 175:8-26.

Boy N, Mühlhausen C, Maier EM, Heringer J, Assmann B, Burgard P, Dixon M, Fleissner S, Greenberg CR, Harting I, Hoffmann GF, Karall D, Koeller DM, Krawinkel MB, Okun JG, Opladen T, Posset R, Sahm K, Zschocke J, Kölker S (2017) Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision. J Inherit Metab Dis 40:75-101.

Hollak CE, Biegstraaten M, Baumgartner MR, Belmatoug N, Bembi B, Bosch A, Brouwers M, Dekker H, Dobbelaere D, Engelen M, Groenendijk MC, Lachmann R, Langendonk JG, Langeveld M, Linthorst G, Morava E, Poll-The BT, Rahman S, Rubio-Gozalbo ME, Spiekerkoetter U, Treacy E, Wanders R, Zschocke J, Hagendijk R (2016) Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet J Rare Dis 11:7.

Marth C, Hubalek M, Petru E, Polterauer S, Reinthaller A, Schauer C, Scholl-Firon T, Singer CF, Zschocke J, Zeimet AG (2015) AGO Austria recommendations for genetic testing of patients with ovarian cancer. Wien Klin Wochenschr 127:652-654.

Zschocke J, Haverkamp T, Moller LB (2012) Clinical utility gene card for: Phenylketonuria. Eur J Hum Genet 20.

Spiekerkoetter U, Lindner M, Santer R, Grotzke M, Baumgartner MR, Boehles H, Das A, Haase C, Hennermann JB, Karall D, de Klerk H, Knerr I, Koch HG, Plecko B, Roschinger W, Schwab KO, Scheible D, Wijburg FA, Zschocke J, Mayatepek E, Wendel U (2009) Treatment recommendations in long-chain fatty acid oxidation defects: consensus from a workshop. J Inherit Metab Dis 32:498-505.

Chalmers RA, Bain MD, Michelakakis H, Zschocke J, Iles RA (2006) Diagnosis and management of trimethylaminuria (FMO3 deficiency) in children. J Inherit Metab Dis 29:162-172.

Reviews (16)

Peters V, Zschocke J, Schmitt CP (2018) Carnosinase, diabetes mellitus and the potential relevance of carnosinase deficiency. J Inherit Metab Dis 41:39-47.

Brady AF, Demirdas S, Fournel-Gigleux S, Ghali N, Giunta C, Kapferer-Seebacher I, Kosho T, Mendoza-Londono R, Pope MF, Rohrbach M, Van Damme T, Vandersteen A, van Mourik C, Voermans N, Zschocke J, Malfait F (2017) The Ehlers-Danlos syndromes, rare types. Am J Med Genet C Semin Med Genet 175:70-115.

Zschocke J (2015) Erbliche Stoffwechselkrankheiten – eine Übersicht. Med Genet (Berlin) 27:268-275.

Wortmann SB, Duran M, Anikster Y, Barth PG, Sperl W, Zschocke J, Morava E, Wevers RA (2013) Inborn errors of metabolism with 3-methylglutaconic aciduria as discriminative feature: proper classification and nomenclature. J Inherit Metab Dis 36:923-928.

Schmuth M, Martinz V, Janecke AR, Fauth C, Schossig A, Zschocke J, Gruber R (2013) Inherited ichthyoses/generalized Mendelian disorders of cornification. Eur J Hum Genet 21:123-133.

Zschocke J (2012) HSD10 disease: clinical consequences of mutations in the HSD17B10 gene. J Inherit Metab Dis 35:81-89.

Zschocke J (2012) [Newborn screening as a predictive genetic test: principles and challenges]. Wien Med Wochenschr 162:168-175.

Schossig A, Wolf NI, Kapferer I, Kohlschutter A, Zschocke J (2012) Epileptic encephalopathy and amelogenesis imperfecta: Kohlschutter-Tonz syndrome. Eur J Med Genet 55:319-322.

Zschocke J (2010) Molecular Basis of Phenylketonuria: From Genotype to Clinical Management. Ann Nestlé 68.

Zschocke J (2008) Dominant versus recessive: molecular mechanisms in metabolic disease. J Inherit Metab Dis 31:599-618.

