Publications of the Mito-Center Salzburg/Munich 2013-2018

2018

Zimmermann, F.A., Neureiter, D., **Sperl, W.**, **Mayr, J.A.**, and **Kofler, B.** (2018). Alterations of Oxidative Phosphorylation Complexes in Papillary Thyroid Carcinoma. Cells 7.

Yepez, V.A., **Kremer, L.S.**, **Iuso, A.**, **Gusic, M.**, **Kopajtich, R.**, **Konarikova, E.**, Nadel, A., Wachutka, L., **Prokisch, H.**, and Gagneur, J. (2018). OCR-Stats: Robust estimation and statistical testing of mitochondrial respiration activities using Seahorse XF Analyzer. PLoS One 13, e0199938.

Xu, Z., Lo, W.S., Beck, D.B., Schuch, L.A., Olahova, M., **Kopajtich, R.**, Chong, Y.E., Alston, C.L., Seidl, E., Zhai, L., Lau, C.F., Timchak, D., LeDuc, C.A., Borczuk, A.C., Teich, A.F., Juusola, J., Sofeso, C., Muller, C., Pierre, G., Hilliard, T., Turnpenny, P.D., **Wagner, M.**, Kappler, M., Brasch, F., Bouffard, J.P., Nangle, L.A., Yang, X.L., Zhang, M., Taylor, R.W., **Prokisch, H.**, Griese, M., Chung, W.K., and Schimmel, P. (2018). Bi-allelic Mutations in Phe-tRNA Synthetase Associated with a Multi-system Pulmonary Disease Support Non-translational Function. Am J Hum Genet 103, 100-114.

Waich, S., Roscher, A., Brunner-Krainz, M., Cortina, G., Kostl, G., **Feichtinger, R.G.**, Entenmann, A., Muller, T., Knisely, A.S., **Mayr, J.A.**, Janecke, A.R., and Vodopiutz, J. (2018). Severe DGUOK Deficiency in Austria: A Six-Patient Series. J Pediatr Gastroenterol Nutr.

van Karnebeek, C.D.M., **Wortmann, S.B.**, Tarailo-Graovac, M., Langeveld, M., Ferreira, C.R., van de Kamp, J.M., Hollak, C.E., Wasserman, W.W., Waterham, H.R., Wevers, R.A., Haack, T.B., Wanders, R.J.A., and Boycott, K.M. (2018). The role of the clinician in the multi-omics era: are you ready? J Inherit Metab Dis 41, 571-582.

Stenton, S.L., and **Prokisch, H.** (2018). Advancing genomic approaches to the molecular diagnosis of mitochondrial disease. Essays Biochem 62, 399-408.

Roeben, B., Schule, R., Ruf, S., Bender, B., **Alhaddad, B.**, Benkert, T., **Meitinger, T.**, Reich, S., Bohringer, J., Langhans, C.D., Vaz, F.M., **Wortmann, S.B.**, Marquardt, T., Haack, T.B., Krageloh-Mann, I., Schols, L., and Synofzik, M. (2018). SERAC1 deficiency causes complicated HSP: evidence from a novel splice mutation in a large family. J Med Genet 55, 39-47.

Repp, B.M., **Mastantuono, E.**, Alston, C.L., Schiff, M., Haack, T.B., Rotig, A., Ardissone, A., Lombes, A., Catarino, C.B., Diodato, D., Schottmann, G., Poulton, J., Burlina, A., Jonckheere, A., Munnich, A., Rolinski, B., Ghezzi, D., Rokicki, D., Wellesley, D., Martinelli, D., Wenhong, D., Lamantea, E., Ostergaard, E., Pronicka, E., Pierre, G., Smeets, H.J.M., Wittig, I., Scurr, I., de Coo, I.F.M., Moroni, I., Smet, J., **Mayr, J.A.**, Dai, L., de Meirleir, L., Schuelke, M., Zeviani, M., Morscher, R.J., McFarland, R., Seneca, S., Klopstock, T., **Meitinger, T.**, Wieland, T., Strom, T.M., Herberg, U., **Ahting, U.**, **Sperl, W.**, Nassogne, M.C., Ling, H., Fang, F., **Freisinger, P.**, Van Coster, R., Strecker, V., Taylor, R.W., Haberle, J., Vockley, J., **Prokisch, H.**, and **Wortmann, S.** (2018). Clinical, biochemical and genetic spectrum of 70 patients with ACAD9 deficiency: is riboflavin supplementation effective? Orphanet J Rare Dis 13, 120.

Piekutowska-Abramczuk, D., Assouline, Z., **Matakovic, L.**, **Feichtinger, R.G.**, **Konarikova, E.**, Jurkiewicz, E., Stawinski, P., **Gusic, M.**, Koller, A., Pollak, A., Gasperowicz, P., Trubicka, J., Ciara,

E., Iwanicka-Pronicka, K., Rokicki, D., Hanein, S., **Wortmann, S.B.**, **Sperl, W.**, Rotig, A., **Prokisch, H.**, Pronicka, E., Ploski, R., Barcia, G., and **Mayr, J.A.** (2018). NDUFB8 Mutations Cause Mitochondrial Complex I Deficiency in Individuals with Leigh-like Encephalomyopathy. Am J Hum Genet 102, 460-467.

Olahova, M., Yoon, W.H., Thompson, K., Jangam, S., Fernandez, L., Davidson, J.M., Kyle, J.E., Grove, M.E., Fisk, D.G., Kohler, J.N., Holmes, M., Dries, A.M., Huang, Y., Zhao, C., Contrepois, K., Zappala, Z., Fresard, L., Waggott, D., Zink, E.M., Kim, Y.M., Heyman, H.M., Stratton, K.G., Webb-Robertson, B.M., Undiagnosed Diseases, N., Snyder, M., Merker, J.D., Montgomery, S.B., Fisher, P.G., **Feichtinger, R.G.**, **Mayr, J.A.**, Hall, J., Barbosa, I.A., Simpson, M.A., Deshpande, C., Waters, K.M., Koeller, D.M., Metz, T.O., Morris, A.A., Schelley, S., Cowan, T., Friederich, M.W., McFarland, R., Van Hove, J.L.K., Enns, G.M., Yamamoto, S., Ashley, E.A., Wangler, M.F., Taylor, R.W., Bellen, H.J., Bernstein, J.A., and Wheeler, M.T. (2018). Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. Am J Hum Genet 102, 494-504.

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

O'Byrne, J.J., Tarailo-Graovac, M., Ghani, A., Champion, M., Deshpande, C., Dursun, A., Ozgul, R.K., **Freisinger, P.**, Garber, I., Haack, T.B., Horvath, R., Baric, I., Husain, R.A., Kluijtmans, L.A.J., Kotzaeridou, U., Morris, A.A., Ross, C.J., Santra, S., Smeitink, J., Tarnopolsky, M., **Wortmann, S.B.**, **Mayr, J.A.**, Brunner-Krainz, M., **Prokisch, H.**, Wasserman, W.W., Wevers, R.A., Engelke, U.F., Rodenburg, R.J., Ting, T.W., McFarland, R., Taylor, R.W., Salvarinova, R., and van Karnebeek, C.D.M. (2018). The genotypic and phenotypic spectrum of MTO1 deficiency. Mol Genet Metab 123, 28-42.

Niedermayr, K., Polzl, G., Scholl-Burgi, S., Fauth, C., Schweigmann, U., Haberlandt, E., Albrecht, U., Zlamy, M., **Sperl, W.**, **Mayr, J.A.**, and Karall, D. (2018). Mitochondrial DNA mutation "m.3243A>G"-Heterogeneous clinical picture for cardiologists ("m.3243A>G": A phenotypic chameleon). Congenit Heart Dis.

Morscher, R.J., Ducker, G.S., Li, S.H., Mayer, J.A., Gitai, Z., **Sperl, W.**, and Rabinowitz, J.D. (2018). Mitochondrial translation requires folate-dependent tRNA methylation. Nature 554, 128-132.

Milev, M.P., Graziano, C., Karall, D., Kuper, W.F.E., Al-Deri, N., Cordelli, D.M., Haack, T.B., Danhauser, K., **Iuso, A.**, Palombo, F., Pippucci, T., **Prokisch, H.**, Saint-Dic, D., Seri, M., Stanga, D., Cenacchi, G., van Gassen, K.L.I., Zschocke, J., Fauth, C., **Mayr, J.A.**, Sacher, M., and van Hasselt, P.M. (2018). Bi-allelic mutations in TRAPPC2L result in a neurodevelopmental disorder and have an impact on RAB11 in fibroblasts. J Med Genet.

Lotz-Havla, A.S., Roschinger, W., Schiergens, K., Singer, K., Karall, D., Konstantopoulou, V., **Wortmann, S.B.**, and Maier, E.M. (2018). Fatal pitfalls in newborn screening for mitochondrial trifunctional protein (MTP)/long-chain 3-Hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency. Orphanet J Rare Dis 13, 122.

Kremer, L.S., **Wortmann, S.B.**, and **Prokisch, H.** (2018). "Transcriptomics": molecular diagnosis of inborn errors of metabolism via RNA-sequencing. J Inherit Metab Dis 41, 525-532.

Kovacs-Nagy, R., Morin, G., Nouri, M.A., Brandau, O., Saadi, N.W., Nouri, M.A., **van den Broek, F.**, **Prokisch, H.**, **Mayr, J.A.**, and **Wortmann, S.B.** (2018). HTRA2 Defect: A Recognizable Inborn Error of Metabolism with 3-Methylglutaconic Aciduria as Discriminating Feature Characterized by Neonatal Movement Disorder and Epilepsy-Report of 11 Patients. Neuropediatrics.

Iuso, A., Wiersma, M., Schuller, H.J., Pode-Shakked, B., Marek-Yagel, D., Grigat, M., Schwarzmayr, T., Berutti, R., **Alhaddad, B.**, Kanon, B., Grzeschik, N.A., Okun, J.G., Perles, Z., Salem, Y., Barel, O., Vardi, A., Rubinshtein, M., Tirosh, T., Dubnov-Raz, G., Messias, A.C., Terrile, C., Barshack, I., Volkov, A., Avivi, C., Eyal, E., **Mastantuono, E.**, Kumbar, M., Abudi, S., Braunisch, M., Strom, T.M., **Meitinger, T.**, Hoffmann, G.F., **Prokisch, H.**, Haack, T.B., Brundel, B., Haas, D., Sibon, O.C.M., and Anikster, Y. (2018). Mutations in PPCS, Encoding Phosphopantothenoylcysteine Synthetase, Cause Autosomal-Recessive Dilated Cardiomyopathy. Am J Hum Genet 102, 1018-1030.

Iuso, A., **Alhaddad, B.**, Weigel, C., Kotzaeridou, U., **Mastantuono, E.**, Schwarzmayr, T., Graf, E., Terrile, C., **Prokisch, H.**, Strom, T.M., Hoffmann, G.F., **Meitinger, T.**, and Haack, T.B. (2018). A Homozygous Splice Site Mutation in SLC25A42, Encoding the Mitochondrial Transporter of Coenzyme A, Causes Metabolic Crises and Epileptic Encephalopathy. JIMD Rep.

Gauthier, J., Meijer, I.A., Lessel, D., Mencacci, N.E., Krainc, D., Hempel, M., Tsiakas, K., **Prokisch, H.**, Rossignol, E., Helm, M.H., Rodan, L.H., Karamchandani, J., Carecchio, M., Lubbe, S.J., Telegrafi, A., Henderson, L.B., Lorenzo, K., Wallace, S.E., Glass, I.A., Hamdan, F.F., Michaud, J.L., Rouleau, G.A., and Campeau, P.M. (2018). Recessive mutations in >VPS13D cause childhood onset movement disorders. Ann Neurol 83, 1089-1095.

Gardeitchik, T., Mohamed, M., Ruzzenente, B., Karall, D., Guerrero-Castillo, S., Dalloyaux, D., van den Brand, M., van Kraaij, S., van Asbeck, E., Assouline, Z., Rio, M., de Lonlay, P., Scholl-Buergi, S., Wolthuis, D., Hoischen, A., Rodenburg, R.J., **Sperl, W.**, Urban, Z., Brandt, U., **Mayr, J.A.**, Wong, S., de Brouwer, A.P.M., Nijtmans, L., Munnich, A., Rotig, A., Wevers, R.A., Metodiev, M.D., and Morava, E. (2018). Bi-allelic Mutations in the Mitochondrial Ribosomal Protein MRPS2 Cause Sensorineural Hearing Loss, Hypoglycemia, and Multiple OXPHOS Complex Deficiencies. Am J Hum Genet 102, 685-695.

Feichtinger, R.G., Lang, R., Geilberger, R., Rathje, F., **Mayr, J.A.**, **Sperl, W.**, Bauer, J.W., Hauser-Kronberger, C., **Kofler, B.**, and Emberger, M. (2018). Melanoma tumors exhibit a variable but distinct metabolic signature. Exp Dermatol 27, 204-207.

Coene, K.L.M., Kluijtmans, L.A.J., van der Heeft, E., Engelke, U.F.H., de Boer, S., Hoegen, B., Kwast, H.J.T., van de Vorst, M., Huigen, M., Keularts, I., Schreuder, M.F., van Karnebeek, C.D.M., **Wortmann, S.B.**, de Vries, M.C., Janssen, M.C.H., Gilissen, C., Engel, J., and Wevers, R.A. (2018). Next-generation metabolic screening: targeted and untargeted metabolomics for the diagnosis of inborn errors of metabolism in individual patients. J Inherit Metab Dis 41, 337-353.

