

2026

Repeat-associated ataxias in a German patient cohort analysed by targeted parallel long-read sequencing.

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Next-Generation Sequencing in Congenital Eye Malformations: Identification of Genetic Causes and Comparison of Different Panel-Based Diagnostic Strategies.

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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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Umfassende Charakterisierung des D4Z4-Repeatarrays mittels Long-Read-Sequenzierung für eine präzise Diagnostik der Fazioskapulohumeralen Muskeldystrophie.

Scharf F, Erdmann H, Lucas MC, Gehling S, Benet-Pagès A, Schäfer J, Hallermayr A, Schönrock V, Köhler U, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.

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Die Ränder des diagnostischen Spektrums der FSHD – Komplexe genetische Befunde in der Diagnostik der Fazioskapulohumeralen Muskeldystrophie (FSHD) und ihre Implikation für das molekulargenetische Modell der Erkrankung.

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Fassrainer DG, Klein W, Neuhann T, Nevinny-Stickel-Hinzpeter C.

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Updated Structure of CNBP Repeat Expansions in Patients With Myotonic Dystrophy Type 2 and Its Implication for Standard Diagnostics.
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Delineation of the adult phenotype of Coffin-Siris syndrome in 35 individuals.

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[Human genetic diagnostics in hereditary eye diseases : What does the ophthalmologist need to know]

Teresa M Neuhann, Lukas Neuhann

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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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Parallel in-depth analysis of repeat expansions in ataxia patients by long-read sequencing.

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Methylation of the 4q35 D4Z4 repeat defines disease status in facioscapulohumeral muscular dystrophy

Hannes Erdmann, Florentine Scharf, Stefanie Gehling, Anna Benet-Pagès, Sibylle Jakubiczka, Kerstin Becker, Maria Seipelt, Felix Kleefeld, Karl Christian Knop, Eva Christina Prott, Miriam Hiebeler, Federica Montagnese, Dieter Gläser, Matthias Vorgerd, Tim Hagenacker, Maggie C Walter, Peter Reilich, Teresa Neuhann, Martin Zenker, Elke Holinski-Feder, Benedikt Schoser, Angela Abicht

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Case Report: DPM1-CDG: Novel Variant with Severe Phenotype and Literature Review

Hanna Lausmann, Martin Zacharias, Teresa M Neuhann, Melanie K Locher, Karl F Schettler

<https://pubmed.ncbi.nlm.nih.gov/35910228/>

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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Initial Clinical Experience with NIPT for Rare Autosomal Aneuploidies and Large Copy Number Variations

Thomas Harasim, Teresa Neuhann, Anne Behnecke, Miriam Stampfer, Elke Holinski-Feder, Angela Abicht

<https://doi.org/10.3390/jcm11020372>

2021

Elucidating the clinical spectrum and molecular basis of HYAL₂ deficiency

James Fasham, Siying Lin, Promita Ghosh, Francesca Clementina Radio, Emily G Farrow, Isabelle Thiffault, Jennifer Kussman, Dihong Zhou, Rick Hemming, Kenneth Zahka, Barry A Chioza, Lettie E Rawlins, Olivia K Wenger, Adam C Gunning, Simone Pizzi, Roberta Onesimo, Giuseppe Zampino, Emily Barker, Natasha Osawa, Megan Christine Rodriguez, Teresa M Neuhann, Elaine H Zackai, Beth Keena, Jenina Capasso, Alex V Levin, Elizabeth Bhoj, Dong Li, Hakon Hakonarson, Ingrid M Wentzensen, Adam Jackson, Kate E Chandler, Zeynep H Coban-Akdemir, Jennifer E Posey, Siddharth Banka, James R Lupski, Sarah E Sheppard, Marco Tartaglia, Barbara Triggs-Raine, Andrew H Crosby, Emma L Baple

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Mutation spectrum of the OPA1 gene in a large cohort of patients with suspected dominant optic atrophy: Identification and classification of 48 novel variants.

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