Zschocke J, Penzien JM, Bielen R, Casals N, Aledo R, Pie J, Hoffmann GF, Hegardt FG, Mayatepek E (2002) The diagnosis of mitochondrial HMG-CoA synthase deficiency. J Pediatr 140:778-780.

Prietsch V, Lindner M, Zschocke J, Nyhan WL, Hoffmann GF (2002) Emergency management of inherited metabolic diseases. J Inherit Metab Dis 25:531-546.

Schrage NF, Langefeld S, Zschocke J, Kuckelkorn R, Redbrake C, Reim M (2000) Eye burns: an emergency and continuing problem. Burns 26:689-699.

Hoffmann GF, Zschocke J (1999) Glutaric aciduria type I: from clinical, biochemical and molecular diversity to successful therapy. J Inherit Metab Dis 22:381-391.

Baric I, Zschocke J, Christensen E, Duran M, Goodman SI, Leonard JV, Muller E, Morton DH, Superti-Furga A, Hoffmann GF (1998) Diagnosis and management of glutaric aciduria type I. J Inherit Metab Dis 21:326-340.

Hoffmann GF, Athanassopoulos S, Burlina AB, Duran M, de Klerk JB, Lehnert W, Leonard JV, Monavari AA, Muller E, Muntau AC, Naughten ER, Plecko-Starting B, Superti-Furga A, Zschocke J, Christensen E (1996) Clinical course, early diagnosis, treatment, and prevention of disease in glutaryl-CoA dehydrogenase deficiency. Neuropediatrics 27:115-123.

Case Reports (56)

Pölsler L, Schatz UA, Simma B, Zschocke J, Rudnik-Schöneborn S (2020) A Syrian patient with Steel syndrome due to compound heterozygous COL27A1 mutations with colobomata of the eye. Am J Med Genet A 182:730-734.

Lepperdinger U, Maurer E, Witsch-Baumgartner M, Stigler R, Zschocke J, Lussi A, Kapferer-Seebacher I (2020) Expanding the phenotype of hypomaturation amelogenesis imperfecta due to a novel SLC24A4 variant. Clin Oral Investig.

Unterberger I, Dobesberger J, Schober H, Krabichler B, Lamina C, Schatz U, Zschocke J, Luef G, Kotzot D, Fauth C (2019) A further case of familial ring chromosome 20 mosaicism - molecular characterization of the ring and review of the literature. Eur J Med Genet 62:103564.

Seidl-Philipp M, Schossig AS, Moosbrugger-Martinz V, Zschocke J, Schmuth M, Gruber R (2019) Impaired epidermal barrier in autosomal recessive congenital ichthyosis (ARCI) caused by missense mutations in SDR9C7 in two Austrian sisters. J Dtsch Dermatol Ges 17:742-745.

Tavasoli AR, Parvaneh N, Ashrafi MR, Rezaei Z, Zschocke J, Rostami P (2018) Clinical presentation and outcome in infantile Sandhoff disease: a case series of 25 patients from Iranian neurometabolic bioregistry with five novel mutations. Orphanet J Rare Dis 13:130.

Alhaddad B, Schossig A, Haack TB, Kovács-Nagy R, Braunisch MC, Makowski C, Senderek J, Vill K, Müller-Felber W, Strom TM, Krabichler B, Freisinger P, Deshpande C, Polster T, Wolf NI, Desguerre I, Wörmann F, Rötig A, Ahting U, Kopajtich R, Prokisch H, Meitinger T, Feichtinger RG, Mayr JA, Jungbluth H, Hubmann M, Zschocke J, Distelmaier F, Koch J (2018) PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics 49:330-338.

Zschocke J, Schossig A, Bosshardt DD, Karall D, Glueckert R, Kapferer-Seebacher I (2017) Variable expressivity of TCTEX1D2 mutations and a possible pathogenic link of molar-incisor malformation to ciliary dysfunction. Arch Oral Biol 80:222-228.

Wimmer K, Beilken A, Nustede R, Ripperger T, Lamottke B, Ure B, Steinmann D, Reineke-Plaass T, Lehmann U, Zschocke J, Valle L, Fauth C, Kratz CP (2017) A novel germline POLE mutation causes an early onset cancer prone syndrome mimicking constitutional mismatch repair deficiency. Fam Cancer 16:67-71.

