X-linked adrenoleukodystrophy presenting as attention deficit hyperactivity disorder

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ABSTRACT

X-linked adrenoleukodystrophy (X-ALD) is one of the leukodystrophies causing a progressive decline in neurological function mainly affecting the children. The most common symptoms are changes in behavior, including social withdrawal or aggression, poor memory or poor scholastic performance. Here, we present a 7-year-old boy who presented with symptoms of inattention and hyperactivity and later turned out to be a case of X-ALD.

Key words: Attention deficit hyperactivity disorder, X-linked adrenoleukodystrophy, Seizures

INTRODUCTION

Leukodystrophies are congenital demyelinating disorders causing damage to the myelin sheath, an insulating membrane that surrounds nerve cells in the brain. There are forty different types and adrenoleukodystrophy (ALD) is an X-linked inherited disorder with a prevalence of 1 in 20,000-50,000 individuals worldwide. This condition occurs with a similar frequency in all populations. People with X-linked adrenoleukodystrophy (X-ALD) accumulate high levels of saturated, very long chain fatty acids (VLCFA) in the brain and adrenal cortex.

The loss of myelin and the progressive dysfunction of the adrenal gland are the primary characteristics of X-ALD. While nearly all patients with X-ALD suffer from adrenal insufficiency, also known as Addison’s disease, the neurological symptoms can begin either in childhood or in adulthood.

The childhood cerebral form is the most severe, with onset between ages 4 and 10. The most common symptoms are usually behavioral changes such as abnormal withdrawal or aggression, poor memory, and poor school performance. Other symptoms include visual loss, learning disabilities, seizures, poorly articulated speech, difficulty swallowing, deafness, disturbances of gait and coordination, fatigue, intermittent vomiting, increased skin pigmentation and progressive dementia.

There are a couple of case reports in India on this disorder, but none relating the symptoms to attention deficit hyperactivity disorder (ADHD). We are presenting this interesting case of X-ALD, childhood onset, initially presenting to psychiatric outpatients department with features of ADHD.

CASE REPORT

A 7-year-old boy was brought to the psychiatry outpatient department by his mother. He was born to nonconsanguineous parents, full term normal delivery. Antenatal, intranatal and postnatal period was uneventful. His developmental milestones were normal. He started kinder garden schooling at the age of three. No concerns noted in the school or at home until 5 years of age.

When he progressed to year one in school, problems started. Teachers complained that he had been inattentive,
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doesn't sit at one place and disruptive in the class. His scholastic performance declined, and mother started having difficult time at home as well. He showed no interest in homework, couldn't remember what has been taught, more importantly started to forget what he has learnt in the past 2 years. e.g., nursery rhymes, alphabets, and numbers.

The teachers have advised consulting a psychiatrist and that he may need a special school for further education as his behavior was unmanageable. She however tried changing the schools twice before bringing him for consultation, but his behavior problems got worse.

During the assessment the boy was very impatient, constantly trying to leave the interview room, running up and down the stairs, distractible, and uncooperative. The assessment was done on two occasions, and neuropsychologist was involved who administered rating scales to screen and assess the severity of behavioral problems. After a comprehensive assessment, a diagnosis of ADHD was made. The boy had hyper-pigmented lesions in his face, but it was overlooked at that point. He was started on methylphenidate 5 mg initially and titrated to 10 mg. As his behavior was very disruptive, putting himself at risk of accidental harm, he was started on risperidone 0.5 mg.

**DISCUSSION**

Literature search has shown a couple of case reports from United States where ALD was misdiagnosed as ADHD. This is the first time we hear about this rare neurological condition presenting as ADHD in India. It never struck in our mind that symptoms of ADHD could be secondary to another neurological disorder in this boy given the typical nature and onset of the presentation. Furthermore, we overlooked the hyperpigmentation in the face initially until he was seen by endocrinologist after the MRI helped with the diagnosis.

The presence of facial hyperpigmentation and occurrence of tonic clonic seizures during the course of the illness should alert the clinicians look for these rare but important genetic cause.

We explained the course and prognosis of the condition to the parents and given appropriate psychological support. The boy was also referred to special education and occupational therapy as part of the treatment plan.

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