Charif, M., Nasca, A., Thompson, K., Gerber, S., **Makowski, C.**, Mazaheri, N., Bris, C., Goudenege, D., Legati, A., Maroofian, R., Shariati, G., Lamantea, E., Hopton, S., Ardissone, A., Moroni, I., Giannotta, M., Siegel, C., Strom, T.M., **Prokisch, H.**, Vignal-Clermont, C., Derrien, S., Zanlonghi, X., Kaplan, J., Hamel, C.P., Leruez, S., Procaccio, V., Bonneau, D., Reynier, P., White, F.E., Hardy, S.A., Barbosa, I.A., Simpson, M.A., Vara, R., Perdomo Trujillo, Y., Galehdari, H., Deshpande, C., Haack, T.B., Rozet, J.M., Taylor, R.W., Ghezzi, D., Amati-Bonneau, P., and Lenaers, G. (2018). Neurologic Phenotypes Associated With Mutations in RTN4IP1 (OPA10) in Children and Young Adults. JAMA Neurol 75, 105-113.

Bruni, F., Di Meo, I., Bellacchio, E., Webb, B.D., McFarland, R., Chrzanowska-Lightowlers, Z.M.A., He, L., Skorupa, E., Moroni, I., Ardissone, A., Walczak, A., Tyynismaa, H., Isohanni, P., Mandel, H., **Prokisch, H.**, Haack, T., Bonnen, P.E., Enrico, B., Pronicka, E., Ghezzi, D., Taylor, R.W., and Diodato, D. (2018). Clinical, biochemical, and genetic features associated with VARS2-related mitochondrial disease. Hum Mutat 39, 563-578.

Alston, C.L., Heidler, J., Dibley, M.G., **Kremer, L.S.**, Taylor, L.S., Fratter, C., French, C.E., Glasgow, R.I.C., **Feichtinger, R.G.**, Delon, I., Pagnamenta, A.T., Dolling, H., Lemonde, H., Aiton, N., Bjornstad, A., Henneke, L., Gartner, J., Thiele, H., Tauchmannova, K., Quaghebeur, G., Houstek, J., **Sperl, W.**, Raymond, F.L., **Prokisch, H.**, **Mayr, J.A.**, McFarland, R., Poulton, J., Ryan, M.T., Wittig, I., Henneke, M., and Taylor, R.W. (2018). Bi-allelic Mutations in NDUFA6 Establish Its Role in Early-Onset Isolated Mitochondrial Complex I Deficiency. Am J Hum Genet 103, 592-601.

Alhaddad, B., Schossig, A., Haack, T.B., **Kovacs-Nagy, R.**, Braunisch, M.C., **Makowski, C.**, Senderek, J., Vill, K., Muller-Felber, W., Strom, T.M., Krabichler, B., **Freisinger, P.**, Deshpande, C., Polster, T., Wolf, N.I., Desguerre, I., Wormann, F., Rotig, A., **Ahting, U.**, **Kopajtich, R.**, **Prokisch, H.**, **Meitinger, T.**, **Feichtinger, R.G.**, **Mayr, J.A.**, Jungbluth, H., Hubmann, M., Zschocke, J., Distelmaier, F., and **Koch, J.** (2018). PRUNE1 Deficiency: Expanding the Clinical and Genetic Spectrum. Neuropediatrics 49, 330-338.

2017

Wortmann, S.B., Timal, S., Venselaar, H., Wintjes, L.T., **Kopajtich, R.**, **Feichtinger, R.G.**, Onnekink, C., Muhlmeister, M., Brandt, U., Smeitink, J.A., Veltman, J.A., **Sperl, W.**, Lefeber, D., Pruijn, G., Stojanovic, V., **Freisinger, P.**, F, V.S., Derks, T.G., Veenstra-Knol, H.E., **Mayr, J.A.**, Rotig, A., Tarnopolsky, M., **Prokisch, H.**, and Rodenburg, R.J. (2017). Biallelic variants in WARS2 encoding mitochondrial tryptophanyl-tRNA synthase in six individuals with mitochondrial encephalopathy. Hum Mutat 38, 1786-1795.

Wortmann, S.B., **Mayr, J.A.**, Nuoffer, J.M., **Prokisch, H.**, and **Sperl, W.** (2017). A Guideline for the Diagnosis of Pediatric Mitochondrial Disease: The Value of Muscle and Skin Biopsies in the Genetics Era. Neuropediatrics 48, 309-314.

Wortmann, S.B., Chen, M.A., Colombo, R., Pontoglio, A., **Alhaddad, B.**, Botto, L.D., Yuzyuk, T., Coughlin, C.R., Descartes, M., Grunewald, S., Maranda, B., Mills, P.B., Pitt, J., Potente, C., Rodenburg, R., Kluijtmans, L.A., Sampath, S., Pai, E.F., Wevers, R.A., Tiller, G.E., and additional

individual, c. (2017). Mild orotic aciduria in UMPS heterozygotes: a metabolic finding without clinical consequences. J Inherit Metab Dis 40, 423-431.

Ucar, S.K., **Mayr, J.A.**, **Feichtinger, R.G.**, Canda, E., Coker, M., and **Wortmann, S.B.** (2017). Previously Unreported Biallelic Mutation in DNAJC19: Are Sensorineural Hearing Loss and Basal Ganglia Lesions Additional Features of Dilated Cardiomyopathy and Ataxia (DCMA) Syndrome? JIMD Rep 35, 39-45.

Sommerville, E.W., Ng, Y.S., Alston, C.L., Dallabona, C., Gilberti, M., He, L., Knowles, C., Chin, S.L., Schaefer, A.M., Falkous, G., Murdoch, D., Longman, C., de Visser, M., Bindoff, L.A., Rawles, J.M., Dean, J.C.S., Petty, R.K., Farrugia, M.E., Haack, T.B., **Prokisch, H.**, McFarland, R., Turnbull, D.M., Donnini, C., Taylor, R.W., and Gorman, G.S. (2017). Clinical Features, Molecular Heterogeneity, and Prognostic Implications in YARS2-Related Mitochondrial Myopathy. JAMA Neurol 74, 686-694.

Sequeira, S., Rodrigues, M., Jacinto, S., Wevers, R.A., and **Wortmann, S.B.** (2017). MEGDEL Syndrome: Expanding the Phenotype and New Mutations. Neuropediatrics 48, 382-384.

Rokicki, D., Pajdowska, M., Trubicka, J., Thong, M.K., Ciara, E., Piekutowska-Abramczuk, D., Pronicki, M., Sikora, R., Haidar, R., Oltarzewski, M., Jablonska, E., Muthukumarasamy, P., Sthaneswar, P., Gan, C.S., Krajewska-Walasek, M., Carrozzo, R., Verrigni, D., Semeraro, M., Rizzo, C., Taurisano, R., **Alhaddad, B.**, **Kovacs-Nagy, R.**, Haack, T.B., Dionisi-Vici, C., Pronicka, E., and **Wortmann, S.B.** (2017). 3-Methylglutaconic aciduria, a frequent but underrecognized finding in carbamoyl phosphate synthetase I deficiency. Clin Chim Acta 471, 95-100.

Pronicka, E., Ropacka-Lesiak, M., Trubicka, J., Pajdowska, M., Linke, M., Ostergaard, E., Saunders, C., Horsch, S., van Karnebeek, C., Yaplito-Lee, J., Distelmaier, F., Ounap, K., Rahman, S., Castelle, M., Kelleher, J., Baris, S., Iwanicka-Pronicka, K., Steward, C.G., Ciara, E., **Wortmann, S.B.**, and Additional individual, c. (2017). A scoring system predicting the clinical course of CLPB defect based on the foetal and neonatal presentation of 31 patients. J Inherit Metab Dis 40, 853-860.

Maas, R.R., Iwanicka-Pronicka, K., Kalkan Ucar, S., **Alhaddad, B.**, AlSayed, M., Al-Owain, M.A., Al-Zaidan, H.I., Balasubramaniam, S., Baric, I., Bubshait, D.K., Burlina, A., Christodoulou, J., Chung, W.K., Colombo, R., Darin, N., **Freisinger, P.**, Garcia Silva, M.T., Grunewald, S., Haack, T.B., van Hasselt, P.M., Hikmat, O., Horster, F., Isohanni, P., Ramzan, K., **Kovacs-Nagy, R.**, Krumina, Z., Martin-Hernandez, E., **Mayr, J.A.**, McClean, P., De Meirleir, L., Naess, K., Ngu, L.H., Pajdowska, M., Rahman, S., Riordan, G., Riley, L., Roeben, B., Rutsch, F., Santer, R., Schiff, M., Seders, M., Sequeira, S., **Sperl, W.**, Staufner, C., Synofzik, M., Taylor, R.W., Trubicka, J., Tsiakas, K., Unal, O., Wassmer, E., Wedatilake, Y., Wolff, T., **Prokisch, H.**, Morava, E., Pronicka, E., Wevers, R.A., de Brouwer, A.P., and **Wortmann, S.B.** (2017). Progressive deafness-dystonia due to SERAC1 mutations: A study of 67 cases. Ann Neurol 82, 1004-1015.

Luna-Sanchez, M., Hidalgo-Gutierrez, A., Hildebrandt, T.M., Chaves-Serrano, J., Barriocanal-Casado, E., Santos-Fandila, A., Romero, M., Sayed, R.K., Duarte, J., **Prokisch, H.**, Schuelke, M., Distelmaier, F., Escames, G., Acuna-Castroviejo, D., and Lopez, L.C. (2017). CoQ deficiency causes disruption of mitochondrial sulfide oxidation, a new pathomechanism associated with this syndrome. EMBO Mol Med 9, 78-95.

Kremer, L.S., and **Prokisch, H.** (2017). Identification of Disease-Causing Mutations by Functional Complementation of Patient-Derived Fibroblast Cell Lines. Methods Mol Biol 1567, 391-406.

Kremer, L.S., Bader, D.M., Mertes, C., **Kopajtich, R.**, Pichler, G., **Iuso, A.**, Haack, T.B., Graf, E., Schwarzmayr, T., Terrile, C., **Konarikova, E.**, **Repp, B.**, Kastenmuller, G., Adamski, J., Lichtner, P., Leonhardt, C., Funalot, B., Donati, A., Tiranti, V., Lombes, A., Jardel, C., Glaser, D., Taylor, R.W., Ghezzi, D., **Mayr, J.A.**, Rotig, A., **Freisinger, P.**, Distelmaier, F., Strom, T.M., **Meitinger, T.**, Gagneur, J., and **Prokisch, H.** (2017). Genetic diagnosis of Mendelian disorders via RNA sequencing. Nat Commun 8, 15824.

Kopajtich, R., **Mayr, J.A.**, and **Prokisch, H.** (2017). Analysis of Mitochondrial RNA-Processing Defects in Patient-Derived Tissues by qRT-PCR and RNAseq. Methods Mol Biol 1567, 379-390.

Koch, J., **Mayr, J.A.**, **Alhaddad, B.**, Rauscher, C., Bierau, J., **Kovacs-Nagy, R.**, Coene, K.L., Bader, I., Holzhacker, M., **Prokisch, H.**, Venselaar, H., Wevers, R.A., Distelmaier, F., Polster, T., Leiz, S., Betzler, C., Strom, T.M., **Sperl, W.**, **Meitinger, T.**, **Wortmann, S.B.**, and Haack, T.B. (2017). CAD mutations and uridine-responsive epileptic encephalopathy. Brain 140, 279-286.

Iuso, A., **Repp, B.**, Biagosch, C., Terrile, C., and **Prokisch, H.** (2017). Assessing Mitochondrial Bioenergetics in Isolated Mitochondria from Various Mouse Tissues Using Seahorse XF96 Analyzer. Methods Mol Biol 1567, 217-230.

Herebian, D., Seibt, A., Smits, S.H.J., Bunning, G., Freyer, C., **Prokisch, H.**, Karall, D., Wredenberg, A., Wedell, A., Lopez, L.C., Mayatepek, E., and Distelmaier, F. (2017). Detection of 6-demethoxyubiquinone in CoQ10 deficiency disorders: Insights into enzyme interactions and identification of potential therapeutics. Mol Genet Metab 121, 216-223.

Hempel, M., **Kremer, L.S.**, Tsiakas, K., **Alhaddad, B.**, Haack, T.B., Lobel, U., **Feichtinger, R.G.**, **Sperl, W.**, **Prokisch, H.**, **Mayr, J.A.**, and Santer, R. (2017). LYRM7 - associated complex III deficiency: A clinical, molecular genetic, MR tomographic, and biochemical study. Mitochondrion 37, 55-61.

Habarou, F., Hamel, Y., Haack, T.B., **Feichtinger, R.G.**, Lebigot, E., Marquardt, I., Busiah, K., Laroche, C., Madrange, M., Grisel, C., Pontoizeau, C., Eisermann, M., Boutron, A., Chretien, D., Chadefaux-Vekemans, B., Barouki, R., Bole-Feysot, C., Nitschke, P., Goudin, N., Boddaert, N., Nemazanyy, I., Delahodde, A., Kolker, S., Rodenburg, R.J., Korenke, G.C., **Meitinger, T.**, Strom, T.M., **Prokisch, H.**, Rotig, A., Ottolenghi, C., **Mayr, J.A.**, and de Lonlay, P. (2017). Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. Am J Hum Genet 101, 283-290.

Glasgow, R.I.C., Thompson, K., Barbosa, I.A., He, L., Alston, C.L., Deshpande, C., Simpson, M.A., Morris, A.A.M., Neu, A., Lobel, U., Hall, J., **Prokisch, H.**, Haack, T.B., Hempel, M., McFarland, R., and Taylor, R.W. (2017). Novel GFM2 variants associated with early-onset neurological presentations of mitochondrial disease and impaired expression of OXPHOS subunits. Neurogenetics 18, 227-235.

