MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

2021

No Difference in Penetrance between Truncating and Missense/Aberrant Splicing Pathogenic Variants in MLH1 and MSH2: A Prospective Lynch Syndrome Database Study. M. Dominguez-Valentin, J.-P. Plazzer, J. R. Sampson, C. Engel, S. Aretz, M. A. Jenkins, L. Sunde, I. Bernstein, G. Capella, F. Balaguer, F. Macrae, I. M. Winship, H. Thomas, D. G. Evans, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, R. H. Sijmons, M. Nielsen, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, A. Lindblom, A. D. Valle, F. Lopez-Kostner, K. Alvarez, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, S. Nakken, E. Hovig, K. Green, F. Lalloo, J. Hill, H. F. A. Vasen, C. Perne, R. Büttner, H. Görgens, E. Holinski-Feder, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, J. Weitz, **V. Steinke-Lange,** W. Schmiegel, D. Vangala, E. J. Crosbie, M. Pineda, M. Navarro, J. Brunet, L. Moreira, A. Sánchez, M. Serra-Burriel, M. Mints, R. Kariv, G. Rosner, T. A. Piñero, W. H. Pavicic, P. Kalfayan, S. W. T. Broeke, J.-P. Mecklin, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, P. Peltomäki, J. L. Hopper, A. K. Win, D. D. Buchanan, N. M. Lindor, S. Gallinger, L. L. Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, T. V. O. Hansen, L. Lindberg, E. A. Rødland, F. Neffa, P. Esperon, D. Tjandra, G. Möslein, T. T. Seppälä, & P. Møller, Journal of Clinical Medicine, 10 (2021) 2856. https://doi.org/10.3390/jcm10132856

Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases. B. Zurek, K. Ellwanger, L. E. L. M. Vissers, R. Schüle, M. Synofzik, A. Töpf, R. M. de Voer, S. Laurie, L. Matalonga, C. Gilissen, S. Ossowski, P. A. C. 't Hoen, A. Vitobello, J. M. Schulze-Hentrich, O. Riess, H. G. Brunner, A. J. Brookes, A. Rath, G. Bonne, G. Gumus, A. Verloes, N. Hoogerbrugge, T. Evangelista, T. Harmuth, M. Swertz, D. Spalding, A. Hoischen, S. Beltran, H. Graessner, & **Solve-RD consortium**, European journal of human genetics: EJHG, (2021). https://doi.org/10.1038/s41431-021-00859-0.

A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report. I. B. A. W. Te Paske, J. Garcia-Pelaez, A. K. Sommer, L. Matalonga, T. Starzynska, A. Jakubowska, Solve-RD-GENTURIS group, R. S. van der Post, J. Lubinski, C. Oliveira, N. Hoogerbrugge, & R. M. de Voer, European journal of human genetics: EJHG, (2021). https://doi.org/10.1038/s41431-021-00853-6

Stepwise ABC system for classification of any type of genetic variant. G. Houge, A. Laner, S. Cirak, N. de Leeuw, H. Scheffer, & J. T. den Dunnen, European journal of human genetics: EJHG, (2021). https://doi.org/10.1038/s41431-021-00903-z.

Actionable secondary findings in arrhythmogenic right ventricle cardiomyopathy genes: impact and challenge of genetic counseling. A. Abicht, U. Schön, A. Laner, **E. Holinski-Feder**, & I. Diebold**,** Cardiovascular Diagnosis and Therapy, 11 (2021) 637–649. https://doi.org/10.21037/cdt-20-585.

HPO-driven virtual gene panel: a new efficient approach in molecular autopsy of sudden unexplained death. U. Schön, A. Holzer, A. Laner, S. Kleinle, F. Scharf, A. Benet-Pagès, O. Peschel, **E. Holinski-Feder**, & I. Diebold, BMC medical genomics, 14 (2021) 94. https://doi.org/10.1186/s12920-021-00946-7 ..

Location matters - Genotype-phenotype correlation in LRSAM1 mutations associated with rare Charcot-Marie-Tooth neuropathy CMT2P. P. Reilich, B. Schlotter, F. Montagnese, B. Jordan, F. Stock, M. Schäff-Vogelsang, B. Hotter, K. Eger, I. Diebold, H. Erdmann, **K. Becker**, U. Schön, & A. Abicht, Neuromuscular disorders: NMD, 31 (2021) 123–133. https://doi.org/10.1016/j.nmd.2020.11.011.

Reproduktionsmedizin: Zahlen und Fakten für die Beratung. Kupka M (Hrsg.), Pörksen S, Popovici R, Krüssel JS, de Geyter C, Katzorke T, Koehler U, Würfel W, von Wolf M, Gnoth C, Kainer F, Trappe H, Weiß C; Urban & Fischer Verlag / Elsevier GmbH, 14. April 2021, ISBN-13: 978-3437249440

Mutation spectrum of the OPA1 gene in a large cohort of patients with suspected dominant optic atrophy: Identification and classification of 48 novel variants. N. Weisschuh, S. Schimpf-Linzenbold, P. Mazzola, S. Kieninger, T. Xiao, U. Kellner, **T. Neuhann**, C. Kelbsch, F. Tonagel, H. Wilhelm, S. Kohl, & B. Wissinger, PloS One, (2021) e0253987. https://doi.org/10.1371/journal.pone.0253987

Variation in the risk of colorectal cancer in families with Lynch syndrome: a retrospective cohort study. International Mismatch Repair Consortium. The Lancet. Oncology, (2021) S1470-2045(21)00189–3. https://doi.org/10.1016/S1470-2045(21)00189-3

Hereditary non-polyposis tumor risk syndromes V. Steinke-Lange & E. Holinski-Feder. MMW Fortschritte der Medizin, 163 (2021) 41–44. https://doi.org/10.1007/s15006-021-9960-1

Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. E. Legius, L. Messiaen, P. Wolkenstein, P. Pancza, R. A. Avery, Y. Berman, J. Blakeley, D.

Publikationsliste

Babovic-Vuksanovic, K. S. Cunha, R. Ferner, M. J. Fisher, J. M. Friedman, D. H. Gutmann, H. Kehrer-Sawatzki, B. R. Korf, V.-F. Mautner, S. Peltonen, K. A. Rauen, V. Riccardi, E. Schorry, A. Stemmer-Rachamimov, D. A. Stevenson, G. Tadini, N. J. Ullrich, D. Viskochil, K. Wimmer, K. Yohay, International Consensus Group on Neurofibromatosis Diagnostic Criteria (I-NF-DC), S. M. Huson, D. G. Evans, & S. R. Plotkin, Genetics in Medicine: Official Journal of the American College of Medical Genetics, (2021). https://doi.org/10.1038/s41436-021-01170-5

Uptake of hysterectomy and bilateral salpingo-oophorectomy in carriers of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report. T. T. Seppälä, M. Dominguez-Valentin, E. J. Crosbie, C. Engel, S. Aretz, F. Macrae, I. Winship, G. Capella, H. Thomas, E. Hovig, M. Nielsen, R. H. Sijmons, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, M. Mints, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, K. Green, F. Lalloo, J. Hill, W. Schmiegel, D. Vangala, C. Perne, H.-G. Strauß, J. Tecklenburg, **E. Holinski-Feder**, V. Steinke-Lange, J.-P. Mecklin, J.-P. Plazzer, M. Pineda, M. Navarro, J. B. Vida, R. Kariv, G. Rosner, T. A. Piñero, W. Pavicic, P. Kalfayan, S. W. Ten Broeke, M. A. Jenkins, L. Sunde, I. Bernstein, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, A. Della Valle, F. Lopez-Koestner, K. Alvarez, R. Büttner, H. Görgens, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, S. Redler, J. Weitz, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, J. L. Hopper, A. K. Win, N. M. Lindor, S. Gallinger, L. Le Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, K. A. W. Wadt, M. J. E. Mourits, Z. Ketabi, O. G. Denton, E. A. Rødland, H. Vasen, F. Neffa, P. Esperon, D. Tjandra, G. Möslein, E. Rokkones, J. R. Sampson, D. G. Evans, & P. Møller, European Journal of Cancer (Oxford, England: 1990), 148 (2021) 124–133. https://doi.org/10.1016/j.ejca.2021.02.022 .

Risk-reducing hysterectomy and bilateral salpingo-oophorectomy in female heterozygotes of pathogenic mismatch repair variants: a Prospective Lynch Syndrome Database report.
M. Dominguez-Valentin, E. J. Crosbie, C. Engel, S. Aretz, F. Macrae, I. Winship, G. Capella, H. Thomas, S. Nakken, E. Hovig, M. Nielsen, R. H. Sijmons, L. Bertario, B. Bonanni, M. G. Tibiletti, G. M. Cavestro, M. Mints, N. Gluck, L. Katz, K. Heinimann, C. A. Vaccaro, K. Green, F. Lalloo, J. Hill, W. Schmiegel, D. Vangala, C. Perne, H.-G. Strauß, J. Tecklenburg, **E. Holinski-Feder**, V. Steinke-Lange, J.-P. Mecklin, J.-P. Plazzer, M. Pineda, M. Navarro, J. B. Vidal, R. Kariv, G. Rosner, T. A. Piñero, M. L. Gonzalez, P. Kalfayan, N. Ryan, S. W. Ten Broeke, M. A. Jenkins, L. Sunde, I. Bernstein, J. Burn, M. Greenblatt, W. H. de Vos Tot Nederveen Cappel, A. Della Valle, F. Lopez-Koestner, K. Alvarez, R. Büttner, H. Görgens, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, J. Weitz, K. Pylvänäinen, L. Renkonen-Sinisalo, A. Lepistö, A. Auranen, J. L. Hopper, A. K. Win, R. W. Haile, N. M. Lindor, S. Gallinger, L. Le Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, C. Therkildsen, H. Okkels, Z. Ketabi, O. G. Denton, E. A. Rødland, H. Vasen, F. Neffa, P. Esperon, D. Tjandra, G. Möslein, J. R. Sampson, D. G. Evans, T. T. Seppälä, & P. Møller, Genetics in Medicine: Official Journal of the American College of Medical Genetics, 23 (2021) 705–712. https://doi.org/10.1038/s41436-020-01029-1 .

Value of upper gastrointestinal endoscopy for gastric cancer surveillance in patients with Lynch syndrome. S. Ladigan-Badura, D. B. Vangala, C. Engel, K. Bucksch, R. Hueneburg, C. Perne, J. Nattermann, V. Steinke-Lange, N. Rahner, H. K. Schackert, J. Weitz, M. Kloor, J. Kuhlkamp, H. P. Nguyen, G. Moeslein, C. Strassburg, M. Morak, **E. Holinski-Feder**, R. Buettner, S. Aretz, M. Loeffler, W. Schmiegel, C. Pox, K. Schulmann, & German Consortium for Familial Intestinal Cancer, International Journal of Cancer, 148 (2021) 106–114. https://doi.org/10.1002/ijc.33294

Effect of Discontinuation of Nusinersen Treatment in Long-Standing SMA3. M. Hiebeler, **A. Abicht,** P. Reilich, & M. C. Walter, Journal of Neuromuscular Diseases, (2021). https://content.iospress.com/articles/journal-of-neuromuscular-diseases/jnd210644

Beta-2-microglobulin Mutations Are Linked to a Distinct Metastatic Pattern and a Favorable Outcome in Microsatellite-Unstable Stage IV Gastrointestinal Cancers E. Busch, A. Ahadova, K. Kosmalla, L. Bohaumilitzky, P. L. Pfuderer, A. Ballhausen, J. Witt, J.-N. Wittemann, H. Bläker, **E. Holinski-Feder**, D. Jäger, M. von Knebel Doeberitz, G. M. Haag, & M. Kloor,. Frontiers in Oncology, 11 (2021) 669774. https://doi.org/10.3389/fonc.2021.669774.

The "unnatural" history of colorectal cancer in Lynch syndrome: Lessons from colonoscopy surveillance. A. Ahadova, T. T. Seppälä, C. Engel, R. Gallon, J. Burn, **E. Holinski-Feder**, V. Steinke-Lange, G. Möslein, M. Nielsen, S. W. Ten Broeke, L. Laghi, M. Dominguez-Valentin, G. Capella, F. Macrae, R. Scott, R. Hüneburg, J. Nattermann, M. Hoffmeister, H. Brenner, H. Bläker, M. von Knebel Doeberitz, J. R. Sampson, H. Vasen, J.-P. Mecklin, P. Møller, & M. Kloor, International Journal of Cancer, 148 (2021) 800–811. https://doi.org/10.1002/ijc.33224 .

Analysis in the Prospective Lynch Syndrome Database identifies sarcoma as part of the Lynch syndrome tumor spectrum.M. Dominguez-Valentin, J. R. Sampson, P. Møller, T. T. Seppälä, & PLSD Collaborators, International Journal of Cancer, 148 (2021) 512–513. https://doi.org/10.1002/ijc.33214

Publikationsliste

Dual roles of HSP70 chaperone HSPA1 in quality control of nascent and newly synthesized proteins. G. Tian, C. Hu, Y. Yun, W. Yang, W. Dubiel, Y. Cheng, & **D. A. Wolf**, The EMBO journal, 40 (2021) e106183. https://doi.org/10.15252/embj.2020106183.

CSN7B defines a variant COP9 signalosome complex with distinct function in DNA damage response. J. Wang, D. Dubiel, Y. Wu, Y. Cheng, **D. A. Wolf**, & W. Dubiel, Cell Reports, 34 (2021) 108662. https://doi.org/10.1016/j.celrep.2020.108662.

2020

PPARγ-p53-Mediated Vasculoregenerative Program to Reverse Pulmonary Hypertension. Hennigs JK, Cao A, Li CG, Shi M, Mienert J, Miyagawa K, Körbelin J, Marciano DP, Chen PI, Roughley M, Elliott MV, Harper RL, Bill MA, Chappell J, Moonen JR, **Diebold I**, Wang L, Snyder MP, Rabinovitch**Analysis of 3297 individuals suggests that the pathogenic germline 5'-UTR variant BRCA1 c.-107A > T is not common in south-east Germany. Laner A**, Benet-Pages A, Neitzel B, Holinski-Feder E. *Fam Cancer*. 2020;19(3):211-213. doi:10.1007/s10689-020-00175-4

Analysis of 3297 individuals suggests that the pathogenic germline 5'-UTR variant BRCA1 c.-107A > T is not common in south-east Germany. Laner A, Benet-Pages A, Neitzel B, Holinski-Feder E. *Fam Cancer*. 2020;19(3):211-213. doi:10.1007/s10689-020-00175-4

Critical assessment of secondary findings in genes linked to primary arrhythmia syndromes. Diebold I, Schön U, Scharf F, Benet-Pagès A, **Laner A**, Holinski-Feder E, Abicht A. *Hum Mutat*. 2020;41(5):1025-1032. doi:10.1002/humu.23996

Genotype-phenotype correlation in a novel ABHD12 mutation underlying PHARC syndrome. Thimm A, Rahal A, Schoen U, **Abicht A**, Klebe S, Kleinschnitz C, Hagenacker T, Stettner M. *J Peripher Nerv Syst*. 2020;25(2):112-116. https://doi.org/10.1111/jns.12367

Targeted deep-intronic sequencing in a cohort of unexplained cases of suspected Lynch syndrome. Arnold AM, Morak M, Benet-Pagès A, **Laner A**, Frishman D, Holinski-Feder E. *Eur J Hum Genet*. 2011 2020;28(5):597-608. doi:10.1038/s41431-019-0536-9

Prevalence of CNV-neutral structural genomic rearrangements in MLH1, MSH2, and PMS2 not detectable in routine NGS diagnostics. Morak M, Steinke-Lange V, Massdorf T, Benet-Pages A, Locher M, **Laner A**, Kayser K, Aretz S, Holinski-Feder E. *Fam Cancer*. 2020;19(2):161-167. doi:10.1007/s10689-020-00159-4

Missense mutations in *CASK* interfere with neurexin binding and neurexin-induced oligomerization. Pan YE, Tibbe D, Harms FL, Reißner C, Becker K, Dingmann B, Mirzaa G, Kattentidt-Mouravieva AA, Shoukier M, Aggarwal S, Missler M, Kutsche K, Kreienkamp H. *J Neurochem*. Published online October 14, 2020:jnc.15215. doi:10.1111/jnc.15215

Cancer incidence and spectrum among children with genetically confirmed Beckwith-Wiedemann spectrum in Germany: a retrospective cohort study. Cöktü S, Spix C, Kaiser M, Beygo J, **Kleinle S**, Bachmann N, Kohlschmidt N, Prawitt D, Beckmann A, Klaes R, Nevinny-Stickel-Hinzpeter C, Döhnert S, Kraus C, Kadgien G, Vater I, Biskup S, Kutsche M, Kohlhase J, Eggermann T, Zenker M, Kratz CP. *Br J Cancer*. 2020;123(4):619-623. doi:10.1038/s41416-020-0911-x

Delineating MT-ATP6-associated disease: From isolated neuropathy to early onset neurodegeneration. Stendel C, Neuhofer C, Floride E, Yuqing S, Ganetzky RD, Park J, Freisinger P, Kornblum C, **Kleinle S**, Schöls L, Distelmaier F, Stettner GM, Büchner B, Falk MJ, Mayr JA, Synofzik M, Abicht A, Haack TB, Prokisch H, Wortmann SB, Murayama K, Fang F, Klopstock T. *Neurol Genet*. 2020;6(1):e393. doi:10.1212/NXG.0000000000000393

Bilateral optic nerve atrophy in an 18-year-old female patient with diabetes mellitus J. Hall**, T. Neuhann**, F. Treumer, T. Neuhann, & I. Neuhann, Der Ophthalmologe: Zeitschrift Der Deutschen Ophthalmologischen Gesellschaft, 117 (2020) 934–938. https://doi.org/10.1007/s00347-019-01037-w

Age-dependent performance of BRAF mutation testing in Lynch syndrome diagnostics. Bläker H, Haupt S, Morak M, Holinski-Feder E, Arnold A, Horst D, Sieber-Frank J, Seidler F, von Winterfeld M, Alwers E, Chang-Claude J, Brenner H, Roth W, Engel C, Löffler M, Möslein G, Schackert H-K, Weitz J, Perne C, Aretz S, Hüneburg R, Schmiegel W, Vangala D, Rahner N, **Steinke-Lange V**, Heuveline V, von Knebel Doeberitz M, Ahadova A, Hoffmeister M, Kloor M. *Int J Cancer*. 2020;147(10):2801-2810. doi:10.1002/ijc.33273

Publikationsliste

Value of upper GI endoscopy for gastric cancer surveillance in patients with Lynch syndrome. Ladigan-Badura S, Vangala DB, Engel C, Bucksch K, Hueneburg R, Perne C, Nattermann J, **Steinke-Lange V**, Rahner N, Schackert HK, Weitz J, Kloor M, Kuhlkamp J, Nguyen HP, Moeslein G, Strassburg C, Morak M, Holinski-Feder E, Buettner R, Aretz S, Loeffler M, Schmiegel W, Pox C, Schulmann K. *Int J Cancer*. Published online September 15, 2020. doi:10.1002/ijc.33294

Rare tumors as leading symptom of hereditary tumor syndromes. Perne C, **Steinke-Lange V**, Aretz S, Spier I. *Pathologe*. 2020;41(5):535-549. doi:10.1007/s00292-020-00806-8

The "unnatural" history of colorectal cancer in Lynch syndrome: Lessons from colonoscopy surveillance. Ahadova A, Seppälä TT, Engel C, Gallon R, Burn J, Holinski-Feder E, **Steinke-Lange V**, Möslein G, Nielsen M, Ten Broeke SW, Laghi L, Dominguez-Valentin M, Capella G, Macrae F, Scott R, Hüneburg R, Nattermann J, Hoffmeister M, Brenner H, Bläker H, von Knebel Doeberitz M, Sampson JR, Vasen H, Mecklin J-P, Møller P, Kloor M. *Int J Cancer*. Published online July 19, 2020. doi:10.1002/ijc.33224

Risk-Reducing Gynecological Surgery in Lynch Syndrome: Results of an International Survey from the Prospective Lynch Syndrome Database. Dominguez-Valentin M, Seppälä TT, Engel C, Aretz S, Macrae F, Winship I, Capella G, Thomas H, Hovig E, Nielsen M, Sijmons RH, Bertario L, Bonanni B, Tibiletti MG, Cavestro GM, Mints M, Gluck N, Katz L, Heinimann K, Vaccaro CA, Green K, Lalloo F, Hill J, Schmiegel W, Vangala D, Perne C, Strauß H-G, Tecklenburg J, Holinski-Feder E, **Steinke-Lange V**, Mecklin J-P, Plazzer J-P, Pineda M, Navarro M, Vidal JB, Kariv R, Rosner G, Piñero TA, Gonzalez ML, Kalfayan P, Sampson JR, Ryan NAJ, Evans DG, Møller P, Crosbie EJ. *J Clin Med*. 2020;9(7). doi:10.3390/jcm9072290

Cancer risks in Lynch syndrome, Lynch-like syndrome, and familial colorectal cancer type X: a prospective cohort study. Bucksch K, Zachariae S, Aretz S, Büttner R, Holinski-Feder E, Holzapfel S, Hüneburg R, Kloor M, von Knebel Doeberitz M, Morak M, Möslein G, Nattermann J, Perne C, Rahner N, Schmiegel W, Schulmann K, **Steinke-Lange V**, Strassburg CP, Vangala DB, Weitz J, Loeffler M, Engel C. *BMC Cancer*. 2020;20(1):460. doi:10.1186/s12885-020-06926-x

Associations of Pathogenic Variants in MLH1, MSH2, and MSH6 With Risk of Colorectal Adenomas and Tumors and With Somatic Mutations in Patients With Lynch Syndrome. Engel C, Ahadova A, Seppälä TT, Aretz S, Bigirwamungu-Bargeman M, Bläker H, Bucksch K, Büttner R, de Vos Tot Nederveen Cappel WT, Endris V, Holinski-Feder E, Holzapfel S, Hüneburg R, Jacobs MAJM, Koornstra JJ, Langers AM, Lepistö A, Morak M, Möslein G, Peltomäki P, Pylvänäinen K, Rahner N, Renkonen-Sinisalo L, Schulmann K, **Steinke-Lange V**, Stenzinger A, Strassburg CP, van de Meeberg PC, van Kouwen M, van Leerdam M, Vangala DB, Vecht J, Verhulst M-L, von Knebel Doeberitz M, Weitz J, Zachariae S, Loeffler M, Mecklin J-P, Kloor M, Vasen HF. *Gastroenterology*. 2020;158(5):1326-1333. doi:10.1053/j.gastro.2019.12.032

Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. Dominguez-Valentin M, Sampson JR, Seppälä TT, Ten Broeke SW, Plazzer J-P, Nakken S, Engel C, Aretz S, Jenkins MA, Sunde L, Bernstein I, Capella G, Balaguer F, Thomas H, Evans DG, Burn J, Greenblatt M, Hovig E, de Vos Tot Nederveen Cappel WH, Sijmons RH, Bertario L, Tibiletti MG, Cavestro GM, Lindblom A, Della Valle A, Lopez-Köstner F, Gluck N, Katz LH, Heinimann K, Vaccaro CA, Büttner R, Görgens H, Holinski-Feder E, Morak M, Holzapfel S, Hüneburg R, Knebel Doeberitz M von, Loeffler M, Rahner N, Schackert HK, **Steinke-Lange V**, Schmiegel W, Vangala D, Pylvänäinen K, Renkonen-Sinisalo L, Hopper JL, Win AK, Haile RW, Lindor NM, Gallinger S, Le Marchand L, Newcomb PA, Figueiredo JC, Thibodeau SN, Wadt K, Therkildsen C, Okkels H, Ketabi Z, Moreira L, Sánchez A, Serra-Burriel M, Pineda M, Navarro M, Blanco I, Green K, Lalloo F, Crosbie EJ, Hill J, Denton OG, Frayling IM, Rødland EA, Vasen H, Mints M, Neffa F, Esperon P, Alvarez K, Kariv R, Rosner G, Pinero TA, Gonzalez ML, Kalfayan P, Tjandra D, Winship IM, Macrae F, Möslein G, Mecklin J-P, Nielsen M, Møller P. *Genet Med*. 2020;22(1):15-25. doi:10.1038/s41436-019-0596-9

Long Term Follow-Up on Pediatric Cases With Congenital Myasthenic Syndromes-A Retrospective Single Centre Cohort Study. Della Marina A, Wibbeler E, **Abicht A**, Kölbel H, Lochmüller H, Roos A, Schara U. *Front Hum Neurosci.* 2020 Dec 7;14:560860. https://doi.org/10.3389/fnhum.2020.560860

Thiamine Treatment and Favorable Outcome in an Infant with Biallelic TPK1 Variants. Eckenweiler M, Mayr JA, Grünert S, **Abicht A**, Korinthenberg R. *Neuropediatrics.* 2020 Oct 21. https://doi.org/10.1055/s-0040-1715631

Next Generation Sequencing in Pediatric Epilepsy Using Customized Panels: Size Matters. Willimsky EK, Munzig A, Mayer K, Biskup S, **Abicht A**, Hoertnagel K, Voss HV, Klein HG, Rost I, Larsen LHG, Dahl HA, Hoelz H, Stuelpnagel CV, Borggraefe I. *Neuropediatrics*. 2020 Oct 21. https://doi.org/10.1055/s-0040-1712488

Publikationsliste

Differential diagnosis of vacuolar myopathies in the NGS era. Mair D, Biskup S, Kress W, **Abicht A**, Brück W, Zechel S, Knop KC, Koenig FB, Tey S, Nikolin S, Eggermann K, Kurth I, Ferbert A, Weis J. *Brain Pathol*. 2020;30(5):877-896. https://doi.org/10.1111/bpa.12864

Dilative cardiomyopathy displaying double trouble etiology: Myocarditis and Mcleod syndrome? Montagnese F, Grabmaier U, **Abicht A**, Schoser B. *Clin Neurol Neurosurg*. 2020;197:106122. https://doi.org/10.1016/j.clineuro.2020.106122

Rare intronic mutation between Exon 62 and 63 (c.9225-285A>G) of the dystrophin gene associated with atypical BMD phenotype. Schüssler SC, Gerhalter T, **Abicht A**, Müller-Felber W, Nagel AM, Trollmann R. *Neuromuscul Disord*. 2020;30(8):680-684. https://doi.org/10.1016/j.nmd.2020.06.003

Mutational and phenotypic expansion of ATP1A3-related disorders: Report of nine cases. Boonsimma P, Michael Gasser M, Netbaramee W, Wechapinan T, Srichomthong C, Ittiwut C, Wagner M, Krenn M, Zimprich F, **Abicht A**, Biskup S, Roser T, Borggraefe I, Suphapeetiporn K, Shotelersuk V. *Gene*. 2020;749:144709. https://doi.org/10.1016/j.gene.2020.144709

Genotype-phenotype correlation in a novel ABHD12 mutation underlying PHARC syndrome. Thimm A, Rahal A, Schoen U, **Abicht A**, Klebe S, Kleinschnitz C, Hagenacker T, Stettner M. *J Peripher Nerv Syst*. 2020;25(2):112-116. https://doi.org/10.1111/jns.12367

[Multiple acyl-CoA dehydrogenase deficiency/glutaric aciduria type 2: difficult diagnosis, easy to treat]. Rabenstein M, Weis J, **Abicht A**, Fink GR, Lehmann HC, Wunderlich G. *Nervenarzt*. 2020;91(4):349-352. https://doi.org/10.1007/s00115-020-00886-0

Cooccurrence of Two Different Genetic Diseases: A Case of Valproic Acid Hepatotoxicity in Nicolaides-Baraitser Syndrome (SMARCA2 Mutation)-Due to a POLG1-Related Effect? Hofmeister B, von Stülpnagel C, Berweck S, **Abicht A**, Kluger G, Weber P. *Neuropediatrics*. 2020;51(1):49-52. https://doi.org/10.1055/s-0039-1694976

ATP1A3-related epilepsy: Report of seven cases and literature-based analysis of treatment response. Gasser M, Boonsimma P, Netbaramee W, Wechapinan T, Srichomthomg C, Ittiwut C, Krenn M, Zimprich F, Milenkovic I, **Abicht A**, Biskup S, Roser T, Shotelersuk V, Tacke M, Kuersten M, Wagner M, Borggraefe I, Suphapeetiporn K, von Stülpnagel C. *J Clin Neurosci*. 2020;72:31-38. https://doi.org/10.1016/j.jocn.2020.01.041

Impact on Clinical Decision Making of Next-Generation Sequencing in Pediatric Epilepsy in a Tertiary Epilepsy Referral Center. Hoelz H, Herdl C, Gerstl L, Tacke M, Vill K, von Stuelpnagel C, Rost I, Hoertnagel K, **Abicht A**, Hollizeck S, Larsen LHG, Borggraefe I. *Clin EEG Neurosci*. 2020;51(1):61-69. https://doi.org/10.1177/1550059419876518

Cancer Surveillance Guideline for individuals with PTEN hamartoma tumour syndrome. M. Tischkowitz, C. Colas, S. Pouwels, N. Hoogerbrugge, PHTS Guideline Development Group, & European Reference Network GENTURIS, European journal of human genetics: EJHG, 28 (2020) 1387–1393. https://doi.org/10.1038/s41431-020-0651-7.

