

2026

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

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Parallel in-depth analysis of repeat expansions: an updated Clin-CATS workflow for nanopore R10 flow cells.

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bioRxiv 2024.11.05.622099; doi: <https://doi.org/10.1101/2024.11.05.622099>

Analysis and occurrence of biallelic pathogenic repeat expansions in RFC1 in a German cohort of patients with a main clinical phenotype of motor neuron disease.

Schaub A, Erdmann H, Scholz V, Timmer M, Cordts I, Günther R, Reilich P, Abicht A, Schöberl F.

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

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

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-Pagés 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.

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Sensory neuropathy due to RFC1 in a patient with ALS: more than a coincidence?

Schoeberl F, Abicht A, Kuepper C, Voelk S, Sonnenfeld S, Tonon M, Schaub A, Scholz V, Kleinle S, Erdmann H, Wolf DA, Reilich P.

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