

Personality profile in SCA27B: a single case report

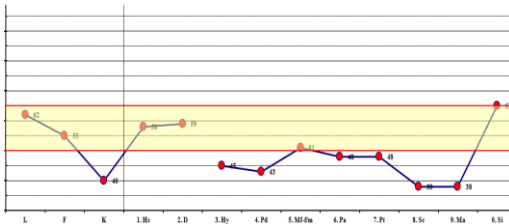
F.Falco, S.Molino; E.Morgillo, M.Migliaccio, A.De Cecca, C.Linguetta, R.De Rosa, V.Nicolella, M.Mocchia, M.Pellecchia, E.Salvatore, C.Criscuolo. (Napoli, Salerno).

Background: Spinocerebellar ataxia 27B (SCA27B) is an autosomal dominant ataxia caused by a GAA repeat expansion in intron 1 of the FGF14 gene. Identified in 2023 as one of the most common late-onset genetic ataxias, SCA27B is characterized by slow progression, down-beat nystagmus, and it is variably associated with psychiatric and/or neurological disorders. Cerebellum is involved not only in motor functioning, but also in cognition and social cognition. These aspects have been explored in the main forms of Spinocerebellar Ataxia (SCA), but never in SCA27B.

Objectives: In this study the aim was to evaluate the personality profile in a SCA27b patient.

Materials and Methods: The Minnesota Multiphasic Personality Inventory-2 (MMPI-II, Ghisi et al., 2006), and a battery to assess theory of mind and social cognition (Reading the mind in the Eyes test- Serafin e Surian., 2004, SET, Sartori's Social Intelligence Battery- Prior et al., 2003) were administered to a 76-year-old man with 13 years of education diagnosed with SCA27B.

Results: The MMPI-2 shows a personality profile characterized by high scores on the clinical scale of social introversion while Theory of mind skills are not impaired.



Discussion: SCA27B could present a personality profile characterized by extreme shyness, insecurity and reticence in social situations, low self-confidence. The social image is that of people who are difficult to know, emotionally cold and detached. They are sensitive to the opinions of others, to rejection and disapproval, and may complain about the lack of emotional involvement in relationships. They may present hyper-control, constrained affectivity, condescension and complacency in interpersonal relationships and an attitude of unconditional acceptance of authority. Subjects who score high are also described as serious, reliable and conventional; they also have difficulty making decisions, tend to worry and are usually irritable.

Conclusions: The study shows that the subject with SCA27B has a personality profile characterized by social introversion which however is not related to theory of mind skills, therefore the personality disorder highlighted does not seem to be related to cognitive deficits but psychological ones. The data from this study should be confirmed by future studies on a larger cohort of subjects with SCA27B.