Feichtinger, R.G., Petervari, E., Zopf, M., Vidali, S., Aminzadeh-Gohari, S., **Mayr, J.A.**, **Kofler, B.**, and Balasko, M. (2017). Effects of alpha-melanocyte-stimulating hormone on mitochondrial energy metabolism in rats of different age-groups. Neuropeptides 64, 123-130.

Feichtinger, R.G., Olahova, M., Kishita, Y., Garone, C., **Kremer, L.S.**, Yagi, M., Uchiumi, T., Jourdain, A.A., Thompson, K., D'Souza, A.R., **Kopajtich, R.**, Alston, C.L., **Koch, J.**, **Sperl, W.**, **Mastantuono, E.**, Strom, T.M., **Wortmann, S.B.**, **Meitinger, T.**, Pierre, G., Chinnery, P.F., Chrzanowska-Lightowlers, Z.M., Lightowlers, R.N., DiMauro, S., Calvo, S.E., Mootha, V.K., Moggio, M., Sciacco, M., Comi, G.P., Ronchi, D., Murayama, K., Ohtake, A., Rebelo-Guiomar, P., Kohda, M., Kang, D., **Mayr, J.A.**, Taylor, R.W., Okazaki, Y., Minczuk, M., and **Prokisch, H.** (2017). Biallelic C1QBP Mutations Cause Severe Neonatal-, Childhood-, or Later-Onset Cardiomyopathy Associated with Combined Respiratory-Chain Deficiencies. Am J Hum Genet 101, 525-538.

Feichtinger, R.G., Neureiter, D., Skaria, T., Wessler, S., Cover, T.L., **Mayr, J.A.**, **Zimmermann, F.A.**, Posselt, G., **Sperl, W.**, and **Kofler, B.** (2017). Oxidative Phosphorylation System in Gastric Carcinomas and Gastritis. Oxid Med Cell Longev 2017, 1320241.

Feichtinger, R.G., Brunner-Krainz, M., **Alhaddad, B.**, **Wortmann, S.B.**, **Kovacs-Nagy, R.**, Stojakovic, T., Erwa, W., Resch, B., Windischhofer, W., Verheyen, S., Uhrig, S., Windpassinger, C., Locker, F., **Makowski, C.**, Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, **Sperl, W.**, Haack, T.B., and **Mayr, J.A.** (2017). Combined Respiratory Chain Deficiency and UQCC2 Mutations in Neonatal Encephalomyopathy: Defective Supercomplex Assembly in Complex III Deficiencies. Oxid Med Cell Longev 2017, 7202589.

El-Hattab, A.W., Dai, H., Almannai, M., Wang, J., Faqeih, E.A., Al Asmari, A., Saleh, M.A.M., Elamin, M.A.O., Alfadhel, M., Alkuraya, F.S., Hashem, M., Aldosary, M.S., Almass, R., Almutairi, F.B., Alsagob, M., Al-Owain, M., Al-Sharfa, S., Al-Hassnan, Z.N., Rahbeeni, Z., Al-Muhaizea, M.A., Makhseed, N., Foskett, G.K., Stevenson, D.A., Gomez-Ospina, N., Lee, C., Boles, R.G., Schrier Vergano, S.A., **Wortmann, S.B.**, **Sperl, W.**, Opladen, T., Hoffmann, G.F., Hempel, M., **Prokisch, H.**, **Alhaddad, B.**, **Mayr, J.A.**, Chan, W., Kaya, N., and Wong, L.C. (2017). Molecular and clinical spectra of FBXL4 deficiency. Hum Mutat 38, 1649-1659.

Distelmaier, F., Haack, T.B., **Wortmann, S.B.**, **Mayr, J.A.**, and **Prokisch, H.** (2017). Treatable mitochondrial diseases: cofactor metabolism and beyond. Brain 140, e11.

Catarino, C.B., **Ahting, U.**, **Gusic, M.**, **Iuso, A.**, **Repp, B.**, Peters, K., Biskup, S., von Livonius, B., **Prokisch, H.**, and Klopstock, T. (2017). Characterization of a Leber's hereditary optic neuropathy (LHON) family harboring two primary LHON mutations m.11778G>A and m.14484T>C of the mitochondrial DNA. Mitochondrion 36, 15-20.

Carecchio, M., Picillo, M., Valletta, L., Elia, A.E., Haack, T.B., Cozzolino, A., Vitale, A., Garavaglia, B., **Iuso, A.**, Bagella, C.F., Pappata, S., Barone, P., **Prokisch, H.**, Romito, L., and Tiranti, V. (2017). Rare causes of early-onset dystonia-parkinsonism with cognitive impairment: a de novo PSEN-1 mutation. Neurogenetics 18, 175-178.

Biagosch, C., Ediga, R.D., Hensler, S.V., Faerberboeck, M., Kuehn, R., Wurst, W., **Meitinger, T.**, Kolker, S., Sauer, S., and **Prokisch, H.** (2017). Elevated glutaric acid levels in Dhtkd1-/Gcdh-

double knockout mice challenge our current understanding of lysine metabolism. Biochim Biophys Acta Mol Basis Dis 1863, 2220-2228.

Bellingrath, J.S., Ochakovski, G.A., Seitz, I.P., Kohl, S., Zrenner, E., Hanig, N., **Prokisch, H.**, Weber, B.H., Downes, S.M., Ramsden, S., MacLaren, R.E., and Fischer, M.D. (2017). High Symmetry of Visual Acuity and Visual Fields in RPGR-Linked Retinitis Pigmentosa. Invest Ophthalmol Vis Sci 58, 4457-4466.

Baertling, F., **Alhaddad, B.**, Seibt, A., Budaeus, S., **Meitinger, T.**, Strom, T.M., Mayatepek, E., Schaper, J., **Prokisch, H.**, Haack, T.B., and Distelmaier, F. (2017). Neonatal encephalocardiomyopathy caused by mutations in VARS2. Metab Brain Dis 32, 267-270.

Anikster, Y., Haack, T.B., Vilboux, T., Pode-Shakked, B., Thony, B., Shen, N., Guarani, V., Meissner, T., Mayatepek, E., Trefz, F.K., Marek-Yagel, D., Martinez, A., Huttlin, E.L., Paulo, J.A., Berutti, R., Benoist, J.F., Imbard, A., Dorboz, I., Heimer, G., Landau, Y., Ziv-Strasser, L., Malicdan, M.C.V., Gemperle-Britschgi, C., Cremer, K., Engels, H., Meili, D., Keller, I., Bruggmann, R., Strom, T.M., **Meitinger, T.**, Mullikin, J.C., Schwartz, G., Ben-Zeev, B., Gahl, W.A., Harper, J.W., Blau, N., Hoffmann, G.F., **Prokisch, H.**, Opladen, T., and Schiff, M. (2017). Biallelic Mutations in DNAJC12 Cause Hyperphenylalaninemia, Dystonia, and Intellectual Disability. Am J Hum Genet 100, 257-266.

Aminzadeh-Gohari, S., **Feichtinger, R.G.**, Vidali, S., Locker, F., Rutherford, T., O'Donnel, M., Stoger-Kleiber, A., **Mayr, J.A.**, **Sperl, W.**, and **Kofler, B.** (2017). A ketogenic diet supplemented with medium-chain triglycerides enhances the anti-tumor and anti-angiogenic efficacy of chemotherapy on neuroblastoma xenografts in a CD1-nu mouse model. Oncotarget 8, 64728-64744.

Ait-El-Mkadem, S., Dayem-Quere, M., **Gusic, M.**, Chaussenot, A., Bannwarth, S., Francois, B., Genin, E.C., Fragaki, K., Volker-Touw, C.L.M., Vasnier, C., Serre, V., van Gassen, K.L.I., Lespinasse, F., Richter, S., Eisenhofer, G., Rouzier, C., Mochel, F., De Saint-Martin, A., Abi Warde, M.T., de Sain-van der Velde, M.G.M., Jans, J.J.M., Amiel, J., Avsec, Z., Mertes, C., Haack, T.B., Strom, T., **Meitinger, T.**, Bonnen, P.E., Taylor, R.W., Gagneur, J., van Hasselt, P.M., Rotig, A., Delahodde, A., **Prokisch, H.**, Fuchs, S.A., and Paquis-Flucklinger, V. (2017). Mutations in MDH2, Encoding a Krebs Cycle Enzyme, Cause Early-Onset Severe Encephalopathy. Am J Hum Genet 100, 151-159.

2016

Zimmermann, F.A., Neureiter, D., **Feichtinger, R.G.**, Trost, A., **Sperl, W.**, **Kofler, B.**, and **Mayr, J.A.** (2016). Deficiency of respiratory chain complex I in Hashimoto thyroiditis. Mitochondrion 26, 1-6.

Van Haute, L., Dietmann, S., Kremer, L., Hussain, S., Pearce, S.F., Powell, C.A., Rorbach, J., Lantaff, R., Blanco, S., Sauer, S., Kotzaeridou, U., Hoffmann, G.F., Memari, Y., Kolb-Kokocinski, A., Durbin, R., **Mayr, J.A.**, Frye, M., **Prokisch, H.**, and Minczuk, M. (2016). Deficient methylation and formylation of mt-tRNA(Met) wobble cytosine in a patient carrying mutations in NSUN3. Nat Commun 7, 12039.

Tuschl, K., Meyer, E., Valdivia, L.E., Zhao, N., Dadswell, C., Abdul-Sada, A., Hung, C.Y., Simpson, M.A., Chong, W.K., Jacques, T.S., Woltjer, R.L., Eaton, S., Gregory, A., Sanford, L., Kara, E., Houlden, H., Cuno, S.M., **Prokisch, H.**, Valletta, L., Tiranti, V., Younis, R., Maher, E.R., Spencer, J., Straatman-Iwanowska, A., Gissen, P., Selim, L.A., Pintos-Morell, G., Coroleu-Lletget, W., Mohammad, S.S., Yoganathan, S., Dale, R.C., Thomas, M., Rihel, J., Bodamer, O.A., Enns, C.A., Hayflick, S.J., Clayton, P.T., Mills, P.B., Kurian, M.A., and Wilson, S.W. (2016). Mutations in SLC39A14 disrupt manganese homeostasis and cause childhood-onset parkinsonism-dystonia. Nat Commun 7, 11601.

Thiels, C., Fleger, M., Huemer, M., Rodenburg, R.J., Vaz, F.M., Houtkooper, R.H., Haack, T.B., **Prokisch, H.**, **Feichtinger, R.G.**, Lucke, T., **Mayr, J.A.**, and **Wortmann, S.B.** (2016). Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation? JIMD Rep 29, 89-93.

Teumer, A., Schurmann, C., Schillert, A., Schramm, K., Ziegler, A., and **Prokisch, H.** (2016). Analyzing Illumina Gene Expression Microarray Data Obtained From Human Whole Blood Cell and Blood Monocyte Samples. Methods Mol Biol 1368, 85-97.

Staufner, C., Haack, T.B., Kopke, M.G., Straub, B.K., Kolker, S., Thiel, C., **Freisinger, P.**, Baric, I., McKiernan, P.J., Dikow, N., Harting, I., Beisse, F., Burgard, P., Kotzaeridou, U., Lenz, D., Kuhr, J., Himbert, U., Taylor, R.W., Distelmaier, F., Vockley, J., Ghaloul-Gonzalez, L., Ozolek, J.A., Zschocke, J., Kuster, A., Dick, A., Das, A.M., Wieland, T., Terrile, C., Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, and Hoffmann, G.F. (2016). Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. J Inherit Metab Dis 39, 3-16.

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

Ortigoza-Escobar, J.D., Molero-Luis, M., Arias, A., Oyarzabal, A., Darin, N., Serrano, M., Garcia-Cazorla, A., Tondo, M., Hernandez, M., Garcia-Villoria, J., Casado, M., Gort, L., **Mayr, J.A.**, Rodriguez-Pombo, P., Ribes, A., Artuch, R., and Perez-Duenas, B. (2016). Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain 139, 31-38.

Olsen, R.K.J., **Konarikova, E.**, Giancaspero, T.A., Mosegaard, S., Boczonadi, V., **Matakovic, L.**, Veauville-Merllie, A., Terrile, C., Schwarzmayr, T., Haack, T.B., Auranen, M., Leone, P., Galluccio, M., Imbard, A., Gutierrez-Rios, P., Palmfeldt, J., Graf, E., Vianey-Saban, C., Oppenheim, M., Schiff, M., Pichard, S., Rigal, O., Pyle, A., Chinnery, P.F., Konstantopoulou, V., Moslinger, D., **Feichtinger, R.G.**, Talim, B., Topaloglu, H., Coskun, T., Gucer, S., Botta, A., Pegoraro, E., Malena, A., Vergani, L., Mazza, D., Zollino, M., Ghezzi, D., Acquaviva, C., Tyni, T., Boneh, A., **Meitinger, T.**, Strom, T.M., Gregersen, N., **Mayr, J.A.**, Horvath, R., Barile, M., and **Prokisch, H.** (2016). Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency. Am J Hum Genet 98, 1130-1145.

Marzi, C., Holdt, L.M., Fiorito, G., Tsai, P.C., Kretschmer, A., Wahl, S., Guarrera, S., Teupser, D., Spector, T.D., Iacoviello, L., Sacerdote, C., Strauch, K., Lee, S., Thasler, W.E., Peters, A., Thorand, B., Wolf, P., **Prokisch, H.**, Tumino, R., Gieger, C., Krogh, V., Panico, S., Bell, J.T., Matullo, G., Waldenberger, M., Grallert, H., and Koenig, W. (2016). Epigenetic Signatures at AQP3 and SOCS3 Engage in Low-Grade Inflammation across Different Tissues. PLoS One 11, e0166015.

