Publikationsliste
Dr. rer. nat. Andreas Laner

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Medizinisch Genetisches Zentrum

## 2026

**Validation structures for sequence variants of uncertain significance in hereditary cancer.**
Lucas MC, Keßler T, Benet-Pagès A, Holinski-Feder E, Laner A, Klink B.
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**Mutational Landscape of Colorectal Tumors From Individuals With Unexplained Adenomatous or Serrated Colorectal Polyposis.**
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## 2025

**Improving genetic diagnosis of hereditary tumor syndromes: From expanded gene panels to functional genomics.**
Sauer M, Lucas MC, Prokosch V, Keßler T, Risch T, Laner A, Henkel J, Benet-Pagès A, Hallermayr A, Steinke-Lange V, Holinski-Feder E, Klink B.
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**Next-Generation Sequencing in Congenital Eye Malformations: Identification of Genetic Causes and Comparison of Different Panel-Based Diagnostic Strategies.**
Neuhann L, Laner A, Holinski-Feder E, Neuhann T.
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**From Expert Judgment to Structured Guidelines: A Brief History and Bright Future of DNA Variant Interpretation.**
Laner A, Zilfalil BA, Hamed SA, Do H, Lopes-Cendes I, Shrestha T, Tobias E, Solano A, Hamosh A, Kumar D.
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**A series of reviews in familial cancer: genetic cancer risk in context variants of uncertain significance in MMR genes: which procedures should be followed?**
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**Joint analysis of germline genetic data from over 29,000 cases with suspected hereditary breast and ovarian cancer (HBOC) as part of the NASGE initiative.**
Henkel J, Laner A, Locher M, Wohlfrom T, Neitzel B, Becker K, Neuhann T, Abicht A, Steinke-Lange V, Klink B, Eichhorn B, Schmidt W, Berner D, Teubert A, Holtorf A, Heinrich S, Wildhardt G, Schulze M, von der Heyden L, Hörtnagel K, Steinberger D, Kleier S, Lorenz P, Glaubitz R, Biskup S, Holinski-Feder E.
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**Blood biomarker fingerprints in a cohort of patients with CHRNE-related congenital myasthenic syndrome.**
Della Marina A, Koutsoulidou A, Natera-de Benito D, Tykocinski LO, Tomazou M, Georgiou K, Laner A, Kölbel H, Nascimento A, Ortez C, Abicht A, Thakur BK, Lochmüller H, Phylactou LA, Ruck T, Schara-Schmidt U, Kale D, Hentschel A, Roos A.
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**Reclassification of VUS in *BRCA1* and *BRCA2* using the new *BRCA1/ BRCA2* ENIGMA track set demonstrates the superiority of ClinGen ENIGMA Expert Panel specifications over the standard ACMG/AMP classification system.**
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## 2024