Tavasoli AR, Shervin Badv R, Zschocke J, Ashrafi MR, Rostami P (2017) Early infantile presentation of 3-methylglutaconic aciduria type 1 with a novel mutation in AUH gene: A case report and literature review. Brain Dev 39:714-716.

Rommer PS, Zschocke J, Fowler B, Födinger M, Konstantopoulou V, Möslinger D, Stögmann E, Suess E, Baumgartner M, Auff E, Sunder-Plassmann G (2017) Manifestations of neurological symptoms and thromboembolism in adults with MTHFR-deficiency. J Neurol Sci 383:123-127.

Gruber R, Rainer G, Weiss A, Udvardi A, Thiele H, Eckl KM, Schupart R, Nürnberg P, Zschocke J, Schmuth M, Volc-Platzer B, Hennies HC (2017) Morphological alterations in two siblings with autosomal recessive congenital ichthyosis associated with CYP4F22 mutations. Br J Dermatol 176:1068-1073.

Rudnicki M, Mayr JA, Zschocke J, Antretter H, Regele H, Feichtinger RG, Windpessl M, Mayer G, Pölzl G (2016) MELAS Syndrome and Kidney Disease Without Fanconi Syndrome or Proteinuria: A Case Report. Am J Kidney Dis 68:949-953.

Del Rizzo M, Galderisi A, Celato A, Furlan F, Giordano L, Cazzorla C, Fasan I, Moretti C, Zschocke J, Burlina AB (2016) The long-term treatment of a patient with type 1 diabetes mellitus and glutaric aciduria type 1: the effect of insulin. Eur J Pediatr 175:1123-1128.

Höck M, Wegleiter K, Ralser E, Kiechl-Kohlendorfer U, Scholl-Bürgi S, Fauth C, Steichen E, Pichler K, Lefeber DJ, Matthjis G, Keldermans L, Maurer K, Zschocke J, Karall D (2015) ALG8-CDG: novel patients and review of the literature. Orphanet J Rare Dis 10:73.

Opladen T, Ebinger F, Zschocke J, Sengupta D, Ben-Omran T, Shahbeck N, Moog U, Fischer C, Burger F, Haas D, Ruef P, Harting I, Al-Rifai H, Hoffmann GF (2014) Aspartylglucosaminuria: unusual neonatal presentation in qatari twins with a novel aspartylglucosaminidase gene mutation and 3 new cases in a Turkish family. J Child Neurol 29:36-42.

Haberlandt E, Zotter S, Witsch-Baumgartner M, Zschocke J, Kotzot D (2014) Don't miss patients with atypical FMR1 mutations: dysmorphism and clinical features in a boy with a partially methylated FMR1 full mutation. Eur J Pediatr 173:1257-1261.

Tönz O, Steiner B, Schossig A, Zschocke J, Kohlschütter A (2013) Hereditäre epileptische Enzephalopathie mit Amelogenesis imperfect: Kohlschütter-Tönz-Syndrom (KTZS). Epileptologie 30:167-174.

Gruber R, Schossig AS, Alnutaifi KA, Martinz V, Blunder S, Zschocke J, Schmuth M, Tinschert S (2013) Slow growth of hair and nails, craniofacial abnormalities and brachyphalangy. J Dtsch Dermatol Ges.

Fruhmesser A, Erdel M, Duba HC, Fauth C, Amberger A, Utermann G, Zschocke J, Kotzot D (2013) Combined Dup(7)(q22.1q32.2), Inv(7)(q31.31q31.33), and Ins(7;19)(q22.1;p13.2p13.2) in a 12-year-old boy with developmental delay and various dysmorphism. Eur J Med Genet 56:383-388.

Fruhmesser A, Blake J, Haberlandt E, Baying B, Raeder B, Runz H, Spreiz A, Fauth C, Benes V, Utermann G, Zschocke J, Kotzot D (2013) Disruption of EXOC6B in a patient with developmental delay, epilepsy, and a de novo balanced t(2;8) translocation. Eur J Hum Genet 21:1177-1180.

Blassnig-Ezeh A, Bandelier C, Fruhmesser A, Revencu N, Krabichler B, Beauloye V, Ravoet M, Fauth C, Zschocke J, Simma B, Kotzot D (2013) Severe growth retardation, delayed bone age, and facial dysmorphism in two patients with microduplications in 2p16 --> p22. Am J Med Genet A 161A:3176-3181.