Maas, R.R., Marina, A.D., de Brouwer, A.P., Wevers, R.A., Rodenburg, R.J., and **Wortmann, S.B.** (2016). SUCLA2 Deficiency: A Deafness-Dystonia Syndrome with Distinctive Metabolic Findings (Report of a New Patient and Review of the Literature). JIMD Rep 27, 27-32.

Kriebel, J., Herder, C., Rathmann, W., Wahl, S., Kunze, S., Molnos, S., Volkova, N., Schramm, K., Carstensen-Kirberg, M., Waldenberger, M., Gieger, C., Peters, A., Illig, T., **Prokisch, H.**, Roden, M., and Grallert, H. (2016). Association between DNA Methylation in Whole Blood and Measures of Glucose Metabolism: KORA F4 Study. PLoS One 11, e0152314.

Kremer, L.S., L'Hermitte-Stead, C., Lesimple, P., Gilleron, M., Filaut, S., Jardel, C., Haack, T.B., Strom, T.M., **Meitinger, T.**, Azzouz, H., Tebib, N., Ogier de Baulny, H., Touati, G., **Prokisch, H.**, and Lombes, A. (2016). Severe respiratory complex III defect prevents liver adaptation to prolonged fasting. J Hepatol 65, 377-385.

Kremer, L.S., Distelmaier, F., **Alhaddad, B.**, Hempel, M., **Iuso, A.**, Kupper, C., Muhlhausen, C., **Kovacs-Nagy, R.**, Satanovskij, R., Graf, E., Berutti, R., Eckstein, G., Durbin, R., Sauer, S., Hoffmann, G.F., Strom, T.M., Santer, R., **Meitinger, T.**, Klopstock, T., **Prokisch, H.**, and Haack, T.B. (2016). Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. Am J Hum Genet 98, 358-362.

Kremer, L.S., Danhauser, K., Herebian, D., Petkovic Ramadza, D., Piekutowska-Abramczuk, D., Seibt, A., Muller-Felber, W., Haack, T.B., Ploski, R., Lohmeier, K., Schneider, D., Klee, D., Rokicki, D., Mayatepek, E., Strom, T.M., **Meitinger, T.**, Klopstock, T., Pronicka, E., **Mayr, J.A.**, Baric, I., Distelmaier, F., and **Prokisch, H.** (2016). NAXE Mutations Disrupt the Cellular NAD(P)HX Repair System and Cause a Lethal Neurometabolic Disorder of Early Childhood. Am J Hum Genet 99, 894-902.

Kopajtich, R., Murayama, K., Janecke, A.R., Haack, T.B., Breuer, M., Knisely, A.S., Harting, I., Ohashi, T., Okazaki, Y., Watanabe, D., Tokuzawa, Y., Kotzaeridou, U., Kolker, S., Sauer, S., Carl, M., Straub, S., Entenmann, A., Gizewski, E., **Feichtinger, R.G.**, **Mayr, J.A.**, Lackner, K., Strom, T.M., **Meitinger, T.**, Muller, T., Ohtake, A., Hoffmann, G.F., **Prokisch, H.**, and Staufner, C. (2016). Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. Am J Hum Genet 99, 414-422.

Koch, J., **Feichtinger, R.G.**, **Freisinger, P.**, Pies, M., Schrodl, F., **Iuso, A.**, **Sperl, W.**, **Mayr, J.A.**, **Prokisch, H.**, and Haack, T.B. (2016). Disturbed mitochondrial and peroxisomal dynamics due to loss of MFF causes Leigh-like encephalopathy, optic atrophy and peripheral neuropathy. J Med Genet 53, 270-278.

Klopstock, T., Klopstock, B., and **Prokisch, H.** (2016). Mitochondrial replacement approaches: challenges for clinical implementation. Genome Med 8, 126.

Kennedy, H., Haack, T.B., Hartill, V., **Matakovic, L.**, Baumgartner, E.R., Potter, H., Mackay, R., Alston, C.L., O'Sullivan, S., McFarland, R., Connolly, G., Gannon, C., King, R., Mead, S., Crozier, I., Chan, W., Florkowski, C.M., Sage, M., Hofken, T., **Alhaddad, B.**, **Kremer, L.S.**, **Kopajtich, R.**, **Feichtinger, R.G.**, **Sperl, W.**, Rodenburg, R.J., Minet, J.C., Dobbie, A., Strom, T.M., **Meitinger, T.**, George, P.M., Johnson, C.A., Taylor, R.W., **Prokisch, H.**, Doudney, K., and **Mayr, J.A.** (2016). Sudden Cardiac Death Due to Deficiency of the Mitochondrial Inorganic Pyrophosphatase PPA2. Am J Hum Genet 99, 674-682.

Holzerova, E., Danhauser, K., Haack, T.B., **Kremer, L.S.**, Melcher, M., Ingold, I., Kobayashi, S., Terrile, C., Wolf, P., Schaper, J., Mayatepek, E., Baertling, F., Friedmann Angeli, J.P., Conrad, M., Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, and Distelmaier, F. (2016). Human thioredoxin 2 deficiency impairs mitochondrial redox homeostasis and causes early-onset neurodegeneration. Brain 139, 346-354.

Haack, T.B., Ignatius, E., Calvo-Garrido, J., **Iuso, A.**, Isohanni, P., Maffezzini, C., Lonnqvist, T., Suomalainen, A., Gorza, M., **Kremer, L.S.**, Graf, E., Hartig, M., Berutti, R., Paucar, M., Svenningsson, P., Stranneheim, H., Brandberg, G., Wedell, A., Kurian, M.A., Hayflick, S.A., Venco, P., Tiranti, V., Strom, T.M., Dichgans, M., Horvath, R., Holinski-Feder, E., Freyer, C., **Meitinger, T.**, **Prokisch, H.**, Senderek, J., Wredenberg, A., Carroll, C.J., and Klopstock, T. (2016). Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. Am J Hum Genet 99, 735-743.

Franko, A., Huypens, P., Neschen, S., Irmler, M., Rozman, J., Rathkolb, B., Neff, F., Prehn, C., Dubois, G., Baumann, M., Massinger, R., Gradinger, D., Przemeck, G.K., **Repp, B.**, Aichler, M., Feuchtinger, A., Schommers, P., Stohr, O., Sanchez-Lasheras, C., Adamski, J., Peter, A., **Prokisch, H.**, Beckers, J., Walch, A.K., Fuchs, H., Wolf, E., Schubert, M., Wiesner, R.J., and Hrabe de Angelis, M. (2016). Bezafibrate Improves Insulin Sensitivity and Metabolic Flexibility in STZ-Induced Diabetic Mice. Diabetes 65, 2540-2552.

Floyd, B.J., Wilkerson, E.M., Veling, M.T., Minogue, C.E., Xia, C., Beebe, E.T., Wrobel, R.L., Cho, H., **Kremer, L.S.**, Alston, C.L., Gromek, K.A., Dolan, B.K., Ulbrich, A., Stefely, J.A., Bohl, S.L., Werner, K.M., Jochem, A., Westphall, M.S., Rensvold, J.W., Taylor, R.W., **Prokisch, H.**, Kim, J.P., Coon, J.J., and Pagliarini, D.J. (2016). Mitochondrial Protein Interaction Mapping Identifies Regulators of Respiratory Chain Function. Mol Cell 63, 621-632.

Feichtinger, R.G., Weis, S., **Mayr, J.A.**, **Zimmermann, F.A.**, Bogner, B., **Sperl, W.**, and **Kofler, B.** (2016). Alterations of oxidative phosphorylation in meningiomas and peripheral nerve sheath tumors. Neuro Oncol 18, 184-194.

Danhauser, K., Herebian, D., Haack, T.B., Rodenburg, R.J., Strom, T.M., **Meitinger, T.**, Klee, D., Mayatepek, E., **Prokisch, H.**, and Distelmaier, F. (2016). Fatal neonatal encephalopathy and lactic acidosis caused by a homozygous loss-of-function variant in COQ9. Eur J Hum Genet 24, 450-454.

Danhauser, K., Haack, T.B., **Alhaddad, B.**, Melcher, M., Seibt, A., Strom, T.M., **Meitinger, T.**, Klee, D., Mayatepek, E., **Prokisch, H.**, and Distelmaier, F. (2016). EARS2 mutations cause fatal

neonatal lactic acidosis, recurrent hypoglycemia and agenesis of corpus callosum. Metab Brain Dis 31, 717-721.

Collet, M., Assouline, Z., Bonnet, D., Rio, M., Iserin, F., Sidi, D., Goldenberg, A., Lardennois, C., Metodiev, M.D., Haberberger, B., Haack, T., Munnich, A., **Prokisch, H.**, and Rotig, A. (2016). High incidence and variable clinical outcome of cardiac hypertrophy due to ACAD9 mutations in childhood. Eur J Hum Genet 24, 1112-1116.

Colin, E., Daniel, J., Ziegler, A., Wakim, J., Scrivo, A., Haack, T.B., Khiati, S., Denomme, A.S., Amati-Bonneau, P., Charif, M., Procaccio, V., Reynier, P., Aleck, K.A., Botto, L.D., Herper, C.L., Kaiser, C.S., Nabbout, R., N'Guyen, S., Mora-Lorca, J.A., Assmann, B., Christ, S., **Meitinger, T.**, Strom, T.M., **Prokisch, H.**, Consortium, F., Miranda-Vizuete, A., Hoffmann, G.F., Lenaers, G., Bomont, P., Liebau, E., and Bonneau, D. (2016). Biallelic Variants in UBA5 Reveal that Disruption of the UFM1 Cascade Can Result in Early-Onset Encephalopathy. Am J Hum Genet 99, 695-703.

Ciara, E., Rokicki, D., Halat, P., Karkucinska-Wieckowska, A., Piekutowska-Abramczuk, D., **Mayr, J.**, Trubicka, J., Szymanska-Debinska, T., Pronicki, M., Pajdowska, M., Dudzinska, M., Gizewska, M., Krajewska-Walasek, M., Ksiazyk, J., **Sperl, W.**, Ploski, R., and Pronicka, E. (2016). Difficulties in recognition of pyruvate dehydrogenase complex deficiency on the basis of clinical and biochemical features. The role of next-generation sequencing. Mol Genet Metab Rep 7, 70-76.

Blommaert, D., van Hulst, K., Hoogen, F.J., Erasmus, C.E., and **Wortmann, S.B.** (2016). Diagnosis and Management of Drooling in Children With Progressive Dystonia: A Case Series of Patients With MEGDEL Syndrome. J Child Neurol 31, 1220-1226.

Baertling, F., Klee, D., Haack, T.B., **Prokisch, H.**, **Meitinger, T.**, Mayatepek, E., Schaper, J., and Distelmaier, F. (2016). The many faces of paediatric mitochondrial disease on neuroimaging. Childs Nerv Syst 32, 2077-2083.

Altmann, J., Buchner, B., Nadaj-Pakleza, A., Schafer, J., Jackson, S., Lehmann, D., Deschauer, M., **Kopajtich, R.**, Lautenschlager, R., Kuhn, K.A., Karle, K., Schols, L., Schulz, J.B., Weis, J., **Prokisch, H.**, Kornblum, C., Claeys, K.G., and Klopstock, T. (2016). Expanded phenotypic spectrum of the m.8344A>G "MERRF" mutation: data from the German mitoNET registry. J Neurol 263, 961-972.

Alston, C.L., Howard, C., Olahova, M., Hardy, S.A., He, L., Murray, P.G., O'Sullivan, S., Doherty, G., Shield, J.P., Hargreaves, I.P., Monavari, A.A., Knerr, I., McCarthy, P., Morris, A.A., Thorburn, D.R., **Prokisch, H.**, Clayton, P.E., McFarland, R., Hughes, J., Crushell, E., and Taylor, R.W. (2016). A recurrent mitochondrial p.Trp22Arg NDUFB3 variant causes a distinctive facial appearance, short stature and a mild biochemical and clinical phenotype. J Med Genet 53, 634-641.

Alston, C.L., Compton, A.G., Formosa, L.E., Strecker, V., Olahova, M., Haack, T.B., Smet, J., Stouffs, K., Diakumis, P., Ciara, E., Cassiman, D., Romain, N., Yarham, J.W., He, L., De Paepe, B., Vanlander, A.V., Seneca, S., **Feichtinger, R.G.**, Ploski, R., Rokicki, D., Pronicka, E., Haller, R.G., Van Hove, J.L., Bahlo, M., **Mayr, J.A.**, Van Coster, R., **Prokisch, H.**, Wittig, I., Ryan, M.T.,

Thorburn, D.R., and Taylor, R.W. (2016). Biallelic Mutations in TMEM126B Cause Severe Complex I Deficiency with a Variable Clinical Phenotype. Am J Hum Genet 99, 217-227.

2015

Wortmann, S.B., Zietkiewicz, S., Kousi, M., Szklarczyk, R., Haack, T.B., Gersting, S.W., Muntau, A.C., Rakovic, A., Renkema, G.H., Rodenburg, R.J., Strom, T.M., **Meitinger, T.**, Rubio-Gozalbo, M.E., Chrusciel, E., Distelmaier, F., Golzio, C., Jansen, J.H., van Karnebeek, C., Lillquist, Y., Lucke, T., Ounap, K., Zordania, R., Yaplito-Lee, J., van Bokhoven, H., Spelbrink, J.N., Vaz, F.M., Pras-Raves, M., Ploski, R., Pronicka, E., Klein, C., Willemsen, M.A., de Brouwer, A.P., **Prokisch, H.**, Katsanis, N., and Wevers, R.A. (2015). CLPB mutations cause 3-methylglutaconic aciduria, progressive brain atrophy, intellectual disability, congenital neutropenia, cataracts, movement disorder. Am J Hum Genet 96, 245-257.