Guidelines for the Li-Fraumeni and heritable TP53-related cancer syndromes. T. Frebourg, S. Bajalica Lagercrantz, C. Oliveira, R. Magenheim, D. G. Evans, & European Reference Network GENTURIS, European journal of human genetics: EJHG, 28 (2020) 1379–1386. https://doi.org/10.1038/s41431-020-0638-4.

Correction: Cancer risks by gene, age, and gender in 6350 carriers of pathogenic mismatch repair variants: findings from the Prospective Lynch Syndrome Database. M. Dominguez-Valentin, J. R. Sampson, T. T. Seppälä, S. W. Ten Broeke, J.-P. Plazzer, S. Nakken, C. Engel, S. Aretz, M. A. Jenkins, L. Sunde, I. Bernstein, G. Capella, F. Balaguer, H. Thomas, D. G. Evans, J. Burn, M. Greenblatt, E. Hovig, W. H. de Vos Tot Nederveen Cappel, R. H. Sijmons, L. Bertario, M. G. Tibiletti, G. M. Cavestro, A. Lindblom, A. Della Valle, F. Lopez-Köstner, N. Gluck, L. H. Katz, K. Heinimann, C. A. Vaccaro, R. Büttner, H. Görgens, **E. Holinski-Feder**, M. Morak, S. Holzapfel, R. Hüneburg, M. von Knebel Doeberitz, M. Loeffler, N. Rahner, H. K. Schackert, V. Steinke-Lange, W. Schmiegel, D. Vangala, K. Pylvänäinen, L. Renkonen-Sinisalo, J. L. Hopper, A. K. Win, R. W. Haile, N. M. Lindor, S. Gallinger, L. Le Marchand, P. A. Newcomb, J. C. Figueiredo, S. N. Thibodeau, K. Wadt, C. Therkildsen, H. Okkels, Z. Ketabi, L. Moreira, A. Sánchez, M. Serra-Burriel, M. Pineda, M. Navarro, I. Blanco, K. Green, F. Lalloo, E. J. Crosbie, J. Hill, O. G. Denton, I. M. Frayling, E. A. Rødland, H. Vasen, M. Mints, F. Neffa, P. Esperon, K. Alvarez, R. Kariv, G. Rosner, T. A. Pinero, M. L. Gonzalez, P. Kalfayan, D. Tjandra, I. M. Winship, F. Macrae, G. Möslein, J.-P. Mecklin, M. Nielsen, & P. Møller, Genetics in Medicine: Official Journal of the American College of Medical Genetics, 22 (2020) 1569. https://doi.org/10.1038/s41436-020-0892-4

Value of upper GI endoscopy for gastric cancer surveillance in patients with Lynch syndrome. Ladigan-Badura S, Vangala DB, Engel C, Bucksch K, Hueneburg R, Perne C, Nattermann J, Steinke-Lange V, Rahner N,

Publikationsliste

Schackert HK, Weitz J, Kloor M, Kuhlkamp J, Nguyen HP, Moeslein G, Strassburg C, Morak M, **Holinski-Feder E**, Buettner R, Aretz S, Loeffler M, Schmiegel W, Pox C, Schulmann K. Int J Cancer. Published online September 15, 2020. https://10.1002/ijc.33294

Contribution of mRNA Splicing to Mismatch Repair Gene Sequence Variant Interpretation. B. A. Thompson, R. Walters, M. T. Parsons, T. Dumenil, M. Drost, Y. Tiersma, N. M. Lindor, S. V. Tavtigian, N. de Wind, A. B. Spurdle, & InSiGHT Variant Interpretation Committee, Frontiers in Genetics, 11 (2020) 798. https://doi.org/10.3389/fgene.2020.00798.

eIF3 Associates with 80S Ribosomes to Promote Translation Elongation, Mitochondrial Homeostasis, and Muscle Health. Lin Y, Li F, Huang L, Polte C, Duan H, Fang J, Sun L, Xing X, Tian G, Cheng Y, Ignatova Z, Yang X, **Wolf DA**. *Molecular Cell*. 2020;79(4):575-587.e7. doi:10.1016/j.molcel.2020.06.003

eIF-Three to Tango: emerging functions of translation initiation factor eIF3 in protein synthesis and disease. **Wolf DA**, Lin Y, Duan H, Cheng Y. *Journal of Molecular Cell Biology*. 2020;12(6):403-409. doi:10.1093/jmcb/mjaa018

CSN7B Defines a Variant COP9 Signalosome Complex with Distinct Function in DNA Damage Response. Wang J, Dubiel D, Wu Y, Cheng Y, **Wolf DA**, Dubiel W. *SSRN Journal*. Published online 2020. doi:10.2139/ssrn.3593525

2019

Full-length transcript amplification and sequencing as universal method to test mRNA integrity and biallelic expression in mismatch repair genes. Morak M, Schaefer K, Steinke-Lange V, Koehler U, Keinath S, Massdorf T, Mauracher B, Rahner N, Bailey J, Kling C, Haeusser T, **Laner A**, Holinski-Feder E. *Eur J Hum Genet*. 2019;27(12):1808-1820. doi:10.1038/s41431-019-0472-8

Hereditary neuralgic amyotrophy in childhood caused by duplication within the SEPT9 gene: A family study. Neubauer K, Boeckelmann D, **Koehler U**, Kracht J, Kirschner J, Pendziwiat M, Zieger B. *Cytoskeleton (Hoboken)*. 2019;76(1):131-136. doi:10.1002/cm.21479

Miniature integrated micro-spectrometer array for snap shot multispectral sensing. Danz N, Höfer B, Förster E, Flügel-Paul T, Harzendorf T, Dannberg P, Leitel R, **Kleinle S**, Brunner R. *Opt Express*. 2019;27(4):5719-5728. doi:10.1364/OE.27.005719

HADHA and HADHB gene associated phenotypes - Identification of rare variants in a patient cohort by Next Generation Sequencing. Diebold I, Schön U, Horvath R, Schwartz O, Holinski-Feder E, Kölbel H, Abicht A. *Mol Cell Probes*. 2019;44:14-20. doi:10.1016/j.mcp.2019.01.003

PPARγ Interaction with UBR5/ATMIN Promotes DNA Repair to Maintain Endothelial Homeostasis. Li CG, Mahon C, Sweeney NM, Verschueren E, Kantamani V, Li D, Hennigs JK, Marciano DP, **Diebold I**, Abu-Halawa O, Elliott M, Sa S, Guo F, Wang L, Cao A, Guignabert C, Sollier J, Nickel NP, Kaschwich M, Cimprich KA, Rabinovitch M. *Cell Rep*. 2019;26(5):1333-1343.e7. doi:10.1016/j.celrep.2019.01.013

Lack of association between screening interval and cancer stage in Lynch syndrome may be accounted for by over-diagnosis; a prospective Lynch syndrome database report. Seppälä TT, Ahadova A, Dominguez-Valentin M, Macrae F, Evans DG, Therkildsen C, Sampson J, Scott R, Burn J, Möslein G, Bernstein I, Holinski-Feder E, Pylvänäinen K, Renkonen-Sinisalo L, Lepistö A, Lautrup CK, Lindblom A, Plazzer J-P, Winship I, Tjandra D, Katz LH, Aretz S, Hüneburg R, Holzapfel S, Heinimann K, Valle AD, Neffa F, Gluck N, de Vos tot Nederveen Cappel WH, Vasen H, Morak M, **Steinke-Lange V**, Engel C, Rahner N, Schmiegel W, Vangala D, Thomas H, Green K, Lalloo F, Crosbie EJ, Hill J, Capella G, Pineda M, Navarro M, Blanco I, ten Broeke S, Nielsen M, Ljungmann K, Nakken S, Lindor N, Frayling I, Hovig E, Sunde L, Kloor M, Mecklin J-P, Kalager M, Møller P. *Hered Cancer Clin Pract*. 2019;17(1):8. doi:10.1186/s13053-019-0106-8

Current recommendations for surveillance, risk reduction and therapy in Lynch syndrome patients. Hüneburg R, Aretz S, Büttner R, Daum S, Engel C, Fechner G, Habermann JK, Heling D, Hoffmann K, Holinski-Feder E, Kloor M, von Knebel-Döberitz M, Loeffler M, Möslein G, Perne C, Redler S, Rieß O, Schmiegel W, Seufferlein T, Siebers-Renelt U, **Steinke-Lange V**, Tecklenburg J, Vangala D, Vilz T, Weitz J, Wiedenmann B, Strassburg CP, Nattermann J. *Z Gastroenterol*. 2019;57(11):1309-1320. doi:10.1055/a-1008-9827

Genetic Screening and Personalized Prevention in Colorectal Cancer. Steinke-Lange V, Holinski-Feder E. *Visc Med*. 2019;35(4):226-230. doi:10.1159/000501941

Publikationsliste

The Apparent Genetic Anticipation in PMS2-Associated Lynch Syndrome Families Is Explained by Birth-cohort Effect. Ten Broeke SW, Rodríguez-Girondo M, Suerink M, Aretz S, Bernstein I, Capellá G, Engel C, Gomez-Garcia EB, van Hest LP, von Knebel Doeberitz M, Lagerstedt-Robinson K, Letteboer TGW, Moller P, van Os TA, Pineda M, Rahner N, Olderode-Berends MJW, von Salomé J, Schackert HK, Spruijt L, **Steinke-Lange V**, Wagner A, Tops CMJ, Nielsen M. *Cancer Epidemiol Biomarkers Prev*. 2019;28(6):1010-1014. doi:10.1158/1055-9965.EPI-18-0576

Survival by colon cancer stage and screening interval in Lynch syndrome: a prospective Lynch syndrome database report. Dominguez-Valentin M, Seppälä TT, Sampson JR, Macrae F, Winship I, Evans DG, Scott RJ, Burn J, Möslein G, Bernstein I, Pylvänäinen K, Renkonen-Sinisalo L, Lepistö A, Lindblom A, Plazzer J-P, Tjandra D, Thomas H, Green K, Lalloo F, Crosbie EJ, Hill J, Capella G, Pineda M, Navarro M, Vidal JB, Rønlund K, Nielsen RT, Yilmaz M, Elvang LL, Katz L, Nielsen M, Ten Broeke SW, Nakken S, Hovig E, Sunde L, Kloor M, Knebel Doeberitz MV, Ahadova A, Lindor N, **Steinke-Lange V**, Holinski-Feder E, Mecklin J-P, Møller P. *Hered Cancer Clin Pract*. 2019;17:28. doi:10.1186/s13053-019-0127-3

Cross Talk Between p22phox and ATF4 in the Endothelial Unfolded Protein Response. Petry A, Zhang Z, Trautz B, Rieß F, Görlach A. *Antioxid Redox Signal*. 2019;30(1):40-55. doi:10.1089/ars.2017.7481

VLDLR-associated Pontocerebellar Hypoplasia with Nonprogressive Congenital Ataxia and a Diagnostic Neuroimaging Pattern. Wilker M, Christen H-J, Schuster S, **Abicht A**, Boltshauser E. *Neuropediatrics*. 2019;50(6):404-405. https://doi.org/10.1055/s-0039-1688953

Charcot-Marie-Tooth disease type 2CC due to a frameshift mutation of the neurofilament heavy polypeptide gene in an Austrian family. Ikenberg E, Reilich P, **Abicht A**, Heller C, Schoser B, Walter MC. *Neuromuscul Disord*. 2019;29(5):392-397. https://doi.org/10.1016/j.nmd.2019.02.007

Congenital myasthenic syndrome caused by novel COL13A1 mutations. Dusl M, Moreno T, Munell F, Macaya A, Gratacòs M, **Abicht A**, Strom TM, Lochmüller H, Senderek J. *J Neurol*. 2019;266(5):1107-1112. https://doi.org/10.1007/s00415-019-09239-7

Characteristic clinical and ultrastructural findings in nesprinopathies. Kölbel H, **Abicht A**, Schwartz O, Katona I, Paulus W, Neuen-Jacob E, Weis J, Schara U. *Eur J Paediatr Neurol*. 2019;23(2):254-261. https://doi.org/10.1016/j.ejpn.2018.12.011

Congenital myasthenic syndromes in adulthood : Challenging, rare but treatable. Wunderlich G, **Abicht A**, Brunn A, Daimagüler H-S, Schroeter M, Fink GR, Lehmann HC, Cirak S. *Nervenarzt*. 2019;90(2):148-159. https://doi.org/10.1007/s00115-018-0562-9

A Novel Gain-of-Function Nav1.9 Mutation in a Child With Episodic Pain. Huang J, Estacion M, Zhao P, Dib-Hajj FB, Schulman B, **Abicht A**, Kurth I, Brockmann K, Waxman SG, Dib-Hajj SD. *Front Neurosci*. 2019;13:918. https://doi.org/10.3389/fnins.2019.00918

Results of multigene panel testing in familial cancer cases without genetic cause demonstrated by single gene testing .Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, Morak M, Holinski-Feder E, Holth A, Capella G, Davidson B, Evans DG, Martins A, Møller P, Hovig E. Sci Rep. 2019;9(1):18555. https://doi.org/10.1038/s41598-019-54517-z

Colorectal cancer prevention and early detection: what is the best strategy? Gross M, **Holinski-Feder E.** MMW Fortschr Med. 2019;161(7):43-48. https://doi.org/10.1007/s15006-019-0407-x

Reducing Mcl-1 gene dosage induces dopaminergic neuronal loss and motor impairments in Park2 knockout mice. Ekholm-Reed S, Baker R, Campos AR, Stouffer D, Henze M, **Wolf DA**, Loring JF, Thomas EA, Reed SI. *Commun Biol*. 2019;2(1):125. doi:10.1038/s42003-019-0366-x

Cross Talk between eIF2α and eEF2 Phosphorylation Pathways Optimizes Translational Arrest in Response to Oxidative Stress. Sanchez M, Lin Y, Yang C-C, McQuary P, Rosa Campos A, Aza Blanc P, **Wolf DA**. *iScience*. 2019;20:466-480. doi:10.1016/j.isci.2019.09.031

2018

Extending the critical regions for mutations in the non-coding gene RNU4ATAC in another patient with Roifman Syndrome. Hallermayr A, Graf J, **Koehler U**, Laner A, Schönfeld B, Benet-Pagès A, Holinski-Feder E. *Clin Case Rep*. 2018;6(11):2224-2228. doi:10.1002/ccr3.1830

LAMA2 gene mutation update: Toward a more comprehensive picture of the laminin-α2 variome and its related phenotypes. Oliveira J, Gruber A, Cardoso M, Taipa R, Fineza I, Gonçalves A, **Laner A**, Winder TL,

Publikationsliste

Schroeder J, Rath J, Oliveira ME, Vieira E, Sousa AP, Vieira JP, Lourenço T, Almendra L, Negrão L, Santos M, Melo-Pires M, Coelho T, den Dunnen JT, Santos R, Sousa M. *Hum Mutat*. 2018;39(10):1314-1337. doi:10.1002/humu.23599

Comprehensive analysis of the MLH1 promoter region in 480 patients with colorectal cancer and 1150 controls reveals new variants including one with a heritable constitutional MLH1 epimutation. Morak M, Ibisler A, Keller G, Jessen E, **Laner A**, Gonzales-Fassrainer D, Locher M, Massdorf T, Nissen AM, Benet-Pagès A, Holinski-Feder E. *J Med Genet*. 2018;55(4):240-248. doi:10.1136/jmedgenet-2017-104744

Comprehensive mutations associated with rare Charcot-Marie-Tooth neuropathy CMT2P. P. Reilich,**analysis of the MLH1 promoter region in 480 patients with colorectal cancer and 1150 controls reveals new variants including one with a heritable constitutional MLH1 epimutation.** Morak M, Ibisler A, Keller G, Jessen E, **Laner A**, Gonzales-Fassrainer D, Locher M, Massdorf T, Nissen AM, Benet-Pagès A, Holinski-Feder E. *J Med Genet*. 2018;55(4):240-248. doi:10.1136/jmedgenet-2017-104744

Variants in exons 5 and 6 of ACTB cause syndromic thrombocytopenia. Latham SL, Ehmke N, Reinke PYA, Taft MH, Eicke D, Reindl T, Stenzel W, Lyons MJ, Friez MJ, Lee JA, Hecker R, Frühwald MC, **Becker K**, Neuhann TM, Horn D, Schrock E, Niehaus I, Sarnow K, Grützmann K, Gawehn L, Klink B, Rump A, Chaponnier C, Figueiredo C, Knöfler R, Manstein DJ, Di Donato N. *Nat Commun*. 2018;9(1):4250. doi:10.1038/s41467-018-06713-0

Mitochondrial and nuclear disease panel (Mito-aND-Panel): Combined sequencing of mitochondrial and nuclear DNA by a cost-effective and sensitive NGS-based method. Abicht A, Scharf F, Kleinle S, Schön U, Holinski-Feder E, Horvath R, Benet-Pagès A, **Diebold I**. *Mol Genet Genomic Med*. ;6(6):1188-1198. doi:10.1002/mgg3.500

Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Trifunov S, Pyle A, Valentino ML, Liguori R, Yu-Wai-Man P, Burté F, Duff J, Kleinle S, **Diebold I**, Rugolo M, Horvath R, Carelli V. *Sci Rep*. 2018;8(1):11682. doi:10.1038/s41598-018-30143-z

Diagnosis of deficiency of adenosine deaminase 2 with early onset polyarteritis nodosa in an adult patient with a novel compound heterozygous CECR1 mutation. Lamprecht P, Humrich JY, **Diebold I**, Riemekasten*Clin Exp Rheumatol*. 2018;36 Suppl 111(2):177.

Extension of the phenotype of biallelic loss-of-function mutations in SLC25A46 to the severe form of pontocerebellar hypoplasia type I. Braunisch MC, Gallwitz H, Abicht A, **Diebold I**, Holinski-Feder E, Van Maldergem L, Lammens M, Kovács-Nagy R, Alhaddad B, Strom TM, Meitinger T, Senderek J, Rudnik-Schöneborn S, Haack TB2018;93(2):255-265. doi:10.1111/cge.13084

Clonal expansion of mtDNA deletions: different disease models assessed by digital droplet PCR in single muscle cells. Trifunov S, Pyle A, Valentino ML, Liguori R, Yu-Wai-Man P, Burté F, Duff J, **Kleinle S**, Diebold I, Rugolo M, Horvath R, Carelli V. *Sci Rep*. 2018;8(1):11682. doi:10.1038/s41598-018-30143-z

Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Begemann M, Rezwan FI, Beygo J, Docherty LE, Kolarova J, Schroeder C, Buiting K, Chokkalingam K, Degenhardt F, Wakeling EL, **Kleinle S**, González Fassrainer D, Oehl-Jaschkowitz B, Turner CLS, Patalan M, Gizewska M, Binder G, Bich Ngoc CT, Chi Dung V, Mehta SG, Baynam G, Hamilton-Shield JP, Aljareh S, Lokulo-Sodipe O, Horton R, Siebert R, Elbracht M, Temple IK, Eggermann T, Mackay DJG. *J Med Genet*. 2018;55(7):497-504. doi:10.1136/jmedgenet-2017-105190

A novel mechanism causing imbalance of mitochondrial fusion and fission in human myopathies. Bartsakoulia M, Pyle A, Troncoso-Chandía D, Vial-Brizzi J, Paz-Fiblas MV, Duff J, Griffin H, Boczonadi V, Lochmüller H, **Kleinle S**, Chinnery PF, Grünert S, Kirschner J, Eisner V, Horvath R. *Hum Mol Genet*. 2018;27(7):1186-1195. doi:10.1093/hmg/ddy033

Molekulare Autopsie nach plötzlichem Herztod. I. Diebold, J. Pickl, · **U. Schön,** · S. Kleinle, · A. Laner, · A. Benet-Pages, A. Abicht, G. Skopp, F. Musshoff, E. Holinski-Feder, *Rechtsmedizin* 2018; (28): 317–329 https://link.springer.com/article/10.1007%2Fs00194-018-0245-7

Revisiting mitochondrial diagnostic criteria in the new era of genomics. Witters P, Saada A, Honzik T, Tesarova M, **Kleinle S**, Horvath R, Goldstein A, Morava E. *Genet Med*. 2018;20(4):444-451. doi:10.1038/gim.2017.125

Copy number variation analysis and targeted NGS in 77 families with suspected Lynch syndrome reveals novel potential causative genes. Kayser K, Degenhardt F, Holzapfel S, Horpaopan S, Peters S, Spier I, Morak M, Vangala D, Rahner N, von Knebel-Doeberitz M, Schackert HK, Engel C, Büttner R, Wijnen J, Doerks T, Bork P,

Moebus S, Herms S, Fischer S, Hoffmann P, Aretz S, **Steinke-Lange V**. *Int J Cancer*. 2018;143(11):2800-2813. doi:10.1002/ijc.31725

No Difference in Colorectal Cancer Incidence or Stage at Detection by Colonoscopy Among 3 Countries With Different Lynch Syndrome Surveillance Policies. Engel C, Vasen HF, Seppälä T, Aretz S, Bigirwamungu-Bargeman M, de Boer SY, Bucksch K, Büttner R, Holinski-Feder E, Holzapfel S, Hüneburg R, Jacobs MAJM, Järvinen H, Kloor M, von Knebel Doeberitz M, Koornstra JJ, van Kouwen M, Langers AM, van de Meeberg PC, Morak M, Möslein G, Nagengast FM, Pylvänäinen K, Rahner N, Renkonen-Sinisalo L, Sanduleanu S, Schackert HK, Schmiegel W, Schulmann K, **Steinke-Lange V**, Strassburg CP, Vecht J, Verhulst M-L, de Vos Tot Nederveen Cappel W, Zachariae S, Mecklin J-P, Loeffler M. *Gastroenterology*. 2018;155(5):1400-1409.e2. doi:10.1053/j.gastro.2018.07.030

Interdisciplinary Diagnosis, Therapy and Follow-up of Patients with Endometrial Cancer. Guideline (S3-Level, AWMF Registry Number 032/034-OL, April 2018) - Part 2 with Recommendations on the Therapy and Follow-up of Endometrial Cancer, Palliative Care, Psycho-oncological/Psychosocial Care/Rehabilitation/Patient Information and Healthcare Facilities. Emons G, Steiner E, Vordermark D, Uleer C, Bock N, Paradies K, Ortmann O, Aretz S, Mallmann P, Kurzeder C, Hagen V, van Oorschot B, Höcht S, Feyer P, Egerer G, Friedrich M, Cremer W, Prott F-J, Horn L-C, Prömpeler H, Langrehr J, Leinung S, Beckmann MW, Kimmig R, Letsch A, Reinhardt M, Alt-Epping B, Kiesel L, Menke J, Gebhardt M, **Steinke-Lange V**, Rahner N, Lichtenegger W, Zeimet A, Hanf V, Weis J, Mueller M, Henscher U, Schmutzler RK, Meindl A, Hilpert F, Panke JE, Strnad V, Niehues C, Dauelsberg T, Niehoff P, Mayr D, Grab D, Kreißl M, Witteler R, Schorsch A, Mustea A, Petru E, Hübner J, Rose AD, Wight E, Tholen R, Bauerschmitz GJ, Fleisch M, Juhasz-Boess I, Lax S, Runnebaum I, Tempfer C, Nothacker MJ, Blödt S, Follmann M, Langer T, Raatz H, Wesselmann S, Erdogan S. *Geburtshilfe Frauenheilkd*. 2018;78(11):1089-1109. doi:10.1055/a-0715-2964

Cancer Risks for PMS2-Associated Lynch Syndrome. Ten Broeke SW, van der Klift HM, Tops CMJ, Aretz S, Bernstein I, Buchanan DD, de la Chapelle A, Capella G, Clendenning M, Engel C, Gallinger S, Gomez Garcia E, Figueiredo JC, Haile R, Hampel HL, Hopper JL, Hoogerbrugge N, von Knebel Doeberitz M, Le Marchand L, Letteboer TGW, Jenkins MA, Lindblom A, Lindor NM, Mensenkamp AR, Møller P, Newcomb PA, van Os TAM, Pearlman R, Pineda M, Rahner N, Redeker EJW, Olderode-Berends MJW, Rosty C, Schackert HK, Scott R, Senter L, Spruijt L, **Steinke-Lange V**, Suerink M, Thibodeau S, Vos YJ, Wagner A, Winship I, Hes FJ, Vasen HFA, Wijnen JT, Nielsen M, Win AK. *J Clin Oncol*. 2018;36(29):2961-2968. doi:10.1200/JCO.2018.78.4777