Baas AF, Gabbett M, Rimac M, Kansikas M, Raphael M, Nievelstein RA, Nicholls W, Offerhaus J, Bodmer D, Wernstedt A, Krabichler B, Strasser U, Nystrom M, Zschocke J, Robertson SP, van Haelst MM, Wimmer K (2013) Agenesis of the corpus callosum and gray matter heterotopia in three patients with constitutional mismatch repair deficiency syndrome. Eur J Hum Genet 21:55-61.

Mayr JA, Koch J, Fauth C, Zimmermann FA, Rauscher C, Zschocke J, Sperl W (2012) A 1.1 million base pair X-chromosomal deletion covering the PDHA1 and CDKL5 genes in a female patient with West syndrome and pyruvate oxidation deficiency. Neuropediatrics 43:130-134.

Haberlandt E, Spreiz A, Sigl SB, Janetschek C, Rothlisberger B, Zotter S, Rostasy K, Zschocke J, Kotzot D (2012) Microdeletion 19p13.2 in an almost 5-year-old boy. Am J Med Genet A 158A:1190-1194.

Fruhmesser A, Haberlandt E, Judmaier W, Schinzel A, Utermann B, Erdel M, Fauth C, Utermann G, Zschocke J, Kotzot D (2012) Effects of deletion and duplication in a patient with a 46,XX,der(7)t(7;17)(q36;p13)mat karyotype. Am J Med Genet A 158A:2239-2244.

Laccone F, Schoner K, Krabichler B, Kluge B, Schwerdtfeger R, Schulze B, Zschocke J, Rehder H (2011) Desbuquois dysplasia type I and fetal hydrops due to novel mutations in the CANT1 gene. Eur J Hum Genet 19:1133-1137.

Spreiz A, Muller D, Zotter S, Albrecht U, Baumann M, Fauth C, Erdel M, Zschocke J, Utermann G, Kotzot D (2010) Phenotypic variability of a deletion and duplication 6q16.1 --> q21 due to a paternal balanced ins(7;6)(p15;q16.1q21). Am J Med Genet A 152A:2762-2767.

Binkert F, Spreiz A, Hockner M, Miny P, von Dach Leu B, Erdel M, Zschocke J, Utermann G, Kotzot D (2010) Parental origin and mechanism of formation of a 46,X,der(X)(pter-->q21.1::p11.4-->pter)/45,X karyotype in a woman with mild Turner syndrome. Fertil Steril 94:350 e312-355.

Kohlschutter B, Ellerbrok M, Merkel M, Tchirikov M, Zschocke J, Santer R, Ullrich K (2009) Phenylalanine tolerance in three phenylketonuric women pregnant with fetuses of different genetic PKU status. J Inherit Metab Dis 32 Suppl 1:S1-4.

Grossmann V, Muller D, Muller W, Fresser F, Erdel M, Janecke AR, Zschocke J, Utermann G, Kotzot D (2009) "Essentially" pure trisomy 3q27 --> qter: further delineation of the partial trisomy 3q phenotype. Am J Med Genet A 149A:2522-2526.

Bagci S, Zschocke J, Hoffmann GF, Bast T, Klepper J, Muller A, Heep A, Bartmann P, Franz AR (2009) Pyridoxal phosphate-dependent neonatal epileptic encephalopathy. BMJ Case Rep 2009.

Ruiz A, Garcia-Villoria J, Ormazabal A, Zschocke J, Fiol M, Navarro-Sastre A, Artuch R, Vilaseca MA, Ribes A (2008) A new fatal case of pyridox(am)ine 5'-phosphate oxidase (PNPO) deficiency. Mol Genet Metab 93:216-218.

Gotthardt D, Runz H, Keitel V, Fischer C, Flechtenmacher C, Wirtenberger M, Weiss KH, Imparato S, Braun A, Hemminki K, Stremmel W, Ruschendorf F, Stiehl A, Kubitz R, Burwinkel B, Schirmacher P, Knisely AS, Zschocke J, Sauer P (2008) A mutation in the canalicular phospholipid transporter gene, ABCB4, is associated with cholestasis, ductopenia, and cirrhosis in adults. Hepatology 48:1157-1166.