Wortmann, S.B., van Hasselt, P.M., Baric, I., Burlina, A., Darin, N., Horster, F., Coker, M., Ucar, S.K., Krumina, Z., Naess, K., Ngu, L.H., Pronicka, E., Riordan, G., Santer, R., Wassmer, E., Zschocke, J., Schiff, M., de Meirleir, L., Alowain, M.A., Smeitink, J.A., Morava, E., Kozicz, T., Wevers, R.A., Wolf, N.I., and Willemsen, M.A. (2015). Eyes on MEGDEL: distinctive basal ganglia involvement in dystonia deafness syndrome. Neuropediatrics 46, 98-103.

Wortmann, S.B., Koolen, D.A., Smeitink, J.A., van den Heuvel, L., and Rodenburg, R.J. (2015). Whole exome sequencing of suspected mitochondrial patients in clinical practice. J Inherit Metab Dis 38, 437-443.

Wortmann, S.B., Espeel, M., Almeida, L., Reimer, A., Bosboom, D., Roels, F., de Brouwer, A.P., and Wevers, R.A. (2015). Inborn errors of metabolism in the biosynthesis and remodelling of phospholipids. J Inherit Metab Dis 38, 99-110.

Westra, H.J., Arends, D., Esko, T., Peters, M.J., Schurmann, C., Schramm, K., Kettunen, J., Yaghootkar, H., Fairfax, B.P., Andiappan, A.K., Li, Y., Fu, J., Karjalainen, J., Platteel, M., Visschedijk, M., Weersma, R.K., Kasela, S., Milani, L., Tserel, L., Peterson, P., Reinmaa, E., Hofman, A., Uitterlinden, A.G., Rivadeneira, F., Homuth, G., Petersmann, A., Lorbeer, R., **Prokisch, H.**, **Meitinger, T.**, Herder, C., Roden, M., Grallert, H., Ripatti, S., Perola, M., Wood, A.R., Melzer, D., Ferrucci, L., Singleton, A.B., Hernandez, D.G., Knight, J.C., Melchiotti, R., Lee, B., Poidinger, M., Zolezzi, F., Larbi, A., Wang de, Y., van den Berg, L.H., Veldink, J.H., Rotzschke, O., Makino, S., Salomaa, V., Strauch, K., Volker, U., van Meurs, J.B., Metspalu, A., Wijmenga, C., Jansen, R.C., and Franke, L. (2015). Cell Specific eQTL Analysis without Sorting Cells. PLoS Genet 11, e1005223.

Wahl, S., Vogt, S., Stuckler, F., Krumsiek, J., Bartel, J., Kacprowski, T., Schramm, K., Carstensen, M., Rathmann, W., Roden, M., Jourdan, C., Kangas, A.J., Soininen, P., Ala-Korpela, M., Nothlings, U., Boeing, H., Theis, F.J., Meisinger, C., Waldenberger, M., Suhre, K., Homuth, G., Gieger, C., Kastenmuller, G., Illig, T., Linseisen, J., Peters, A., **Prokisch, H.**, Herder, C., Thorand, B., and Grallert, H. (2015). Multi-omic signature of body weight change: results from a population-based cohort study. BMC Med 13, 48.

Vodopiutz, J., Seidl, R., Prayer, D., Khan, M.I., **Mayr, J.A.**, Streubel, B., Steiss, J.O., Hahn, A., Csaicsich, D., Castro, C., Assoum, M., Muller, T., Wieczorek, D., Mancini, G.M., Sadowski, C.E., Levy, N., Megarbane, A., Godbole, K., Schanze, D., Hildebrandt, F., Delague, V., Janecke, A.R., and Zenker, M. (2015). WDR73 Mutations Cause Infantile Neurodegeneration and Variable Glomerular Kidney Disease. Hum Mutat 36, 1021-1028.

Vidali, S., Aminzadeh, S., Lambert, B., Rutherford, T., **Sperl, W.**, **Kofler, B.**, and **Feichtinger, R.G.** (2015). Mitochondria: The ketogenic diet--A metabolism-based therapy. Int J Biochem Cell Biol 63, 55-59.

Venco, P., Bonora, M., Giorgi, C., Papaleo, E., **Iuso, A.**, **Prokisch, H.**, Pinton, P., and Tiranti, V. (2015). Mutations of C19orf12, coding for a transmembrane glycine zipper containing mitochondrial protein, cause mis-localization of the protein, inability to respond to oxidative stress and increased mitochondrial Ca(2)(+). Front Genet 6, 185.

Tischner, C., Hofer, A., Wulff, V., Stepek, J., Dumitru, I., Becker, L., Haack, T., Kremer, L., Datta, A.N., **Sperl, W.**, Floss, T., Wurst, W., Chrzanowska-Lightowlers, Z., De Angelis, M.H., Klopstock, T., **Prokisch, H.**, and Wenz, T. (2015). MTO1 mediates tissue specificity of OXPHOS defects via tRNA modification and translation optimization, which can be bypassed by dietary intervention. Hum Mol Genet 24, 2247-2266.

Sperl, W., Fleuren, L., **Freisinger, P.**, Haack, T.B., Ribes, A., **Feichtinger, R.G.**, Rodenburg, R.J., **Zimmermann, F.A.**, **Koch, J.**, Rivera, I., **Prokisch, H.**, Smeitink, J.A., and **Mayr, J.A.** (2015). The spectrum of pyruvate oxidation defects in the diagnosis of mitochondrial disorders. J Inherit Metab Dis 38, 391-403.

Seifert, E.L., Ligeti, E., **Mayr, J.A.**, Sondheimer, N., and Hajnoczky, G. (2015). The mitochondrial phosphate carrier: Role in oxidative metabolism, calcium handling and mitochondrial disease. Biochem Biophys Res Commun 464, 369-375.

Schiff, M., Haberberger, B., Xia, C., Mohsen, A.W., Goetzman, E.S., Wang, Y., Uppala, R., Zhang, Y., Karunanidhi, A., Prabhu, D., Alharbi, H., Prochownik, E.V., Haack, T., Haberle, J., Munnich, A., Rotig, A., Taylor, R.W., Nicholls, R.D., Kim, J.J., **Prokisch, H.**, and Vockley, J. (2015). Complex I assembly function and fatty acid oxidation enzyme activity of ACAD9 both contribute to disease severity in ACAD9 deficiency. Hum Mol Genet 24, 3238-3247.

Renkema, G.H., **Wortmann, S.B.**, Smeets, R.J., Venselaar, H., Antoine, M., Visser, G., Ben-Omran, T., van den Heuvel, L.P., Timmers, H.J., Smeitink, J.A., and Rodenburg, R.J. (2015). SDHA mutations causing a multisystem mitochondrial disease: novel mutations and genetic overlap with hereditary tumors. Eur J Hum Genet 23, 202-209.

Powell, C.A., **Kopajtich, R.**, D'Souza, A.R., Rorbach, J., **Kremer, L.S.**, Husain, R.A., Dallabona, C., Donnini, C., Alston, C.L., Griffin, H., Pyle, A., Chinnery, P.F., Strom, T.M., **Meitinger, T.**, Rodenburg, R.J., Schottmann, G., Schuelke, M., Romain, N., Haller, R.G., Ferrero, I., Haack, T.B., Taylor, R.W., **Prokisch, H.**, and Minczuk, M. (2015). TRMT5 Mutations Cause a Defect in Post-transcriptional Modification of Mitochondrial tRNA Associated with Multiple Respiratory-Chain Deficiencies. Am J Hum Genet 97, 319-328.

Peters, M.J., Joehanes, R., Pilling, L.C., Schurmann, C., Conneely, K.N., Powell, J., Reinmaa, E., Sutphin, G.L., Zhernakova, A., Schramm, K., Wilson, Y.A., Kobes, S., Tukiainen, T., Consortium, N.U., Ramos, Y.F., Goring, H.H., Fornage, M., Liu, Y., Gharib, S.A., Stranger, B.E., De Jager, P.L., Aviv, A., Levy, D., Murabito, J.M., Munson, P.J., Huan, T., Hofman, A., Uitterlinden, A.G., Rivadeneira, F., van Rooij, J., Stolk, L., Broer, L., Verbiest, M.M., Jhamai, M., Arp, P., Metspalu, A., Tserel, L., Milani, L., Samani, N.J., Peterson, P., Kasela, S., Codd, V., Peters, A., Ward-Caviness, C.K., Herder, C., Waldenberger, M., Roden, M., Singmann, P., Zeilinger, S., Illig, T., Homuth, G., Grabe, H.J., Volzke, H., Steil, L., Kocher, T., Murray, A., Melzer, D., Yaghootkar, H., Bandinelli, S., Moses, E.K., Kent, J.W., Curran, J.E., Johnson, M.P., Williams-Blangero, S., Westra, H.J., McRae, A.F., Smith, J.A., Kardia, S.L., Hovatta, I., Perola, M., Ripatti, S., Salomaa, V., Henders, A.K., Martin, N.G., Smith, A.K., Mehta, D., Binder, E.B., Nylocks, K.M., Kennedy, E.M., Klengel, T., Ding, J., Suchy-Dicey, A.M., Enquobahrie, D.A., Brody, J., Rotter, J.I., Chen, Y.D., Houwing-Duistermaat, J., Kloppenburg, M., Slagboom, P.E., Helmer, Q., den Hollander, W., Bean, S., Raj, T., Bakhshi, N., Wang, Q.P., Oyston, L.J., Psaty, B.M., Tracy, R.P., Montgomery, G.W., Turner, S.T., Blangero, J., Meulenbelt, I., Ressler, K.J., Yang, J., Franke, L., Kettunen, J., Visscher, P.M., Neely, G.G., Korstanje, R., Hanson, R.L., **Prokisch, H.**, Ferrucci, L., Esko, T., Teumer, A., van Meurs, J.B., and Johnson, A.D. (2015). The transcriptional landscape of age in human peripheral blood. Nat Commun 6, 8570.

Olahova, M., Hardy, S.A., Hall, J., Yarham, J.W., Haack, T.B., Wilson, W.C., Alston, C.L., He, L., Aznauryan, E., Brown, R.M., Brown, G.K., Morris, A.A., Mundy, H., Broomfield, A., Barbosa, I.A., Simpson, M.A., Deshpande, C., Moeslinger, D., **Koch, J.**, Stettner, G.M., Bonnen, P.E., **Prokisch, H.**, Lightowlers, R.N., McFarland, R., Chrzanowska-Lightowlers, Z.M., and Taylor, R.W. (2015). LRPPRC mutations cause early-onset multisystem mitochondrial disease outside of the French-Canadian population. Brain 138, 3503-3519.

Olahova, M., Haack, T.B., Alston, C.L., Houghton, J.A., He, L., Morris, A.A., Brown, G.K., McFarland, R., Chrzanowska-Lightowlers, Z.M., Lightowlers, R.N., **Prokisch, H.**, and Taylor, R.W. (2015). A truncating PET100 variant causing fatal infantile lactic acidosis and isolated cytochrome c oxidase deficiency. Eur J Hum Genet 23, 935-939.

Morscher, R.J., Aminzadeh-Gohari, S., **Feichtinger, R.G.**, **Mayr, J.A.**, Lang, R., Neureiter, D., **Sperl, W.**, and **Kofler, B.** (2015). Inhibition of Neuroblastoma Tumor Growth by Ketogenic Diet and/or Calorie Restriction in a CD1-Nu Mouse Model. PLoS One 10, e0129802.

McCann, B.J., Tuppen, H.A., Kusters, B., Lammens, M., Smeitink, J.A., Taylor, R.W., Rodenburg, R.J., and **Wortmann, S.B.** (2015). A novel mitochondrial DNA m.7507A>G mutation is only pathogenic at high levels of heteroplasmy. Neuromuscul Disord 25, 262-267.

Mayr, J.A., Haack, T.B., **Freisinger, P.**, Karall, D., **Makowski, C.**, **Koch, J.**, **Feichtinger, R.G.**, **Zimmermann, F.A.**, Rolinski, B., **Ahting, U.**, **Meitinger, T.**, **Prokisch, H.**, and **Sperl, W.** (2015). Spectrum of combined respiratory chain defects. J Inherit Metab Dis 38, 629-640.

Mayr, J.A. (2015). Lipid metabolism in mitochondrial membranes. J Inherit Metab Dis 38, 137-144.

Magner, M., Dvorakova, V., Tesarova, M., Mazurova, S., Hansikova, H., Zahorec, M., Brennerova, K., Bzduch, V., Spiegel, R., Horovitz, Y., Mandel, H., Eminoglu, F.T., **Mayr, J.A.**,

Koch, J., Martinelli, D., Bertini, E., Konstantopoulou, V., Smet, J., Rahman, S., Broomfield, A., Stojanovic, V., Dionisi-Vici, C., van Coster, R., Morava, E., **Sperl, W.**, Zeman, J., and Honzik, T. (2015). Erratum to: TMEM70 deficiency: long-term outcome of 48 patients. J Inherit Metab Dis 38, 583-584.

Magner, M., Dvorakova, V., Tesarova, M., Mazurova, S., Hansikova, H., Zahorec, M., Brennerova, K., Bzduch, V., Spiegel, R., Horovitz, Y., Mandel, H., Eminoglu, F.T., **Mayr, J.A.**, **Koch, J.**, Martinelli, D., Bertini, E., Konstantopoulou, V., Smet, J., Rahman, S., Broomfield, A., Stojanovic, V., Dionisi-Vici, C., van Coster, R., Morava, E., **Sperl, W.**, Zeman, J., and Honzik, T. (2015). TMEM70 deficiency: long-term outcome of 48 patients. J Inherit Metab Dis 38, 417-426.

