SCA 17 presenting as a dominant choreic syndrome: a case report

A.Di Paolantonio MD, M. Petracca MD, A. Bentivoglio MD

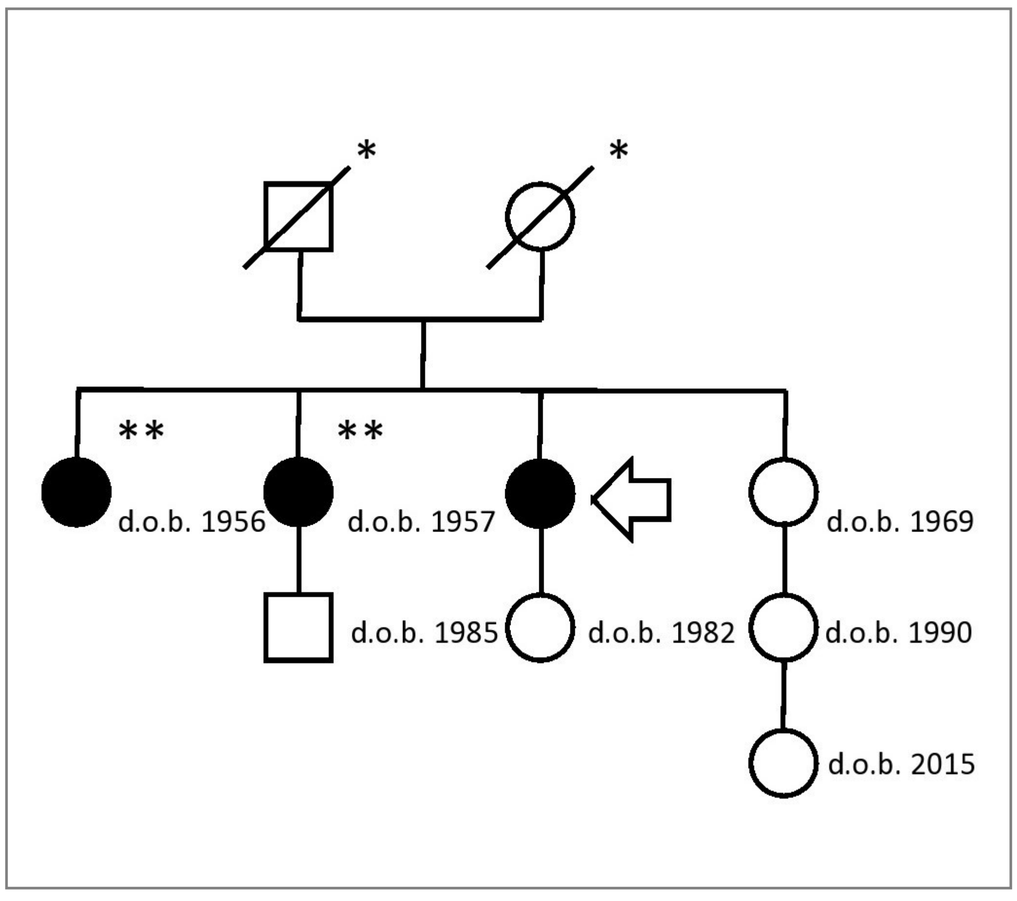
Institute of Neurology – Università Cattolica del Sacro Cuore, Rome

Introduction

SCA 17 is a neurodegenerative disease with dominant inheritance caused by the expansion of a CAG-CAA trinucleotide in the gene TBP. The gene codifies a RNA polymerase II transcription factor and contains a polyglutamine sequence ranging from 25 to 42 repetitions. More than 44 CAG/CAA triplets have been related to SCA 17. The symptomatology consists of cerebellar ataxia, signs of upper motoneuron, dementia and chorea. Some patients have a predominant HD-like syndrome, commonly associated with cerebellar signs. We describe a case of SCA 17 manifested with a dominant choreic syndrome associated to depression and cognitive impairment.

Materials and methods

A 58-year-old woman with a 10-year history of involuntary movements of limbs and face was admitted at our department. On neurological examination she had a severe impairment of saccadic initiation and movements, a mild slowing on fine hand dexterity and intermittent choreic movements of the entire body, involving also the gait and provoking an important impairment of gait and of tandem walking (UHDR scale score: 35). She also complained of depressive mood and her family referred perseveration of ideation and difficulties in management of money. Others two sisters of her had involuntary movements. MRI study identified a mild enlargement of lateral ventricles. Neuropsychological tests were performed on 2017, showing impairment of attention, working memory, executive functions and language (MMSE=20/30). The cognitive dysfunction of our patient was similar to those already described in literature.



Genealogical tree of our patient.

** Unknow state*

*** Involuntary movements referred by the proband.*

Genetic test not performed

d.o.b.: date of birth

Results

HD test was performed and was negative. A genetic panel for SCAs identified a 46 CAG/CAA expansion, diagnostic for SCA 17. Low doses of tetrabenazine were started, with marked reduction of involuntary movement, especially in limbs (UHDR scale score: 29).

Conclusions

Genetic test for SCA 17 should be performed in HD-like symptomatology. Especially in brief CAA-CAG trinucleotide expansions, cerebellar signs could be mild. Cognitive impairment is usually moderate-severe in these forms.
The good response to tetrabenazine in our patient suggests the utility of this drug in the control of choreic movement in SCA 17.

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