Bagci S, Zschocke J, Hoffmann GF, Bast T, Klepper J, Muller A, Heep A, Bartmann P, Franz AR (2008) Pyridoxal phosphate-dependent neonatal epileptic encephalopathy. Arch Dis Child Fetal Neonatal Ed 93:F151-152.

Seel C, Hager HD, Jauch A, Tariverdian G, Zschocke J (2005) Survival up to age 10 years in a patient with partial duplication 6q: case report and review of the literature. Clin Dysmorphol 14:51-54.

Knerr I, Zschocke J, Schellmoser S, Topf HG, Weigel C, Dotsch J, Rascher W (2005) An exceptional Albanian family with seven children presenting with dysmorphic features and mental retardation: maternal phenylketonuria. BMC Pediatr 5:5.

Haas D, Armbrust S, Haas JP, Zschocke J, Muhlmann K, Fusch C, Neumann LM (2005) Smith-Lemli-Opitz syndrome with a classical phenotype, oesophageal achalasia and borderline plasma sterol concentrations. J Inherit Metab Dis 28:1191-1196.

Wolf NI, Willemsen MA, Engelke UF, van der Knaap MS, Pouwels PJ, Harting I, Zschocke J, Sistermans EA, Rating D, Wevers RA (2004) Severe hypomyelination associated with increased levels of N-acetylaspartylglutamate in CSF. Neurology 62:1503-1508.

Sass JO, Sander S, Zschocke J (2004) Isobutyryl-CoA dehydrogenase deficiency: isobutyrylglycinuria and ACAD8 gene mutations in two infants. J Inherit Metab Dis 27:741-745.

Illsinger S, Lucke T, Zschocke J, Gibson KM, Das AM (2004) 3-methylglutaconic aciduria type I in a boy with fever-associated seizures. Pediatr Neurol 30:213-215.

Assmann BE, Robinson RO, Surtees RA, Brautigam C, Heales SJ, Wevers RA, Zschocke J, Hyland K, Sharma R, Hoffmann GF (2004) Infantile Parkinsonism-dystonia and elevated dopamine metabolites in CSF. Neurology 62:1872-1874.

Zschocke J, Schaefer JR (2003) Homozygous familial hypercholesterolaemia in identical twins. Lancet 361:1641.

Wolf NI, Rahman S, Clayton PT, Zschocke J (2003) Mitochondrial HMG-CoA synthase deficiency: identification of two further patients carrying two novel mutations. Eur J Pediatr 162:279-280.

Mazon Ramos A, Gil-Setas A, Berrade Zubiri S, Bandres Echeverri T, Wevers R, Engelke U, Zschocke J (2003) [Primary trimethylaminuria or fish odor syndrome. A novel mutation in the first documented case in Spain]. Med Clin (Barc) 120:219-221.

Zschocke J, Schindler S, Hoffmann GF, Albani M (2002) Nature and nurture in vitamin B12 deficiency. Arch Dis Child 87:75-76.

Lassker U, Zschocke J, Blau N, Santer R (2002) Tetrahydrobiopterin responsiveness in phenylketonuria. Two new cases and a review of molecular genetic findings. J Inherit Metab Dis 25:65-70.

Knerr I, Zschocke J, Trautmann U, Dorland L, de Koning TJ, Muller P, Christensen E, Trefz FK, Wundisch GF, Rascher W, Hoffmann GF (2002) Glutaric aciduria type III: a distinctive non-disease? J Inherit Metab Dis 25:483-490.

Bahr O, Mader I, Zschocke J, Dichgans J, Schulz JB (2002) Adult onset glutaric aciduria type I presenting with a leukoencephalopathy. Neurology 59:1802-1804.

Kolker S, Ramaekers VT, Zschocke J, Hoffmann GF (2001) Acute encephalopathy despite early therapy in a patient with homozygosity for E365K in the glutaryl-coenzyme A dehydrogenase gene. J Pediatr 138:277-279.

Kasim S, Moo LR, Zschocke J, Jinnah HA (2001) Phenylketonuria presenting in adulthood as progressive spastic paraparesis with dementia. J Neurol Neurosurg Psychiatry 71:795-797.