Koch, J., **Freisinger, P.**, **Feichtinger, R.G.**, **Zimmermann, F.A.**, Rauscher, C., Wagentristl, H.P., Konstantopoulou, V., Seidl, R., Haack, T.B., **Prokisch, H.**, **Ahting, U.**, **Sperl, W.**, **Mayr, J.A.**, and Maier, E.M. (2015). Mutations in TTC19: expanding the molecular, clinical and biochemical phenotype. Orphanet J Rare Dis 10, 40.

Kettwig, M., Schubach, M., **Zimmermann, F.A.**, Klinge, L., **Mayr, J.A.**, Biskup, S., **Sperl, W.**, Gartner, J., and Huppke, P. (2015). From ventriculomegaly to severe muscular atrophy: expansion of the clinical spectrum related to mutations in AIFM1. Mitochondrion 21, 12-18.

Huemer, M., Karall, D., Schossig, A., Abdenur, J.E., Al Jasmi, F., Biagosch, C., Distelmaier, F., **Freisinger, P.**, Graham, B.H., Haack, T.B., Hauser, N., Hertecant, J., Ebrahimi-Fakhari, D., Konstantopoulou, V., Leydiker, K., Lourenco, C.M., Scholl-Burgi, S., Wilichowski, E., Wolf, N.I., **Wortmann, S.B.**, Taylor, R.W., **Mayr, J.A.**, Bonnen, P.E., **Sperl, W.**, **Prokisch, H.**, and McFarland, R. (2015). Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. J Inherit Metab Dis 38, 905-914.

Holzerova, E., and **Prokisch, H.** (2015). Mitochondria: Much ado about nothing? How dangerous is reactive oxygen species production? Int J Biochem Cell Biol 63, 16-20.

Haack, T.B., Staufner, C., Kopke, M.G., Straub, B.K., Kolker, S., Thiel, C., **Freisinger, P.**, Baric, I., McKiernan, P.J., Dikow, N., Harting, I., Beisse, F., Burgard, P., Kotzaeridou, U., Kuhr, J., Himbert, U., Taylor, R.W., Distelmaier, F., Vockley, J., Ghaloul-Gonzalez, L., Zschocke, J., **Kremer, L.S.**, Graf, E., Schwarzmayr, T., Bader, D.M., Gagneur, J., Wieland, T., Terrile, C., Strom, T.M., **Meitinger, T.**, Hoffmann, G.F., and **Prokisch, H.** (2015). Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. Am J Hum Genet 97, 163-169.

Haack, T.B., Jackson, C.B., Murayama, K., **Kremer, L.S.**, Schaller, A., Kotzaeridou, U., de Vries, M.C., Schottmann, G., Santra, S., Buchner, B., Wieland, T., Graf, E., **Freisinger, P.**, Eggimann, S., Ohtake, A., Okazaki, Y., Kohda, M., Kishita, Y., Tokuzawa, Y., Sauer, S., Memari, Y., Kolb-Kokocinski, A., Durbin, R., Hasselmann, O., Cremer, K., Albrecht, B., Wieczorek, D., Engels, H., Hahn, D., Zink, A.M., Alston, C.L., Taylor, R.W., Rodenburg, R.J., Trollmann, R., **Sperl, W.**, Strom, T.M., Hoffmann, G.F., **Mayr, J.A.**, **Meitinger, T.**, Bolognini, R., Schuelke, M., Nuoffer, J.M., Kolker, S., **Prokisch, H.**, and Klopstock, T. (2015). Deficiency of ECHS1 causes mitochondrial encephalopathy with cardiac involvement. Ann Clin Transl Neurol 2, 492-509.

Franik, S., Huidekoper, H.H., Visser, G., de Vries, M., de Boer, L., Hermans-Peters, M., Rodenburg, R., Verhaak, C., Vlieger, A.M., Smeitink, J.A., Janssen, M.C., and **Wortmann, S.B.** (2015). High prevalence of complementary and alternative medicine use in patients with genetically proven mitochondrial disorders. J Inherit Metab Dis 38, 477-482.

Flaquer, A., Rospleszcz, S., Reischl, E., Zeilinger, S., **Prokisch, H.**, **Meitinger, T.**, Meisinger, C., Peters, A., Waldenberger, M., Grallert, H., and Strauch, K. (2015). Mitochondrial GWA Analysis of Lipid Profile Identifies Genetic Variants to Be Associated with HDL Cholesterol and Triglyceride Levels. PLoS One 10, e0126294.

Falk, M.J., Shen, L., Gonzalez, M., Leipzig, J., Lott, M.T., Stassen, A.P., Diroma, M.A., Navarro-Gomez, D., Yeske, P., Bai, R., Boles, R.G., Brilhante, V., Ralph, D., DaRe, J.T., Shelton, R., Terry, S.F., Zhang, Z., Copeland, W.C., van Oven, M., **Prokisch, H.**, Wallace, D.C., Attimonelli, M., Krotoski, D., Zuchner, S., Gai, X., Participants, M.S.C., MseqDr Consortium participants: Sherri Bale, Rosanna Clima, Amy Goldstein, Aurora Pujol, Claire Sheldon, and Lee-Jun, W. (2015). Mitochondrial Disease Sequence Data Resource (MSeqDR): a global grass-roots consortium to facilitate deposition, curation, annotation, and integrated analysis of genomic data for the mitochondrial disease clinical and research communities. Mol Genet Metab 114, 388-396.

Evangelisti, C., de Biase, D., Kurelac, I., Ceccarelli, C., **Prokisch, H.**, **Meitinger, T.**, Caria, P., Vanni, R., Romeo, G., Tallini, G., Gasparre, G., and Bonora, E. (2015). A mutation screening of oncogenes, tumor suppressor gene TP53 and nuclear encoded mitochondrial complex I genes in oncocytic thyroid tumors. BMC Cancer 15, 157.

Ebner, S., Mangge, H., Langhof, H., Halle, M., Siegrist, M., Aigner, E., Paulmichl, K., Paulweber, B., Datz, C., **Sperl, W.**, **Kofler, B.**, and Weghuber, D. (2015). Mitochondrial Haplogroup T Is Associated with Obesity in Austrian Juveniles and Adults. PLoS One 10, e0135622.

Distelmaier, F., Haack, T.B., Catarino, C.B., Gallenmuller, C., Rodenburg, R.J., Strom, T.M., Baertling, F., **Meitinger, T.**, Mayatepek, E., **Prokisch, H.**, and Klopstock, T. (2015). MRPL44 mutations cause a slowly progressive multisystem disease with childhood-onset hypertrophic cardiomyopathy. Neurogenetics 16, 319-323.

Diodato, D., Invernizzi, F., Lamantea, E., Fagiolari, G., Parini, R., Menni, F., Parenti, G., Bollani, L., Pasquini, E., Donati, M.A., Cassandrini, D., Santorelli, F.M., Haack, T.B., **Prokisch, H.**, Ghezzi, D., Lamperti, C., and Zeviani, M. (2015). Common and Novel TMEM70 Mutations in a Cohort of Italian Patients with Mitochondrial Encephalocardiomyopathy. JIMD Rep 15, 71-78.

Danhauser, K., Smeitink, J.A., **Freisinger, P.**, **Sperl, W.**, Sabir, H., Hadzik, B., Mayatepek, E., Morava, E., and Distelmaier, F. (2015). Treatment options for lactic acidosis and metabolic crisis in children with mitochondrial disease. J Inherit Metab Dis 38, 467-475.

Brea-Calvo, G., Haack, T.B., Karall, D., Ohtake, A., Invernizzi, F., Carrozzo, R., Kremer, L., Dusi, S., Fauth, C., Scholl-Burgi, S., Graf, E., **Ahting, U.**, Resta, N., Laforgia, N., Verrigni, D., Okazaki, Y., Kohda, M., Martinelli, D., **Freisinger, P.**, Strom, T.M., **Meitinger, T.**, Lamperti, C., Lacson, A., Navas, P., **Mayr, J.A.**, Bertini, E., Murayama, K., Zeviani, M., **Prokisch, H.**, and Ghezzi, D. (2015). COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency. Am J Hum Genet 96, 309-317.

Baertling, F., Haack, T.B., Rodenburg, R.J., Schaper, J., Seibt, A., Strom, T.M., **Meitinger, T.**, Mayatepek, E., Hadzik, B., Selcan, G., **Prokisch, H.**, and Distelmaier, F. (2015). MRPS22 mutation causes fatal neonatal lactic acidosis with brain and heart abnormalities. Neurogenetics 16, 237-240.

Aminzadeh, S., Vidali, S., **Sperl, W.**, **Kofler, B.**, and **Feichtinger, R.G.** (2015). Energy metabolism in neuroblastoma and Wilms tumor. Transl Pediatr 4, 20-32.

Ahting, U., **Mayr, J.A.**, Vanlander, A.V., Hardy, S.A., Santra, S., **Makowski, C.**, Alston, C.L., **Zimmermann, F.A.**, Abela, L., Plecko, B., Rohrbach, M., Spranger, S., Seneca, S., Rolinski, B., Hagendorff, A., Hempel, M., **Sperl, W.**, **Meitinger, T.**, Smet, J., Taylor, R.W., Van Coster, R., **Freisinger, P.**, **Prokisch, H.**, and Haack, T.B. (2015). Clinical, biochemical, and genetic spectrum of seven patients with NFU1 deficiency. Front Genet 6, 123.

Ahlqvist, K.J., Leoncini, S., Pecorelli, A., **Wortmann, S.B.**, Ahola, S., Forsstrom, S., Guerranti, R., De Felice, C., Smeitink, J., Ciccoli, L., Hamalainen, R.H., and Suomalainen, A. (2015). MtDNA mutagenesis impairs elimination of mitochondria during erythroid maturation leading to enhanced erythrocyte destruction. Nat Commun 6, 6494.

2014

Wortmann, S.B., Kluijtmans, L.A., Sequeira, S., Wevers, R.A., and Morava, E. (2014). Leucine Loading Test is Only Discriminative for 3-Methylglutaconic Aciduria Due to AUH Defect. JIMD Rep 16, 1-6.

van der Zee, J., Van Langenhove, T., Kovacs, G.G., Dillen, L., Deschamps, W., Engelborghs, S., Matej, R., Vandenbulcke, M., Sieben, A., Dermaut, B., Smets, K., Van Damme, P., Merlin, C., Laureys, A., Van Den Broeck, M., Mattheijssens, M., Peeters, K., Benussi, L., Binetti, G., Ghidoni, R., Borroni, B., Padovani, A., Archetti, S., Pastor, P., Razquin, C., Ortega-Cubero, S., Hernandez, I., Boada, M., Ruiz, A., de Mendonca, A., Miltenberger-Miltenyi, G., do Couto, F.S., Sorbi, S., Nacmias, B., Bagnoli, S., Graff, C., Chiang, H.H., Thonberg, H., Perneczky, R., Diehl-Schmid, J., Alexopoulos, P., Frisoni, G.B., Bonvicini, C., Synofzik, M., Maetzler, W., vom Hagen, J.M., Schols, L., Haack, T.B., Strom, T.M., **Prokisch, H.**, Dols-Icardo, O., Clarimon, J., Lleo, A., Santana, I., Almeida, M.R., Santiago, B., Heneka, M.T., Jessen, F., Ramirez, A., Sanchez-Valle, R., Llado, A., Gelpi, E., Sarafov, S., Tournev, I., Jordanova, A., Parobkova, E., Fabrizi, G.M., Testi, S., Salmon, E., Strobel, T., Santens, P., Robberecht, W., De Jonghe, P., Martin, J.J., Cras, P., Vandenberghe, R., De Deyn, P.P., Cruts, M., Sleegers, K., and Van Broeckhoven, C. (2014). Rare mutations in SQSTM1 modify susceptibility to frontotemporal lobar degeneration. Acta Neuropathol 128, 397-410.

Synofzik, M., Haack, T.B., **Kopajtich, R.**, Gorza, M., Rapaport, D., Greiner, M., Schonfeld, C., Freiberg, C., Schorr, S., Holl, R.W., Gonzalez, M.A., Fritsche, A., Fallier-Becker, P., Zimmermann, R., Strom, T.M., **Meitinger, T.**, Zuchner, S., Schule, R., Schols, L., and **Prokisch, H.** (2014). Absence of BiP co-chaperone DNAJC3 causes diabetes mellitus and multisystemic neurodegeneration. Am J Hum Genet 95, 689-697.

Synofzik, M., Gonzalez, M.A., Lourenco, C.M., Coutelier, M., Haack, T.B., Rebelo, A., Hannequin, D., Strom, T.M., **Prokisch, H.**, Kernstock, C., Durr, A., Schols, L., Lima-Martinez, M.M., Farooq, A., Schule, R., Stevanin, G., Marques, W., Jr., and Zuchner, S. (2014). PNPLA6 mutations cause Boucher-Neuhauser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. Brain 137, 69-77.

Smith, P.M., Elson, J.L., Greaves, L.C., **Wortmann, S.B.**, Rodenburg, R.J., Lightowlers, R.N., Chrzanowska-Lightowlers, Z.M., Taylor, R.W., and Vila-Sanjurjo, A. (2014). The role of the mitochondrial ribosome in human disease: searching for mutations in 12S mitochondrial rRNA with high disruptive potential. Hum Mol Genet 23, 949-967.

Schulte, E.C., Schramm, K., Schurmann, C., Lichtner, P., Herder, C., Roden, M., Gieger, C., Peters, A., Trenkwalder, C., Hogl, B., Frauscher, B., Berger, K., Fietze, I., Gross, N., Stiasny-Kolster, K., Oertel, W., Bachmann, C.G., Paulus, W., Zimprich, A., Volzke, H., Schminke, U., Nauck, M., Illig, T., **Meitinger, T.**, Muller-Myhsok, B., **Prokisch, H.**, and Winkelmann, J. (2014). Blood cis-eQTL analysis fails to identify novel association signals among sub-threshold candidates from genome-wide association studies in restless legs syndrome. PLoS One 9, e98092.