Interdisciplinary Diagnosis, Therapy and Follow-up of Patients with Endometrial Cancer. Guideline (S3-Level, AWMF Registry Nummer 032/034-OL, April 2018) - Part 1 with Recommendations on the Epidemiology, Screening, Diagnosis and Hereditary Factors of Endometrial Cancer. Emons G, Steiner E, Vordermark D, Uleer C, Bock N, Paradies K, Ortmann O, Aretz S, Mallmann P, Kurzeder C, Hagen V, van Oorschot B, Höcht S, Feyer P, Egerer G, Friedrich M, Cremer W, Prott F-J, Horn L-C, Prömpeler H, Langrehr J, Leinung S, Beckmann MW, Kimmig R, Letsch A, Reinhardt M, Alt-Epping B, Kiesel L, Menke J, Gebhardt M, **Steinke-Lange V**, Rahner N, Lichtenegger W, Zeimet A, Hanf V, Weis J, Mueller M, Henscher U, Schmutzler RK, Meindl A, Hilpert F, Panke JE, Strnad V, Niehues C, Dauelsberg T, Niehoff P, Mayr D, Grab D, Kreißl M, Witteler R, Schorsch A, Mustea A, Petru E, Hübner J, Rose AD, Wight E, Tholen R, Bauerschmitz GJ, Fleisch M, Juhasz-Boess I, Sigurd L, Runnebaum I, Tempfer C, Nothacker MJ, Blödt S, Follmann M, Langer T, Raatz H, Wesselmann S, Erdogan S. *Geburtshilfe Frauenheilkd*. 2018;78(10):949-971. doi:10.1055/a-0713-1218

A nomenclature and classification for the congenital myasthenic syndromes: preparing for FAIR data in the genomic era. Thompson R, **Abicht A**, Beeson D, Engel AG, Eymard B, Maxime E, Lochmüller H. *Orphanet J Rare Dis*. 2018;13(1):211. https://doi.org/10.1186/s13023-018-0955-7

The Curse of Apneic Spells. Radke J, Dreesmann M, Radke M, von Moers A, **Abicht A**, Stenzel W, Goebel HH. *Semin Pediatr Neurol*. 2018;26:56-58. https://doi.org/10.1016/j.spen.2017.03.006

A new case of limb girdle muscular dystrophy 2G in a Greek patient, founder effect and review of the literature. Brusa R, Magri F, Papadimitriou D, Govoni A, Del Bo R, Ciscato P, Savarese M, Cinnante C, Walter MC, **Abicht A**, Bulst S, Corti S, Moggio M, Bresolin N, Nigro V, Comi GP. *Neuromuscul Disord*. 2018;28(6):532-537. https://doi.org/10.1016/j.nmd.2018.04.006

Congenital myasthenic syndrome with episodic apnoea: clinical, neurophysiological and genetic features in the long-term follow-up of 19 patients. McMacken G, Whittaker RG, Evangelista T, **Abicht A**, Dusl M, Lochmüller H. *J Neurol*. 2018;265(1):194-203. https://doi.org/10.1007/s00415-017-8689-3

Role of germline aberrations affecting CTNNA1, MAP3K6 and MYD88 in gastric cancer susceptibility. Weren RDA, van der Post RS, Vogelaar IP, van Krieken JH, Spruijt L, Lubinski J, Jakubowska A, Teodorczyk U, Aalfs CM, van Hest LP, Oliveira C, Kamping EJ, Schackert HK, Ranzani GN, Gómez García EB, Hes FJ, **Holinski-Feder E**,

Publikationsliste

Genuardi M, Ausems MGEM, Sijmons RH, Wagner A, van der Kolk LE, Cats A, Bjørnevoll I, Hoogerbrugge N, Ligtenberg MJL. J Med Genet. 2018;55(10):669-674. https://doi.org/10.1136/jmedgenet-2017-104962

Haplotype analysis suggest that the MLH1 c.2059C > T mutation is a Swedish founder mutation. von Salomé J, Liu T, Keihäs M, Morak M, **Holinski-Feder E**, Berry IR, Moilanen JS, Baert-Desurmont S, Lindblom A, Lagerstedt-Robinson K. Fam Cancer. 2018;17(4):531-537. https://doi.org/10.1007/s10689-017-0067-x

Cancer risk and survival in path_MMR carriers by gene and gender up to 75 years of age: a report from the Prospective Lynch Syndrome Database. Møller P, Seppälä TT, Bernstein I, **Holinski-Feder E**, Sala P, Gareth Evans D, Lindblom A, Macrae F, Blanco I, Sijmons RH, Jeffries J, Vasen HFA, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen JT, Jenkins MA, Green K, Lalloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, Morak M, Renkonen-Sinisalo L, Valentin MD, Frayling IM, Plazzer J-P, Pylvanainen K, Genuardi M, Mecklin J-P, Moeslein G, Sampson JR, Capella G. Gut. 2018;67(7):1306-1316. https://doi.org/10.1136/gutjnl-2017-314057

Identification of genetic variants for clinical management of familial colorectal tumors. Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, Morak M, **Holinski-Feder E**, Martins A, Møller P, Hovig E. BMC Med Genet. 2018;19(1):26. https://doi.org/10.1186/s12881-018-0533-9

Potentially pathogenic germline CHEK2 c.319+2T>A among multiple early-onset cancer families. Dominguez-Valentin M, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, Morak M, Holinski-Feder E, Martins A, Møller P, Hovig E. Fam Cancer. 2018;17(1):141-153. https://doi.org/10.1007/s10689-017-0011-0

Genetic variants of prospectively demonstrated phenocopies in BRCA1/2 kindreds. Dominguez-Valentin M, Evans DGR, Nakken S, Tubeuf H, Vodak D, Ekstrøm PO, Nissen AM, Morak M, **Holinski-Feder E**, Martins A, Møller P, Hovig E. Hered Cancer Clin Pract. 2018;16:4. https://doi.org/10.1186/s13053-018-0086-0

Oxidized analogs of Di(1 H-indol-3-yl)methyl-4-substituted benzenes are NR4A1-dependent UPR inducers with potent and safe anti-cancer activity. Sanchez M, Xia Z, Rico-Bautista E, Cao X, Cuddy M, Castro DJ, Correa RG, Chen L, Yu J, Bobkov A, Ruvolo V, Andreeff M, Oshima RG, Matsuzawa S-I, Reed JC, Zhang X-K, Hansel D, **Wolf DA**, Dawson MI. *Oncotarget*. 2018;9(38):25057-25074. doi:10.18632/oncotarget.25285

Cullin 3-Based Ubiquitin Ligases as Master Regulators of Mammalian Cell Differentiation. Dubiel W, Dubiel D, **Wolf DA**, Naumann M. *Trends in Biochemical Sciences*. 2018;43(2):95-107. doi:10.1016/j.tibs.2017.11.010

2017

Novel mutation in two brothers with Hermansky Pudlak syndrome type 3. Sandrock-Lang K, Bartsch I, Buechele N, **Koehler U**, Simon-Gabriel CP, Eckenweiler M, Zieger B. *Blood Cells Mol Dis*. 2017;67:75-80. doi:10.1016/j.bcmd.2017.03.001

Genetic and phenotypic dissection of 1q43q44 microdeletion syndrome and neurodevelopmental phenotypes associated with mutations in ZBTB18 and HNRNPU. Depienne C, Nava C, Keren B, Heide S, Rastetter A, Passemard S, Chantot-Bastaraud S, Moutard M-L, Agrawal PB, VanNoy G, Stoler JM, Amor DJ, Billette de Villemeur T, Doummar D, Alby C, Cormier-Daire V, Garel C, Marzin P, Scheidecker S, de Saint-Martin A, Hirsch E, Korff C, Bottani A, Faivre L, Verloes A, Orzechowski C, Burglen L, Leheup B, Roume J, Andrieux J, Sheth F, Datar C, Parker MJ, Pasquier L, Odent S, Naudion S, Delrue M-A, Le Caignec C, Vincent M, Isidor B, Renaldo F, Stewart F, Toutain A, **Koehler U**, Häckl B, von Stülpnagel C, Kluger G, Møller RS, Pal D, Jonson T, Soller M, Verbeek NE, van Haelst MM, de Kovel C, Koeleman B, Monroe G, van Haaften G, Attié-Bitach T, Boutaud L, Héron D, Mignot C. *Hum Genet*. 2017;136(4):463-479. doi:10.1007/s00439-017-1772-0

BRCA1/2 missense mutations and the value of in-silico analyses. Sadowski CE, Kohlstedt D, Meisel C, Keller K, **Becker K**, Mackenroth L, Rump A, Schröck E, Wimberger P, Kast K. *Eur J Med Genet*. 2017;60(11):572-577. doi:10.1016/j.ejmg.2017.08.005

Molecular characterization of multifocal glioblastoma proves its monoclonal origin and reveals novel insights into clonal evolution and heterogeneity of glioblastomas. Abou-El-Ardat K, Seifert M, **Becker K**, Eisenreich S, Lehmann M, Hackmann K, Rump A, Meijer G, Carvalho B, Temme A, Schackert G, Schröck E, Krex D, Klink B. *Neuro Oncol*. 2017;19(4):546-557. doi:10.1093/neuonc/now231

Genetic heterogeneity of motor neuropathies. Bansagi B, Griffin H, Whittaker RG, Antoniadi T, Evangelista T, Miller J, Greenslade M, Forester N, Duff J, Bradshaw A, **Kleinle S**, Boczonadi V, Steele H, Ramesh V, Franko E, Pyle A, Lochmüller H, Chinnery PF, Horvath R. *Neurology*. 2017;88(13):1226-1234. doi:10.1212/WNL.0000000000003772

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

Spectrum of genetic variants of BRCA1 and BRCA2 in a German single center study. Meisel C, Sadowski CE, Kohlstedt D, Keller K, Stäritz F, Grübling N, **Becker K**, Mackenroth L, Rump A, Schröck E, Arnold N, Wimberger P, Kast K. *Arch Gynecol Obstet*. 2017;295(5):1227-1238. doi:10.1007/s00404-017-4330-z

Comprehensive molecular characterization of multifocal glioblastoma proves its monoclonal origin and reveals novel insights into clonal evolution and heterogeneity of glioblastomas. Abou-El-Ardat K, Seifert M, **Becker K**, Eisenreich S, Lehmann M, Hackmann K, Rump A, Meijer G, Carvalho B, Temme A, Schackert G, Schröck E, Krex D, Klink B. *Neuro Oncol*. 2017;19(4):546-557. doi:10.1093/neuonc/now231

Autosomal dominant frontometaphyseal dysplasia: Delineation of the clinical phenotype. Wade EM, Jenkins ZA, Daniel PB, Morgan T, Addor MC, Adés LC, Bertola D, Bohring A, Carter E, Cho T-J, de Geus CM, Duba H-C, Fletcher E, Hadzsiev K, Hennekam RCM, Kim CA, Krakow D, Morava E, **Neuhann T**, Sillence D, Superti-Furga A, Veenstra-Knol HE, Wieczorek D, Wilson LC, Markie DM, Robertson SP. *Am J Med Genet A*. 2017;173(7):1739-1746. doi:10.1002/ajmg.a.38267

A de novo 1q23.3-q24.2 deletion combined with a GORAB missense mutation causes a distinctive phenotype with cutis laxa. Al-Bughaili M, **Neuhann TM**, Flöttmann R, Mundlos S, Spielmann M, Kornak U, Fischer-Zirnsak B. *J Hum Genet*. 2017;62(2):325-328. doi:10.1038/jhg.2016.111

Molecular characterization of congenital myasthenic syndromes in Spain. Natera-de Benito D, Töpf A, Vilchez JJ, González-Quereda L, Domínguez-Carral J, Díaz-Manera J, Ortez C, Bestué M, Gallano P, Dusl M, **Abicht A**, Müller JS, Senderek J, García-Ribes A, Muelas N, Evangelista T, Azuma Y, McMacken G, Paipa Merchan A, Rodríguez Cruz PM, Camacho A, Jiménez E, Miranda-Herrero MC, Santana-Artiles A, García-Campos O, Dominguez-Rubio R, Olivé M, Colomer J, Beeson D, Lochmüller H, Nascimento A. *Neuromuscul Disord*. 2017;27(12):1087-1098. https://doi.org/10.1016/j.nmd.2017.08.003

Corrigendum to "Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient" [Neuromuscular Disorders 27 (2017) 856-860]. Ikenberg E, Karin I, Ertl-Wagner B, **Abicht A**, Bulst S, Krause S, Schoser B, Reilich P, Walter MC. *Neuromuscul Disord*. 2017;27(12):e1. https://doi.org/10.1016/j.nmd.2017.10.001

Rare diagnosis of telethoninopathy (LGMD2G) in a Turkish patient. Ikenberg E, Karin I, Ertl-Wagner B, **Abicht A**, Bulst S, Krause S, Schoser B, Reilich P, Walter MC. *Neuromuscul Disord*. 2017;27(9):856-860. https://doi.org/10.1016/j.nmd.2017.05.017

The Increasing Genetic and Phenotypical Diversity of Congenital Myasthenic Syndromes. McMacken G, **Abicht A**, Evangelista T, Spendiff S, Lochmüller H. *Neuropediatrics*. 2017;48(4):294-308. https://doi.org/10.1055/s-0037-1602832

Lifetime exercise intolerance with lactic acidosis as key manifestation of novel compound heterozygous ACAD9 mutations causing complex I deficiency. Schrank B, Schoser B, Klopstock T, Schneiderat P, Horvath R, **Abicht A**, Holinski-Feder E, Augustis S. *Neuromuscul Disord*. 2017;27(5):473-476. https://doi.org/10.1016/j.nmd.2017.02.005

Stroke as Initial Manifestation of Adenosine Deaminase 2 Deficiency. Elbracht M, Mull M, Wagner N, Kuhl C, **Abicht A**, Kurth I, Tenbrock K, Häusler M. *Neuropediatrics*. 2017;48(2):111-114. https://doi.org/10.1055/s-0036-1597611

Unraveling genetic predisposition to familial or early onset gastric cancer using germline whole-exome sequencing. Vogelaar IP, van der Post RS, van Krieken JHJ, Spruijt L, van Zelst-Stams WA, Kets CM, Lubinski J, Jakubowska A, Teodorczyk U, Aalfs CM, van Hest LP, Pinheiro H, Oliveira C, Jhangiani SN, Muzny DM, Gibbs RA, Lupski JR, de Ligt J, Vissers LELM, Hoischen A, Gilissen C, van de Vorst M, Goeman JJ, Schackert HK, Ranzani GN, Molinaro V, Gómez García EB, Hes FJ, Holinski-Feder E, Genuardi M, Ausems MGEM, Sijmons RH, Wagner A, van der Kolk LE, Bjørnevoll I, Høberg-Vetti H, van Kessel AG, Kuiper RP, Ligtenberg MJL, Hoogerbrugge N. Eur J Hum Genet. 2017;25(11):1246-1252. https://doi.org/10.1038/ejhg.2017.138

Elucidating the molecular basis of MSH2-deficient tumors by combined germline and somatic analysis. Vargas-Parra GM, González-Acosta M, Thompson BA, Gómez C, Fernández A, Dámaso E, Pons T, Morak M, Del Valle J, Iglesias S, Velasco À, Solanes A, Sanjuan X, Padilla N, de la Cruz X, Valencia A, Holinski-Feder E, Brunet J, Feliubadaló L, Lázaro C, Navarro M, Pineda M, Capellá G. Int J Cancer. 2017;141(7):1365-1380. https://doi.org/10.1002/ijc.30820

Loss of MSH2 and MSH6 due to heterozygous germline defects in MSH3 and MSH6. Morak M, Käsbauer S, Kerscher M, Laner A, Nissen AM, Benet-Pagès A, Schackert HK, Keller G, Massdorf T, **Holinski-Feder E.** Fam Cancer. 2017;16(4):491-500. https://doi.org/10.1007/s10689-017-9975-z

Publikationsliste

Incidence of and survival after subsequent cancers in carriers of pathogenic MMR variants with previous cancer: a report from the prospective Lynch syndrome database. Møller P, Seppälä T, Bernstein I, **Holinski-Feder E**, Sala P, Evans DG, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Jenkins M, Green K, Lalloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, Morak M, Renkonen-Sinisalo L, Frayling IM, Plazzer J-P, Pylvanainen K, Genuardi M, Mecklin J-P, Möslein G, Sampson JR, Capella G. Gut. 2017;66(9):1657-1664. https://doi.org/10.1136/gutjnl-2016-311403

Cancer incidence and survival in Lynch syndrome patients receiving colonoscopic and gynaecological surveillance: first report from the prospective Lynch syndrome database. Møller P, Seppälä T, Bernstein I, **Holinski-Feder E**, Sala P, Evans DG, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Green K, Lalloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, Morak M, Renkonen-Sinisalo L, Frayling IM, Plazzer J-P, Pylvanainen K, Sampson JR, Capella G, Mecklin J-P, Möslein G. Gut. 2017;66(3):464-472. https://doi.org/10.1136/gutjnl-2015-309675

Colorectal cancer incidence in path_MLH1 carriers subjected to different follow-up protocols: a Prospective Lynch Syndrome Database report. Seppälä T, Pylvänäinen K, Evans DG, Järvinen H, Renkonen-Sinisalo L, Bernstein I, **Holinski-Feder E**, Sala P, Lindblom A, Macrae F, Blanco I, Sijmons R, Jeffries J, Vasen H, Burn J, Nakken S, Hovig E, Rødland EA, Tharmaratnam K, de Vos Tot Nederveen Cappel WH, Hill J, Wijnen J, Jenkins M, Genuardi M, Green K, Lalloo F, Sunde L, Mints M, Bertario L, Pineda M, Navarro M, Morak M, Frayling IM, Plazzer J-P, Sampson JR, Capella G, Möslein G, Mecklin J-P, Møller P. Hered Cancer Clin Pract. 2017;15:18. https://doi.org/10.1186/s13053-017-0078-5

Trade-off and flexibility in the dynamic regulation of the cullin-RING ubiquitin ligase repertoire. Straube R, Shah M, Flockerzi D, **Wolf DA**. Shahrezaei V, ed. *PLoS Comput Biol*. 2017;13(11):e1005869. doi:10.1371/journal.pcbi.1005869

Schizosaccharomyces pombe - Polysome Profile Analysis and RNA Purification. **Wolf DA**, Bähler J, Wise JA. *Cold Spring Harb Protoc*. 2017;2017(4):pdb.prot091637. doi:10.1101/pdb.prot091637

Ubiquitylation-dependent oligomerization regulates activity of Nedd4 ligases. Attali I, Tobelaim WS, Persaud A, Motamedchaboki K, Simpson-Lavy KJ, Mashahreh B, Levin-Kravets O, Keren-Kaplan T, Pilzer I, Kupiec M, Wiener R, **Wolf DA**, Rotin D, Prag G. *EMBO J*. 2017;36(4):425-440. doi:10.15252/embj.201694314

2016

CANPMR syndrome and chromosome 1p32-p31 deletion syndrome coexist in two related individuals affected by simultaneous haplo-insufficiency of CAMTA1 and NIFA genes. Coci EG, **Koehler U**, Liehr T, Stelzner A, Fink C, Langen H, Riedel J. *Mol Cytogenet*. 2016;9:10. doi:10.1186/s13039-016-0219-y

In Pulmonary Arterial Hypertension, Reduced BMPR2 Promotes Endothelial-to-Mesenchymal Transition via HMGA1 and Its Target Slug. Hopper RK, Moonen J-RAJ, **Diebold I**, Cao A, Rhodes CJ, Tojais NF, Hennigs JK, Gu M, Wang L, Rabinovitch M. *Circulation*. 2016;133(18):1783-1794. doi:10.1161/CIRCULATIONAHA.115.020617

Novel genetic and neuropathological insights in neurogenic muscle weakness, ataxia, and retinitis pigmentosa (NARP). Claeys KG, Abicht A, Häusler M, **Kleinle S**, Wiesmann M, Schulz JB, Horvath R, Weis J. *Muscle Nerve*. 2016;54(2):328-333. doi:10.1002/mus.25125

Mitochondrial dysfunction in liver failure requiring transplantation. Lane M, Boczonadi V, Bachtari S, Gomez-Duran A, Langer T, Griffiths A, **Kleinle S**, Dineiger C, Abicht A, Holinski-Feder E, Schara U, Gerner P, Horvath R. *J Inherit Metab Dis*. 2016;39(3):427-436. doi:10.1007/s10545-016-9927-z

Exome Sequencing Identifies Biallelic MSH3 Germline Mutations as a Recessive Subtype of Colorectal Adenomatous Polyposis. Adam R, Spier I, Zhao B, Kloth M, Marquez J, Hinrichsen I, Kirfel J, Tafazzoli A, Horpaopan S, Uhlhaas S, Stienen D, Friedrichs N, Altmüller J, **Laner A**, Holzapfel S, Peters S, Kayser K, Thiele H, Holinski-Feder E, Marra G, Kristiansen G, Nöthen MM, Büttner R, Möslein G, Betz RC, Brieger A, Lifton RP, Aretz S. *Am J Hum Genet*. 2016;99(2):337-351. doi:10.1016/j.ajhg.2016.06.015

Exome sequencing identifies potential novel candidate genes in patients with unexplained colorectal adenomatous polyposis. Spier I, Kerick M, Drichel D, Horpaopan S, Altmüller J, **Laner A**, Holzapfel S, Peters S, Adam R, Zhao B, Becker T, Lifton RP, Holinski-Feder E, Perner S, Thiele H, Nöthen MM, Hoffmann P, Timmermann B, Schweiger MR, Aretz S. *Fam Cancer*. 2016;15(2):281-288. doi:10.1007/s10689-016-9870-z

Publikationsliste

Low-level APC mutational mosaicism is the underlying cause in a substantial fraction of unexplained colorectal adenomatous polyposis cases. Spier I, Drichel D, Kerick M, Kirfel J, Horpaopan S, **Laner A**, Holzapfel S, Peters S, Adam R, Zhao B, Becker T, Lifton RP, Perner S, Hoffmann P, Kristiansen G, Timmermann B, Nöthen MM, Holinski-Feder E, Schweiger MR, Aretz S. *J Med Genet*. 2016;53(3):172-179. doi:10.1136/jmedgenet-2015-103468

Recurrent de novo BICD2 mutation associated with arthrogryposis multiplex congenita and bilateral perisylvian polymicrogyria. Ravenscroft G, Di Donato N, Hahn G, Davis MR, Craven PD, Poke G, Neas KR, **Neuhann TM**, Dobyns WB, Laing NG. *Neuromuscul Disord*. 2016;26(11):744-748. doi:10.1016/j.nmd.2016.09.009

Mutations in MAP3K7 that Alter the Activity of the TAK1 Signaling Complex Cause Frontometaphyseal Dysplasia. Wade EM, Daniel PB, Jenkins ZA, McInerney-Leo A, Leo P, Morgan T, Addor MC, Adès LC, Bertola D, Bohring A, Carter E, Cho T-J, Duba H-C, Fletcher E, Kim CA, Krakow D, Morava E, **Neuhann T**, Superti-Furga A, Veenstra-Knol I, Wieczorek D, Wilson LC, Hennekam RCM, Sutherland-Smith AJ, Strom TM, Wilkie AOM, Brown MA, Duncan EL, Markie DM, Robertson SP. *Am J Hum Genet*. 2016;99(2):392-406. doi:10.1016/j.ajhg.2016.05.024

Mutations in EXOSC2 are associated with a novel syndrome characterised by retinitis pigmentosa, progressive hearing loss, premature ageing, short stature, mild intellectual disability and distinctive gestalt. Di Donato N, **Neuhann T**, Kahlert A-K, Klink B, Hackmann K, Neuhann I, Novotna B, Schallner J, Krause C, Glass IA, Parnell SE, Benet-Pages A, Nissen AM, Berger W, Altmüller J, Thiele H, Weber BHF, Schrock E, Dobyns WB, Bier A, Rump A. *J Med Genet*. 2016;53(6):419-425. doi:10.1136/jmedgenet-2015-103511

The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. Suriyanarayanan S, Auranen M, Toppila J, Paetau A, Shcherbii M, Palin E, Wei Y, Lohioja T, Schlotter-Weigel B**, Schön U,** Abicht A, Rautenstrauss B, Tyynismaa H, Walter MC, Hornemann T, Ylikallio E. *Neuromolecular Med*. 2016;18(1):81-90. doi:10.1007/s12017-015-8379-1

Novel homozygous RARS2 mutation in two siblings without pontocerebellar hypoplasia - further expansion of the phenotypic spectrum. Lühl S, Bode H, Schlötzer W, Bartsakoulia M, Horvath R, **Abicht A**, Stenzel M, Kirschner J, Grünert SC. *Orphanet J Rare Dis*. 2016;11(1):140. https://doi.org/10.1186/s13023-016-0525-9

Identification of mutations in the MYO9A gene in patients with congenital myasthenic syndrome. O'Connor E, Töpf A, Müller JS, Cox D, Evangelista T, Colomer J, **Abicht A**, Senderek J, Hasselmann O, Yaramis A, Laval SH, Lochmüller H. *Brain*. 2016;139(Pt 8):2143-2153. https://doi.org/10.1093/brain/aww130

Thomsen myotonia--A 4-generation family with a new mutation and a mild phenotype. Derevenciuc A-I, **Abicht A**, Hamza S, Roth C, Ferbert A. *Muscle Nerve*. 2016;53(4):653-654. https://doi.org/10.1002/mus.24971

The Variant p.(Arg183Trp) in SPTLC2 Causes Late-Onset Hereditary Sensory Neuropathy. Suriyanarayanan S, Auranen M, Toppila J, Paetau A, Shcherbii M, Palin E, Wei Y, Lohioja T, Schlotter-Weigel B, Schön U, **Abicht A**, Rautenstrauss B, Tyynismaa H, Walter MC, Hornemann T, Ylikallio E. *Neuromolecular Med*. 2016;18(1):81-90. https://doi.org/10.1007/s12017-015-8379-1

KLHL40-related nemaline myopathy with a sustained, positive response to treatment with acetylcholinesterase inhibitors. Natera-de Benito D, Nascimento A, **Abicht A**, Ortez C, Jou C, Müller JS, Evangelista T, Töpf A, Thompson R, Jimenez-Mallebrera C, Colomer J, Lochmüller H. *J Neurol*. 2016;263(3):517-523. https://doi.org/10.1007/s00415-015-8015-x

Absence of the Autophagy Adaptor SQSTM1/p62 Causes Childhood-Onset Neurodegeneration with Ataxia, Dystonia, and Gaze Palsy. Haack TB, Ignatius E, Calvo-Garrido J, Iuso A, Isohanni P, Maffezzini C, Lönnqvist T, Suomalainen A, Gorza M, Kremer LS, Graf E, Hartig M, Berutti R, Paucar M, Svenningsson P, Stranneheim H, Brandberg G, Wedell A, Kurian MA, Hayflick SA, Venco P, Tiranti V, Strom TM, Dichgans M, Horvath R, **Holinski-Feder E**, Freyer C, Meitinger T, Prokisch H, Senderek J, Wredenberg A, Carroll CJ, Klopstock T. Am J Hum Genet. 2016;99(3):735-743. https://doi.org/10.1016/j.ajhg.2016.06.026

