FGF-14 GAA intronic expansion in a cohoort of 37 unsolved late onset cerebellar ataxia brazilian patients.

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Abstract: About 1/3 of the patients with hereditary cerebellar ataxias do not have a molecular diagnosis, which is even more expressive in the subgroup of autosomal dominant cerebellar ataxias and late-onset sporadic cerebellar ataxias. Recently, Pellerin and collaborators, in a study with a population of European and Asian ancestry, demonstrated the presence of GAA expansion in intron 1 of the FGF14 gene as an important cause of late-onset autosomal dominant cerebellar ataxia. Thirty-seven patients from 3 different centers (southeast, south and north of the country), with progressive cerebellar ataxia, over 20 years of age, with an autosomal dominant or sporadic pattern of inheritance were selected. The FGF14 locus was amplified with long range polymerase chain reaction (PCR) in all patients for initial screening. Positive cases were then submitted to Repeat Primed PCR and Sanger sequencing for confirmation of GAA expansion. Five patients (13,5%) were identified with at least 250 GAA repeats in the FGF14 gene. Two patients have American ancestry, which was not described in the original study. FGF-14 related ataxia may be an important etiological cause of unsolved late onset cerebellar ataxia.