

2025

A series of reviews in familial cancer: genetic cancer risk in context variants of uncertain significance in MMR genes: which procedures should be followed?

Lucas MC, Keßler T, Scharf F, Steinke-Lange V, Klink B, Laner A, Holinski-Feder E.

<https://pubmed.ncbi.nlm.nih.gov/40317406/>

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.

Nervenheilkunde 2025; 44(03): 168-169. DOI: [10.1055/s-0044-1801528](https://doi.org/10.1055/s-0044-1801528)

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.

Erdmann H, Gehling S, Scharf F, Lucas MC, Kleefeld F, Becker K, Schönrock V, Saak A, Schäfer J, Neuhoff S, Hagenacker T, Pormann J, Rausch HW, Schirmer L, Berking AC, Neuhann T, Holinski-Feder E, Walter M, Schoser B, Abicht A.

Nervenheilkunde 2025; 44(03): 143. DOI: [10.1055/s-0044-1801447](https://doi.org/10.1055/s-0044-1801447)

2024

Optical Genome Mapping as a Potential Routine Clinical Diagnostic Method.

Barseghyan H, Eisenreich D, Lindt E, Wendlandt M, Scharf F, Benet-Pagès A, Sendelbach K, Neuhann T, Abicht A, Holinski-Feder E, Koehler U.

<https://pubmed.ncbi.nlm.nih.gov/38540401/>

2023

Reply: An epigenetic basis for genetic anticipation in facioscapulohumeral muscular dystrophy type 1.

Erdmann H, Scharf F, Hallermayr A, Barseghyan H, Walter MC, Holinski-Feder E, Schoser B, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/37348868/>

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.

<https://pubmed.ncbi.nlm.nih.gov/36593122/>

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

Erdmann H, Schöberl F, Giurgiu M, Leal Silva RM, Scholz V, Scharf F, Wendlandt M, Kleinle S, Deschauer M, Nübling G, Heide W, Babacan SS, Schneider C, Neuhann T, Hahn K, Schoser B, Holinski-Feder E, Wolf DA, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/36227727/>

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

Erdmann H, Scharf F, Gehling S, Benet-Pagès A, Jakubiczka S, Becker K, Seipelt M, Kleefeld F, Knop KC, Prott EC, Hiebeler M, Montagnese F, Gläser D, Vorgerd M, Hagenacker T, Walter MC, Reilich P, Neuhann T, Zenker M, Holinski-Feder E, Schoser B, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/36100962/>

Closing the Gap - Detection of 5q-Spinal Muscular Atrophy by Short-Read Next-Generation Sequencing and Unexpected Results in a Diagnostic Patient Cohort.

Kleinle S, Scholz V, Benet-Pagès A, Wohlfrom T, Gehling S, Scharf F, Rost S, Prott EC, Grinzinger S, Hotter A, Haug V, Niemeier S, Wiethoff-Ubrig L, Hagenacker T, Goldhahn K, von Moers A, Walter MC, Reilich P, Eggermann K, Kraft F, Kurth I, Erdmann H, Holinski-Feder E, Neuhann T, Abicht A.

<https://pubmed.ncbi.nlm.nih.gov/37424474/>

2022

Highly sensitive liquid biopsy Duplex sequencing complements tissue biopsy to enhance detection of clinically relevant genetic variants.

Hallermayr A, Neuhann TM, Steinke-Lange V, Scharf F, Laner A, Ewald R, Liesfeld B, Holinski-Feder E, Pickl JMA.
<https://pubmed.ncbi.nlm.nih.gov/36636551/>

Constitutional chromothripsis of the APC locus as a cause of genetic predisposition to colon cancer.

Scharf F, Leal Silva RM, Morak M, Hastie A, Pickl JMA, Sendelbach K, Gebhard C, Locher M, Laner A, Steinke-Lange V, Koehler U, Holinski-Feder E, Wolf DA.
<https://pubmed.ncbi.nlm.nih.gov/34911816/>

Somatic copy number alteration and fragmentation analysis in circulating tumor DNA for cancer screening and treatment monitoring in colorectal cancer patients.

Hallermayr A, Wohlfrom T, Steinke-Lange V, Benet-Pagès A, Scharf F, Heitzer E, Mansmann U, Haberl C, de Wit M, Vogelsang H, Rentsch M, Holinski-Feder E, Pickl JMA.
<https://pubmed.ncbi.nlm.nih.gov/36056434/>

2021

HPO-driven virtual gene panel: a new efficient approach in molecular autopsy of sudden unexplained death.

Schön U, Holzer A, Laner A, Kleinle S, Scharf F, Benet-Pagès A, Peschel O, Holinski-Feder E, Diebold I.
<https://pubmed.ncbi.nlm.nih.gov/33789662/>

2020

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.
<https://pubmed.ncbi.nlm.nih.gov/32048431/>

2018

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.
<https://pubmed.ncbi.nlm.nih.gov/30406974/>