Identification and Functional Testing of ERCC2 Mutations in a Multi-national Cohort of Patients with Familial Breast- and Ovarian Cancer. Rump A, Benet-Pages A, Schubert S, Kuhlmann JD, Janavičius R, Macháčková E, Foretová L, Kleibl Z, Lhota F, Zemankova P, Betcheva-Krajcir E, Mackenroth L, Hackmann K, Lehmann J, Nissen A, DiDonato N, Opitz R, Thiele H, Kast K, Wimberger P, **Holinski-Feder E**, Emmert S, Schröck E, Klink B. PLoS Genet. 2016;12(8):e1006248. https://doi.org/10.1371/journal.pgen.1006248

Quantitative Analysis of Human Pluripotency and Neural Specification by In-Depth (Phospho)Proteomic Profiling. Singec I, Crain AM, Hou J, Tobe BTD, Talantova M, Winquist AA, Doctor KS, Choy J, Huang X, La Monaca E, Horn DM, **Wolf DA**, Lipton SA, Gutierrez GJ, Brill LM, Snyder EY. *Stem Cell Reports*. 2016;7(3):527-542. doi:10.1016/j.stemcr.2016.07.019

Publikationsliste

A Transcript-Specific eIF3 Complex Mediates Global Translational Control of Energy Metabolism. Shah M, Su D, Scheliga JS, Pluskal T, Boronat S, Motamedchaboki K, Campos AR, Qi F, Hidalgo E, Yanagida M, **Wolf DA**. *Cell Reports*. 2016;16(7):1891-1902. doi:10.1016/j.celrep.2016.07.006

2015

Genetische Untersuchungen in der Reproduktionsmedizin. Kleinle S, **Koehler U**, Gonzalez Fassreiner D, Holinski-Feder E. *Journal für Reproduktionsmedizin und Endokrinologie* 2015 (2): 57-64. doi: https://www.kup.at/kup/pdf/12848.pdf

Phosphatidylinositol 3-kinase (PI3K) signalling regulates insulin-like-growth factor binding protein-2 (IGFBP-2) production in human adipocytes. Wilhelm F, Kässner F, Schmid G, Kratzsch J, **Laner A**, Wabitsch M, Körner A, Kiess W, Garten A. *Growth Horm IGF Res*. 2015;25(3):115-120. doi:10.1016/j.ghir.2015.03.003

Genome-wide CNV analysis in 221 unrelated patients and targeted high-throughput sequencing reveal novel causative candidate genes for colorectal adenomatous polyposis. Horpaopan S, Spier I, Zink AM, Altmüller J, Holzapfel S, **Laner A**, Vogt S, Uhlhaas S, Heilmann S, Stienen D, Pasternack SM, Keppler K, Adam R, Kayser K, Moebus S, Draaken M, Degenhardt F, Engels H, Hofmann A, Nöthen MM, Steinke V, Perez-Bouza A, Herms S, Holinski-Feder E, Fröhlich H, Thiele H, Hoffmann P, Aretz S. *Int J Cancer*. 2015;136(6):E578-589. doi:10.1002/ijc.29215

Elafin Reverses Pulmonary Hypertension via Caveolin-1-Dependent Bone Morphogenetic Protein Signaling. Nickel NP, Spiekerkoetter E, Gu M, Li CG, Li H, Kaschwich M, **Diebold I**, Hennigs JK, Kim K-Y, Miyagawa K, Wang L, Cao A, Sa S, Jiang X, Stockstill RW, Nicolls MR, Zamanian RT, Bland RD, Rabinovitch M. *Am J Respir Crit Care Med*. 2015;191(11):1273-1286. doi:10.1164/rccm.201412-2291OC

BMPR2 preserves mitochondrial function and DNA during reoxygenation to promote endothelial cell survival and reverse pulmonary hypertension. Diebold I, Hennigs JK, Miyagawa K, Li CG, Nickel NP, Kaschwich M, Cao A, Wang L, Reddy S, Chen P-I, Nakahira K, Alcazar MAA, Hopper RK, Ji L, Feldman BJ, Rabinovitch M. *Cell Metab*. ;21(4):596-608. doi:10.1016/j.cmet.2015.03.010

Early-onset leukoencephalopathy due to a homozygous missense mutation in the DARS2 gene. Köhler C, Heyer C, Hoffjan S, Stemmler S, Lücke T, Thiels C, Kohlschütter A, Löbel U, Horvath R, **Kleinle S**, Benet-Pages A, Abicht A. *Mol Cell Probes*. 2015;29(5):319-322. doi:10.1016/j.mcp.2015.06.005

Whole exome sequencing and the clinician: we need clinical skills and functional validation in variant filtering. Daud D, Griffin H, Douroudis K, **Kleinle S**, Eglon G, Pyle A, Chinnery PF, Horvath R. *J Neurol*. 2015;262(7):1673-1677. doi:10.1007/s00415-015-7755-y

Respiratory chain deficiency in nonmitochondrial disease. Pyle A, Nightingale HJ, Griffin H, Abicht A, Kirschner J, Baric I, Cuk M, Douroudis K, Feder L, Kratz M, Czermin B, **Kleinle S**, Santibanez-Koref M, Karcagi V, Holinski-Feder E, Chinnery PF, Horvath R. *Neurol Genet*. 2015;1(1):e6. doi:10.1212/NXG.0000000000000006

ATP synthase deficiency due to TMEM70 mutation leads to ultrastructural mitochondrial degeneration and is amenable to treatment. Braczynski AK, Vlaho S, Müller K, Wittig I, Blank A-E, Tews DS, Drott U, **Kleinle S**, Abicht A, Horvath R, Plate KH, Stenzel W, Goebel HH, Schulze A, Harter PN, Kieslich M, Mittelbronn M. *Biomed Res Int*. 2015;2015:462592. doi:10.1155/2015/462592

ADAMTSL4-associated isolated ectopia lentis: Further patients, novel mutations and a detailed phenotype description. Neuhann TM, Stegerer A, Riess A, Blair E, Martin T, Wieser S, Kläs R, Bouman A, Kuechler A, Rittinger O. *Am J Med Genet A*. 2015;167A(10):2376-2381. doi:10.1002/ajmg.a.37157
Hereditary ectopia lentis. Neuhann TM. *Klin Monbl Augenheilkd*. 2015;232(3):259-265. doi:10.1055/s-0034-1383330

Frequency and phenotypic spectrum of germline mutations in *POLE* and seven other polymerase genes in 266 patients with colorectal adenomas and carcinomas: *POLE* Mutations in Polyposis. Spier I, Holzapfel S, Altmüller J, Zhao B, Horpaopan S, Vogt S, Chen S, Morak M, Raeder S, Kayser K, Stienen D, Adam R, Nürnberg P, Plotz G, Holinski-Feder E, Lifton RP, Thiele H, Hoffmann P, **Steinke V**, Aretz S. *Int J Cancer*. 2015;137(2):320-331. doi:10.1002/ijc.29396

Functional testing strategy for coding genetic variants of unclear significance in MLH1 in Lynch syndrome diagnosis. Hinrichsen I, Schafer D, Langer D, Koger N, Wittmann M, Aretz S, **Steinke V**, Holzapfel S, Trojan J, Konig R, Zeuzem S, Brieger A, Plotz G. *Carcinogenesis*. 2015;36(2):202-211. doi:10.1093/carcin/bgu239

Publikationsliste

47 patients with FLNA associated periventricular nodular heterotopia. Lange M, Kasper B, Bohring A, Rutsch F, Kluger G, Hoffjan S, Spranger S, **Behnecke A**, Ferbert A, Hahn A, Oehl-Jaschkowitz B, Graul-Neumann L, Diepold K, Schreyer I, Bernhard MK, Mueller F, Siebers-Renelt U, Beleza-Meireles A, Uyanik G, Janssens S, Boltshauser E, Winkler J, Schuierer G, Hehr U. *Orphanet J Rare Dis*. 2015;10:134. doi:10.1186/s13023-015-0331-9

Genetische Syndrome mit Seh- und Hörbehinderungen. Moog U, Burau K, **Behnecke A**, Rohrschneider K. In: *Med Men Geist Mehrf Beh*. Vol 12. ; 2015:15-23.

Long-term follow-up in patients with congenital myasthenic syndrome due to RAPSN mutations. Natera-de Benito D, Bestué M, Vilchez JJ, Evangelista T, Töpf A, García-Ribes A, Trujillo-Tiebas MJ, García-Hoyos M, Ortez C, Camacho A, Jiménez E, Dusl M, **Abicht A**, Lochmüller H, Colomer J, Nascimento A. *Neuromuscul Disord*. 2016;26(2):153-159. https://doi.org/10.1016/j.nmd.2015.10.013

A 3'-UTR mutation creates a microRNA target site in the GFPT1 gene of patients with congenital myasthenic syndrome. Dusl M, Senderek J, Müller JS, Vogel JG, Pertl A, Stucka R, Lochmüller H, David R, **Abicht A**. *Hum Mol Genet*. 2015;24(12):3418-3426. https://doi.org/10.1093/hmg/ddv090

A de novo Mutation in the SCN4A Gene Causing Sodium Channel Myotonia. Ørstavik K, Wallace SC, Torbergsen T, **Abicht A**, Erik Tangsrud S, Kerty E, Rasmussen M. *J Neuromuscul Dis*. 2015;2(2):181-184. https://doi.org/10.3233/JND-150069

Economic evaluation of genetic screening for Lynch syndrome in Germany. Severin F, Stollenwerk B, **Holinski-Feder E**, Meyer E, Heinemann V, Giessen-Jung C, Rogowski W. Genet Med. 2015;17(10):765-773. https://doi.org/10.1038/gim.2014.190

Identification and characterization of a novel ISG15-ubiquitin mixed chain and its role in regulating protein homeostasis. Fan J-B, Arimoto K, Motamedchaboki K, Yan M, **Wolf DA**, Zhang D-E. *Sci Rep*. 2015;5(1):12704. doi:10.1038/srep12704

Downregulation of c-SRC kinase CSK promotes castration resistant prostate cancer and pinpoints a novel disease subclass. Yang C-C, Fazli L, Loguercio S, Zharkikh I, Aza-Blanc P, Gleave ME, **Wolf DA**. *Oncotarget*. 2015;6(26):22060-22071. doi:10.18632/oncotarget.4279

2014

Clinical utility gene card for: Williams-Beuren Syndrome [7q11.23]. Koehler U, Pabst B, Pober B, Kozel B. *Eur J Hum Genet*. 2014;22(9). doi:10.1038/ejhg.2014.28

Studie zur klinischen Etablierung der Präimplantationsdiagnostik. Seifert B, Paulmann B, Seifert D, Brey S, Gaßner C, Kwiatkowski B, Schön U, **Koehler U**, Holinski-Feder E. *Journal für Reproduktionsmedizin und Endokrinologie* 2014; 11(1):12-17. Doi:https://www.kup.at/kup/pdf/12122.pdf

Molecular and clinical analyses of 16q24.1 duplications involving FOXF1 identify an evolutionarily unstable large minisatellite. Dharmadhikari AV, Gambin T, Szafranski P, Cao W, Probst FJ, Jin W, Fang P, Gogolewski K, Gambin A, George-Abraham JK, Golla S, Boidein F, Duban-Bedu B, Delobel B, Andrieux J, Becker K, Holinski-Feder E, Cheung SW, Stankiewicz P. *BMC Med Genet*. 2014;15(1):128. doi:10.1186/s12881-014-0128-z

Biallelic MUTYH mutations can mimic Lynch syndrome. Morak M, Heidenreich B, Keller G, Hampel H, **Laner A**, de la Chapelle A, Holinski-Feder E. *Eur J Hum Genet*. 2014;22(11):1334-1337. doi:10.1038/ejhg.2014.15

Sirolimus treatment of severe PTEN hamartoma tumor syndrome: case report and in vitro studies. Schmid GL, Kässner F, Uhlig HH, Körner A, Kratzsch J, Händel N, Zepp F-P, Kowalzik F, **Laner A**, Starke S, Wilhelm FK, Schuster S, Viehweger A, Hirsch W, Kiess W, Garten A. *Pediatr Res*. 2014;75(4):527-534. doi:10.1038/pr.2013.246

Unexplained infertility: increased risk for 21-hydroxylase-deficiency in parents and offspring? Baumgartner-Parzer S, Vytiska-Binsdorfer E, **Kleinle S**, Heinze G, Vierhapper H. *Eur J Obstet Gynecol Reprod Biol*. 2014;182:258-259. doi:10.1016/j.ejogrb.2014.09.014

Use of whole-exome sequencing to determine the genetic basis of multiple mitochondrial respiratory chain complex deficiencies. Taylor RW, Pyle A, Griffin H, Blakely EL, Duff J, He L, Smertenko T, Alston CL, Neeve VC, Best A, Yarham JW, Kirschner J, Schara U, Talim B, Topaloglu H, Baric I, Holinski-Feder E, Abicht A, Czermin B, **Kleinle S**, Morris AAM, Vassallo G, Gorman GS, Ramesh V, Turnbull DM, Santibanez-Koref M, McFarland R, Horvath R, Chinnery PF. *JAMA*. 2014;312(1):68-77. doi:10.1001/jama.2014.7184

Hereditäre Optikusatrophien. Neuhann TM, Rautenstrauss B. *Medizinische Genetik*. 2014;26(1)

Publikationsliste

Germline variants in the SEMA4A gene predispose to familial colorectal cancer type X. Schulz E, Klampfl P, Holzapfel S, Janecke AR, Ulz P, Renner W, Kashofer K, Nojima S, Leitner A, Zebisch A, Wölfler A, Hofer S, Gerger A, Lax S, Beham-Schmid C, **Steinke V**, Heitzer E, Geigl JB, Windpassinger C, Hoefler G, Speicher MR, Richard Boland C, Kumanogoh A, Sill H. *Nat Commun*. 2014;5(1):5191. doi:10.1038/ncomms6191

Evaluating the performance of clinical criteria for predicting mismatch repair gene mutations in Lynch syndrome: A comprehensive analysis of 3,671 families: Performance of clinical criteria for Lynch syndrome. **Steinke V**, Holzapfel S, Loeffler M, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Pox C, Royer-Pokora B, von Knebel-Doeberitz M, Büttner R, Propping P, Engel C, on behalf of the German HNPCC Consortium. *Int J Cancer*. 2014;135(1):69-77. doi:10.1002/ijc.28650

Genome-wide analysis associates familial colorectal cancer with increases in copy number variations and a rare structural variation at 12p12.3. Yang R, Chen B, Pfutze K, Buch S, **Steinke V**, Holinski-Feder E, Stocker S, von Schonfels W, Becker T, Schackert HK, Royer-Pokora B, Kloor M, Schmiegel WH, Buttner R, Engel C, Lascorz Puertolas J, Forsti A, Kunkel N, Bugert P, Schreiber S, Krawczak M, Schafmayer C, Propping P, Hampe J, Hemminki K, Burwinkel B. *Carcinogenesis*. 2014;35(2):315-323. doi:10.1093/carcin/bgt344

Reduced migration of MLH1 deficient colon cancer cells depends on SPTAN1. Hinrichsen I, Ernst B, Nuber F, Passmann S, Schäfer D, **Steinke V**, Friedrichs N, Plotz G, Zeuzem S, Brieger A. *Mol Cancer*. 2014;13(1):11. doi:10.1186/1476-4598-13-11

Studie zur klinischen Etablierung der Präimplantationsdiagnostik. Seifert B, Paulmann B, Seifert D, Brey S, Gaßner C, Kwiatkowski B, **Schön U**, Koehler U, Holinski-Feder E *J Reproduktionsmed Endokrinol* 2014;

A blinded international study on the reliability of genetic testing for GGGGCC-repeat expansions in C9orf72 reveals marked differences in results among 14 laboratories. Akimoto C, Volk AE, van Blitterswijk M, Van den Broeck M, Leblond CS, Lumbroso S, Camu W, **Neitzel B**, Onodera O, van Rheenen W, Pinto S, Weber M, Smith B, Proven M, Talbot K, Keagle P, Chesi A, Ratti A, van der Zee J, Alstermark H, Birve A, Calini D, Nordin A, Tradowsky DC, Just W, Daoud H, Angerbauer S, DeJesus-Hernandez M, Konno T, Lloyd-Jani A, de Carvalho M, Mouzat K, Landers JE, Veldink JH, Silani V, Gitler AD, Shaw CE, Rouleau GA, van den Berg LH, Van Broeckhoven C, Rademakers R, Andersen PM, Kubisch C. *J Med Genet*. 2014;51(6):419-424. doi:10.1136/jmedgenet-2014-102360

The β3-integrin binding protein β3-endonexin is a novel negative regulator of hypoxia-inducible factor-1. Kračun D, Riess F, Kanchev I, Gawaz M, Görlach A. *Antioxid Redox Signal*. 2014;20(13):1964-1976. doi:10.1089/ars.2013.5286

ATP synthase deficiency due to TMEM70 mutation leads to ultrastructural mitochondrial degeneration and is amenable to treatment. Braczynski AK, Vlaho S, Müller K, Wittig I, Blank A-E, Tews DS, Drott U, Kleinle S, **Abicht A**, Horvath R, Plate KH, Stenzel W, Goebel HH, Schulze A, Harter PN, Kieslich M, Mittelbronn M. *Biomed Res Int*. 2015;2015:462592. https://doi.org/10.1155/2015/462592

Agrin mutations lead to a congenital myasthenic syndrome with distal muscle weakness and atrophy. Nicole S, Chaouch A, Torbergsen T, Bauché S, de Bruyckere E, Fontenille M-J, Horn MA, van Ghelue M, Løseth S, Issop Y, Cox D, Müller JS, Evangelista T, Stålberg E, Ioos C, Barois A, Brochier G, Sternberg D, Fournier E, Hantaï D, **Abicht A**, Dusl M, Laval SH, Griffin H, Eymard B, Lochmüller H. *Brain*. 2014;137(Pt 9):2429-2443. https://doi.org/10.1093/brain/awu160

Novel ETFDH mutation and imaging findings in an adult with glutaric aciduria type II. Rosenbohm A, Süssmuth SD, Kassubek J, Müller H-P, Pontes C, **Abicht A**, Bulst S, Ludolph AC, Pinkhardt E. *Muscle Nerve*. 2014;49(3):446-450. https://doi.org/10.1002/mus.23979

Congenital myasthenic syndrome due to choline acetyltransferase mutations in infants: clinical suspicion and comprehensive electrophysiological assessment are important for early diagnosis. Dilena R, **Abicht A**, Sergi P, Comi GP, Di Fonzo A, Chidini G, Natacci F, Barbieri S, Lochmüller H. *J Child Neurol*. 2014;29(3):389-393. https://doi.org/10.1177/0883073812470000

Novel TPM3 mutation in a family with cap myopathy and review of the literature. Schreckenbach T, Schröder JM, Voit T, **Abicht A**, Neuen-Jacob E, Roos A, Bulst S, Kuhl C, Schulz JB, Weis J, Claeys KG. *Neuromuscul Disord*. 2014;24(2):117-124. https://doi.org/10.1016/j.nmd.2013.10.002

Salbutamol-responsive limb-girdle congenital myasthenic syndrome due to a novel missense mutation and heteroallelic deletion in MUSK. Gallenmüller C, Müller-Felber W, Dusl M, Stucka R, Guergueltcheva V, Blaschek A, von der Hagen M, Huebner A, Müller JS, Lochmüller H, **Abicht A**. *Neuromuscul Disord*. 2014;24(1):31-35. https://doi.org/10.1016/j.nmd.2013.08.002

Publikationsliste

Mutations in the Mitochondrial Citrate Carrier SLC25A1 are Associated with Impaired Neuromuscular Transmission. Chaouch A, Porcelli V, Cox D, Edvardson S, Scarcia P, De Grassi A, Pierri CL, Cossins J, Laval SH, Griffin H, Müller JS, Evangelista T, Töpf A, **Abicht A**, Huebner A, von der Hagen M, Bushby K, Straub V, Horvath R, Elpeleg O, Palace J, Senderek J, Beeson D, Palmieri L, Lochmüller H. *J Neuromuscul Dis*. 2014;1(1):75-90. https://doi.org/10.3233/JND-140021

A pooled analysis of the outcome of prospective colonoscopic surveillance for familial colorectal cancer. Mesher D, Dove-Edwin I, Sasieni P, Vasen H, Bernstein I, Royer-Pokora B, Holinski-Feder E, Lalloo F, Evans DG, Forsberg A, Lindblom A, Thomas H. Int J Cancer. 2014;134(4):939-947. https://doi.org/10.1002/ijc.28397

Application of a 5-tiered scheme for standardized classification of 2,360 unique mismatch repair gene variants in the InSiGHT locus-specific database. Thompson BA, Spurdle AB, Plazzer J-P, Greenblatt MS, Akagi K, Al-Mulla F, Bapat B, Bernstein I, Capellá G, den Dunnen JT, du Sart D, Fabre A, Farrell MP, Farrington SM, Frayling IM, Frebourg T, Goldgar DE, Heinen CD, **Holinski-Feder E**, Kohonen-Corish M, Robinson KL, Leung SY, Martins A, Moller P, Morak M, Nystrom M, Peltomaki P, Pineda M, Qi M, Ramesar R, Rasmussen LJ, Royer-Pokora B, Scott RJ, Sijmons R, Tavtigian SV, Tops CM, Weber T, Wijnen J, Woods MO, Macrae F, Genuardi M. Nat Genet. 2014;46(2):107-115. https://doi.org/10.1038/ng.2854

Is Reliance on Mitochondrial Respiration a "Chink in the Armor" of Therapy-Resistant Cancer? Wolf DA. *Cancer Cell*. 2014;26(6):788-795. doi:10.1016/j.ccell.2014.10.001

Systems analysis of the prostate tumor suppressor NKX3.1 supports roles in DNA repair and luminal cell differentiation. Yang C-C, Chung A, Ku C-Y, Brill LM, Williams R, **Wolf DA**. *F1000Res*. 2014;3:115. doi:10.12688/f1000research.3818.1

2013

X-linked intellectual disability type Nascimento is a clinically distinct, probably underdiagnosed entity. Czeschik JC, Bauer P, Buiting K, Dufke C, Guillén-Navarro E, Johnson DS, **Koehler U**, López-González V, Lüdecke H-J, Male A, Morrogh D, Rieß A, Tzschach A, Wieczorek D, Kuechler A. *Orphanet J Rare Dis*. 2013;8:146. doi:10.1186/1750-1172-8-146

The phenotypic spectrum of duplication 5q35.2-q35.3 encompassing NSD1: is it really a reversed Sotos syndrome? Dikow N, Maas B, Gaspar H, Kreiss-Nachtsheim M, Engels H, Kuechler A, Garbes L, Netzer C, Neuhann TM, **Koehler U**, Casteels K, Devriendt K, Janssen JWG, Jauch A, Hinderhofer K, Moog U. *Am J Med Genet A*. 2013;161A(9):2158-2166. doi:10.1002/ajmg.a.36046

Pregnancy and Birth After a Two-Step PGD: Polar Body Diagnosis for Hemophilia A and Array CGH on Trophectoderm Cells for Chromosomal Aberrations. Würfel W, Suttner R, Shakeshaft D, Mayer V, Schoen U, Sendelbach K, Locher M, **Koehler U**, Fiedler K, Krüsmann G, Holinski-Feder E. *Geburtshilfe Frauenheilkd*. 2013;73(8):812-814. doi:10.1055/s-0033-1350705

Generalized epilepsy in two patients with 5p duplication. Kluger G, **Koehler U**, Neuhann TM, Pieper T, Staudt M, von Stülpnagel C. *Neuropediatrics*. 2013;44(4):225-229. doi:10.1055/s-0033-1333872

Molecular genetic analysis of a patient with moderate hemophilia A and psychomotor developmental delay. Gothwal M, Nakamura L, Hainmann I, **Koehler U**, Schilling F, Rost S, Oldenburg J, Zieger B. *Klin Padiatr*. 2013;225(3):175-176. doi:10.1055/s-0033-1341490

CD133 is a predictor of poor survival in head and neck squamous cell carcinomas. Canis M, Lechner A, Mack B, Zengel P, Laubender RP, **Koehler U**, Heissmeyer V, Gires O. *CBM*. 2013;12(2):97-105. doi:10.3233/CBM-130297

Preimplantation Genetic Diagnosis for Monogenic Disorders and Chromosomal Rearrangements – the German Perspective. Koehler U., Schoen U., Mayer V., Holinski-Feder. E. *Journal für Reproduktionsmedizin und Endokrinologie* 2013; Special Issue 1:36-42. doi:https://www.kup.at/kup/pdf/11278.pdf

Mutations in genes encoding the cadherin receptor-ligand pair DCHS1 and FAT4 disrupt cerebral cortical development. Cappello S, Gray MJ, Badouel C, Lange S, Einsiedler M, Srour M, Chitayat D, Hamdan FF, Jenkins ZA, Morgan T, Preitner N, Uster T, Thomas J, Shannon P, Morrison V, Di Donato N, Van Maldergem L, **Neuhann T**, Newbury-Ecob R, Swinkells M, Terhal P, Wilson LC, Zwijnenburg PJG, Sutherland-Smith AJ, Black MA, Markie D, Michaud JL, Simpson MA, Mansour S, McNeill H, Götz M, Robertson SP. *Nat Genet*. 2013;45(11):1300-1308. doi:10.1038/ng.2765

The White man's burden - a case study caught between bipolar affective disorder and Huntington's disease. Nowidi K, Kunisch R, Bouna-Pyrrou P, Meißner D, Hennig-Fast K, Weindl A, Förster S, **Neuhann TM**, Falkai P, Berger M, Musil R. *Fortschr Neurol Psychiatr*. 2013;81(6):337-345. doi:10.1055/s-0033-1335040

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

A novel germline KIT mutation (p.L576P) in a family presenting with juvenile onset of multiple gastrointestinal stromal tumors, skin hyperpigmentations, and esophageal stenosis. Neuhann TM, Mansmann V, Merkelbach-Bruse S, Klink B, Hellinger A, Höffkes H-G, Wardelmann E, Schildhaus H-U, Tinschert S. *Am J Surg Pathol*. 2013;37(6):898-905. doi:10.1097/PAS.0b013e31827bc071

Combined NGS approaches identify mutations in the intraflagellar transport gene IFT140 in skeletal ciliopathies with early progressive kidney Disease. Schmidts M, Frank V, Eisenberger T, Al Turki S, Bizet AA, Antony D, Rix S, Decker C, Bachmann N, Bald M, Vinke T, Toenshoff B, Di Donato N, **Neuhann T**, Hartley JL, Maher ER, Bogdanović R, Peco-Antić A, Mache C, Hurles ME, Joksić I, Guć-Šćekić M, Dobricic J, Brankovic-Magic M, Bolz HJ, Pazour GJ, Beales PL, Scambler PJ, Saunier S, Mitchison HM, Bergmann C. *Hum Mutat*. 2013;34(5):714-724. doi:10.1002/humu.22294

Genetic and phenotypic variability of optic neuropathies. Neuhann T, Rautenstrauss B. *Expert Rev Neurother*. 2013;13(4):357-367. doi:10.1586/ern.13.19