Zafeiriou DI, Zschocke J, Augoustidou-Savvopoulou P, Mauromatis I, Sewell A, Kontopoulos E, Katzos G, Hoffmann GF (2000) Atypical and variable clinical presentation of glutaric aciduria type I. Neuropediatrics 31:303-306.

Zschocke J, Quak E, Knauer A, Fritz B, Aslan M, Hoffmann GF (1999) Large heterozygous deletion masquerading as homozygous missense mutation: a pitfall in diagnostic mutation analysis. J Inherit Metab Dis 22:687-692.

Zschocke J, Koch G, Hoffmann GF (1999) Urinary excretion of Krebs cycle metabolites in a 13-year-old girl. J Inherit Metab Dis 22:945.

Nyhan WL, Zschocke J, Hoffmann G, Stein DE, Bao L, Goodman S (1999) Glutaryl-CoA dehydrogenase deficiency presenting as 3-hydroxyglutaric aciduria. Mol Genet Metab 66:199-204.

Weglage J, van Teefelen-Heithoff A, Zschocke J, Zass R, Marquardt T, Koch HG (1998) Delayed onset of phenylketonuria. Eur J Pediatr 157:171.

Tyfield LA, Zschocke J, Stephenson A, Cockburn F, Harvie A, Bidwell JL, Wood NA, Hunt LP (1995) Discordant phenylketonuria phenotypes in one family: the relationship between genotype and clinical outcome is a function of multiple effects. J Med Genet 32:867-870.

Editorials, Letters (8)

Burgard P, Ullrich K, Ballhausen D, Hennermann JB, Hollak CEM, Langeveld M, Karall D, Konstantopoulou V, Maier EM, Lang F, Lachmann R, Murphy E, Garbade S, Hoffmann GF, Kölker S, Lindner M, Zschocke J (2017) Issues with European guidelines for phenylketonuria. Lancet Diabetes Endocrinol 5:681-683.

Zschocke J, Baumgartner MR, Morava E, Patterson MC, Peters V, Rahman S (2016) Recommendations and guidelines in the JIMD: suggested procedures and avoidance of conflicts of interest. J Inherit Metab Dis 39:327-329.

Rahman S, Baumgartner MR, Morava E, Patterson M, Peters V, Zschocke J (2016) Peer review fraud-it's not big and it's not clever. J Inherit Metab Dis 39:1-2.

Zschocke J (2015) Erbliche Stoffwechselkrankheiten. Med Genet (Berlin) 27:267.

Morava E, Rahman S, Peters V, Baumgartner MR, Patterson M, Zschocke J (2015) Quo vadis: the re-definition of "inborn metabolic diseases". J Inherit Metab Dis 38:1003-1006.

Karall D, Scholl-Bürgi S, Zschocke J (2015) "Crossing borders" SSIEM 2014 annual symposium in Innsbruck. J Inherit Metab Dis 38:619.

Zschocke J (2003) Focus on the molecular genetics of phenylketonuria. Hum Mutat 21:331-332.

Zschocke J, Hoffmann GF (2000) PAH gene mutation analysis in clinical practice--comments on mutation analysis anticipates dietary requirements in phenylketonuria. Eur J Pediatr 159 Suppl 2:S154-155.

Book Chapters, Electronic Media (13)

Zschocke J (2020) Therapie genetisch bedingter Krankheiten. In: Pädiatrie: Grundlagen und Praxis, Hoffmann GF, Lentze MJ, Spranger J, Zepp F, Berner R, eds. Heidelberg, Springer, in press.

Zschocke J, Tinschert S (2014) Genetic counselling of Inborn Errors of Metabolism. In: Physician’s Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases, Blau N, Duran M, Gibson KM, Dinosi-Vici C, eds. Heidelberg, Springer, 56-75.

Zschocke J (2014) SSIEM Classification of Inborn Errors of Metabolism. In: Physician’s Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases, Blau N, Duran M, Gibson KM, Dinosi-Vici C, eds. Heidelberg, Springer, 817-830.

Zschocke J (2014) Grundlagen der Vererbung. In: Medizinische Genetik für die Praxis: Diagnostik, Beratung, Fallbeispiele, Moog U, Ries O, eds. Stuttgart, Thieme, 56-75.

