CACNA1A-related disorders

https://pubmed.ncbi.nlm.nih.gov/35041321/

Four cases of autosomal dominant CNS disorders related to

CACNA1A

mutations and detected by massive parallel sequencing are reported: a non-familial case of episodic ataxia type 2 (EA2) with the previously reported mutation c.269_270insA (p.Tyr90Ter) in a 35-year-old man; familial hemiplegic migraine type 1 (FHM1) in a girl aged 3 years 10 months and her mother aged 38 yrs with a novel mutation 1829C>T (p.Ser610Phe), members of a family with 4 patients and incomplete penetrance; developmental and epileptic encephalopathy 42 (DEE42) in a 9-year-old girl and a 5-year-old boy from different families with the identical

de novo

mutation c.2137G>A (p.Ala713Thr) reported earlier. Clinical and genetic characteristics are analyzed compared to literature.