Role of the Oxidative DNA Damage Repair Gene OGG1 in Colorectal Tumorigenesis. Smith CG, West H, Harris R, Idziaszczyk S, Maughan TS, Kaplan R, Richman S, Quirke P, Seymour M, Moskvina V, **Steinke V**, Propping P, Hes FJ, Wijnen J, Cheadle JP. *JNCI: Journal of the National Cancer Institute*. 2013;105(16):1249-1253. doi:10.1093/jnci/djt183

Hereditary Nonpolyposis Colorectal Cancer (HNPCC)/Lynch Syndrome. **Steinke V**, Engel C, Büttner R, Schackert HK, Schmiegel WH, Propping P. *Deutsches Aerzteblatt Online*. Published online January 18, 2013. doi:10.3238/arztebl.2013.0032

Delayed primary diagnosis of LEOPARD syndrome type 1. Staub J, **Behnecke A**, Leverkus M. *J Am Acad Dermatol*. 2013;68(2):e58-60. doi:10.1016/j.jaad.2012.08.037

Novel CACNA1A mutation(s) associated with slow saccade velocities. Kipfer S, Jung S, Lemke JR, Kipfer-Kauer A, Howell JP, Kaelin-Lang A, Nyffeler T, Gutbrod K, **Abicht A**, Müri RM. *J Neurol*. 2013;260(12):3010-3014. https://doi.org/10.1007/s00415-013-7099-4

Quinine sulfate as a therapeutic option in a patient with slow channel congenital myasthenic syndrome. Peyer A-K, **Abicht A**, Heinimann K, Sinnreich M, Fischer D. *Neuromuscul Disord*. 2013;23(7):571-574. https://doi.org/10.1016/j.nmd.2013.04.001

DOK7 limb-girdle myasthenic syndrome mimicking congenital muscular dystrophy. Mahjneh I, Lochmüller H, Muntoni F, **Abicht A**. *Neuromuscul Disord*. 2013;23(1):36-42. https://doi.org/10.1016/j.nmd.2012.06.355

NDUFS8-related Complex I Deficiency Extends Phenotype from "PEO Plus" to Leigh Syndrome. Marina AD, Schara U, Pyle A, Möller-Hartmann C, Holinski-Feder E, **Abicht A**, Czermin B, Lochmüller H, Griffin H, Santibanez-Koref M, Chinnery PF, Horvath R. *JIMD Rep*. 2013;10:17-22. https://doi.org/10.1007/8904_2012_195

Case study of sporadic mitochondrial disease with myotonic discharges and optic atrophy. Soysal A, Yuksel B, **Czermin B**, Aydemir S, Tugcu B, Aysal F, Arpaci B. *Muscle Nerve*. 2013;47(2):308-309. doi:10.1002/mus.23641

Early muscle and brain ultrastructural changes in polymerase gamma 1-related encephalomyopathy. Nolte KW, Trepels-Kottek S, Honnef D, Weis J, Bien CG, van Baalen A, Ritter K, **Czermin B**, Rudnik-Schoneborn S, Wagner N, Hausler M. *Neuropathology*. 2013;33(1):59-67. doi:10.1111/j.1440-1789.2012.01317.x

Small molecule-induced mitochondrial disruption directs prostate cancer inhibition via UPR signaling. Rico-Bautista E, Zhu W, Kitada S, Ganapathy S, Lau E, Krajewski S, Ramirez J, Bush JA, Yuan Z, **Wolf DA**. *Oncotarget*. 2013;4(8):1212-1229. doi:10.18632/oncotarget.1130

CAND1 controls in vivo dynamics of the cullin 1-RING ubiquitin ligase repertoire. Wu S, Zhu W, Nhan T, Toth JI, Petroski MD, **Wolf DA**. *Nat Commun*. 2013;4(1):1642. doi:10.1038/ncomms2636

Relative impact of 3- and 5-hydroxyl groups of cytosporone B on cancer cell viability. Xia Z, Cao X, Rico-Bautista E, Yu J, Chen L, Chen J, Bobkov A, **Wolf DA**, Zhang X-K, Dawson MI. *Med Chem Commun*. 2013;4(2):332-339. doi:10.1039/C2MD20243C

Combined Total Proteomic and Phosphoproteomic Analysis of Human Pluripotent Stem Cells. Hou J, Tobe BTD, Lo F, Blethrow JD, Crain AM, **Wolf DA**, Snyder EY, Singec I, Brill LM. In: Zavazava N, ed. *Embryonic Stem Cell Immunobiology*. Vol 1029. Methods in Molecular Biology. Humana Press; 2013:163-189. doi:10.1007/978-1-62703-478-4_12

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

2012

NADPH oxidases as a source of oxidative stress and molecular target in ischemia/reperfusion injury. Kleikers PWM, Wingler K, Hermans JJR, **Diebold I**, Altenhöfer S, Radermacher KA, Janssen B, Görlach A, Schmidt HHHW. *J Mol Med (Berl)*. 2012;90(12):1391-1406. doi:10.1007/s00109-012-0963-3

The HIF1 target gene NOX2 promotes angiogenesis through urotensin-II. Diebold I, Petry A, Sabrane K, Djordjevic T, Hess J, Görlach A. *J Cell Sci*. 2012;125(Pt 4):956-964. doi:10.1242/jcs.094060

STEAP1 is associated with the invasive and oxidative stress phenotype of Ewing tumors. Grunewald TGP, **Diebold I**, Esposito I, Plehm S, Hauer K, Thiel U, da Silva-Buttkus P, Neff F, Unland R, Müller-Tidow C, Zobywalski C, Lohrig K, Lewandrowski U, Sickmann A, Prazeres da Costa O, Görlach A, Cossarizza A, Butt E, Richter GHS, Burdach S. *Mol Cancer Res*. 2012;10(1):52-65. doi:10.1158/1541-7786.MCR-11-0524

Germline truncating-mutations in BRCA1 and MSH6 in a patient with early onset endometrial cancer. Kast K, Neuhann TM, Görgens H, **Becker K**, Keller K, Klink B, Aust D, Distler W, Schröck E, Schackert HK. *BMC Cancer*. 2012;12:531. doi:10.1186/1471-2407-12-531

BRCA1/2 testing: uptake, phenocopies, and strategies to improve detection rates in initially negative families. Fischer C, Engel C, Sutter C, Zachariae S, Schmutzler R, Meindl A, Heidemann S, Grimm T, Goecke T, Debatin I, Horn D, Wieacker P, Gadzicki D, Becker K, Schäfer D, Stock F, Voigtländer T, on behalf of the German Consortium for Hereditary Breast and Ovarian Cancer. *Clinical Genetics*. 2012;82(5):478-483. doi:10.1111/j.1399-0004.2011.01788.x

De novo microdeletions of chromosome 6q14.1-q14.3 and 6q12.1-q14.1 in two patients with intellectual disability - further delineation of the 6q14 microdeletion syndrome and review of the literature. Becker K, Di Donato N, Holder-Espinasse M, Andrieux J, Cuisset J-M, Vallée L, Plessis G, Jean N, Delobel B, Thuresson A-C, Annerén G, Ravn K, Tümer Z, Tinschert S, Schrock E, Jønch AE, Hackmann K. *Eur J Med Genet*. 2012;55(8-9):490-497. doi:10.1016/j.ejmg.2012.03.003

First experiences with PGD after trophectoderm biopsy at Kinderwunsch Centrum Munich (KCM), Germany. Suttner R, Shakeshaft D, **Koehler U**, Schön U, Hararsim T, Wagner A, Holinski-Feder E, Rost I, Würfel W. *Reprod BioMed Online* 2012; 24:48.

Increased pregnancy rates after trophectoderm biopsy for PGD of monogenic diseases and chromosomal aberrrations. U. Schön, U. Koehler, V. Mayer, A. Stegerer, K. Sendelbach, M. Locher, B. Paulmann, D. Shakeshaft, R. Suttner, B. Seifert, W. Würfel, E. Holinski-Feder. *Reproductive BioMedicine Online* 2012; 24:55-56.

Steigerung der Schwangerschaftsrate durch Präimplantationsdiagnostik für monogene Erkrankungen und chromosomale Aberrationen nach Trophektodermbiopsie. Koehler U, Mayer V, Schön U, Stegerer A, Sendelbach K, Locher M, Paulmann B, Shakeshaft D, Suttner R, Seifert B, Würfel W, Holinski-Feder E. *Arch Gynecol Obstet* 2012; 286:49-279.

Preimplantation genetic diagnosis for monogenic diseases and chromosomal aberrations after trophectoderm biopsy- results from 2010 to 2011. Koehler U, Schön U, Mayer V, Stegerer A, Sendelbach K, Locher M, Paulmann B, Shakeshaft, D. Suttner R, Seifert B, Würfel W, Holinski-Feder E. *European Journal of Human Genetics* 2012; 20:140.

Cardiac malformation of partial trisomy 7p/monosomy 18p and partial trisomy 18p/monosomy 7p in siblings as a result of reciprocal unbalanced malsegregation--and review of the literature. Schmidt B, Udink ten Cate F, Weiss M, **Koehler U**. *Eur J Pediatr*. 2012;171(7):1047-1053. doi:10.1007/s00431-012-1682-z

Macrocerebellum: significance and pathogenic considerations. Poretti A, Mall V, Smitka M, Grunt S, Risen S, Toelle SP, Benson JE, Yoshida S, Jung NH, Tinschert S, **Neuhann TM**, Rauch A, Steinlin M, Meoded A, Huisman TAGM, Boltshauser E. *Cerebellum*. 2012;11(4):1026-1036. doi:10.1007/s12311-012-0379-1

Germline truncating-mutations in BRCA1 and MSH6 in a patient with early onset endometrial cancer. Kast K, **Neuhann TM**, Görgens H, Becker K, Keller K, Klink B, Aust D, Distler W, Schröck E, Schackert HK. *BMC Cancer*. 2012;12:531. doi:10.1186/1471-2407-12-531

A further patient with van Maldergem syndrome. Neuhann TM, Müller D, Hackmann K, Holzinger S, Schrock E, Di Donato N. *Eur J Med Genet*. 2012;55(6-7):423-428. doi:10.1016/j.ejmg.2012.02.012

Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2, EPCAM) - update 2012. Rahner N, **Steinke V**, Schlegelberger B, Eisinger F, Hutter P, Olschwang S. *Eur J Hum Genet*. 2013;21(1):118-118. doi:10.1038/ejhg.2012.164

Publikationsliste

Risks of Less Common Cancers in Proven Mutation Carriers With Lynch Syndrome. Engel C, Loeffler M, **Steinke V**, Rahner N, Holinski-Feder E, Dietmaier W, Schackert HK, Goergens H, von Knebel Doeberitz M, Goecke TO, Schmiegel W, Buettner R, Moeslein G, Letteboer TGW, García EG, Hes FJ, Hoogerbrugge N, Menko FH, van Os TAM, Sijmons RH, Wagner A, Kluijt I, Propping P, Vasen HFA. *JCO*. 2012;30(35):4409-4415. doi:10.1200/JCO.2012.43.2278

Mutation and association analyses of the candidate genes ESR1, ESR2, MAX, PCNA, and KAT2A in patients with unexplained MSH2-deficient tumors. The Dutch Cancer Genetics Group, Rahner N, Brockschmidt FF, **Steinke V**, Kahl P, Becker T, Vasen HFA, Wijnen JT, Tops CJM, Holinski-Feder E, Ligtenberg MJL, Spruijt L, Görgens H, Stemmler S, Kloor M, Dietmaier W, Schumacher J, Nöthen MM, Propping P. *Familial Cancer*. 2012;11(1):19-26.
doi:10.1007/s10689-011-9489-z

Silver-Russell syndrome due to maternal uniparental disomy 7 and a familial reciprocal translocation t(7;13). Behnecke A, Hinderhofer K, Jauch A, Janssen JWG, Moog U. *Clin Genet*. 2012;82(5):494-498. doi:10.1111/j.1399-0004.2011.01792.x

Molecular and clinical characterization of an in frame deletion of uncertain clinical significance in the BRCA2 gene. Rath MG, Fathali-Zadeh F, Langheinz A, Tchatchou S, Voigtländer T, Heil J, Golatta M, Schott S, Drasseck T, **Behnecke A**, Burgemeister A-L, Evers C, Bugert P, Junkermann H, Schneeweiss A, Bartram CR, Sohn C, Sutter C, Burwinkel B. *Breast Cancer Res Treat*. 2012;133(2):725-734. doi:10.1007/s10549-011-1917-0

How fingers and face can be the clue? Behnecke A, Dikow N, Moog U. *Eur J Neurol*. 2012;19(3):e27-28. doi:10.1111/j.1468-1331.2011.03622.x

Humangenetische Diagnostik und Beratung: Seltene Erkrankungen in der Pränatalmedizin. Moog U, Dikow N, Gaspar H, **Behnecke A**, Schenk JP, Flechtenmacher C, Beedgen B, Elsässer M. In: *Gynäkologe*. Vol 45. ; 2012:376–382.

A new phenotype of brain iron accumulation with dystonia, optic atrophy, and peripheral neuropathy. Horvath R, Holinski-Feder E, Neeve VCM, Pyle A, Griffin H, Ashok D, Foley C, Hudson G, Rautenstrauss B, Nürnberg G, Nürnberg P, Kortler J, **Neitzel B**, Bässmann I, Rahman T, Keavney B, Loughlin J, Hambleton S, Schoser B, Lochmüller H, Santibanez-Koref M, Chinnery PF. *Mov Disord*. 2012;27(6):789-793. doi:10.1002/mds.24980

Exercise-induced myalgia and rhabdomyolysis in a patient with the rare m.3243A>T mtDNA mutation. Czell D, **Abicht A**, Hench J, Weber M. *BMJ Case Rep*. 2012;2012. https://doi.org/10.1136/bcr-2012-006980

What is influencing the phenotype of the common homozygous polymerase-γ mutation p.Ala467Thr? Neeve VCM, Samuels DC, Bindoff LA, van den Bosch B, Van Goethem G, Smeets H, Lombès A, Jardel C, Hirano M, Dimauro S, De Vries M, Smeitink J, Smits BW, de Coo IFM, Saft C, Klopstock T, Keiling B-C, Czermin B, **Abicht A**, Lochmüller H, Hudson G, Gorman GG, Turnbull DM, Taylor RW, Holinski-Feder E, Chinnery PF, Horvath R. *Brain*. 2012;135(Pt 12):3614-3626. https://doi.org/10.1093/brain/aws298

Congenital myasthenic syndromes: achievements and limitations of phenotype-guided gene-after-gene sequencing in diagnostic practice: a study of 680 patients. Abicht A, Dusl M, Gallenmüller C, Guergueltcheva V, Schara U, Della Marina A, Wibbeler E, Almaras S, Mihaylova V, von der Hagen M, Huebner A, Chaouch A, Müller JS, Lochmüller H. *Hum Mutat*. 2012;33(10):1474-1484. https://doi.org/10.1002/humu.22130

In vitro supplementation with deoxynucleoside monophosphates rescues mitochondrial DNA depletion. Bulst S, Holinski-Feder E, Payne B, **Abicht A**, Krause S, Lochmüller H, Chinnery PF, Walter MC, Horvath R. *Mol Genet Metab*. 2012;107(1-2):95-103. https://doi.org/10.1016/j.ymgme.2012.04.022

Febrile infection-related epilepsy syndrome without detectable autoantibodies and response to immunotherapy: a case series and discussion of epileptogenesis in FIRES. van Baalen A, Häusler M, Plecko-Startinig B, Strautmanis J, Vlaho S, Gebhardt B, Rohr A, **Abicht A**, Kluger G, Stephani U, Probst C, Vincent A, Bien CG. *Neuropediatrics*. 2012;43(4):209-216. https://doi.org/10.1055/s-0032-1323848

Congenital myasthenic syndromes: current diagnostic and therapeutic approaches. Schara U, Della Marina A, **Abicht A**. *Neuropediatrics*. 2012;43(4):184-193. https://doi.org/10.1055/s-0032-1323850

Congenital myasthenic syndrome with tubular aggregates caused by GFPT1 mutations. Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizberea A, Soler DM, Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, **Abicht A**, Lochmüller H. *J Neurol*. 2012;259(5):838-850. https://doi.org/10.1007/s00415-011-6262-z

Publikationsliste

Classical MERRF phenotype associated with mitochondrial tRNA(Leu) (m.3243A>G) mutation. Brackmann F, **Abicht A**, Ahting U, Schröder R, Trollmann R. *Eur J Pediatr*. 2012;171(5):859-862. https://doi.org/10.1007/s00431-011-1662-8

A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. Chaouch A, Müller JS, Guergueltcheva V, Dusl M, Schara U, Rakocević-Stojanović V, Lindberg C, Scola RH, Werneck LC, Colomer J, Nascimento A, Vilchez JJ, Muelas N, Argov Z, **Abicht A**, Lochmüller H. *J Neurol*. 2012;259(3):474-481. https://doi.org/10.1007/s00415-011-6204-9

OPA1 mutations induce mtDNA proliferation in leukocytes of patients with dominant optic atrophy. Sitarz KS, Almind GJ, Horvath R, **Czermin B**, Gronskov K, Pyle A, Taylor RW, Larsen M, Chinnery PF, Yu-Wai-Man P. *Neurology*. 2012;79(14):1515-1517. doi:10.1212/WNL.0b013e31826d5f60

Adult-onset cerebellar ataxia due to mutations in CABC1/ADCK3. Horvath R, **Czermin B**, Gulati S, Demuth S, Houge G, Pyle A, Dineiger C, Blakely EL, Hassani A, Foley C, Brodhun M, Storm K, Kirschner J, Gorman GS, Lochmuller H, Holinski-Feder E, Taylor RW, Chinnery PF. *J Neurol Neurosurg Psychiatry*. 2012;83(2):174-178. doi:10.1136/jnnp-2011-301258

Deep intronic APC mutations explain a substantial proportion of patients with familial or early-onset adenomatous polyposis. Spier I, Horpaopan S, Vogt S, Uhlhaas S, Morak M, Stienen D, Draaken M, Ludwig M, **Holinski-Feder E**, Nöthen MM, Hoffmann P, Aretz S. Hum Mutat. 2012;33(7):1045-1050. https://doi.org/10.1002/humu.22082

[18F]Fluorodeoxyglucose positron emission tomography/computed tomography-positive gastric adenocarcinoma in a 12-year-old girl with Peutz-Jeghers syndrome. Schneider C, Simon T, Hero B, Uphoff US, Drebber U, Alakus H, **Holinski-Feder E**, Berthold F, Dietlein M, Schmidt MC. J Clin Oncol. 2012;30(14):e140-143. https://doi.org/10.1200/JCO.2011.39.7422

An American founder mutation in MLH1. Tomsic J, Liyanarachchi S, Hampel H, Morak M, Thomas BC, Raymond VM, Chittenden A, Schackert HK, Gruber SB, Syngal S, Viel A, **Holinski-Feder E**, Thibodeau SN, de la Chapelle A. Int J Cancer. 2012;130(9):2088-2095. https://doi.org/10.1002/ijc.26233

The prevalence of familial hyperaldosteronism in apparently sporadic primary aldosteronism in Germany: a single center experience. Pallauf A, Schirpenbach C, Zwermann O, Fischer E, Morak M, **Holinski-Feder E**, Hofbauer L, Beuschlein F, Reincke M. Horm Metab Res. 2012;44(3):215-220. https://doi.org/10.1055/s-0031-1299730

Skipping Cancer: Small Molecule Inhibitors of SKP2-Mediated p27 Degradation. Rico-Bautista E, **Wolf DA**. *Chemistry & Biology*. 2012;19(12):1497-1498. doi:10.1016/j.chembiol.2012.12.001

Translational Control of Cell Division by Elongator. Bauer F, Matsuyama A, Candiracci J, Dieu M, Scheliga J, **Wolf DA**, Yoshida M, Hermand D. *Cell Reports*. 2012;1(5):424-433. doi:10.1016/j.celrep.2012.04.001

Synthetic biology approach to reconstituting the ubiquitylation cascade in bacteria: Reconstituted ubiquitylation cascade in bacteria. Keren-Kaplan T, Attali I, Motamedchaboki K, Davis BA, Tanner N, Reshef Y, Laudon E, Kolot M, Levin-Kravets O, Kleifeld O, Glickman M, Horazdovsky BF, **Wolf DA**, Prag G. *The EMBO Journal*. 2012;31(2):378-390. doi:10.1038/emboj.2011.397

Regulation of transcriptome, translation, and proteome in response to environmental stress in fission yeast. Lackner DH, Schmidt MW, Wu S, **Wolf DA**, Bahler J. *Genome Biol*. 2012;13(4):R25. doi:10.1186/gb-2012-13-4-r25

2011

PID für monogene Erkrankungen nach Polkörper- und/oder Trophektodermbiopsie. Hehr A, Paulmann P, **Koehler U.,** Gassner C, Bals-Pratsch M, Holinski-Feder E, et al**. J Reproduktionsmed Endokrinol 2011; 8:319.**

Diagnostik der spinalen Muskelatrophie Typ 1 (SMA1) durch Trophektodermbiopsie von Blastozysten. Seifert B, Schön U, Paulmann B, Seifert D, Hehr A, **Koehler U**, Holinski-Feder E. J Reproduktionsmed Endokrinol 2011; 8:335.

Phenotypic spectrum associated with CASK loss-of-function mutations. Moog U, Kutsche K, Kortüm F, Chilian B, Bierhals T, Apeshiotis N, Balg S, Chassaing N, Coubes C, Das S, Engels H, Van Esch H, Grasshoff U, Heise M, Isidor B, Jarvis J, **Koehler U**, Martin T, Oehl-Jaschkowitz B, Ortibus E, Pilz DT, Prabhakar P, Rappold G, Rau I, Rettenberger G, Schlüter G, Scott RH, Shoukier M, Wohlleber E, Zirn B, Dobyns WB, Uyanik G. *J Med Genet*. 2011;48(11):741-751. doi:10.1136/jmedgenet-2011-100218

Publikationsliste

Biallelic MLH1 SNP cDNA expression or constitutional promoter methylation can hide genomic rearrangements causing Lynch syndrome. Morak M, **Koehler U**, Schackert HK, Steinke V, Royer-Pokora B, Schulmann K, Kloor M, Höchter W, Weingart J, Keiling C, Massdorf T, Holinski-Feder E. *J Med Genet*. 2011;48(8):513-519. doi:10.1136/jmedgenet-2011-100050

NOX4 mediates activation of FoxO3a and matrix metalloproteinase-2 expression by urotensin-II. Diebold I, Petry A, Burger M, Hess J, Görlach A. *Mol Biol Cell*. 2011;22(22):4424-4434. doi:10.1091/mbc.E10-12-0971

A new autosomal dominant syndrome of distinctive face showing ptosis and prominent eyes associated with cleft palate, ear anomalies, and learning disability. Tyshchenko N, **Neuhann TM**, Gerlach E, Hahn G, Heisch K, Rump A, Schrock E, Tinschert S, Hackmann K. *Am J Med Genet A*. 2011;155A(9):2060-2065. doi:10.1002/ajmg.a.34159

Expanding the clinical and neuroradiological phenotype of 6q27 microdeletion: olfactory bulb aplasia and anosmia. Gerber JC, **Neuhann TM**, Tyshchenko N, Smitka M, Hackmann K. *Am J Med Genet A*. 2011;155A(8):1981-1986. doi:10.1002/ajmg.a.34079

A homozygous microdeletion within ADAMTSL4 in patients with isolated ectopia lentis: evidence of a founder mutation. Neuhann TM, Artelt J, **Neuhann TF**, Tinschert S, Rump A. *Invest Ophthalmol Vis Sci*. 2011;52(2):695-700. doi:10.1167/iovs.10-5740

FHL2 expression in peritumoural fibroblasts correlates with lymphatic metastasis in sporadic but not in HNPCC-associated colon cancer. Gullotti L, Czerwitzki J, Kirfel J, Propping P, Rahner N, **Steinke V**, Kahl P, Engel C, Schüle R, Buettner R, Friedrichs N. *Lab Invest*. 2011;91(12):1695-1705. doi:10.1038/labinvest.2011.109

Association Between TAS2R38 Gene Polymorphisms and Colorectal Cancer Risk: A Case-Control Study in Two Independent Populations of Caucasian Origin. Carrai M, **Steinke V**, Vodicka P, Pardini B, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Stemmler S, Betz B, Kloor M, Engel C, Büttner R, Naccarati A, Vodickova L, Novotny J, Stein A, Hemminki K, Propping P, Försti A, Canzian F, Barale R, Campa D. Song Y, ed. *PLoS ONE*. 2011;6(6):e20464. doi:10.1371/journal.pone.0020464

Missense variants in hMLH1 identified in patients from the German HNPCC consortium and functional studies. Peter Propping and the German HNPCC consortium, Hardt K, Heick SB, Betz B, Goecke T, Yazdanparast H, Küppers R, Servan K, **Steinke V**, Rahner N, Morak M, Holinski-Feder E, Engel C, Möslein G, Schackert H-K, von Knebel Doeberitz M, Pox C, Hegemann JH, Royer-Pokora B. *Familial Cancer*. 2011;10(2):273-284. doi:10.1007/s10689-011-9431-4

Recurrence and variability of germline *EPCAM* deletions in Lynch syndrome. Kuiper RP, Vissers LELM, Venkatachalam R, Bodmer D, Hoenselaar E, Goossens M, Haufe A, Kamping E, Niessen RC, Hogervorst FBL, Gille JJP, Redeker B, Tops CMJ, van Gijn ME, van den Ouweland AMW, Rahner N, **Steinke V**, Kahl P, Holinski-Feder E, Morak M, Kloor M, Stemmler S, Betz B, Hutter P, Bunyan DJ, Syngal S, Culver JO, Graham T, Chan TL, Nagtegaal ID, van Krieken JHJM, Schackert HK, Hoogerbrugge N, van Kessel AG, Ligtenberg MJL. *Hum Mutat*. 2011;32(4):407-414. doi:10.1002/humu.21446

Risk of colorectal and endometrial cancers in EPCAM deletion-positive Lynch syndrome: a cohort study. Kempers MJ, Kuiper RP, Ockeloen CW, Chappuis PO, Hutter P, Rahner N, Schackert HK, **Steinke V**, Holinski-Feder E, Morak M, Kloor M, Büttner R, Verwiel ET, van Krieken JH, Nagtegaal ID, Goossens M, van der Post RS, Niessen RC, Sijmons RH, Kluijt I, Hogervorst FB, Leter EM, Gille JJ, Aalfs CM, Redeker EJ, Hes FJ, Tops CM, van Nesselrooij BP, van Gijn ME, García EBG, Eccles DM, Bunyan DJ, Syngal S, Stoffel EM, Culver JO, Palomares MR, Graham T, Velsher L, Papp J, Oláh E, Chan TL, Leung SY, van Kessel AG, Kiemeney LA, Hoogerbrugge N, Ligtenberg MJ. *The Lancet Oncology*. 2011;12(1):49-55. doi:10.1016/S1470-2045(10)70265-5

Diagnostik der spinalen Muskelatrophie Typ 1 (SMA1) durch Trophektodermbiopsie von Blastozysten. Seifert B, **Schön U,** Paulmann B, Seifert D, Hehr A, Köhler U, Holinski-Feder E. Reproduktionsmed Endokrinol; (2011) 8:335

