A case of Spinocerebellar Ataxia from ethnic tribe of Assam

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Abstract

Here we present the case of a 17-year-old girl belonging to an ethnic tribe (Bodo tribe) of Assam, presenting with bilateral cerebellar signs and with history suggestive of an autosomal dominant pattern of inheritance, who was found to have spinocerebellar ataxia 7 on genetic testing. This case throws light on the probability of more such cases in the multi-ethnic society of the North-Eastern Indian states, which are not studied or reported till date.

Key Words

Autosomal dominant, Spinocerebellar Ataxia, Inherited Ataxia,

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Introduction

Spinocerebellar ataxia (SCA) has a worldwide distribution, but some cases are more prevalent in one region than the other. SCA 2, SCA 3, and SCA6 appear to be the most common and together account for nearly half of all families worldwide. SCA 2 is typically the most common among the SCAs in India,[1] and the next most common SCA after SCA 3 worldwide. SCA type 7 (SCA7) is a rare autosomal dominant, late onset, slowly progressive disorder, primarily characterized by gradual loss of motor coordination, resulting from dysfunction and degeneration of the cerebellum and its connecting pathways. The disease is caused by expansion of a CAG trinucleotide repeat within the SCA7 gene on chromosome 3. This is the only SCA with prominent visual loss related to a maculopathy. SCA7 is the first of the neurodegenerative disorders caused by an expanded trinucleotide repeat in which the degenerative process also affects the retina. SCA 7 has rarely been reported from India. Further, no case of SCA has yet been reported from any ethnic tribe of any of the North-Eastern state of India. The detection of a case of SCA 7 amongst them may reveal interesting data for future studies.

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Case Report

Case

A 17-year-old unmarried girl from Bodo community (an ethnic community of Assam), resident of Barpeta district of Assam, born of nonconsanguineous marriage, with normal birth and developmental history, presented with imbalance while walking since around 2 years ago, with tendency to reel and fall in an unpredictable direction, along with in-coordination of both hands since same duration, causing difficulty in writing, eating, and operating appliances. The above symptoms were progressive in nature. Her medical history was otherwise unremarkable. There was no relevant past history. There was no history of exposure to alcohol, drugs, or toxins. There was history of similar symptoms in her mother and maternal uncle, suggestive of autosomal dominant inheritance; however, this could not be confirmed by molecular studies. Examination: She had normal vitals and general examination; mental status and speech were normal. Cranial nerves examination including fundii did not reveal any abnormality. Motor system examination revealed normal bulk and power, normal tone and deep tendon reflexes in all four limbs, and plantars bilateral flexor. All modalities of sensation were intact. There were cerebellar signs in both upper and lower limbs with ataxic gait.

Investigation

Normal routine blood, liver function test, renal function test, no K-F ring seen on slit lamp examination. MRI brain revealed cerebellar atrophy. On DNA-PCR testing for SCA by agarose gel electrophoresis: Detected CAG nucleotide repeat number 74 copies for SCA-7 [Figure 1].

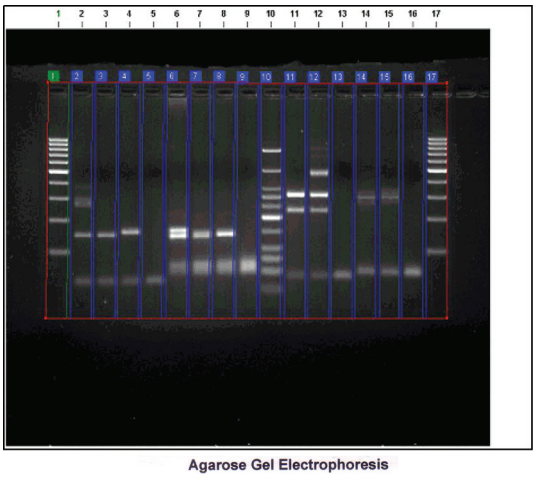

Figure 1: Gel picture of expanded repeats

Discussion

Diverse rates of SCA have been observed since molecular testing became available over the past decade. Whereas SCA7 is considered to be one of the most rare forms of genetically verified autosomal dominant cerebellar ataxia, it is the most frequent subtype in Sweden and Finland in a survey of hereditary ataxias in Scandinavia.[2] The first symptom is insidious, progressive visual loss caused by macular degeneration. Another early sign is slow saccades.[3] Gradually progressing cerebellar dysfunction and pyramidal signs develop. In early onset cases, visual loss may precede ataxia, and in later onset cases ataxia may precede visual loss. Subclinical visual impairment can be detected by a tritan axis defect found on the Farnsworth D15 color vision test or by electroretinogram. Anticipation is very common, and is greater in paternal than in maternal transmissions, and has a more rapid clinical course in successive generations.[4] A report of analysis of CAG repeat in SCA patients in nine ethnic populations of eastern India, did not detect any patient with expansion in the SCA 7 and DRPLA loci.[5] In a study of 235 Indian families with clinically diagnosed ataxia, only 2 were detected to have expansion at SCA 7 locus.[6] A recent study of clinical and molecular study of SCA has shown that SCA 12 was the most common cause of SCA in Agarwal ethnic population (56.6%) followed by SCA 2 (16.7%), SCA 3 (3%), SCA 7 (3%), and SCA due to unidentified gene (20%).[7] An analysis of CAG repeats in nine ethnic populations in eastern India revealed repeat expansion at SCA 1 locus 10.5%, SCA-2 locus 17.5%, SCA-3 locus 7%, SCA-6 locus 1.8%.[5] SCA-2 is probably the most common type of adult onset SCA seen in India, and it has considerable frequency in eastern India. SCA-3 is slightly more frequent in ethnic Bengali families.[8,9] In a study,[10] 12 families and a total of 21 patients were tested positive for SCA-12 locus, 11 of these belonged to a specific ethnic caste, the Agarwals.

Conclusions

Although SCA 2 is the most common SCA in India, due to the multi ethnicity and multi religious pattern of society other uncommon SCAs may also be detected in presently unreported populations of the country. The clinical phenotype suggestive of an SCA may sometimes vary on genetic testing.

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