Publikationsverzeichnis Praxis für Humangenetik Wuppertal

ANKRD11 variants: KBG syndrome and beyond I. Parenti, M. B. Mallozzi, I. Hüning, C. Gervasini, A. Kuechler, E. Agolini, B. Albrecht, C. Baquero-Montoya, A. Bohring, N. C. Bramswig, A. Busche, A. Dalski, Y. Guo, B. Hanker, Y. Hellenbroich, D. Horn, A. M. Innes, C. Leoni, Y. R. Li, S. A. Lynch, M. Mariani, L. Medne, B. Mikat, D. Milani, R. Onesimo, X. Ortiz-Gonzalez, **E. C. Prott**, H. Reutter, E. Rossier, A. Selicorni, P. Wieacker, A. Wilkens, D. Wieczorek, E. H. Zackai, G. Zampino, B. Zirn, H. Hakonarson, M. A. Deardorff, G. Gillessen-Kaesbach, & F. J. Kaiser,**.** *Clinical Genetics*, (2021). https://doi.org/10.1111/cge.13977.

Two cancer-predisposing variants in one family: Incidental finding of a fumarate hydrogenase (FH) germline variant in a family with Li-Fraumeni syndrome. L. Pahl, R. Beier, N. von Neuhoff, B. Auber, M. Höfs, E.-C. Prott, B. Schlegelberger, D. Reinhardt, & D. Steinemann*, Pediatric Blood & Cancer*, 65 (2018) e27254. https://doi.org/10.1002/pbc.27254.

Compound heterozygosity of low-frequency promoter deletions and rare loss-of-function mutations in TXNL4A causes Burn-McKeown syndrome. D. Wieczorek, W. G. Newman, T. Wieland, T. Berulava, M. Kaffe, D. Falkenstein, C. Beetz, E. Graf, T. Schwarzmayr, S. Douzgou, J. Clayton-Smith, S. B. Daly, S. G. Williams, S. S. Bhaskar, J. E. Urquhart, B. Anderson, J. O'Sullivan, O. Boute, J. Gundlach, J. C. Czeschik, A. J. van Essen, F. Hazan, S. Park, A. Hing, A. Kuechler, D. R. Lohmann, K. U. Ludwig, E. Mangold, L. Steenpaß, M. Zeschnigk, J. R. Lemke, C. M. Lourenco, U. Hehr, **E.-C. Prott**, M. Waldenberger, A. C. Böhmer, B. Horsthemke, R. T. O'Keefe, T. Meitinger, J. Burn, H.-J. Lüdecke, & T. M. Strom, American Journal of Human Genetics, 95 (2014) 698–707. https://doi.org/10.1016/j.ajhg.2014.10.014.

Behavioral phenotype in five individuals with de novo mutations within the GRIN2B gene. I. Freunscht, B. Popp, R. Blank, S. Endele, U. Moog, H. Petri, **E.-C. Prott**, A. Reis, J. Rübo, B. Zabel, M. Zenker, J. Hebebrand, & D. Wieczorek, Behavioral and brain functions: BBF, 9 (2013) 20. https://doi.org/10.1186/1744-9081-9-20.

A Novel Homozygous WDR72 Mutation in Two Siblings with Amelogenesis Imperfecta and Mild Short Stature. A. Kuechler, J. Hentschel, I. Kurth, B. Stephan, **E.-C. Prott**, B. Schweiger, A. Schuster, D. Wieczorek, & H.-J. Lüdecke, Molecular Syndromology, 3 (2012) 223–229. https://doi.org/10.1159/000343746.

Targeted next generation sequencing as a diagnostic tool in epileptic disorders. J. R. Lemke, E. Riesch, T. Scheurenbrand, M. Schubach, C. Wilhelm, I. Steiner, J. Hansen, C. Courage, S. Gallati, S. Bürki, S. Strozzi, B. G. Simonetti, S. Grunt, M. Steinlin, M. Alber, M. Wolff, T. Klopstock, **E. C. Prott**, R. Lorenz, C. Spaich, S. Rona, M. Lakshminarasimhan, J. Kröll, T. Dorn, G. Krämer, M. Synofzik, F. Becker, Y. G. Weber, H. Lerche, D. Böhm, & S. Biskup, *Epilepsia*, **53** (2012) 1387–1398. https://doi.org/10.1111/j.1528-1167.2012.03516.x.