A family with dystrophic alopecia and keratosis pilaris. Scholz IM, Hausser I, **Behnecke A**, Helmbold P. *J Dtsch Dermatol Ges*. 2011;9(12):1055-1056. doi:10.1111/j.1610-0387.2011.07792.x

Intragenic deletions of IL1RAPL1: Report of two cases and review of the literature. Behnecke A, Hinderhofer K, Bartsch O, Nümann A, Ipach M-L, Damatova N, Haaf T, Dufke A, Riess O, Moog U. *Am J Med Genet A*. 2011;155A(2):372-379. doi:10.1002/ajmg.a.33656

Chorea-acanthocytosis genotype in the original critchley kentucky neuroacanthocytosis kindred. Velayos-Baeza A, Holinski-Feder E, **Neitzel B**, Bader B, Critchley EMR, Monaco AP, Danek A, Walker RH. *Arch Neurol*. 2011;68(10):1330-1333. doi:10.1001/archneurol.2011.239

Publikationsliste

Neuromuscular signal transmission in adulthood. Current facets of acquired and hereditary disorders. **Abicht A**, Kröger S, Schoser B. *Nervenarzt*. 2011;82(6):707-711. https://doi.org/10.1007/s00115-010-2969-9

Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, Urtizberea A, von der Hagen M, Hübner A, Palace J, Bushby K, Straub V, Beeson D, **Abicht A**, Lochmüller H. *Am J Hum Genet*. 2011;88(2):162-172. https://doi.org/10.1016/j.ajhg.2011.01.008

Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. Schara U, von Kleist-Retzow J-C, Lainka E, Gerner P, Pyle A, Smith PM, Lochmüller H, Czermin B, **Abicht A**, Holinski-Feder E, Horvath R. *J Inherit Metab Dis*. 2011;34(1):197-201. https://doi.org/10.1007/s10545-010-9250-z

Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. Kemp JP, Smith PM, Pyle A, Neeve VCM, Tuppen HAL, Schara U, Talim B, Topaloglu H, Holinski-Feder E, **Abicht A**, Czermin B, Lochmüller H, McFarland R, Chinnery PF, Chrzanowska-Lightowlers ZMA, Lightowlers RN, Taylor RW, Horvath R. *Brain*. 2011;134(Pt 1):183-195. https://doi.org/10.1093/brain/awq320

The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Reilich P, Horvath R, Krause S, Schramm N, Turnbull DM, Trenell M, Hollingsworth KG, Gorman GS, Hans VH, Reimann J, MacMillan A, Turner L, Schollen A, Witte G, **Czermin B**, Holinski-Feder E, Walter MC, Schoser B, Lochmuller H. *J Neurol*. 2011;258(11):1987-1997. doi:10.1007/s00415-011-6055-4

Deficiency of the mitochondrial phosphate carrier presenting as myopathy and cardiomyopathy in a family with three affected children. Mayr JA, Zimmermann FA, Horvath R, Schneider H-C, Schoser B, Holinski-Feder E, **Czermin B**, Freisinger P, Sperl W. *Neuromuscul Disord*. 2011;21(11):803-808. doi:10.1016/j.nmd.2011.06.005

RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions. Fratter C, Raman P, Alston CL, Blakely EL, Craig K, Smith C, Evans J, Seller A, **Czermin B**, Hanna MG, Poulton J, Brierley C, Staunton TG, Turnpenny PD, Schaefer AM, Chinnery PF, Horvath R, Turnbull DM, Gorman GS, Taylor RW. *Neurology*. 2011;76(23):2032-2034. doi:10.1212/WNL.0b013e31821e558b

Predicting the contribution of novel POLG mutations to human disease through analysis in yeast model. Baruffini E, Horvath R, Dallabona C, **Czermin B**, Lamantea E, Bindoff L, Invernizzi F, Ferrero I, Zeviani M, Lodi T. *Mitochondrion*. 2011;11(1):182-190. doi:10.1016/j.mito.2010.09.007

The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. Reilich P, Horvath R, Krause S, Schramm N, Turnbull DM, Trenell M, Hollingsworth KG, Gorman GS, Hans VH, Reimann J, MacMillan A, Turner L, Schollen A, Witte G, Czermin B, **Holinski-Feder E**, Walter MC, Schoser B, Lochmüller H. J Neurol. 2011;258(11):1987-1997. https://doi.org/10.1007/s00415-011-6055-4

First evidence for digenic inheritance in hereditary colorectal cancer by mutations in the base excision repair genes. Morak M, Massdorf T, Sykora H, Kerscher M, **Holinski-Feder E**. Eur J Cancer. 2011;47(7):1046-1055. https://doi.org/10.1016/j.ejca.2010.11.016

Deciphering the colon cancer genes--report of the InSiGHT-Human Variome Project Workshop, UNESCO, Paris 2010. Kohonen-Corish MRJ, Macrae F, Genuardi M, Aretz S, Bapat B, Bernstein IT, Burn J, Cotton RGH, den Dunnen JT, Frebourg T, Greenblatt MS, Hofstra R, **Holinski-Feder E**, Lappalainen I, Lindblom A, Maglott D, Møller P, Morreau H, Möslein G, Sijmons R, Spurdle AB, Tavtigian S, Tops CMJ, Weber TK, de Wind N, Woods MO. Hum Mutat. 2011;32(4):491-494. https://doi.org/10.1002/humu.21450

Polymorphisms in CTNNBL1 in relation to colorectal cancer with evolutionary implications. Huhn S, Ingelfinger D, Bermejo JL, Bevier M, Pardini B, Naccarati A, Steinke V, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Pox CP, Goecke T, Kloor M, Loeffler M, Büttner R, Vodickova L, Novotny J, Demir K, Cruciat C-M, Renneberg R, Huber W, Niehrs C, Boutros M, Propping P, Vodièka P, Hemminki K, Försti A. Int J Mol Epidemiol Genet. 2011;2(1):36-50.

Primary mucinous adenocarcinoma of the vermiform appendix with high grade microsatellite instability. Komm M, Kronawitter-Fesl M, Kremer M, Lutz L, **Holinski-Feder E**, Kopp R. J Cancer. 2011;2:302-306. https://doi.org/10.7150/jca.2.302

Urm1 couples sulfur transfer to ubiquitin-like protein function in oxidative stress: Fig. 1. Petroski MD, Salvesen GS, **Wolf DA**. *Proc Natl Acad Sci USA*. 2011;108(5):1749-1750. doi:10.1073/pnas.1019043108

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

2010

Three-dimensional cell growth confers radioresistance by chromatin density modification. Storch K, Eke I, Borgmann K, Krause M, Richter C, **Becker K**, Schröck E, Cordes N. *Cancer Res*. 2010;70(10):3925-3934. doi:10.1158/0008-5472.CAN-09-3848

Stress-mediated nuclear stabilization of p53 is regulated by ubiquitination and importin-αlpha3 binding. Marchenko ND, Hanel W, Li D, Becker K, Reich N, Moll UM. *Cell Death Differ*. 2010;17(2):255-267. doi:10.1038/cdd.2009.173

A novel 1p31.3p32.2 deletion involving the NFIA gene detected by array CGH in a patient with macrocephaly and hypoplasia of the corpus callosum. Koehler U, Holinski-Feder E, Ertl-Wagner B, Kunz J, von Moers A, von Voss H, Schell-Apacik C. *Eur J Pediatr*. 2010;169(4):463-468. doi:10.1007/s00431-009-1057-2

Selbstschädigendes Verhalten bei zwei männlichen Patienten als typisches Syndrom des Smith-Magenis-Syndrom – guter therapeutischer Effekt von Aripiprazol. Martin P., **Koehler U.** Medizin für Menschen mit geistiger oder mehrfacher Behinderung; Edition Benthein 2010(2), 45-51.

Interstitial microduplication of Xp22.31: Causative of intellectual disability or benign copy number variant? Li F, Shen Y, **Köhler U,** Sharkey FH, Menon D, Coulleaux L, Malan V, Rio M, McMullan DJ, Cox H, Fagan KA, Gaunt L, Metcalfe K, Heinrich U, Hislop G, Maye U, Sutcliffe M, Wu B-L, Thiel BD, Mulchandani S, Conlin LK, Spinner NB, Murphy KM, Batista DAS. European Journal of Medical Genetics. 2010;53(2):93-99. doi:10.1016/j.ejmg.2010.01.004

Humangenetische Untersuchungen in der Pränataldiagnostik Frauenheilkunde. Koehler U., Holinski-Feder E. up2date 2010(5) 278-283.

MUTYH-associated polyposis - variability of the clinical phenotype in patients with biallelic and monoallelic MUTYH mutations and report on novel mutations. Morak M, **Laner A**, Bacher U, Keiling C, Holinski-Feder E. *Clin Genet*. ;78(4):353-363. doi:10.1111/j.1399-0004.2010.01478.x

PHD3 regulates differentiation, tumour growth and angiogenesis in pancreatic cancer. Su Y, Loos M, Giese N, Hines OJ, **Diebold I**, Görlach A, Metzen E, Pastorekova S, Friess H, Büchler P. *Br J Cancer*. 2010;103(10):1571-1579. doi:10.1038/sj.bjc.6605936

Reciprocal regulation of Rac1 and PAK-1 by HIF-1alpha: a positive-feedback loop promoting pulmonary vascular remodeling. Diebold I, Petry A, Djordjevic T, Belaiba RS, Fineman J, Black S, Schreiber C, Fratz S, Hess J, Kietzmann T, Görlach A. *Antioxid Redox Signal*. 2010;13(4):399-412. doi:10.1089/ars.2009.3013

The hypoxia-inducible factor-2alpha is stabilized by oxidative stress involving NOX4. Diebold I, Flügel D, Becht S, Belaiba RS, Bonello S, Hess J, Kietzmann T, Görlach A. *Antioxid Redox Signal*. 2010;13(4):425-436. doi:10.1089/ars.2009.3014

The NADPH oxidase subunit NOX4 is a new target gene of the hypoxia-inducible factor-1. Diebold I, Petry A, Hess J, Görlach A. *Mol Biol Cell*. 2010;21(12):2087-2096. doi:10.1091/mbc.e09-12-1003

Escherichia coli-cloned CFTR loci relevant for human artificial chromosome therapy. Rocchi L, Braz C, Cattani S, Ramalho A, Christan S, Edlinger M, Ascenzioni F, **Laner A**, Kraner S, Amaral M, Schindelhauer D. *Hum Gene Ther*. 2010;21(9):1077-1092. doi:10.1089/hum.2009.225

Clopidogrel and proton pump inhibitor (PPI) interaction: separate intake and a non-omeprazole PPI the solution? Kenngott S, Olze R, Kollmer M, Bottheim H, **Laner A**, Holinski-Feder E, Gross M. *Eur J Med Res*. 2010;15(5):220-224. doi:10.1186/2047-783x-15-5-220

Chemotherapeutic drugs inhibit ribosome biogenesis at various levels. Burger K, Mühl B, Harasim T, Rohrmoser M, Malamoussi A, Orban M, Kellner M, Gruber-Eber A, Kremmer E, Hölzel M, Eick D. *J Biol Chem*. 2010;285(16):12416-12425. doi:10.1074/jbc.M109.074211

Defects in 18 S or 28 S rRNA processing activate the p53 pathway. Hölzel M, Orban M, Hochstatter J, Rohrmoser M, Harasim T, Malamoussi A, Kremmer E, Längst G, Eick D. *J Biol Chem*. 2010;285(9):6364-6370. doi:10.1074/jbc.M109.054734

Homozygous loss of CHRNA7 on chromosome 15q13.3 causes severe encephalopathy with seizures and hypotonia. Endris V, Hackmann K, **Neuhann TM**, Grasshoff U, Bonin M, Haug U, Hahn G, Schallner JC, Schröck E, Tinschert S, Rappold G, Moog U. *Am J Med Genet A*. 2010;152A(11):2908-2911. doi:10.1002/ajmg.a.33692

Publikationsliste

Polymorphisms of genes coding for ghrelin and its receptor in relation to colorectal cancer risk: a two-step gene-wide case-control study. Campa D, Pardini B, Naccarati A, Vodickova L, Novotny J, **Steinke V**, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Kötting J, Betz B, Kloor M, Engel C, Büttner R, Propping P, Försti A, Hemminki K, Barale R, Vodicka P, Canzian F. *BMC Gastroenterol*. 2010;10(1):112. doi:10.1186/1471-230X-10-112

Clinical utility gene card for: Lynch syndrome (MLH1, MSH2, MSH6, PMS2). Rahner N, **Steinke V**, Schlegelberger B, Olschwang S, Eisinger F, Hutter P. *Eur J Hum Genet*. 2010;18(9):1071-1071. doi:10.1038/ejhg.2009.232

Genome-wide association study for colorectal cancer identifies risk polymorphisms in German familial cases and implicates MAPK signalling pathways in disease susceptibility. Lascorz J, Försti A, Chen B, Buch S, **Steinke V**, Rahner N, Holinski-Feder E, Morak M, Schackert HK, Görgens H, Schulmann K, Goecke T, Kloor M, Engel C, Büttner R, Kunkel N, Weires M, Hoffmeister M, Pardini B, Naccarati A, Vodickova L, Novotny J, Schreiber S, Krawczak M, Bröring CD, Völzke H, Schafmayer C, Vodicka P, Chang-Claude J, Brenner H, Burwinkel B, Propping P, Hampe J, Hemminki K. *Carcinogenesis*. 2010;31(9):1612-1619. doi:10.1093/carcin/bgq146

Efficacy of Annual Colonoscopic Surveillance in Individuals With Hereditary Nonpolyposis Colorectal Cancer. Engel C, Rahner N, Schulmann K, Holinski–Feder E, Goecke TO, Schackert HK, Kloor M, **Steinke V**, Vogelsang H, Möslein G, Görgens H, Dechant S, von Knebel Doeberitz M, Rüschoff J, Friedrichs N, Büttner R, Loeffler M, Propping P, Schmiegel W. *Clinical Gastroenterology and Hepatology*. 2010;8(2):174-182. doi:10.1016/j.cgh.2009.10.003

Clinical and neuropathological findings in patients with TACO1 mutations. Seeger J, Schrank B, Pyle A, Stucka R, Lörcher U, Müller-Ziermann S, **Abicht A**, Czermin B, Holinski-Feder E, Lochmüller H, Horvath R. *Neuromuscul Disord*. 2010;20(11):720-724. https://doi.org/10.1016/j.nmd.2010.06.017

Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. Walter MC, Czermin B, Muller-Ziermann S, Bulst S, Stewart JD, Hudson G, Schneiderat P, **Abicht A**, Holinski-Feder E, Lochmüller H, Chinnery PF, Klopstock T, Horvath R. *J Neurol*. 2010;257(9):1517-1523. https://doi.org/10.1007/s00415-010-5565-9

Molecular characterisation of congenital myasthenic syndromes in Southern Brazil. Mihaylova V, Scola RH, Gervini B, Lorenzoni PJ, Kay CK, Werneck LC, Stucka R, Guergueltcheva V, von der Hagen M, Huebner A, **Abicht A**, Müller JS, Lochmüller H. *J Neurol Neurosurg Psychiatry*. 2010;81(9):973-977. https://doi.org/10.1136/jnnp.2009.177816

Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. Schara U, Christen H-J, Durmus H, Hietala M, Krabetz K, Rodolico C, Schreiber G, Topaloglu H, Talim B, Voss W, Pihko H, **Abicht A**, Müller JS, Lochmüller H. *Eur J Paediatr Neurol*. 2010;14(4):326-333. https://doi.org/10.1016/j.ejpn.2009.09.009

The prevalence and natural history of dominant optic atrophy due to OPA1 mutations. Yu-Wai-Man P, Griffiths PG, Burke A, Sellar PW, Clarke MP, Gnanaraj L, Ah-Kine D, Hudson G, **Czermin B**, Taylor RW, Horvath R, Chinnery PF. *Ophthalmology*. 2010;117(8):1538-1546, 1546.e1. doi:10.1016/j.ophtha.2009.12.038

Long-term survival of neonatal mitochondrial complex III deficiency associated with a novel BCS1L gene mutation. Tuppen HAL, Fehmi J, **Czermin B**, Goffrini P, Meloni F, Ferrero I, He L, Blakely EL, McFarland R, Horvath R, Turnbull DM, Taylor RW. *Mol Genet Metab*. 2010;100(4):345-348. doi:10.1016/j.ymgme.2010.04.010

The clinical, histochemical, and molecular spectrum of PEO1 (Twinkle)-linked adPEO. Fratter C, Gorman GS, Stewart JD, Buddles M, Smith C, Evans J, Seller A, Poulton J, Roberts M, Hanna MG, Rahman S, Omer SE, Klopstock T, Schoser B, Kornblum C, **Czermin B**, Lecky B, Blakely EL, Craig K, Chinnery PF, Turnbull DM, Horvath R, Taylor RW. *Neurology*. 2010;74(20):1619-1626. doi:10.1212/WNL.0b013e3181df099f

Multi-system neurological disease is common in patients with OPA1 mutations. Yu-Wai-Man P, Griffiths PG, Gorman GS, Lourenco CM, Wright AF, Auer-Grumbach M, Toscano A, Musumeci O, Valentino ML, Caporali L, Lamperti C, Tallaksen CM, Duffey P, Miller J, Whittaker RG, Baker MR, Jackson MJ, Clarke MP, Dhillon B, **Czermin B**, Stewart JD, Hudson G, Reynier P, Bonneau D, Marques WJ, Lenaers G, McFarland R, Taylor RW, Turnbull DM, Votruba M, Zeviani M, Carelli V, Bindoff LA, Horvath R, Amati-Bonneau P, Chinnery PF. *Brain*. 2010;133(Pt 3):771-786. doi:10.1093/brain/awq007

Whole-body high-field MRI shows no skeletal muscle degeneration in young patients with recessive myotonia congenita. Kornblum C, Lutterbey GG, **Czermin B**, Reimann J, von Kleist-Retzow J-C, Jurkat-Rott K, Wattjes MP. *Acta Neurol Scand*. 2010;121(2):131-135. doi:10.1111/j.1600-0404.2009.01228.x

Publikationsliste

Laminopathy presenting as familial atrial fibrillation. Beckmann BM, Holinski-Feder E, Walter MC, Haserück N, Reithmann C, Hinterseer M, Wilde AA, Kääb S. Int J Cardiol. 2010;145(2):394-396. https://doi.org/10.1016/j.ijcard.2010.04.024

Leiden Open Variation Database of the MUTYH gene. Out AA, Tops CMJ, Nielsen M, Weiss MM, van Minderhout IJHM, Fokkema IFAC, Buisine M-P, Claes K, Colas C, Fodde R, Fostira F, Franken PF, Gaustadnes M, Heinimann K, Hodgson SV, Hogervorst FBL, **Holinski-Feder E**, Lagerstedt-Robinson K, Olschwang S, van den Ouweland AMW, Redeker EJW, Scott RJ, Vankeirsbilck B, Grønlund RV, Wijnen JT, Wikman FP, Aretz S, Sampson JR, Devilee P, den Dunnen JT, Hes FJ. Hum Mutat. 2010;31(11):1205-1215. https://doi.org/10.1002/humu.21343

Serum antibodies against frameshift peptides in microsatellite unstable colorectal cancer patients with Lynch syndrome. Reuschenbach M, Kloor M, Morak M, Wentzensen N, Germann A, Garbe Y, Tariverdian M, Findeisen P, Neumaier M, **Holinski-Feder E**, von Knebel Doeberitz M. Fam Cancer. 2010;9(2):173-179. https://doi.org/10.1007/s10689-009-9307-z

The role of genetic predisposition in early detection of cancer. Holinski-Feder E. MMW Fortschr Med. 2010;152(4):35-36. https://doi.org/10.1007/BF03365972

Recovery of systolic dysfunction in duchenne muscular dystrophy due to the point mutation c.4213C>T. Finsterer J, Stöllberger C, **Holinski-Feder E.** Cardiology. 2010;117(4):265-267. https://doi.org/10.1159/000323515

Chemical genetics approach to restoring p27Kip1 reveals novel compounds with antiproliferative activity in prostate cancer cells. Rico-Bautista E, Yang C-C, Lu L, Roth GP, **Wolf DA**. *BMC Biol*. 2010;8(1):153. doi:10.1186/1741-7007-8-153

2009

Phosphodiesterase 2 mediates redox-sensitive endothelial cell proliferation and angiogenesis by thrombin via Rac1 and NADPH oxidase 2. Diebold I, Djordjevic T, Petry A, Hatzelmann A, Tenor H, Hess J, Görlach A. *Circ Res*. 2009;104(10):1169-1177. doi:10.1161/CIRCRESAHA.109.196592

In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. Bulst S, Abicht A, Holinski-Feder E, Müller-Ziermann S, **Koehler U**, Thirion C, Walter MC, Stewart JD, Chinnery PF, Lochmüller H, Horvath R. *Hum Mol Genet*. 2009;18(9):1590-1599. doi:10.1093/hmg/ddp074

Single cell analysis of mutations in the APC gene. Mayer V, Schoen U, Holinski-Feder E, **Koehler U**, Thalhammer S. *Fetal Diagn Ther*. 2009;26(3):148-156. doi:10.1159/000248721

Single particle adsorbing transfer system. Woide D, Mayer V, Wachtmeister T, Hoehn N, Zink A, **Koehler U**, Thalhammer S. Biomed Microdevices. 2009 11(3): 609-614.

Duplications of the functional CYP21A2 gene are primarily restricted to Q318X alleles: evidence for a founder effect. Kleinle S, Lang R, Fischer GF, Vierhapper H, Waldhauser F, Födinger M, Baumgartner-Parzer SM. *J Clin Endocrinol Metab*. 2009;94(10):3954-3958. doi:10.1210/jc.2009-0487

1.6Mb deletion in chromosome band 3q29 associated with eye abnormalities. Tyshchenko N, Hackmann K, Gerlach E-M, **Neuhann T**, Schrock E, Tinschert S. *Eur J Med Genet*. 2009;52(2-3):128-130. doi:10.1016/j.ejmg.2009.03.002

Pontine tegmental cap dysplasia: the severe end of the clinical spectrum. Rauscher C, Poretti A, **Neuhann TM**, Forstner R, Hahn G, Koch J, Tinschert S, Boltshauser E. *Neuropediatrics*. 2009;40(1):43-46. doi:10.1055/s-0029-1224100

Expanded Extracolonic Tumor Spectrum in MUTYH-Associated Polyposis. Vogt S, Jones N, Christian D, Engel C, Nielsen M, Kaufmann A, **Steinke V**, Vasen HF, Propping P, Sampson JR, Hes FJ, Aretz S. *Gastroenterology*. 2009;137(6):1976-1985.e10. doi:10.1053/j.gastro.2009.08.052

Deciphering the genetics of hereditary non-syndromic colorectal cancer. The CORGI Consortium, Papaemmanuil E, Carvajal-Carmona L, Sellick GS, Kemp Z, Webb E, Spain S, Sullivan K, Barclay E, Lubbe S, Jaeger E, Vijayakrishnan J, Broderick P, Gorman M, Martin L, Lucassen A, Bishop DT, Evans DG, Maher ER, **Steinke V**, Rahner N, Schackert HK, Goecke TO, Holinski-Feder E, Propping P, Van Wezel T, Wijnen J, Cazier J-B, Thomas H, Houlston RS, Tomlinson I. *Eur J Hum Genet*. 2008;16(12):1477-1486. doi:10.1038/ejhg.2008.129

Hereditary Cancer Syndromes. Rahner N, **Steinke V**. *Deutsches Aerzteblatt Online*. Published online October 10, 2008. doi:10.3238/arztebl.2008.0706

Publikationsliste

Compound heterozygosity for twoMSH6 mutations in a patient with early onset colorectal cancer, vitiligo and systemic lupus erythematosus. Rahner N, Höefler G, Högenauer C, Lackner C, **Steinke V**, Sengteller M, Friedl W, Aretz S, Propping P, Mangold E, Walldorf C. *Am J Med Genet*. 2008;146A(10):1314-1319. doi:10.1002/ajmg.a.32210

No association between MUTYH and MSH6 germline mutations in 64 HNPCC patients.for The German HNPCC Consortium, **Steinke V**, Rahner N, Morak M, Keller G, Schackert HK, Görgens H, Schmiegel W, Royer-Pokora B, Dietmaier W, Kloor M, Engel C, Propping P, Aretz S. *Eur J Hum Genet*. 2008;16(5):587-592. doi:10.1038/ejhg.2008.26

Nine novel pathogenic germline mutations in *MLH1* , *MSH2* , *MSH6* and *PMS2* in families with Lynch syndrome. Rahner N, Friedrichs N, Wehner M, **Steinke V**, Aretz S, Friedl W, Buettner R, Mangold E, Propping P, Walldorf C. *Acta Oncologica*. 2007;46(6):763-769. doi:10.1080/02841860701230217

Human endogenous retroviral long terminal repeat sequences as cell type-specific promoters in retroviral vectors. Schön U, Diem O, Leitner L, Günzburg WH, Mager DL, Salmons B, Leib-Mösch C. *J Virol*. 2009;83(23):12643-12650. doi:10.1128/JVI.00858-09

IgE-mediated allergen gene vaccine platform targeting human antigen-presenting cells through the high-affinity IgE receptor. Behnecke A, Li W, Chen L, Saxon A, Zhang K. *J Allergy Clin Immunol*. 2009;124(1):108-113. doi:10.1016/j.jaci.2009.03.020

A novel multi-potential dendritic cell targeted gene vaccination platform; application to food allergy and beyond. Zhang K, **Behnecke A**, Li W, Saxon A. *Arb Paul Ehrlich Inst Bundesinstitut Impfstoffe Biomed Arzneim Langen Hess*. 2009;96:338-347; discussion 348.

