Schizophrenia in a patient with Spinocerebellar Ataxia-6: a case report

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ABSTRACT

Spinocerebellar ataxias (SCAs) are a group of heterogeneous genetic disorders involving neurodegeneration of the cerebellum. Almost 30 distinct genetic causes of SCA are known, numbered chronologically in order of discovery. Here we present a case of schizophrenia in a 46 years old male diagnosed with SCA-6 with onset of psychotic symptoms 2 years after the onset of motor symptoms and treated with olanzapine. The purpose of this case report is to explore the relationship between schizophrenia and a neurodegenerative disease like SCA-6 and whether schizophrenia is a concomitant diagnosis or it is a part of the neurodegenerative process.

Key words: spinocerebellar ataxia, schizophrenia, neurodegenerative illness

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INTRODUCTION

SCAs are progressive neurodegenerative disorders primarily involving the cerebellum with additional involvement of the spinal cord, brainstem, basal ganglia and other parts of the nervous system. They are Autosomal Dominant disorders and 27 have been discovered till date with possibility of rarer variants [1]. SCAs display classic cerebellar signs along with the presence of disabling noncerebellar signs, most commonly brainstem dysfunction. Symptoms typically begin with gait imbalance followed by appendicular ataxia. Following this, dysarthria begins and visual problems can occur. SCA-6 which constitutes nearly 15% of SCAs typically occurring in the 40s-50s represents a milder form, most often manifesting as a pure cerebellar ataxia accompanied by dysarthria and gaze-evoked nystagmus. It is caused by a uniquely small CAG repeat expansion (19-30) in the CACNA1 gene encoding a voltage-dependent calcium channel alpha subunit [2]. Significant cognitive impairment among SCA patients has been noted in 25–50% of cases and depression in up to 30%. There exist case reports of psychosis co-occurring with SCA 2/3/7/10/17 but there is lack of such an occurrence with SCA-6 [2]. While the co-occurrence of both psychiatric and neurodegenerative conditions in our patient may be coincidental, it may also indicate to an underlying neurodegenerative process.

CASE REPORT

A 46 year old male was brought to our outpatient department by his wife with complaints of suspiciousness towards his neighbours, fearfulness and muttering to self since 2 months. On further interview the wife

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claimed that the symptoms started without any apparent stressor, with the patient expressing concerns regarding his neighbour's activities. He would claim that one of his neighbours was keeping a watch on him so as to kill him at the right moment. On being confronted, the patient would point to the newly installed dish antenna at their house which he claimed was used for spying on him. Gradually the patient stopped going out, interacting with friends or family, would ask the wife to keep the windows shut and doors bolted. He would further question the wife whether she was involved in their plans. On being questioned about the cause for spying he would claim that his neighbour was under the orders of some higher authority and carrying out their plans. He would further claim to hear voices of those people who would threaten him, often mocking him for not doing enough to save himself. The patient would consequently be seen talking to them back, perceived as self muttering by the wife. The patient would be firm on his belief and not be influenced by any reasoning. The symptoms worsened over the next 2 months with the patient isolating himself in a room, remaining awake throughout the night. The patient did not have any past history of similar complaints. No history of any other psychiatric illness or substance use in a dependent pattern was noted.

The wife revealed that the patient quit his job around 2 years back due to difficulty in maintaining balance, stiffness, repeated falls, and tremulousness of both hands which made it difficult for him to work on ship. The symptoms developed over a period of 6 months, with gradual progression following which Neurology opinion was taken. Magnetic Resonance Imaging (MRI) of the Brain revealed moderate cerebellar atrophy, most prominently in the superior vermis noted with no abnormal signal intensity and blood investigations including genetic testing were done leading to a diagnosis of SCA-6. Symptomatic management with Propranolol 40mg and Baclofen 30mg was given and the patient was explained about the prognosis of his illness. Apart from the motor symptoms, the patient was apparently alright till the onset of suspiciousness 2 months back.

He had no other, medical or surgical illness. In his family history, patient recollected his maternal grandfather of having similar symptoms of incoordination, loss of balance and dysarthria. On neurological examination, the patient had mild scanning dysarthria, dysmetria, and dysdiadochokinesia. His gait was wide-based and tandem walking was impaired. There was diminished joint position and vibration sense in his toes and fingers bilaterally. Deep tendon reflexes were absent throughout, and plantar responses were bilaterally flexor. His eye movements revealed nystagmus.

On Mental Status Examination, the patient was well oriented to time, place & person. His active attention was arousable but ill sustained and passive attention increased. He conveyed his mood as fearful and had a hyper vigilant affect. Speech was scanning, and thinking revealed persecutory and referential delusions. He further conveyed 2nd and 3rd person auditory hallucinations, which were continuous and threatening in nature. He had no memory impairments.

A provisional diagnosis of Paranoid Schizophrenia was made as per ICD10 and he was started on Tab. Olanzapine 5mg, which was gradually up-titrated to 15mg over the next 14 days. Presently the patient is well maintained on Olanzapine 15mg and denies suspiciousness or hallucinations and has good functional recovery. He has further started teaching arithmetic to a group of high school students at his home.

DISCUSSION

SCA-6 is an Autosomal Dominant disorder caused by a pathological expansion of a CAG repeat sequence on chromosome 19. It presents clinically as a pure cerebellar ataxia accompanied by dysarthria and gaze-evoked nystagmus. While most of the 27 types of SCAs reported to date involve some degree of neuropsychological deficit including depression and personality change, psychosis appears to be less common [3-4]. Although it is seen that SCA-17 may appear with psychosis with or without chorea, however, to our knowledge, there are no cases of SCA-6 in which psychosis was either the presenting symptom or a development during the course of the disease [5]. In this case hence we must ask whether SCA-6 is responsible for the development of psychotic symptoms or whether a concomitant diagnosis of schizophrenia is warranted. Neurodegenerative diseases which predominantly affect the striatum like Huntington's disease often have predominant psychiatric disturbances [6-7].

When psychiatric symptoms develop in cerebellar disorders, they are often associated with neurological symptoms suggestive of striatal involvement. This association suggests the possibility that psychosis in cerebellar disorders may actually be due to extra cerebellar pathology (striatal involvement). Alternately, the cerebellar atrophy seen in our patient may directly contribute to the psychotic features as cerebellar abnormalities have been implicated in various psychiatric disorders including psychosis, possibly by modulating the fronto-striatal circuits [8-10].

Points in favour of a concomitant diagnosis includes the symptomatology and course of illness, although the age of onset of symptoms were later than typically seen in schizophrenia. Thus, it is difficult to ascertain a relationship, if any, between a molecular diagnosis of SCA-6 and development of psychotic symptoms. Further study is warranted.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. The patient has consented for; his clinical information will be used to report in a journal. The patient also understands that his name and other initials will not be disclosed, and due efforts will be made to conceal his identity.

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