Zschocke J (2009) Molecular Testing for Endocrine & Metabolic Disorders. In: Pediatric Endocrinology and Inborn Errors of Metabolism, Sarafoglou K, Hoffmann GF, Roth KS, eds. New York, McGraw Hill, 33-36.

Zschocke J (2007) Genetik in der Pädiatrie. In: Lehrbuch Pädiatrie, Mayatepek E, ed. München, Elsevier, 57-71.

Zschocke J (2003) Fallbeiträge und Expertenkommentare In: Campus Pädiatrie interaktiv [Elektronische Ressource]: interaktives fallbasiertes Lernen in der Kinderheilkunde, Tönshoff B, Köpf S, Singer R, Hoffmann GF, eds. Berlin, Springer.

Hoffmann GF, Zschocke J (2002) Erbliche proteinabhängige Stoffwechselstörungen - Aminosäurenstoffwechsel und Harnstoffzyklus. In: Angeborene Stoffwechselkrankheiten: Genetik, Diagnostik, Therapie, Strohmeyer G, Stremmel W, Niederau C, eds. Landsberg, Ecomed, 53-93.

Zschocke J, Hoffmann GF (2001) Cycle Sequencing of Polymerase Chain Reaction-Amplified Genomic DNA with Dye-Labeled Universal Primers. In: DNA Sequencing Protocols, Graham CA, Hill AJM, eds. Totowa, Humana Press, 113-117.

Wanders RJA, Ruiter JPN, Kuiper S, Brand J, Lindner M, Hoffmann GF, Mayatepek E, Zschocke J (2001) 2-Methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, a new disorder of isoleucine and branched-chain fatty acid degradation: studies on the forward and reverse reactions. In: New Avenues of Research in Fatty Acid Oxidation and Ketone Body Metabolism, Eaton S, Quant PA, eds. London, FAOXK Press, 59-61.

Kölker S, Liesert M, Zschocke J, Hoffmann GF (2001) Glutaryl-CoA dehydrogenase. In: Encyclopedia of Molecular Medicine, Creighton TE, ed. New York, John Wiley & Sons, 1468-1470.

Zschocke J, Hoffmann GF (1999) Organoazidurien. In: Handbuch der Molekularen Medizin, Band 6: Ausgewählte monogene Erkrankungen, Ganten D, Ruckpaul K, eds. Berlin, Springer, 253-277.

Zschocke J, Hoffmann GF (1998) Erbliche Stoffwechselkrankheiten. In: Kompendium Endokrinopathien und Stoffwechselkrankheiten, Köbberling J, Tillil H, von Boxberg C, eds. Berlin, Multimedica / Online-Dienst bsmedic.

Books

Zschocke, J., Hoffmann, G. F. (2020). Vademecum Metabolicum: Diagnosis and Treatment of Inherited Metabolic Disorders. 5th Edition. Stuttgart: Thieme

- 1st Edition 1997 (German), Friedrichsdorf: Milupa; Translations: Hungarian, Italian
- 2nd Edition 1999 (German), Stuttgart: Schattauer; Translations: English, Italian
- 3rd Edition 2004 (German), Stuttgart: Schattauer; Translations: English, Italian, French, Polish, Portuguese, Japanese, Chinese
- 4th Edition 2011 (German), Stuttgart: Schattauer; Translations: English, Italian, French, Japanese, Farsi/Persian, Russian

Schaaf, C. P., Zschocke, J. (2018). Basiswissen Humangenetik, 3rd Edition. Heidelberg: Springer

- 1st Edition 2008
- 2nd Edition 2013

Hoffmann, G. F., Zschocke, J., Nyhan, W. L., eds. (2017) Inherited metabolic diseases – A clinical approach. 2nd Edition. Heidelberg: Springer. (Various own chapters)

- 1st Edition 2010

Schaaf, C. P., Zschocke, J., Potocki, L. (2012) Human Genetics: From Molecules to Medicine. Baltimore, Lippincott Williams & Wilkins

Zschocke, J. (2005) Erbliche Stoffwechselkrankheiten: Vom Genotyp zum Phänotyp. Heilbronn: SPS Verlagsgesellschaft

Hoffmann, G. F., Nyhan, W. L., Zschocke, J., Kahler, S. G., Mayatepek, E. (2001) Inherited metabolic diseases. Baltimore: Lippincott Williams & Wilkins.

- Translations: Czech, Russian