Severe Myoclonic Epilepsy in Infancy - Adult Phenotype with Bradykinesia, Hypomimia, and Perseverative Behavior: Report of Five Cases. Martin P, Rautenstrauß B, **Abicht A**, Fahrbach J, Koster S. *Mol Syndromol*. 2010;1(5):231-238. https://doi.org/10.1159/000326746

Refinement of the clinical phenotype in musk-related congenital myasthenic syndromes. Mihaylova V, Salih MAM, Mukhtar MM, Abuzeid HA, El-Sadig SM, von der Hagen M, Huebner A, Nürnberg G, **Abicht A**, Müller JS, Lochmüller H, Guergueltcheva V. *Neurology*. 2009;73(22):1926-1928. https://doi.org/10.1212/WNL.0b013e3181c3fce9

Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. Schara U, Barisic N, Deschauer M, Lindberg C, Straub V, Strigl-Pill N, Wendt M, **Abicht A**, Müller JS, Lochmüller H. *Neuromuscul Disord*. 2009;19(12):828-832. https://doi.org/10.1016/j.nmd.2009.09.008

In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. Bulst S, **Abicht A**, Holinski-Feder E, Müller-Ziermann S, Koehler U, Thirion C, Walter MC, Stewart JD, Chinnery PF, Lochmüller H, Horvath R. *Hum Mol Genet*. 2009;18(9):1590-1599. https://doi.org/10.1093/hmg/ddp074

Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNA(Val) causing MNGIE-like gastrointestinal dysmotility and cachexia. Horváth R, Bender A, **Abicht A**, Holinski-Feder E, Czermin B, Trips T, Schneiderat P, Lochmüller H, Klopstock T. *J Neurol*. 2009;256(5):810-815. https://doi.org/10.1007/s00415-009-5023-8

Mitochondrial neurogastrointestinal encephalomyopathy mimicking anorexia nervosa. Feddersen B, DE LA Fontaine L, Sass JO, Lutz J, **Abicht A**, Klopstock T, Verma IC, Meisenzahl E, Pogarell O. *Am J Psychiatry*. 2009;166(4):494-495. https://doi.org/10.1176/appi.ajp.2008.08101525

Collated mutations in mitochondrial DNA (mtDNA) depletion syndrome (excluding the mitochondrial gamma polymerase, POLG1). Poulton J, Hirano M, Spinazzola A, Arenas Hernandez M, Jardel C, Lombes A, **Czermin B**, Horvath R, Taanman JW, Rotig A, Zeviani M, Fratter C. *Biochim Biophys Acta*. 2009;1792(12):1109-1112. doi:10.1016/j.bbadis.2009.08.016

Fragile X-associated tremor/ataxia syndrome. Finke C, Horváth R, Holinski-Feder E, Ploner CJ. Nervenarzt. 2009;80(12):1473-1479. https://doi.org/10.1007/s00115-009-2846-6

Molecular basis of infantile reversible cytochrome c oxidase deficiency myopathy. Horvath R, Kemp JP, Tuppen HAL, Hudson G, Oldfors A, Marie SKN, Moslemi A-R, Servidei S, Holme E, Shanske S, Kollberg G, Jayakar P, Pyle A, Marks HM, **Holinski-Feder E**, Scavina M, Walter MC, Coku J, Günther-Scholz A, Smith PM, McFarland R, Chrzanowska-Lightowlers ZMA, Lightowlers RN, Hirano M, Lochmüller H, Taylor RW, Chinnery PF, Tulinius M, DiMauro S. Brain. 2009;132(Pt 11):3165-3174. https://doi.org/10.1093/brain/awp221

Publikationsliste

The eIF3 Interactome Reveals the Translasome, a Supercomplex Linking Protein Synthesis and Degradation Machineries. Sha Z, Brill LM, Cabrera R, Kleifeld O, Scheliga JS, Glickman MH, Chang EC, **Wolf DA**. *Molecular Cell*. 2009;36(1):141-152. doi:10.1016/j.molcel.2009.09.026

Destruction of RhoA CULtivates Actin. Wu S, **Wolf DA**. *Molecular Cell*. 2009;35(6):735-736. doi:10.1016/j.molcel.2009.09.012

F-Box-Directed CRL Complex Assembly and Regulation by the CSN and CAND1. Schmidt MW, McQuary PR, Wee S, Hofmann K, **Wolf DA**. *Molecular Cell*. 2009;35(5):586-597. doi:10.1016/j.molcel.2009.07.024

Zcchc11-dependent uridylation of microRNA directs cytokine expression. Jones MR, Quinton LJ, Blahna MT, Neilson JR, Fu S, Ivanov AR, **Wolf DA**, Mizgerd JP. *Nat Cell Biol*. 2009;11(9):1157-1163. doi:10.1038/ncb1931

Comprehensive proteomic analysis of Schizosaccharomyces pombe by two-dimensional HPLC-tandem mass spectrometry. Brill LM, Motamedchaboki K, Wu S, **Wolf DA**. *Methods*. 2009;48(3):311-319. doi:10.1016/j.ymeth.2009.02.023

Rfu1: stimulus for the ubiquitin economy. **Wolf DA**, Petroski MD. *Cell*. 2009;137(3):397-398. doi:10.1016/j.cell.2009.04.032

Inflamed Snail Speeds Metastasis. Yang C-C, **Wolf DA**. *Cancer Cell*. 2009;15(5):355-357. doi:10.1016/j.ccr.2009.04.003

Snail puts melanoma on the fast track. Herzinger T, **Wolf DA**. *Pigment Cell & Melanoma Research*. 2009;22(2):150-151. doi:10.1111/j.1755-148X.2009.00552.x

2008

The "PAI-1 paradox" in vascular remodeling. Diebold I, Kraicun D, Bonello S, Görlach A. *Thromb Haemost*. 2008;100(6):984-991.

Rac-1 promotes pulmonary artery smooth muscle cell proliferation by upregulation of plasminogen activator inhibitor-1: role of NFkappaB-dependent hypoxia-inducible factor-1alpha transcription. Diebold I, Djordjevic T, Hess J, Görlach A. *Thromb Haemost*. 2008;100(6):1021-1028.

Impact of heart rate variability in patients with normal sinus rhythm on image quality in coronary magnetic angiography. Tangcharoen T, Jahnke C, **Koehler U**, Schnackenburg B, Klein C, Fleck E, Nagel E. *J Magn Reson Imaging*. 2008;28(1):74-79. doi:10.1002/jmri.21426

A role of HAUSP in tumor suppression in a human colon carcinoma xenograft model. Becker K, Marchenko ND, Palacios G, Moll UM. *Cell Cycle*. 2008;7(9):1205-1213. doi:10.4161/cc.7.9.5756

The CALM and CALM/AF10 interactor CATS is a marker for proliferation. Archangelo LF, Greif PA, Hölzel M, Harasim T, Kremmer E, Przemeck GKH, Eick D, Deshpande AJ, Buske C, de Angelis MH, Saad STO, Bohlander SK. *Mol Oncol*. 2008;2(4):356-367. doi:10.1016/j.molonc.2008.08.001

The nucleolar SUMO-specific protease SENP3 reverses SUMO modification of nucleophosmin and is required for rRNA processing. Haindl M, Harasim T, Eick D, Muller S. *EMBO Rep*. 2008;9(3):273-279. doi:10.1038/embor.2008.3

Report on de-novo mutation in the MSH2 gene as a rare event in hereditary nonpolyposis colorectal cancer. Morak M, **Laner A**, Scholz M, Madorf T, Holinski-Feder E. *Eur J Gastroenterol Hepatol*. 2008;20(11):1101-1105. doi:10.1097/MEG.0b013e328305e185

Septal perforation and palatal defect. Behnecke A, Aigner T, Rösler W, Schick B. *HNO*. 2008;56(6):623-626. doi:10.1007/s00106-008-1668-3

Evaluation of ECP release from intact tissue biopsies from patients with nasal polyps. Behnecke A, Mayr S, Schick B, Iro H, Raithel M. *Inflamm Res*. 2008;57 Suppl 1:S65-66. doi:10.1007/s00011-007-0632-0

Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. Mihaylova V, Müller JS, Vilchez JJ, Salih MA, Kabiraj MM, D'Amico A, Bertini E, Wölfle J, Schreiner F, Kurlemann G, Rasic VM, Siskova D, Colomer J, Herczegfalvi A, Fabriciova K, Weschke B, Scola R, Hoellen F, Schara U, **Abicht A**, Lochmüller H. *Brain*. 2008;131(Pt 3):747-759. https://doi.org/10.1093/brain/awm325

ESC, ESCL and their roles in Polycomb Group mechanisms. Ohno K, McCabe D, **Czermin B**, Imhof A, Pirrotta V. *Mech Dev*. 2008;125(5-6):527-541. doi:10.1016/j.mod.2008.01.002

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

2007

Interdependence of Pes1, Bop1, and WDR12 controls nucleolar localization and assembly of the PeBoW complex required for maturation of the 60S ribosomal subunit. Rohrmoser M, Hölzel M, Grimm T, Malamoussi A, Harasim T, Orban M, Pfisterer I, Gruber-Eber A, Kremmer E, Eick D. *Mol Cell Biol*. 2007;27(10):3682-3694. doi:10.1128/MCB.00172-07

I**Hyperubiquitylation of wild-type p53 contributes to cytoplasmic sequestration in neuroblastoma**. **Becker K,** Marchenko ND, Maurice M, Moll UM. *Cell Death Differ*. 2007;14(7):1350-1360. doi:10.1038/sj.cdd.4402126

Monoubiquitylation promotes mitochondrial p53 translocation. Marchenko ND, Wolff S, Erster S, **Becker K**, Moll UM. *EMBO J*. 2007;26(4):923-934. doi:10.1038/sj.emboj.7601560

A new interphase fluorescence in situ hybridization approach for genomic rearrangements involving MLH1 and MSH6 in hereditary nonpolyposis colorectal cancer-suspected mutation-negative patients. Koehler U, Grabowski M, Bacher U, Holinski-Feder E. *Cancer Genet Cytogenet*. 2007;175(1):81-84. doi:10.1016/j.cancergencyto.2007.01.008

Programmierbares, zytogenetisches Submikroliter-Chiplabor für molekular-diagnostische Anwendungen. S. Thalhammer S., **Koehler U**., et al. GenomXPress 2007 ISBN: 1617-562X, 1.07: 29-31.

Targeting of the glucocorticoid hormone receptor with plasmid DNA comprising glucocorticoid response elements improves nonviral gene transfer efficiency in the lungs of mice. Dames P, **Laner A**, Maucksch C, Aneja MK, Rudolph C. *J Gene Med*. 2007;9(9):820-829. doi:10.1002/jgm.1082

Rapid conditional knock-down-knock-in system for mammalian cells. Hölzel M, Rohrmoser M, Orban M, Hömig C, Harasim T, Malamoussi A, Gruber-Eber A, Heissmeyer V, Bornkamm G, Eick D. *Nucleic Acids Res*. 2007;35(3):e17. doi:10.1093/nar/gkl1055

The BRCT domain of mammalian Pes1 is crucial for nucleolar localization and rRNA processing. Hölzel M, Grimm T, Rohrmoser M, Malamoussi A, Harasim T, Gruber-Eber A, Kremmer E, Eick D. *Nucleic Acids Res*. 2007;35(3):789-800. doi:10.1093/nar/gkl1058

The potential of retroviral vectors to cotransfer human endogenous retroviruses (HERVs) from human packaging cell lines. Zeilfelder U, Frank O, Sparacio S, **Schön U**, Bosch V, Seifarth W, Leib-Mösch C. *Gene*. 2007;390(1-2):175-179. doi:10.1016/j.gene.2006.08.019

Low volume amplification and sequencing of mitochondrial DNA on a chemically structured chip. Lutz-Bonengel S, Sänger T, Heinrich M, **Schön U,** Schmidt U. *Int J Legal Med*. 2007;121(1):68-73. doi:10.1007/s00414-006-0125-7

The potential of retroviral vectors to cotransfer endogenous retroviruses (HERVs) from human packaging cell lines. Udo Zeilfelder, Oliver Frank, Sandra Sparacio, **Ulrike Schön,** Valerie Bosch, Wolfgang Seifarth, Christine Leib-Mösch, Gene. 2007 Apr 1;390(1-2):175-9.

Congenital myasthenic syndromes: spotlight on genetic defects of neuromuscular transmission. Müller JS, Mihaylova V, **Abicht A**, Lochmüller H. *Expert Rev Mol Med*. 2007;9(22):1-20. https://doi.org/10.1017/S1462399407000427

Phenotypical spectrum of DOK7 mutations in congenital myasthenic syndromes. Müller JS, Herczegfalvi A, Vilchez JJ, Colomer J, Bachinski LL, Mihaylova V, Santos M, Schara U, Deschauer M, Shevell M, Poulin C, Dias A, Soudo A, Hietala M, Aärimaa T, Krahe R, Karcagi V, Huebner A, Beeson D, **Abicht A**, Lochmüller H. *Brain*. 2007;130(Pt 6):1497-1506. https://doi.org/10.1093/brain/awm068

Comparative proteomic and transcriptomic profiling of the fission yeast *Schizosaccharomyces pombe*. Schmidt MW, Houseman A, Ivanov AR, **Wolf DA**. *Mol Syst Biol*. 2007;3(1):79. doi:10.1038/msb4100117

2006

Patterns of p73 N-terminal isoform expression and p53 status have prognostic value in gynecological cancers. Becker K, Pancoska P, Concin N, Vanden Heuvel K, Slade N, Fischer M, Chalas E, Moll UM. *Int J Oncol*. 2006;29(4):889-902.

Interaction of polyamine gene vectors with RNA leads to the dissociation of plasmid DNA-carrier complexes. Huth S, Hoffmann F, von Gersdorff K, **Laner A**, Reinhardt D, Rosenecker J, Rudolph C. *J Gene Med*. 2006;8(12):1416-1424. doi:10.1002/jgm.975

Publikationsliste

Leigh syndrome caused by mutations in the flavoprotein (Fp) subunit of succinate dehydrogenase (SDHA). Horváth R, Abicht A, Holinski-Feder E, **Laner A**, Gempel K, Prokisch H, Lochmüller H, Klopstock T, Jaksch M. *J Neurol Neurosurg Psychiatry*. 2006;77(1):74-76. doi:10.1136/jnnp.2005.067041

Dominant-negative Pes1 mutants inhibit ribosomal RNA processing and cell proliferation via incorporation into the PeBoW-complex. Grimm T, Hölzel M, Rohrmoser M, Harasim T, Malamoussi A, Gruber-Eber A, Kremmer E, Eick D. *Nucleic Acids Res*. 2006;34(10):3030-3043. doi:10.1093/nar/gkl378

Low-volume amplification on chemically structured chips using the PowerPlex16 DNA amplification kit. Schmidt U, Lutz-Bonengel S, Weisser H-J, Sänger T, Pollak S**, Schön U**, Zacher T, Mann W. *Int J Legal Med*. 2006;120(1):42-48. doi:10.1007/s00414-005-0041-2

Impaired receptor clustering in congenital myasthenic syndrome with novel RAPSN mutations. Müller JS, Baumeister SK, Rasic VM, Krause S, Todorovic S, Kugler K, Müller-Felber W, **Abicht A**, Lochmüller H. *Neurology*. 2006;67(7):1159-1164. https://doi.org/10.1212/01.wnl.0000233837.79459.40

CHRND mutation causes a congenital myasthenic syndrome by impairing co-clustering of the acetylcholine receptor with rapsyn. Müller JS, Baumeister SK, Schara U, Cossins J, Krause S, von der Hagen M, Huebner A, Webster R, Beeson D, Lochmüller H, **Abicht A**. *Brain*. 2006;129(Pt 10):2784-2793. https://doi.org/10.1093/brain/awl188

Long-term improvement of slow-channel congenital myasthenic syndrome with fluoxetine. Colomer J, Müller JS, Vernet A, Nascimento A, Pons M, Gonzalez V, **Abicht A**, Lochmüller H. *Neuromuscul Disord*. 2006;16(5):329-333. https://doi.org/10.1016/j.nmd.2006.02.009

Facing the genetic heterogeneity in neuromuscular disorders: linkage analysis as an economic diagnostic approach towards the molecular diagnosis. von der Hagen M, Schallner J, Kaindl AM, Koehler K, Mitzscherling P, **Abicht A**, Grieben U, Korinthenberg R, Kress W, von Moers A, Müller JS, Schara U, Vorgerd M, Walter MC, Müller-Reible C, Hübner C, Lochmüller H, Huebner A. *Neuromuscul Disord*. 2006;16(1):4-13. https://doi.org/10.1016/j.nmd.2005.10.001

2005

Bacterial transfer of large functional genomic DNA into human cells. Laner A, Goussard S, Ramalho AS, Schwarz T, Amaral MD, Courvalin P, Schindelhauer D, Grillot-Courvalin C. *Gene Ther*. 2005;12(21):1559-1572. doi:10.1038/sj.gt.3302576

Deletions account for 17% of pathogenic germline alterations in MLH1 and MSH2 in hereditary nonpolyposis colorectal cancer (HNPCC) families. Grabowski M, Mueller-Koch Y, Grasbon-Frodl E, **Koehler U**, Keller G, Vogelsang H, Dietmaier W, Kopp R, Siebers U, Schmitt W, Neitzel B, Gruber M, Doerner C, Kerker B, Ruemmele P, Henke G, Holinski-Feder E. *Genet Test*. 2005;9(2):138-146. doi:10.1089/gte.2005.9.138

Mammalian WDR12 is a novel member of the Pes1-Bop1 complex and is required for ribosome biogenesis and cell proliferation. Hölzel M, Rohrmoser M, Schlee M, Grimm T, Harasim T, Malamoussi A, Gruber-Eber A, Kremmer E, Hiddemann W, Bornkamm GW, Eick D. *J Cell Biol*. 2005;170(3):367-378. doi:10.1083/jcb.200501141

Molekularbiologische Analysen von Einzelzellen. K. Hagen-Mann, Th. Zacher, O. Khanaga, V. Baukloh, W. Mann**, U. Schön** Medgen (2005)

Hereditary non-polyposis colorectal cancer: clinical and molecular evidence for a new entity of hereditary colorectal cancer. Y. Mueller-Koch, H. Vogelsang, R. Kopp, P. Lohse, G. Keller, D. Aust, M. Muders, M. Gross, J. Daum, U. Schiemann, M. Grabowski, M. Scholz, B. Kerker, I. Becker, G. Henke, & E. Holinski-Feder, Gut, 54 (2005) 1733–1740. https://doi.org/10.1136/gut.2004.060905.

Spectrum and frequencies of mutations in MSH2 and MLH1 identified in 1,721 German families suspected of hereditary nonpolyposis colorectal cancer. Mangold E, Pagenstecher C, Friedl W, Mathiak M, Buettner R, Engel C, Loeffler M, Holinski-Feder E, **Müller-Koch Y**, Keller G, Schackert HK, Krüger S, Goecke T, Moeslein G, Kloor M, Gebert J, Kunstmann E, Schulmann K, Rüschoff J, Propping P. *Int J Cancer*. 2005;116(5):692-702. doi:10.1002/ijc.20863

Detection of occult high graded microsatellite instabilities in MMR gene mutation negative HNPCC tumors by addition of complementary marker analysis. Schiemann U, **Müller-Koch Y**, Gross M, Glas J, Baretton G, Muders M, Mussack T, Holinski-Feder E. *Eur J Med Res*. 2005;10(1):23-28.

Preoperative serum levels of the carcinoembryonic antigen in hereditary non-polyposis colorectal cancer compared to levels in sporadic colorectal cancer. Schiemann U, Günther S, Gross M, Henke G, **Müller-Koch Y**, König A, Muders M, Folwaczny C, Mussack T, Holinski-Feder E. *Cancer Detect Prev*. 2005;29(4):356-360. doi:10.1016/j.cdp.2005.04.003

Publikationsliste

An intronic base alteration of the CHRNE gene leading to a congenital myasthenic syndrome. Müller JS, Stucka R, Neudecker S, Zierz S, Schmidt C, Huebner A, Lochmüller H, **Abicht A**. *Neurology*. 2005;65(3):463-465. https://doi.org/10.1212/01.wnl.0000172346.26219.fd

An Iranian family with congenital myasthenic syndrome caused by a novel acetylcholine receptor mutation (CHRNE K171X). Soltanzadeh P, Müller JS, Ghorbani A, **Abicht A**, Lochmüller H, Soltanzadeh A. *J Neurol Neurosurg Psychiatry*. 2005;76(7):1039-1040. https://doi.org/10.1136/jnnp.2004.059436

Clinical variability of CMS-EA (congenital myasthenic syndrome with episodic apnea) due to identical CHAT mutations in two infants. Barisic N, Müller JS, Paucic-Kirincic E, Gazdik M, Lah-Tomulic K, Pertl A, Sertic J, Zurak N, Lochmüller H, **Abicht A**. *Eur J Paediatr Neurol*. 2005;9(1):7-12. https://doi.org/10.1016/j.ejpn.2004.10.008

PCI proteins eIF3e and eIF3m define distinct translation initiation factor 3 complexes. Zhou C, Arslan F, Wee S, Krishnan S, Ivanov AR, Oliva A, Leatherwood J, **Wolf DA**. *BMC Biol*. 2005;3:14. doi:10.1186/1741-7007-3-14

CSN facilitates Cullin–RING ubiquitin ligase function by counteracting autocatalytic adapter instability. Wee S, Geyer RK, Toda T, **Wolf DA**. *Nat Cell Biol*. 2005;7(4):387-391. doi:10.1038/ncb1241

2004

Transdominant DeltaTAp73 Isoforms are frequently up-regulated in ovarian cancer. Evidence for their role as epigenetic p53 inhibitors in vivo .Concin N, Becker K, Slade N, Erster S, Mueller-Holzner E, Ulmer H, Daxenbichler G, Zeimet A, Zeillinger R, Marth C, Moll UM. *Cancer Res*. 2004;64(7):2449-2460. doi:10.1158/0008-5472.CAN-03-1060

High-throughput analysis of genome-wide receptor tyrosine kinase expression in human cancers identifies potential novel drug targets. Müller-Tidow C, Schwäble J, Steffen B, Tidow N, Brandt B, **Becker K**, Schulze-Bahr E, Halfter H, Vogt U, Metzger R, Schneider PM, Büchner T, Brandts C, Berdel WE, Serve H. *Clin Cancer Res*. 2004;10(4):1241-1249. doi:10.1158/1078-0432.ccr-0954-03

Gene delivery systems--gene therapy vectors for cystic fibrosis. Klink D, Schindelhauer D, **Laner A**, Tucker T, Bebok Z, Schwiebert EM, Boyd AC, Scholte BJ. *J Cyst Fibros*. 2004;3 Suppl 2:203-212. doi:10.1016/j.jcf.2004.05.042

Rapid identification of female carriers of DMD/BMD by quantitative real-time PCR. Joncourt F, Neuhaus B, Jostarndt-Foegen K, **Kleinle S**, Steiner B, Gallati S. *Hum Mutat*. 2004;23(4):385-391. doi:10.1002/humu.20007

Suitability of a CMV/EGFP cassette to monitor stable expression from human artificial chromosomes but not transient transfer in the cells forming viable clones. Laner A, Schwarz T, Christan S, Schindelhauer D. *Cytogenet Genome Res*. 2004;107(1-2):9-13. doi:10.1159/000079564

Influence of human endogenous retroviruses on cellular gene expression. In: "Retroviruses and Primate Genome Evolution" Leib-Mösch, C., Seifarth, W., Schön, U. *Landes Bioscience* (E. Sverdlov, Ed.), Georgetown, Texas (2004)

Mutation analysis of the MLH1, MSH2 and MSH6 genes in patients with double primary cancers of the colorectum and the endometrium: a population-based study in northern Sweden. Cederquist K, Emanuelsson M, Göransson I, Holinski-Feder E, **Müller-Koch Y**, Golovleva I, Grönberg H. *Int J Cancer*. 2004;109(3):370-376. doi:10.1002/ijc.11718

Extended microsatellite analysis in microsatellite stable, MSH2 and MLH1 mutation-negative HNPCC patients: genetic reclassification and correlation with clinical features. Schiemann U, **Müller-Koch Y**, Gross M, Daum J, Lohse P, Baretton G, Muders M, Mussack T, Kopp R, Holinski-Feder E. *Digestion*. 2004;69(3):166-176. doi:10.1159/000078223

A newly identified chromosomal microdeletion of the rapsyn gene causes a congenital myasthenic syndrome. Müller JS, **Abicht A**, Christen H-J, Stucka R, Schara U, Mortier W, Huebner A, Lochmüller H. *Neuromuscul Disord*. 2004;14(11):744-749. https://doi.org/10.1016/j.nmd.2004.06.010

Mutation history of the roma/gypsies. Morar B, Gresham D, Angelicheva D, Tournev I, Gooding R, Guergueltcheva V, Schmidt C, **Abicht A**, Lochmuller H, Tordai A, Kalmar L, Nagy M, Karcagi V, Jeanpierre M, Herczegfalvi A, Beeson D, Venkataraman V, Warwick Carter K, Reeve J, de Pablo R, Kucinskas V, Kalaydjieva L. *Am J Hum Genet*. 2004;75(4):596-609. https://doi.org/10.1086/424759

The congenital myasthenic syndrome mutation RAPSN N88K derives from an ancient Indo-European founder. Müller JS, **Abicht A**, Burke G, Cossins J, Richard P, Baumeister SK, Stucka R, Eymard B, Hantaï D, Beeson D, Lochmüller H. *J Med Genet*. 2004;41(8):e104. https://doi.org/10.1136/jmg.2004.021139

Publikationsliste

Synaptic congenital myasthenic syndrome in three patients due to a novel missense mutation (T441A) of the COLQ gene. Müller JS, Petrova S, Kiefer R, Stucka R, König C, Baumeister SK, Huebner A, Lochmüller H, **Abicht A**. *Neuropediatrics*. 2004;35(3):183-189. https://doi.org/10.1055/s-2004-820996

The N-terminus of Drosophila SU(VAR)3-9 mediates dimerization and regulates its methyltransferase activity. Eskeland R, **Czermin B**, Boeke J, Bonaldi T, Regula JT, Imhof A. *Biochemistry*. 2004;43(12):3740-3749. doi:10.1021/bi035964s

Rapid prefractionation of complex protein lysates with centrifugal membrane adsorber units improves the resolving power of 2D-PAGE-based proteome analysis. Doud MK, Schmidt MW, Hines D, Naumann C, Kocourek A, Kashani-Poor N, Zeidler R, **Wolf DA**. *BMC Genomics*. 2004;5(1):25. doi:10.1186/1471-2164-5-25

Multidimensional proteomic analysis of proteolytic pathways involved in cell cycle control. Schmidt MW, Jain A, **Wolf DA**. *Methods Mol Biol*. 2004;241:235-245. doi:10.1385/1-59259-646-0:235

2003

Mutations in NSD1 are responsible for Sotos syndrome, but are not a frequent finding in other overgrowth phenotypes. Türkmen S, Gillessen-Kaesbach G, Meinecke P, Albrecht B, Neumann LM, Hesse V, Palanduz S, Balg S, Majewski F, Fuchs S, Zschieschang P, Greiwe M, Mennicke K, Kreuz FR, Dehmel HJ, Rodeck B, Kunze J, Tinschert S, Mundlos S, Horn D. *Eur J Hum Genet*. 2003;11(11):858-865. doi:10.1038/sj.ejhg.5201050

Problems in detecting mosaic DNA methylation in Angelman syndrome. Horsthemke B, Lich C, Buiting K, Achmann R, Aulehla-Scholz C, Baumer A, Bürger J, Dworniczak B, Gläser D, Holinski-Feder E, Janssen B, **Kleinle S**, Kochhan L, Krasemann E, Kraus C, Kroisel P, Plendl H, Purmann S, Sander G, Skladny H, Spitzer E, Thamm-Mücke B, Varon-Mateeva R, Weinhäusel A, Weirich H. *Eur J Hum Genet*. 2003;11(12):913-915. doi:10.1038/sj.ejhg.5201078

N-methyl-norsalsolinol modulates serotonin metabolism in the rat caudate nucleus: correlation with behavioural changes. Thümen A, **Behnecke A**, Qadri F, Moser A. *Int J Neuropsychopharmacol*. 2003;6(1):35-40. doi:10.1017/S1461145702003206

Easy, accurate and reliable screening for SNPs by ion pair/reverse phase HPLC: simultaneous detection of factor V G1691A, prothrombin G20210A and methylenetetrahydrofolate reductase C677T variants. Neitzel B, Matern C, Holinski-Feder E. *Clin Lab*. 2003;49(7-8):313-318.