Expanding the clinical spectrum associated with defects in CNTNAP2 and NRXN1. A. Gregor, B. Albrecht, I. Bader, E. K. Bijlsma, A. B. Ekici, H. Engels, K. Hackmann, D. Horn, J. Hoyer, J. Klapecki, J. Kohlhase, I. Maystadt, S. Nagl, **E. Prott**, S. Tinschert, R. Ullmann, E. Wohlleber, G. Woods, A. Reis, A. Rauch, & C. Zweier, *BMC medical genetics*, **12** (2011) 106. https://doi.org/10.1186/1471-2350-12-106.

IGF2/H19 hypomethylation in Silver-Russell syndrome and isolated hemihypoplasia. M. Zeschnigk, B. Albrecht, K. Buiting, D. Kanber, T. Eggermann, G. Binder, J. Gromoll, **E.-C. Prott**, S. Seland, & B. Horsthemke. European journal of human genetics: EJHG, 16 (2008) 328–334. https://doi.org/10.1038/sj.ejhg.5201974.

Towards mapping phenotypical traits in 18p- syndrome by array-based comparative genomic hybridisation and fluorescent in situ hybridisation. C. H. Brenk, **E.-C. Prott**, D. Trost, A. Hoischen, C. Walldorf, B. Radlwimmer, D. Wieczorek, P. Propping, G. Gillessen-Kaesbach, R. G. Weber, & H. Engels, European journal of human genetics: EJHG, 15 (2007) 35–44. https://doi.org/10.1038/sj.ejhg.5201718.

Is there a higher incidence of maternal uniparental disomy 14 [upd(14)mat]? Detection of 10 new patients by methylation-specific PCR D. Mitter, K. Buiting, F. von Eggeling, A. Kuechler, T. Liehr, U. A. Mau-Holzmann, **E.-C. Prott**, D. Wieczorek, & G. Gillessen-Kaesbach,. American Journal of Medical Genetics. Part A, 140 (2006) 2039–2049. https://doi.org/10.1002/ajmg.a.31414.

The first missense alteration in the MCPH1 gene causes autosomal recessive microcephaly with an extremely mild cellular and clinical phenotype. M. Trimborn, R. Richter, N. Sternberg, I. Gavvovidis, D. Schindler, A. P. Jackson, **E.-C. Prott**, K. Sperling, G. Gillessen-Kaesbach, & H. Neitzel, Human Mutation, 26 (2005) 496. https://doi.org/10.1002/humu.9382.

Childhood overgrowth in patients with common NF1 microdeletions. M. Spiegel, K. Oexle, D. Horn, E. Windt, A. Buske, B. Albrecht, **E.-C. Prott**, E. Seemanová, J. Seidel, T. Rosenbaum, D. Jenne, H. Kehrer-Sawatzki, & S. Tinschert, European journal of human genetics: EJHG, 13 (2005) 883–888. https://doi.org/10.1038/sj.ejhg.5201419.

Genotyping in 46 patients with tentative diagnosis of Treacher Collins syndrome revealed unexpected phenotypic variation. O. A. Teber, G. Gillessen-Kaesbach, S. Fischer, S. Böhringer, B. Albrecht, A. Albert, M. Arslan-Kirchner, E. Haan, M. Hagedorn-Greiwe, C. Hammans, W. Henn, G. K. Hinkel, R. König, E. Kunstmann, J. Kunze, L. M. Neumann, E.-C. Prott, A. Rauch, H.-D. Rott, H. Seidel, S. Spranger, M. Sprengel, B. Zoll, D. R. Lohmann, & D. Wieczorek,. European journal of human genetics: EJHG, 12 (2004) 879–890. https://doi.org/10.1038/sj.ejhg.5201260.

Prenatally detected trisomy 4 and 6 mosaicism--cytogenetic results and clinical phenotype. D. Wieczorek**, E. C. Prott**, W. P. Robinson, E. Passarge, & G. Gillessen-Kaesbach, *Prenatal Diagnosis*, **23** (2003) 128–133. https://doi.org/10.1002/pd.557.