Psychometric properties of the Friedreich Ataxia Rating Scale

Objective To investigate the psychometric properties of the Friedreich Ataxia Rating Scale neurologic examination (FARSn) and its subscores, as well as the influence of the modifications resulting in the now widely used modified FARS (mFARS) examination.

Methods Based on cross-sectional FARS data from the FA–Clinical Outcome Measures cohort, we conducted correlation-based psychometric analyses to investigate the interplay of items and subscores within the FARSn/mFARS constructs.

Results The results provide support for both the FARSn and the mFARS constructs, as well as individually for their upper limb and lower limb coordination components. The omission of the peripheral nervous system subscore (D) and 2 items of the bulbar subscore (A) in the mFARS strengthens the overall construct compared with the complete FARS.

Conclusions A correlation-based psychometric analysis of the neurologic FARSn score justifies the overall validity of the scale. In addition, omission of items of limited functional significance as created in the mFARS improves the features of the measures. Such information is crucial to the ongoing application of the mFARS in natural history studies and clinical trials. Additional analyses of longitudinal changes will be necessary to fully ascertain its utility, especially in nonambulant patients.

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GNA11 brain somatic pathogenic variant in an individual with phacomatosis pigmentovascularis

Objective To describe the findings of histopathology and genotyping studies in affected brain tissue from an individual with phacomatosis pigmentovascularis (PPV).

Methods A retrospective chart review of a 2-year 10-month-old male with a clinical diagnosis of PPV cesionmarmorata (or type V) was performed. Clinical features, brain imaging and histopathology findings, and genotyping studies in his affected brain tissue are summarized.

Results The proband had a clinically severe neurologic phenotype characterized by global developmental delay, generalized hypotonia, and recurrent episodes of cardiac asystole in the setting of status epilepticus. A somatic pathogenic variant in GNA11 (c.547C>T, p.Arg183Cys) was detected in his skin tissue but not in blood (previously published). He underwent an urgent left posterior quadrantectomy for his life-threatening seizures. Histopathology of resected brain tissue showed an increase in leptomeningeal melanocytes and abnormal vasculature, and the exact pathogenic variant in GNA11 (c.547C>T, p.Arg183Cys), previously isolated from his skin tissue but not blood, was detected in his resected brain tissue.

Conclusions The finding of this variant in affected skin and brain tissue of our patient with PPV supports a unifying genetic diagnosis of his neurocutaneous features.

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