FMR1 gene deletion/reversion: a pitfall of fragile X carrier testing. Gasteiger M, Grasbon-Frodl E, **Neitzel B**, Kooy F, Holinski-Feder E. *Genet Test*. 2003;7(4):303-308. doi:10.1089/109065703322783653

Rapsyn N88K is a frequent cause of congenital myasthenic syndromes in European patients. Müller JS, Mildner G, Müller-Felber W, Schara U, Krampfl K, Petersen B, Petrova S, Stucka R, Mortier W, Bufler J, Kurlemann G, Huebner A, Merlini L, Lochmüller H, **Abicht A**. *Neurology*. 2003;60(11):1805-1810. https://doi.org/10.1212/01.wnl.0000072262.14931.80

Congenital myasthenic syndrome due to a novel missense mutation in the gene encoding choline acetyltransferase. Schmidt C, **Abicht A**, Krampfl K, Voss W, Stucka R, Mildner G, Petrova S, Schara U, Mortier W, Bufler J, Huebner A, Lochmüller H. *Neuromuscul Disord*. 2003;13(3):245-251. https://doi.org/10.1016/s0960-8966(02)00273-0

The sounds of silence--histone deacetylation meets histone methylation. Czermin B, Imhof A. *Genetica*. 2003;117(2-3):159-164. doi:10.1023/a:1022927725945

The COP9 signalosome: an assembly and maintenance platform for cullin ubiquitin ligases? Wolf DA, Zhou C, Wee S. *Nat Cell Biol*. 2003;5(12):1029-1033. doi:10.1038/ncb1203-1029

Dynamic Release of Cdc34 from SCF. **Wolf DA**, Geyer R. *Cell*. 2003;114(5):532-533. doi:10.1016/S0092-8674(03)00682-2

BTB/POZ Domain Proteins Are Putative Substrate Adaptors for Cullin 3 Ubiquitin Ligases. Geyer R, Wee S, Anderson S, Yates J, **Wolf DA**. *Molecular Cell*. 2003;12(3):783-790. doi:10.1016/S1097-2765(03)00341-1

Fission Yeast COP9/Signalosome Suppresses Cullin Activity through Recruitment of the Deubiquitylating Enzyme Ubp12p. Zhou C, Wee S, Rhee E, Naumann M, Dubiel W, **Wolf DA**. *Molecular Cell*. 2003;11(4):927-938. doi:10.1016/S1097-2765(03)00136-9

MGZ
Medizinisch Genetisches Zentrum

Publikationsliste

2002

NADPH oxidase mediates tissue factor-dependent surface procoagulant activity by thrombin in human vascular smooth muscle cells. Herkert O, **Diebold I**, Brandes RP, Hess J, Busse R, Görlach A. *Circulation*. 2002;105(17):2030-2036. doi:10.1161/01.cir.0000014611.28864.1e

Visible transient expression of EGFP requires intranuclear injection of large copy numbers. Schindelhauer D, **Laner A**. *Gene Ther*. 2002;9(11):727-730. doi:10.1038/sj.gt.3301755

Chromosome painting reveals that galagos have highly derived karyotypes. Stanyon R, **Koehler U**, Consigliere S. *Am J Phys Anthropol*. 2002;117(4):319-326. doi:10.1002/ajpa.10047

N-methyl-norsalsolinol, a putative dopaminergic neurotoxin, passes through the blood-brain barrier in vivo. Thümen A, **Behnecke A**, Qadri F, Bäuml E, Thümen A, Behnecke CA, Qadri F, Bäuml E, Moser A. *Neuroreport*. 2002;13(1):25-28. doi:10.1097/00001756-200201210-00010

What's in the serum of seronegative MG and LEMS?: MuSK et al. Abicht A, Lochmüller H. *Neurology*. 2002;59(11):1672-1673. https://doi.org/10.1212/01.wnl.0000041026.90947.79

Congenital myasthenic syndrome (CMS) in three European kinships due to a novel splice mutation (IVS7 - 2 A/G) in the epsilon acetylcholine receptor (AChR) subunit gene. Barisic N, Schmidt C, Sidorova OP, Herczegfalvi A, Gekht BM, Song I-H, Stucka R, Karcagi V, **Abicht A**, Lochmüller H. *Neuropediatrics*. 2002;33(5):249-254. https://doi.org/10.1055/s-2002-36738

A newly identified chromosomal microdeletion and an N-box mutation of the AChR epsilon gene cause a congenital myasthenic syndrome. Abicht A, Stucka R, Schmidt C, Briguet A, Höpfner S, Song I-H, Pongratz D, Müller-Felber W, Ruegg MA, Lochmüller H. *Brain*. 2002;125(Pt 5):1005-1013. https://doi.org/10.1093/brain/awf095

Drosophila enhancer of Zeste/ESC complexes have a histone H3 methyltransferase activity that marks chromosomal Polycomb sites. Czermin B, Melfi R, McCabe D, Seitz V, Imhof A, Pirrotta V. *Cell*. 2002;111(2):185-196. doi:10.1016/s0092-8674(02)00975-3

Conservation of the COP9/signalosome in budding yeast. Wee S, Hetfeld B, Dubiel W, **Wolf DA**. *BMC Genet*. 2002;3:15. doi:10.1186/1471-2156-3-15

The F-box protein SKP2 mediates androgen control of p27 stability in LNCaP human prostate cancer cells. Lu L, Schulz H, **Wolf DA**. *BMC Cell Biol*. 2002;3:22. doi:10.1186/1471-2121-3-22

Combinatorial diversity of fission yeast SCF ubiquitin ligases by homo- and heterooligomeric assemblies of the F-box proteins Pop1p and Pop2p. Seibert V, Prohl C, Schoultz I, Rhee E, Lopez R, Abderazzaq K, Zhou C, **Wolf DA**. *BMC Biochem*. 2002;3:22. doi:10.1186/1471-2091-3-22

2001

Thrombin activates the hypoxia-inducible factor-1 signaling pathway in vascular smooth muscle cells: Role of the p22(phox)-containing NADPH oxidase. Görlach A, **Diebold I**, Schini-Kerth VB, Berchner-Pfannschmidt U, Roth U, Brandes RP, Kietzmann T, Busse R. *Circ Res*. 2001;89(1):47-54. doi:10.1161/hh1301.092678

GTG banding pattern on human metaphase chromosomes revealed by high resolution atomic-force microscopy. Thalhammer S, **Koehler U**, Stark RW, Heckl WM. *J Microsc*. 2001;202(Pt 3):464-467. doi:10.1046/j.1365-2818.2001.00909.x

Frequency of mitochondrial transfer RNA mutations and deletions in 225 patients presenting with respiratory chain deficiencies. Jaksch M, **Kleinle S**, Scharfe C, Klopstock T, Pongratz D, Müller-Höcker J, Gerbitz KD, Liechti-Gallati S, Lochmuller H, Horvath R. *J Med Genet*. 2001;38(10):665-673. doi:10.1136/jmg.38.10.665

Cell type-specific expression and promoter activity of human endogenous retroviral long terminal repeats. Schön U, Baust, C, Seifarth W, Hohenadl C, Erfle V and Leib-Mösch C. *Virology* (2001) 279(1): 280-289

The R-region of HERV-K-T47D related LTRs contains signals for modulation of HERV promoter activity. Baust, C., Seifarth, W., **Schön, U.,** Hehlmann, R. and Leib-Mösch, C. *Virology* (2001) 283(2): 262-272

Sixteen rare sequence variants of the hMLH1 and hMSH2 genes found in a cohort of 254 suspected HNPCC (hereditary non-polyposis colorectal cancer) patients: mutations or polymorphisms? Müller-Koch Y, Kopp R, Lohse P, Baretton G, Stoetzer A, Aust D, Daum J, Kerker B, Gross M, Dietmeier W, Holinski-Feder E. *Eur J Med Res*. 2001;6(11):473-482.

Publikationsliste

DHPLC mutation analysis of the hereditary nonpolyposis colon cancer (HNPCC) genes hMLH1 and hMSH2. Holinski-Feder E, **Müller-Koch Y**, Friedl W, Moeslein G, Keller G, Plaschke J, Ballhausen W, Gross M, Baldwin-Jedele K, Jungck M, Mangold E, Vogelsang H, Schackert HK, Lohsea P, Murken J, Meitinger T. *J Biochem Biophys Methods*. 2001;47(1-2):21-32. doi:10.1016/s0165-022x(00)00148-2

Deactivation and desensitization of mouse embryonic- and adult-type nicotinic receptor channel currents. Jahn K, Mohammadi B, Krampfl K, **Abicht A**, Lochmüller H, Bufler J. *Neurosci Lett*. 2001;307(2):89-92. https://doi.org/10.1016/s0304-3940(01)01929-2

Physical and functional association of SU(VAR)3-9 and HDAC1 in Drosophila. Czermin B, Schotta G, Hulsmann BB, Brehm A, Becker PB, Reuter G, Imhof A. *EMBO Rep*. 2001;2(10):915-919. doi:10.1093/embo-reports/kve210

Promotion of NEDD8-CUL1 Conjugate Cleavage by COP9 Signalosome. Lyapina S. *Science*. 2001;292(5520):1382-1385. doi:10.1126/science.1059780

The fission yeast COP9/signalosome is involved in cullin modification by ubiquitin-related Ned8p. Zhou C, Seibert V, Geyer R, Rhee E, Lyapina S, Cope G, Deshaies RJ, **Wolf DA**. *BMC Biochem*. 2001;2:7. doi:10.1186/1471-2091-2-7

2000

Video technology--a medium for milieu therapy?. Wormstall H, Günthner A, Balg S, Schwärzler F. *Psychiatr Prax*. 2000;27(5):235-238.

A second case of inv(4)pat with both recombinants in the offspring: rec dup(4q) in a girl with Wolf-Hirschhorn syndrome and rec dup(4p). Dufke A, Eggermann K, Balg S, Stengel-Rutkowski S, Enders H, Kaiser P2000;91(1-4):85-89. doi:10.1159/000056824

Mitochondrial diseases represent a risk factor for valproate-induced fulminant liver failure. Krähenbühl S, Brandner S, **Kleinle S**, Liechti S, Straumann D. *Liver*. 2000;20(4):346-348. doi:10.1034/j.1600-0676.2000.020004346.x

Hyperventilation due to mitochondrial myopathy. Moosmann P, Brandner S, **Kleinle S**, Frauchiger B. *J R Soc Med*. 2000;93(1):25-26. doi:10.1177/014107680009300108

HERV-IP-T47D, a novel type C-related human endogenous retroviral sequence derived from T47D particles. Seifarth, W., Baust, C., **Schön, U.,** Reichert, A., Hehlmann, R. and Leib-Mösch, C., *AIDS Res Hum Retroviruses* 16, 471-80

Retroviral vectors based on human endogenous LTR sequences, Schön, U., Seifarth, W., Baust, C., Erfle, V. and Leib-Mösch, C. J. Gene Medicine 2 (2000) , 54

Analysis of human endogenous retrovirus activity in normal and malignant tissues of patients with breast cancer. 1. Schön, U., Seifarth, W., Baust, C., Erfle, V. and Leib-Mösch, C. (2000) Seifarth, W., Schön, U., Krause, U.,Schwarz, N., Verbeke, C., Hehlmann, R. and Leib-Mösch, C. *Infection* 28, (2000) 53

Technology evaluation: edrecolomab, Centocor Inc. Abicht A, Lochmüller H. *Curr Opin Mol Ther*. 2000;2(5):593-600.

A modified alignment of human and rodent 5' untranslated sequences of the acetylcholine receptor epsilon subunit gene reveals additional regions of high homology. Stucka R, **Abicht A**, Song IH, Bönsch D, Deufel T, Lochmüller H. *Neuromuscul Disord*. 2000;10(3):213-214. https://doi.org/10.1016/s0960-8966(99)00112-1

Leber's hereditary optic neuropathy presenting as multiple sclerosis-like disease of the CNS. Horváth R, **Abicht A**, Shoubridge EA, Karcagi V, Rózsa C, Komoly S, Lochmüller H. *J Neurol*. 2000;247(1):65-67. https://doi.org/10.1007/s004150050015

1999

Microvesicular steatosis, hemosiderosis and rapid development of liver cirrhosis in a patient with Pearson's syndrome. Krähenbühl S, **Kleinle S**, Henz S, Leibundgut K, Liechti S, Zimmermann A, Wiesmann U. *J Hepatol*. 1999;31(3):550-555. doi:10.1016/s0168-8278(99)80050-6

Construction of tissue specific retroviral vectors using HERV LTRs. Schön, U., Erfle, V. and Leib-Mösch, C. J. *Gene Medicine* 1, (1999) 12

Publikationsliste

A common mutation (epsilon1267delG) in congenital myasthenic patients of Gypsy ethnic origin. Abicht A, Stucka R, Karcagi V, Herczegfalvi A, Horváth R, Mortier W, Schara U, Ramaekers V, Jost W, Brunner J, Janssen G, Seidel U, Schlotter B, Müller-Felber W, Pongratz D, Rüdel R, Lochmüller H. *Neurology*. 1999;53(7):1564-1569. https://doi.org/10.1212/wnl.53.7.1564

Technology evaluation: CRIB (CNTF delivery) CytoTherapeutics Inc. Abicht A, Lochmüller H. *Curr Opin Mol Ther*. 1999;1(5):645-650.

Localization of transforming growth factor beta in association with neuromuscular junctions in adult human muscle. Toepfer M, Fischer P, **Abicht A**, Lochmüller H, Pongratz D, Müller-Felber W. *Cell Mol Neurobiol*. 1999;19(2):297-300. https://doi.org/10.1023/a:1006989530148

Budding yeast Cdc6p induces re-replication in fission yeast by inhibition of SCFPop-mediated proteolysis. **Wolf DA**, McKeon F, Jackson PK. *Mol Gen Genet*. 1999;262(3):473-480. doi:10.1007/s004380051108

F-box/WD-repeat proteins Pop1p and Sud1p/Pop2p form complexes that bind and direct the proteolysis of Cdc18p. **Wolf DA**, McKeon F, Jackson PK. *Current Biology*. 1999;9(7):373-377. doi:10.1016/S0960-9822(99)80165-1

Tumor Necrosis Factor-α Contributes to Ischemia- and Reperfusion-Induced Endothelial Activation in Isolated Hearts. Kupatt C, Habazettl H, Goedecke A, **Wolf DA**, Zahler S, Boekstegers P, Kelly RA, Becker BF. *Circulation Research*. 1999;84(4):392-400. doi:10.1161/01.RES.84.4.392

Regulation of c-myc and immunoglobulin kappa gene transcription by promoter-proximal pausing of RNA polymerase II. Schneider EE, Albert T, **Wolf DA**, Eick D. *Curr Top Microbiol Immunol*. 1999;246:225-231. doi:10.1007/978-3-642-60162-0_28

1998

In situ hybridization (FISH) maps chromosomal homologies between Alouatta belzebul (Platyrrhini, Cebidae) and other primates and reveals extensive interchromosomal rearrangements between howler monkey genomes. Consigliere S, Stanyon R, **Koehler U**, Arnold N, Wienberg J. *Am J Primatol*. 1998;46(2):119-133. doi:10.1002/(SICI)1098-2345(1998)46:2<119::AID-AJP2>3.0.CO;2-Z

A systematic mutation screen of 10 nuclear and 25 mitochondrial candidate genes in 21 patients with cytochrome c oxidase (COX) deficiency shows tRNA(Ser)(UCN) mutations in a subgroup with syndromal encephalopathy. Jaksch M, Hofmann S, **Kleinle S**, Liechti-Gallati S, Pongratz DE, Müller-Höcker J, Jedele KB, Meitinger T, Gerbitz KD. *J Med Genet*. 1998;35(11):895-900. doi:10.1136/jmg.35.11.895

Progressive myoclonus epilepsy and mitochondrial myopathy associated with mutations in the tRNA(Ser(UCN)) gene. Jaksch M, Klopstock T, Kurlemann G, Dörner M, Hofmann S, **Kleinle S**, Hegemann S, Weissert M, Müller-Höcker J, Pongratz D, Gerbitz KD. *Ann Neurol*. 1998;44(4):635-640. doi:10.1002/ana.410440409

A novel mitochondrial tRNA(Phe) mutation inhibiting anticodon stem formation associated with a muscle disease. Kleinle S, Schneider V, Moosmann P, Brandner S, Krähenbühl S, Liechti-Gallati S. *Biochem Biophys Res Commun*. 1998;247(1):112-115. doi:10.1006/bbrc.1998.8729

Myotonic ADR-MDX mutant mice show less severe muscular dystrophy than MDX mice. Krämer R, Lochmüller H, **Abicht A**, Rüdel R, Brinkmeier H. *Neuromuscul Disord*. 1998;8(8):542-550. https://doi.org/10.1016/s0960-8966(98)00078-9

Cell cycle: Oiling the gears of anaphase. **Wolf DA**, Jackson PK. *Current Biology*. 1998;8(18):R636-R639. doi:10.1016/S0960-9822(07)00410-1

1997

Fluorescene in situ hybridization establishes homology between human and silvered leaf monkey chromosomes, reveals reciprocal translocations between chromosomes homologous to human Y/5, 1/9, and 6/16, and delineates an X1X2Y1Y2/X1X1X2X2 sex-chromosome system. Bigoni F, **Koehler U**, Stanyon R, Ishida T, Wienberg J. *Am J Phys Anthropol*. 1997;102(3):315-327. doi:10.1002/(SICI)1096-8644(199703)102:3<315::AID-AJPA2>3.0.CO;2-U

Mapping homology between human and black and white colobine monkey chromosomes by fluorescent in situ hybridization. Bigoni F, Stanyon R, **Koehler U**, Morescalchi AM, Wienberg J. *Am J Primatol*. 1997;42(4):289-298. doi:10.1002/(SICI)1098-2345(1997)42:4<289::AID-AJP4>3.0.CO;2-T

Publikationsliste

Detection and characterization of mitochondrial DNA rearrangements in Pearson and Kearns-Sayre syndromes by long PCR. Kleinle S, Wiesmann U, Superti-Furga A, Krähenbühl S, Boltshauser E, Reichen J, Liechti-Gallati S. *Hum Genet*. 1997;100(5-6):643-650. doi:10.1007/s004390050567

Tissue specifi expression and promoter activity of different HERV-LTRs. Schön, U., Baust, C., Hohenadl, C., Erfle, V., Hehlmann, R. and Leib-Mösch, C. *J. Mol. Med.* 75 (1997), 228

Congenital myasthenic syndromes: clinical and genetic analysis of 18 patients. Abicht A, Müller-Felber W, Fischer P, Jakob I, Kürz L, Rudel R, Mortier W, Pongratz D, Lochmüller H. *Eur J Med Res*. 1997;2(12):515-522

1996

Craniosynostosis suggestive of Saethre-Chotzen syndrome: clinical description of a large kindred and exclusion of candidate regions on 7p. von Gernet S, Schuffenhauer S, Golla A, Lichtner P, Balg S, Mühlbauer W, Murken J, Fairley J, Meitinger T. *Am J Med Genet*. 1996;63(1):177-184. doi:10.1002/(SICI)1096-8628(19960503)63:1<177::AID-AJMG31>3.0.CO;2-J

Analysis of the organisation and localisation of the FSHD-associated tandem array in primates: implications for the origin and evolution of the 3.3 kb repeat family. Clark LN, **Koehler U**, Ward DC, Wienberg J, Hewitt JE. *Chromosoma*. 1996;105(3):180-189. doi:10.1007/BF02509499

Chromosome painting defines genomic rearrangements between red howler monkey subspecies. Consigliere S, Stanyon R, **Koehler U**, Agoramoorthy G, Wienberg J. *Chromosome Res*. 1996;4(4):264-270. doi:10.1007/BF02263675

Comparative fluorescence in situ hybridization mapping of primate chromosomes with Alu polymerase chain reaction generated probes from human/rodent somatic cell hybrids. Müller S, **Koehler U**, Weinberg J, Marzella R, Finelli P, Antonacci R, Rocchi M, Archidiacono N. *Chromosome Res*. 1996;4(1):38-42. doi:10.1007/BF02254943

Combined 3-Methylglutaconic and 3-Hydroxy-3-Methylglutaric Aciduria with EndocardialFibroelastosis and Dilatative Cardiomyopathy inMale and Female Siblings with Partial Deficiencyof Complex II/III in Fibroblasts. Ruesch S, Krähenbühl S, **Kleinle S**, Liechti-Gallati S, Schaffner T, Wermuth B, Weber J, Wiesmann UN. *Enzyme Protein*. 1996;49(5-6):321-329. doi:10.1159/000468642

Cloning and sequencing of the dnaK operon of B. stearothermophilus. Herbort, M., Schön, U., Angermann, K., Lang, J. and Schumann, W. *FEMS-Microbiol-Lett.* 139 (1996), 183-188

Disruption of Re-replication Control by Overexpression of Human *ORC1* in Fission Yeast. **Wolf DA**, Wu D, McKeon F. *J Biol Chem*. 1996;271(51):32503-32506. doi:10.1074/jbc.271.51.32503

1995

Genomic reorganization in the concolor gibbon (Hylobates concolor) revealed by chromosome painting. Koehler U, Bigoni F, Wienberg J, Stanyon R. *Genomics*. 1995;30(2):287-292. doi:10.1006/geno.1995.9875

Genomic reorganization and disrupted chromosomal synteny in the siamang (Hylobates syndactylus) revealed by fluorescence in situ hybridization. Koehler U, Arnold N, Wienberg J, Tofanelli S, Stanyon R. *Am J Phys Anthropol*. 1995;97(1):37-47. doi:10.1002/ajpa.1330970104

Chromosomal painting shows that "marked chromosomes" in lesser apes and Old World monkeys are not homologous and evolved by convergence. Stanyon R, Arnold N, **Koehler U**, Bigoni F, Wienberg J. *Cytogenet Cell Genet*. 1995;68(1-2):74-78. doi:10.1159/000133894

Mulibrey nanism. Balg S, Stengel-Rutkowski S, Döhlemann C, Boergen K. *Clin Dysmorphol*. 1995;4(1):63-69.

A novel type of unstable homogeneously staining region with a head-to-tail arrangement: spontaneous decay and reintegration of DNA elements into a plethora of new chromosomal sites. Koehler U, Abken H, Grummt F, Wienberg J, Weidle UH. *Cytogenet Cell Genet*. 1995;68(1-2):33-38. doi:10.1159/000133883

Distinct mouse DNA sequences enable establishment and persistence of plasmid DNA polymers in mouse cells. Zastrow G, **Koehler U**, Müller F, Klavinius A, Wegner M, Wienberg J, Weidle UH, Grummt F. *Nucleic Acids Res*. 1989;17(5):1867-1879. doi:10.1093/nar/17.5.1867

Overproduction, purification and characterization of GroES and GroEL from the thermophilic B. stearothermophilus. **Schön, U.** and Schumann, W. (1995) *Gene* 170, 81-84

Ultraviolet B irradiation-induced G2 cell cycle arrest in human keratinocytes by inhibitory phosphorylation of the cdc2 cell cycle kinase. Herzinger T, Funk JO, Hillmer K, Eick D, **Wolf DA**, Kind P. *Oncogene*. 1995;11(10):2151-2156.

Publikationsliste

Variable pause positions of RNA polymerase II lie proximal to the c-myc promoter irrespective of transcriptional activity. **Wolf DA**, Strobl LJ, Pullner A, Eick D. *Nucleic Acids Res*. 1995;23(17):3373-3379. doi:10.1093/nar/23.17.3373

A complex between E2F and the pRb-related protein p130 is specifically targeted by the simian virus 40 large T antigen during cell transformation. **Wolf DA**, Hermeking H, Albert T, Herzinger T, Kind P, Eick D. *Oncogene*. 1995;10(11):2067-2078.

The pRb-related protein p130 is a possible effector of transforming growth factor beta 1 induced cell cycle arrest in keratinocytes. Herzinger T, **Wolf DA**, Eick D, Kind P. *Oncogene*. 1995;10(11):2079-2084.

The role of immunoglobulin kappa elements in c-myc activation. Hörtnagel K, Mautner J, Strobl LJ, **Wolf DA**, Christoph B, Geltinger C, Polack A. *Oncogene*. 1995;10(7):1393-1401.

1994

Construction of His6-tagging vectors allowing single-step purification of GroES and other polypeptides produced in Bacillus subtilis. Schön, U. and Schumann, W. *Gene 147,* (1994) 91-94

Role of c-myc in simian virus 40 large tumor antigen-induced DNA synthesis in quiescent 3T3-L1 mouse fibroblasts. Hermeking H, **Wolf DA**, Kohlhuber F, Dickmanns A, Billaud M, Fanning E, Eick D. *Proc Natl Acad Sci U S A*. 1994;91(22):10412-10416. doi:10.1073/pnas.91.22.10412

Activation of pausing RNA polymerases by nuclear run-on experiments. Eick D, Kohlhuber F, **Wolf DA**, Strobl LJ. *Anal Biochem*. 1994;218(2):347-351. doi:10.1006/abio.1994.1190

1993

Sequencing and characterization of the groE-locus from Bacillus stearothermophilus. Schön, U. and Schumann, W. *BioEngineering.* 9, (1993) 28

Molecular Cloning, Sequencing, and Transcriptional Analysis of the groESL Operon from Bacillus stearothermophilus. Schön, U. and Schumann, W. J. *Bacteriol*. 175, (1993) 2465-2469

Congenital Myasthenic Syndromes. Abicht A, Müller JS, Lochmüller H. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJ, Stephens K, Amemiya A, eds. *GeneReviews(®)*. University of Washington, Seattle; 1993.

Transcriptional and posttranscriptional regulation of human androgen receptor expression by androgen. **Wolf DA**, Herzinger T, Hermeking H, Blaschke D, Hörz W. *Mol Endocrinol*. 1993;7(7):924-936. doi:10.1210/mend.7.7.8413317

1992

Isolation and sequenzing of the groE-homolog of B. stearothermophilus. **Schön, U.** and Schumann, W. *BioEngineering*. 8, (1992) 73

Transcriptional regulation of prostate kallikrein-like genes by androgen. **Wolf DA**, Schulz P, Fittler F. *Mol Endocrinol*. 1992;6(5):753-762. doi:10.1210/mend.6.5.1376410

Transcriptional down-regulation of c-myc in human prostate carcinoma cells by the synthetic androgen mibolerone. **Wolf DA**, Kohlhuber F, Schulz P, Fittler F, Eick D. *Br J Cancer*. 1992;65(3):376-382. doi:10.1038/bjc.1992.76

1991

Synthetic androgens suppress the transformed phenotype in the human prostate carcinoma cell line LNCaP. **Wolf DA**, Schulz P, Fittler F. *Br J Cancer*. 1991;64(1):47-53. doi:10.1038/bjc.1991.237