**Large-scale application of ClinGen-InSiGHT APC-specific ACMG/AMP variant classification criteria leads to substantial reduction in VUS.**
Yin X, Richardson M, Laner A, Shi X, Ognedal E, Vasta V, Hansen TVO, Pineda M, Ritter D, de Dunnen J, Hassanin E, Lin WL, Borras E, Krahn K, Nordling M, Martins A, Mahmood K, Nadeau E, Beshay V, Tops C, Genuardi M, Pesaran T, Frayling IM, Capellá G, Latchford A, Tavtigian SV, Maj C, Plon SE, Greenblatt MS, Macrae FA, Spier I, Aretz S.
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**Comprehensive reanalysis for CNVs in ES data from unsolved rare disease cases results in new diagnoses.**
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**Comparison of the ABC and ACMG systems for variant classification.**
Houge G, Bratland E, Aukrust I, Tveten K, Žukauskaitė G, Sansovic I, Brea-Fernández AJ, Mayer K, Paakkola T, McKenna C, Wright W, Markovic MK, Lildballe DL, Konecny M, Smol T, Alhopuro P, Gouttenoire EA, Obeid K, Todorova A, Jankovic M, Lubieniecka JM, Stojiljkovic M, Buisine MP, Haukanes BI, Lorans M, Roomere H, Petit FM, Haanpää MK, Beneteau C, Pérez B, Plaseska-Karanfilska D, Rath M, Fuhrmann N, Ferreira BI, Stephanou C, Sjursen W, Maver A, Rouzier C, Chirita-Emandi A, Gonçalves J, Kuek WCD, Broly M, Haer-Wigman L, Thong MK, Tae SK, Hyblova M, den Dunnen JT, Laner A.
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**Mismatch repair gene specifications to the ACMG/AMP classification criteria: Consensus recommendations from the InSiGHT ClinGen Hereditary Colorectal Cancer / Polyposis Variant Curation Expert Panel.**
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**Systematic large-scale application of ClinGen InSiGHT APC-specific ACMG/AMP variant classification criteria substantially alleviates the burden of variants of uncertain significance in ClinVar and LOVD databases.**
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**Novel Genetic and Biochemical Insights into the Spectrum of NEFL-Associated Phenotypes.**
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**Gene-specific ACMG/AMP classification criteria for germline APC variants: recommendations from the ClinGen InSiGHT Hereditary Colorectal Cancer / Polyposis Variant Curation Expert Panel.**
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**Novel Homozygous FA2H Variant Causing the Full Spectrum of Fatty Acid Hydroxylase-Associated Neurodegeneration (SPG35).**
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**Diagnostic yield and clinical relevance of expanded germline genetic testing for nearly 7000 suspected HBOC patients**
Jan Henkel, Andreas Laner, Melanie Locher, Tobias Wohlfrom, Birgit Neitzel, Kerstin Becker, Teresa Neuhann, Angela Abicht, Verena Steinke-Lange, Elke Holinski-Feder
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**Highly sensitive liquid biopsy Duplex sequencing complements tissue biopsy to enhance detection of clinically relevant genetic variants.**
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**Transcript capture and ultradeep long-read RNA sequencing (CAPLRseq) to diagnose HNPCC/Lynch syndrome.**
Schwenk V, Leal Silva RM, Scharf F, Knaust K, Wendlandt M, Häusser T, Pickl JMA, Steinke-Lange V, Laner A, Morak M, Holinski-Feder E, Wolf DA.
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**Biallelic PRMT7 pathogenic variants are associated with a recognizable syndromic neurodevelopmental disorder with short stature, obesity, and craniofacial and digital abnormalities.**
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**Long-term chemoprevention in patients with adenomatous polyposis coli: an observational study**
Teresa M Neuhann, Katharina Haub, Verena Steinke-Lange, Monika Morak, Andreas Laner, Melanie Locher, Elke Holinski-Feder
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**Constitutional chromothripsis of the *APC* locus as a cause of genetic predisposition to colon cancer**
Florentine Scharf, Rafaela Magalhaes Leal Silva, Monika Morak, Alex Hastie, Julia M A Pickl, Kai

Sendelbach, Christian Gebhard, Melanie Locher, Andreas Laner, Verena Steinke-Lange, Udo Koehler, Elke Holinski-Feder, Dieter A Wolf
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**Human Mutation special issue on "Variant Effect Prediction"**
Andreas Laner, Ales Maver, Johan T den Dunnen
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**Splicing analyses for variants in MMR genes: best practice recommendations from the European Mismatch Repair Working Group**
Monika Morak, Marta Pineda, Alexandra Martins, Pascaline Gaildrat, Hélène Tubeuf, Aurélie Drouet, Carolina Gómez, Estela Dámaso, Kerstin Schaefer, Verena Steinke-Lange, Udo Koehler, Andreas Laner, Julie Hauchard , Karine Chauris, Elke Holinski-Feder, Gabriel Capellá
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**Long-term chemoprevention in patients with adenomatous polyposis coli: an observational study**
Teresa M Neuhann, Katharina Haub, Verena Steinke-Lange , Monika Morak , Andreas Laner , Melanie Locher , Elke Holinski-Feder
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**Solving the genetic aetiology of hereditary gastrointestinal tumour syndromes- a collaborative multicentre endeavour within the project Solve-RD**
Anna K Sommer, Iris B A W Te Paske, José Garcia-Pelaez, Andreas Laner, Elke Holinski-Feder, Verena Steinke-Lange, Sophia Peters, Laura Valle, Isabel Spier, David Huntsman, Solve-RD-GENTURIS group; Carla Oliveira, Richarda M de Voer, Nicoline Hoogerbrugge, Stefan Aretz
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## 2021

**Solve-RD: systematic pan-European data sharing and collaborative analysis to solve rare diseases.**
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**A mosaic PIK3CA variant in a young adult with diffuse gastric cancer: case report.**
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**HPO-driven virtual gene panel: a new efficient approach in molecular autopsy of sudden unexplained death.**
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## 2020

**Analysis of 3297 individuals suggests that the pathogenic germline 5'-UTR variant BRCA1 c.-107A > T is not common in south-east Germany.**
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**Targeted deep-intronic sequencing in a cohort of unexplained cases of suspected Lynch syndrome.**
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**Loss of MSH2 and MSH6 due to heterozygous germline defects in MSH3 and MSH6.**
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