Rodriguez, E., Baurecht, H., Wahn, A.F., Kretschmer, A., Hotze, M., Zeilinger, S., Klopp, N., Illig, T., Schramm, K., **Prokisch, H.**, Kuhnel, B., Gieger, C., Harder, J., Cifuentes, L., Novak, N., and Weidinger, S. (2014). An integrated epigenetic and transcriptomic analysis reveals distinct tissue-specific patterns of DNA methylation associated with atopic dermatitis. J Invest Dermatol 134, 1873-1883.

Reuter, M.S., Sass, J.O., Leis, T., Kohler, J., **Mayr, J.A.**, **Feichtinger, R.G.**, Rauh, M., Schanze, I., Bahr, L., Trollmann, R., Uebe, S., Ekici, A.B., and Reis, A. (2014). HIBCH deficiency in a patient with phenotypic characteristics of mitochondrial disorders. Am J Med Genet A 164A, 3162-3169.

Muller-Hocker, J., Schafer, S., Krebs, S., Blum, H., Zsurka, G., Kunz, W.S., **Prokisch, H.**, Seibel, P., and Jung, A. (2014). Oxyphil cell metaplasia in the parathyroids is characterized by somatic mitochondrial DNA mutations in NADH dehydrogenase genes and cytochrome c oxidase activity-impairing genes. Am J Pathol 184, 2922-2935.

Melchionda, L., Haack, T.B., Hardy, S., Abbink, T.E., Fernandez-Vizarra, E., Lamantea, E., Marchet, S., Morandi, L., Moggio, M., Carrozzo, R., Torraco, A., Diodato, D., Strom, T.M., **Meitinger, T.**, Tekturk, P., Yapici, Z., Al-Murshedi, F., Stevens, R., Rodenburg, R.J., Lamperti, C., Ardissone, A., Moroni, I., Uziel, G., **Prokisch, H.**, Taylor, R.W., Bertini, E., van der Knaap, M.S., Ghezzi, D., and Zeviani, M. (2014). Mutations in APOPT1, encoding a mitochondrial protein, cause cavitating leukoencephalopathy with cytochrome c oxidase deficiency. Am J Hum Genet 95, 315-325.

Medici, M., Porcu, E., Pistis, G., Teumer, A., Brown, S.J., Jensen, R.A., Rawal, R., Roef, G.L., Plantinga, T.S., Vermeulen, S.H., Lahti, J., Simmonds, M.J., Husemoen, L.L., Freathy, R.M., Shields, B.M., Pietzner, D., Nagy, R., Broer, L., Chaker, L., Korevaar, T.I., Plia, M.G., Sala, C., Volker, U., Richards, J.B., Sweep, F.C., Gieger, C., Corre, T., Kajantie, E., Thuesen, B., Taes, Y.E., Visser, W.E., Hattersley, A.T., Kratzsch, J., Hamilton, A., Li, W., Homuth, G., Lobina, M.,

Mariotti, S., Soranzo, N., Cocca, M., Nauck, M., Spielhagen, C., Ross, A., Arnold, A., van de Bunt, M., Liyanarachchi, S., Heier, M., Grabe, H.J., Masciullo, C., Galesloot, T.E., Lim, E.M., Reischl, E., Leedman, P.J., Lai, S., Delitala, A., Bremner, A.P., Philips, D.I., Beilby, J.P., Mulas, A., Vocale, M., Abecasis, G., Forsen, T., James, A., Widen, E., Hui, J., **Prokisch, H.**, Rietzschel, E.E., Palotie, A., Feddema, P., Fletcher, S.J., Schramm, K., Rotter, J.I., Kluttig, A., Radke, D., Traglia, M., Surdulescu, G.L., He, H., Franklyn, J.A., Tiller, D., Vaidya, B., de Meyer, T., Jorgensen, T., Eriksson, J.G., O'Leary, P.C., Wichmann, E., Hermus, A.R., Psaty, B.M., Ittermann, T., Hofman, A., Bosi, E., Schlessinger, D., Wallaschofski, H., Pirastu, N., Aulchenko, Y.S., de la Chapelle, A., Netea-Maier, R.T., Gough, S.C., Meyer Zu Schwabedissen, H., Frayling, T.M., Kaufman, J.M., Linneberg, A., Raikkonen, K., Smit, J.W., Kiemeney, L.A., Rivadeneira, F., Uitterlinden, A.G., Walsh, J.P., Meisinger, C., den Heijer, M., Visser, T.J., Spector, T.D., Wilson, S.G., Volzke, H., Cappola, A., Toniolo, D., Sanna, S., Naitza, S., and Peeters, R.P. (2014). Identification of novel genetic Loci associated with thyroid peroxidase antibodies and clinical thyroid disease. PLoS Genet 10, e1004123.

Mayr, J.A., **Feichtinger, R.G.**, Tort, F., Ribes, A., and **Sperl, W.** (2014). Lipoic acid biosynthesis defects. J Inherit Metab Dis 37, 553-563.

Kopajtich, R., Nicholls, T.J., Rorbach, J., Metodiev, M.D., **Freisinger, P.**, Mandel, H., Vanlander, A., Ghezzi, D., Carrozzo, R., Taylor, R.W., Marquard, K., Murayama, K., Wieland, T., Schwarzmayr, T., **Mayr, J.A.**, Pearce, S.F., Powell, C.A., Saada, A., Ohtake, A., Invernizzi, F., Lamantea, E., Sommerville, E.W., Pyle, A., Chinnery, P.F., Crushell, E., Okazaki, Y., Kohda, M., Kishita, Y., Tokuzawa, Y., Assouline, Z., Rio, M., Feillet, F., Mousson de Camaret, B., Chretien, D., Munnich, A., Menten, B., Sante, T., Smet, J., Regal, L., Lorber, A., Khoury, A., Zeviani, M., Strom, T.M., **Meitinger, T.**, Bertini, E.S., Van Coster, R., Klopstock, T., Rotig, A., Haack, T.B., Minczuk, M., and **Prokisch, H.** (2014). Mutations in GTPBP3 cause a mitochondrial translation defect associated with hypertrophic cardiomyopathy, lactic acidosis, and encephalopathy. Am J Hum Genet 95, 708-720.

Jackson, C.B., Nuoffer, J.M., Hahn, D., **Prokisch, H.**, Haberberger, B., Gautschi, M., Haberli, A., Gallati, S., and Schaller, A. (2014). Mutations in SDHD lead to autosomal recessive encephalomyopathy and isolated mitochondrial complex II deficiency. J Med Genet 51, 170-175.

Iuso, A., Sibon, O.C., Gorza, M., Heim, K., Organisti, C., **Meitinger, T.**, and **Prokisch, H.** (2014). Impairment of Drosophila orthologs of the human orphan protein C19orf12 induces bang sensitivity and neurodegeneration. PLoS One 9, e89439.

Herder, C., Nuotio, M.L., Shah, S., Blankenberg, S., Brunner, E.J., Carstensen, M., Gieger, C., Grallert, H., Jula, A., Kahonen, M., Kettunen, J., Kivimaki, M., Koenig, W., Kristiansson, K., Langenberg, C., Lehtimaki, T., Luotola, K., Marzi, C., Muller, C., Peters, A., **Prokisch, H.**, Raitakari, O., Rathmann, W., Roden, M., Salmi, M., Schramm, K., Swerdlow, D., Tabak, A.G., Thorand, B., Wareham, N., Wild, P.S., Zeller, T., Hingorani, A.D., Witte, D.R., Kumari, M., Perola, M., and Salomaa, V. (2014). Genetic determinants of circulating interleukin-1 receptor antagonist levels and their association with glycemic traits. Diabetes 63, 4343-4359.

Haghighi, A., Haack, T.B., Atiq, M., Mottaghi, H., Haghighi-Kakhki, H., Bashir, R.A., **Ahting, U.**, **Feichtinger, R.G.**, **Mayr, J.A.**, Rotig, A., Lebre, A.S., Klopstock, T., Dworschak, A., Pulido, N.,

Saeed, M.A., Saleh-Gohari, N., **Holzerova, E.**, Chinnery, P.F., Taylor, R.W., and **Prokisch, H.** (2014). Sengers syndrome: six novel AGK mutations in seven new families and review of the phenotypic and mutational spectrum of 29 patients. Orphanet J Rare Dis 9, 119.

Haack, T.B., Klee, D., Strom, T.M., Mayatepek, E., **Meitinger, T.**, **Prokisch, H.**, and Distelmaier, F. (2014). Infantile Leigh-like syndrome caused by SLC19A3 mutations is a treatable disease. Brain 137, e295.

Haack, T.B., Gorza, M., Danhauser, K., **Mayr, J.A.**, Haberberger, B., Wieland, T., Kremer, L., Strecker, V., Graf, E., Memari, Y., **Ahting, U.**, **Kopajtich, R.**, **Wortmann, S.B.**, Rodenburg, R.J., Kotzaeridou, U., Hoffmann, G.F., **Sperl, W.**, Wittig, I., Wilichowski, E., Schottmann, G., Schuelke, M., Plecko, B., Stephani, U., Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, and **Freisinger, P.** (2014). Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Mol Genet Metab 111, 342-352.

Feichtinger, R.G., Weis, S., **Mayr, J.A.**, Zimmermann, F., Geilberger, R., **Sperl, W.**, and **Kofler, B.** (2014). Alterations of oxidative phosphorylation complexes in astrocytomas. Glia 62, 514-525.

Feichtinger, R.G., **Sperl, W.**, Bauer, J.W., and **Kofler, B.** (2014). Mitochondrial dysfunction: a neglected component of skin diseases. Exp Dermatol 23, 607-614.

Dusi, S., Valletta, L., Haack, T.B., Tsuchiya, Y., Venco, P., Pasqualato, S., Goffrini, P., Tigano, M., Demchenko, N., Wieland, T., Schwarzmayr, T., Strom, T.M., Invernizzi, F., Garavaglia, B., Gregory, A., Sanford, L., Hamada, J., Bettencourt, C., Houlden, H., Chiapparini, L., Zorzi, G., Kurian, M.A., Nardocci, N., **Prokisch, H.**, Hayflick, S., Gout, I., and Tiranti, V. (2014). Exome sequence reveals mutations in CoA synthase as a cause of neurodegeneration with brain iron accumulation. Am J Hum Genet 94, 11-22.

Diodato, D., Melchionda, L., Haack, T.B., Dallabona, C., Baruffini, E., Donnini, C., Granata, T., Ragona, F., Balestri, P., Margollicci, M., Lamantea, E., Nasca, A., Powell, C.A., Minczuk, M., Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, Lamperti, C., Zeviani, M., and Ghezzi, D. (2014). VARS2 and TARS2 mutations in patients with mitochondrial encephalomyopathies. Hum Mutat 35, 983-989.

Deutschmann, A.J., Amberger, A., Zavadil, C., Steinbeisser, H., **Mayr, J.A.**, **Feichtinger, R.G.**, Oerum, S., Yue, W.W., and Zschocke, J. (2014). Mutation or knock-down of 17beta-hydroxysteroid dehydrogenase type 10 cause loss of MRPP1 and impaired processing of mitochondrial heavy strand transcripts. Hum Mol Genet 23, 3618-3628.

Dallabona, C., Diodato, D., Kevelam, S.H., Haack, T.B., Wong, L.J., Salomons, G.S., Baruffini, E., Melchionda, L., Mariotti, C., Strom, T.M., **Meitinger, T.**, **Prokisch, H.**, Chapman, K., Colley, A., Rocha, H., Ounap, K., Schiffmann, R., Salsano, E., Savoiardo, M., Hamilton, E.M., Abbink, T.E., Wolf, N.I., Ferrero, I., Lamperti, C., Zeviani, M., Vanderver, A., Ghezzi, D., and van der Knaap, M.S. (2014). Novel (ovario) leukodystrophy related to AARS2 mutations. Neurology 82, 2063-2071.

Becker, L., Kling, E., Schiller, E., Zeh, R., Schrewe, A., Holter, S.M., Mossbrugger, I., Calzada-Wack, J., Strecker, V., Wittig, I., Dumitru, I., Wenz, T., Bender, A., Aichler, M., Janik, D., Neff, F., Walch, A., Quintanilla-Fend, L., Floss, T., Bekeredjian, R., Gailus-Durner, V., Fuchs, H., Wurst, W., **Meitinger, T.**, **Prokisch, H.**, de Angelis, M.H., and Klopstock, T. (2014). MTO1-deficient mouse model mirrors the human phenotype showing complex I defect and cardiomyopathy. PLoS One 9, e114918.

Banka, S., de Goede, C., Yue, W.W., Morris, A.A., von Bremen, B., Chandler, K.E., **Feichtinger, R.G.**, Hart, C., Khan, N., **Lunzer, V.**, **Matakovic, L.**, Marquardt, T., **Makowski, C.**, **Prokisch, H.**, Debus, O., Nosaka, K., Sonwalkar, H., **Zimmermann, F.A.**, **Sperl, W.**, and **Mayr, J.A.** (2014). Expanding the clinical and molecular spectrum of thiamine pyrophosphokinase deficiency: a treatable neurological disorder caused by TPK1 mutations. Mol Genet Metab 113, 301-306.

Aulbert, W., Weigt-Usinger, K., Thiels, C., Kohler, C., Vorgerd, M., Schreiner, A., Hoffjan, S., Rothoeft, T., **Wortmann, S.B.**, Heyer, C.M., Podskarbi, T., and Lucke, T. (2014). Long survival in Leigh syndrome: new cases and review of literature. Neuropediatrics 45, 346-353.

2013

Wortmann, S.B., Kluijtmans, L.A., Rodenburg, R.J., Sass, J.O., Nouws, J., van Kaauwen, E.P., Kleefstra, T., Tranebjaerg, L., de Vries, M.C., Isohanni, P., Walter, K., Alkuraya, F.S., Smuts, I., Reinecke, C.J., van der Westhuizen, F.H., Thorburn, D., Smeitink, J.A., Morava, E., and Wevers, R.A. (2013). 3-Methylglutaconic aciduria--lessons from 50 genes and 977 patients. J Inherit Metab Dis 36, 913-921.

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

Westra, H.J., Peters, M.J., Esko, T., Yaghootkar, H., Schurmann, C., Kettunen, J., Christiansen, M.W., Fairfax, B.P., Schramm, K., Powell, J.E., Zhernakova, A., Zhernakova, D.V., Veldink, J.H., Van den Berg, L.H., Karjalainen, J., Withoff, S., Uitterlinden, A.G., Hofman, A., Rivadeneira, F., Hoen, P.A.C., Reinmaa, E., Fischer, K., Nelis, M., Milani, L., Melzer, D., Ferrucci, L., Singleton, A.B., Hernandez, D.G., Nalls, M.A., Homuth, G., Nauck, M., Radke, D., Volker, U., Perola, M., Salomaa, V., Brody, J., Suchy-Dicey, A., Gharib, S.A., Enquobahrie, D.A., Lumley, T., Montgomery, G.W., Makino, S., **Prokisch, H.**, Herder, C., Roden, M., Grallert, H., **Meitinger, T.**, Strauch, K., Li, Y., Jansen, R.C., Visscher, P.M., Knight, J.C., Psaty, B.M., Ripatti, S., Teumer, A., Frayling, T.M., Metspalu, A., van Meurs, J.B.J., and Franke, L. (2013). Systematic identification of trans eQTLs as putative drivers of known disease associations. Nat Genet 45, 1238-1243.

Stojanovic, V., **Mayr, J.A.**, **Sperl, W.**, Barisic, N., Doronjski, A., and Milak, G. (2013). Infantile peripheral neuropathy, deafness, and proximal tubulopathy associated with a novel mutation of the RRM2B gene: case study. Croat Med J 54, 579-584.

Schulte, E.C., Claussen, M.C., Jochim, A., Haack, T., Hartig, M., Hempel, M., **Prokisch, H.**, Haun-Junger, U., Winkelmann, J., Hemmer, B., Forschler, A., and Ilg, R. (2013). Mitochondrial

membrane protein associated neurodegenration: a novel variant of neurodegeneration with brain iron accumulation. Mov Disord 28, 224-227.

Oexle, K., Schormair, B., Ried, J.S., Czamara, D., Heim, K., Frauscher, B., Hogl, B., Trenkwalder, C., Martin Fiedler, G., Thiery, J., Lichtner, P., **Prokisch, H.**, Specht, M., Muller-Myhsok, B., Doring, A., Gieger, C., Peters, A., Wichmann, H.E., **Meitinger, T.**, and Winkelmann, J. (2013). Dilution of candidates: the case of iron-related genes in restless legs syndrome. Eur J Hum Genet 21, 410-414.

Neveling, K., Feenstra, I., Gilissen, C., Hoefsloot, L.H., Kamsteeg, E.J., Mensenkamp, A.R., Rodenburg, R.J., Yntema, H.G., Spruijt, L., Vermeer, S., Rinne, T., van Gassen, K.L., Bodmer, D., Lugtenberg, D., de Reuver, R., Buijsman, W., Derks, R.C., Wieskamp, N., van den Heuvel, B., Ligtenberg, M.J., Kremer, H., Koolen, D.A., van de Warrenburg, B.P., Cremers, F.P., Marcelis, C.L., Smeitink, J.A., **Wortmann, S.B.**, van Zelst-Stams, W.A., Veltman, J.A., Brunner, H.G., Scheffer, H., and Nelen, M.R. (2013). A post-hoc comparison of the utility of sanger sequencing and exome sequencing for the diagnosis of heterogeneous diseases. Hum Mutat 34, 1721-1726.

Mehta, D., Heim, K., Herder, C., Carstensen, M., Eckstein, G., Schurmann, C., Homuth, G., Nauck, M., Volker, U., Roden, M., Illig, T., Gieger, C., **Meitinger, T.**, and **Prokisch, H.** (2013). Impact of common regulatory single-nucleotide variants on gene expression profiles in whole blood. Eur J Hum Genet 21, 48-54.

Mayerle, J., den Hoed, C.M., Schurmann, C., Stolk, L., Homuth, G., Peters, M.J., Capelle, L.G., Zimmermann, K., Rivadeneira, F., Gruska, S., Volzke, H., de Vries, A.C., Volker, U., Teumer, A., van Meurs, J.B., Steinmetz, I., Nauck, M., Ernst, F., Weiss, F.U., Hofman, A., Zenker, M., Kroemer, H.K., **Prokisch, H.**, Uitterlinden, A.G., Lerch, M.M., and Kuipers, E.J. (2013). Identification of genetic loci associated with Helicobacter pylori serologic status. JAMA 309, 1912-1920.

Kornblum, C., Nicholls, T.J., Haack, T.B., Scholer, S., Peeva, V., Danhauser, K., Hallmann, K., Zsurka, G., Rorbach, J., **Iuso, A.**, Wieland, T., Sciacco, M., Ronchi, D., Comi, G.P., Moggio, M., Quinzii, C.M., DiMauro, S., Calvo, S.E., Mootha, V.K., Klopstock, T., Strom, T.M., **Meitinger, T.**, Minczuk, M., Kunz, W.S., and **Prokisch, H.** (2013). Loss-of-function mutations in MGME1 impair mtDNA replication and cause multisystemic mitochondrial disease. Nat Genet 45, 214-219.

Koene, S., **Wortmann, S.B.**, de Vries, M.C., Jonckheere, A.I., Morava, E., de Groot, I.J., and Smeitink, J.A. (2013). Developing outcome measures for pediatric mitochondrial disorders: which complaints and limitations are most burdensome to patients and their parents? Mitochondrion 13, 15-24.

Jakobs, B.S., van den Heuvel, L.P., Smeets, R.J., de Vries, M.C., Hien, S., Schaible, T., Smeitink, J.A., Wevers, R.A., **Wortmann, S.B.**, and Rodenburg, R.J. (2013). A novel mutation in COQ2 leading to fatal infantile multisystem disease. J Neurol Sci 326, 24-28.

Hildick-Smith, G.J., Cooney, J.D., Garone, C., **Kremer, L.S.**, Haack, T.B., Thon, J.N., Miyata, N., Lieber, D.S., Calvo, S.E., Akman, H.O., Yien, Y.Y., Huston, N.C., Branco, D.S., Shah, D.I.,

Freedman, M.L., Koehler, C.M., Italiano, J.E., Jr., Merkenschlager, A., Beblo, S., Strom, T.M., **Meitinger, T.**, **Freisinger, P.**, Donati, M.A., **Prokisch, H.**, Mootha, V.K., DiMauro, S., and Paw, B.H. (2013). Macrocytic anemia and mitochondriopathy resulting from a defect in sideroflexin 4. Am J Hum Genet 93, 906-914.

Hayflick, S.J., Kruer, M.C., Gregory, A., Haack, T.B., Kurian, M.A., Houlden, H.H., Anderson, J., Boddaert, N., Sanford, L., Harik, S.I., Dandu, V.H., Nardocci, N., Zorzi, G., Dunaway, T., Tarnopolsky, M., Skinner, S., Holden, K.R., Frucht, S., Hanspal, E., Schrander-Stumpel, C., Mignot, C., Heron, D., Saunders, D.E., Kaminska, M., Lin, J.P., Lascelles, K., Cuno, S.M., Meyer, E., Garavaglia, B., Bhatia, K., de Silva, R., Crisp, S., Lunt, P., Carey, M., Hardy, J., **Meitinger, T.**, **Prokisch, H.**, and Hogarth, P. (2013). beta-Propeller protein-associated neurodegeneration: a new X-linked dominant disorder with brain iron accumulation. Brain 136, 1708-1717.

Hartig, M., **Prokisch, H.**, **Meitinger, T.**, and Klopstock, T. (2013). Mitochondrial membrane protein-associated neurodegeneration (MPAN). Int Rev Neurobiol 110, 73-84.

Haack, T.B., Rolinski, B., Haberberger, B., Zimmermann, F., Schum, J., Strecker, V., Graf, E., Athing, U., Hoppen, T., Wittig, I., **Sperl, W.**, **Freisinger, P.**, **Mayr, J.A.**, Strom, T.M., **Meitinger, T.**, and **Prokisch, H.** (2013). Homozygous missense mutation in BOLA3 causes multiple mitochondrial dysfunctions syndrome in two siblings. J Inherit Metab Dis 36, 55-62.

Haack, T.B., **Kopajtich, R.**, **Freisinger, P.**, Wieland, T., Rorbach, J., Nicholls, T.J., Baruffini, E., Walther, A., Danhauser, K., **Zimmermann, F.A.**, Husain, R.A., Schum, J., Mundy, H., Ferrero, I., Strom, T.M., **Meitinger, T.**, Taylor, R.W., Minczuk, M., **Mayr, J.A.**, and **Prokisch, H.** (2013). ELAC2 mutations cause a mitochondrial RNA processing defect associated with hypertrophic cardiomyopathy. Am J Hum Genet 93, 211-223.

Haack, T.B., Hogarth, P., Gregory, A., **Prokisch, H.**, and Hayflick, S.J. (2013). BPAN: the only X-linked dominant NBIA disorder. Int Rev Neurobiol 110, 85-90.

Grunert, S.C., Mullerleile, S., De Silva, L., Barth, M., Walter, M., Walter, K., Meissner, T., Lindner, M., Ensenauer, R., Santer, R., Bodamer, O.A., Baumgartner, M.R., Brunner-Krainz, M., Karall, D., Haase, C., Knerr, I., Marquardt, T., Hennermann, J.B., Steinfeld, R., Beblo, S., Koch, H.G., Konstantopoulou, V., Scholl-Burgi, S., van Teeffelen-Heithoff, A., Suormala, T., **Sperl, W.**, Kraus, J.P., Superti-Furga, A., Schwab, K.O., and Sass, J.O. (2013). Propionic acidemia: clinical course and outcome in 55 pediatric and adolescent patients. Orphanet J Rare Dis 8, 6.

Gai, X., Ghezzi, D., Johnson, M.A., Biagosch, C.A., Shamseldin, H.E., Haack, T.B., Reyes, A., Tsukikawa, M., Sheldon, C.A., Srinivasan, S., Gorza, M., **Kremer, L.S.**, Wieland, T., Strom, T.M., Polyak, E., Place, E., Consugar, M., Ostrovsky, J., Vidoni, S., Robinson, A.J., Wong, L.J., Sondheimer, N., Salih, M.A., Al-Jishi, E., Raab, C.P., Bean, C., Furlan, F., Parini, R., Lamperti, C., **Mayr, J.A.**, Konstantopoulou, V., Huemer, M., Pierce, E.A., **Meitinger, T.**, **Freisinger, P.**, **Sperl, W.**, **Prokisch, H.**, Alkuraya, F.S., Falk, M.J., and Zeviani, M. (2013). Mutations in FBXL4, encoding a mitochondrial protein, cause early-onset mitochondrial encephalomyopathy. Am J Hum Genet 93, 482-495.

Finkenstedt, A., Schranz, M., Bosch, S., Karall, D., Burgi, S.S., Ensinger, C., Drach, M., **Mayr, J.A.**, Janecke, A.R., Vogel, W., Nachbaur, D., and Zoller, H. (2013). MNGIE Syndrome: Liver Cirrhosis Should Be Ruled Out Prior to Bone Marrow Transplantation. JIMD Rep 10, 41-44.

Baruffini, E., Dallabona, C., Invernizzi, F., Yarham, J.W., Melchionda, L., Blakely, E.L., Lamantea, E., Donnini, C., Santra, S., Vijayaraghavan, S., Roper, H.P., Burlina, A., **Kopajtich, R.**, Walther, A., Strom, T.M., Haack, T.B., **Prokisch, H.**, Taylor, R.W., Ferrero, I., Zeviani, M., and Ghezzi, D. (2013). MTO1 mutations are associated with hypertrophic cardiomyopathy and lactic acidosis and cause respiratory chain deficiency in humans and yeast. Hum Mutat 34, 1501-1509.

Baric, I., Fumic, K., Petkovic Ramadza, D., **Sperl, W.**, **Zimmermann, F.A.**, Muacevic-Katanec, D., Mitrovic, Z., Pazanin, L., Cvitanovic Sojat, L., Kekez, T., Reiner, Z., and **Mayr, J.A.** (2013). Mitochondrial myopathy associated with a novel 5522G>A mutation in the mitochondrial tRNA(Trp) gene. Eur J Hum Genet 21, 871-875.

Ashraf, S., Gee, H.Y., Woerner, S., Xie, L.X., Vega-Warner, V., Lovric, S., Fang, H., Song, X., Cattran, D.C., Avila-Casado, C., Paterson, A.D., Nitschke, P., Bole-Feysot, C., Cochat, P., Esteve-Rudd, J., Haberberger, B., Allen, S.J., Zhou, W., Airik, R., Otto, E.A., Barua, M., Al-Hamed, M.H., Kari, J.A., Evans, J., Bierzynska, A., Saleem, M.A., Bockenhauer, D., Kleta, R., El Desoky, S., Hacihamdioglu, D.O., Gok, F., Washburn, J., Wiggins, R.C., Choi, M., Lifton, R.P., Levy, S., Han, Z., Salviati, L., **Prokisch, H.**, Williams, D.S., Pollak, M., Clarke, C.F., Pei, Y., Antignac, C., and Hildebrandt, F. (2013). ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. J Clin Invest 123, 5179-5189.

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