Mania with Aarskog-Scott Syndrome

RAGHAVENDRA B NAYAK, *L AMBIKA, GS BHOGALE AND A PANDURANGI
From the Departments of Psychiatry, JN Medical College, KLE University, Belgaum, Karnataka; and *Department of Oral Medicine and Radiology, School of Dental Sciences, Krishna Institute of Medical Sciences University, Karad, Maharashtra; India.

Correspondence to: Dr Raghavendra B Nayak, Assistant Professor, Department of Psychiatry, JN Medical College, KLE University, Belgaum 10, Karnataka, India. rbn.psych@gmail.com
Received: May 5, 2011; Initial review: June 13, 2011; Accepted: July 26, 2011.

Aarskog-Scott syndrome is transmitted as an X-linked trait and affects males. We report a 10-year-old boy presenting with complaints of increased temper tantrums, demanding behavior, grandiose ideas, overfamiliarity, abusive assaultive behavior and tobacco abuse. On examination, patient had most of the physical characteristics of Aarskog-Scott Syndrome. He also had global developmental delay and attention deficit hyperactivity disorder. This is the first case report of Aarskog Scott syndrome combined with mania.

Key words: ADHD, Aarskog-Scott Syndrome, Mania.

Aarskog-Scott syndrome is characterized by embryonic ocular hypertelorism, antverted nostrils, broad upper lip, cryptorchidism, sternal deformity, protruding umbilicus, pulmonary stenosis, ventricular septal defect, peculiar penoscrotal relations (‘saddle-bag scrotum’ or ‘shawl scrotum’) and ligamentous laxity manifest by hyperextensibility of the fingers, genu recurvatum, and flat feet. The neuropsychiatric manifestations include epilepsy, ADHD, mental retardation and Asperger’s syndrome [1].

CASE REPORT

A 10-year-old boy presented to the Department of Psychiatry with mild global developmental delay and behavioral problems for evaluation. Patient was the second of the two siblings born out of consanguineous marriage. Apart from developmental delay patient was found to be restless, not sitting at one place, impulsive behavior, difficulty in completing the given work, not able to wait for his turn while playing and while at queue. He also had temper tantrums which started from early childhood continuing till date. For two months prior to presentation, parents noticed increasing temper tantrums, demanding money, tobacco habit, ideas of buying more cattle, abusive and assaultive behavior, overfamiliarity, intrusive behavior, increased speech, irritable and ideas of grandiosity. His baseline laboratory investigations were within normal limits. X-ray imaging of skull and bilateral wrist, echocardiography did not reveal any abnormalities. Orthopantomogram revealed broad right central incisor and impacted left central incisor. The patient was screened with MINI-Kid (MINI International Neuropsychiatric Interview for Children and Adolescents English Version 5.0) for presence of psychiatric disorders. A diagnosis of first episode Mania with Aarskog-Scott Syndrome and mild Mental Retardation with Attention Deficit Hyperactivity Disorder (ADHD) was made. Initially patient was started with oral Olanzapine 2.5 mg/day and gradually dose was increased upto 10mg/day. Lorazepam was given to target agitation. Manic symptoms responded in two weeks with persistence of ADHD symptoms. Clonidine was started to target ADHD symptoms and patient was subsequently discharged from the hospital. After six months of regular therapy with olanzapine and clonidine, there was absence of manic symptoms and some improvement in ADHD symptoms. The improvement in ADHD symptoms in comparison to premorbid state were reported by his father. The IQ assessment of the patient was done after the control of Manic symptoms. His IQ was 60 and falls in the mild mental retardation category.

DISCUSSION

According to the Fryns [2] the incidence of mental handicap in Aarskog syndrome may be as high as 30%. Later studies tested this observation with clinically confirmed Aarskog syndrome and found their IQs to lie...
within the normal range. So, Aarskog syndrome may or may not be associated with mental handicap [1]. The behavioural abnormalities described mainly include ADHD [1] and none of the other specific psychiatric disorders are mentioned with this syndrome. ADHD may be more common in some of the genetic disorders, including Fragile X Syndrome, Neurofibromatosis 1, DiGeorge Syndrome, Tuberous Sclerosis Complex, Turner Syndrome, Williams Syndrome and Klinefelter Syndrome [3]. ADHD is also commonly associated with Aarskog-Scott syndrome [4]. Presence of ADHD with Aarskog-Scott syndrome might be due to common underlying genetic problem that is FGD1 mutation [1]. Genetic analysis was not performed in this case.

We can make three hypotheses for the presence of mania with Aarskog-Scott Syndrome in this case. First is that it could be related to common underlying genetics for Mania, ADHD and Aarskog-Scott Syndrome (X-linked inheritance or FGD1 gene). Second hypothesis is that mania may be related to the presence of ADHD. More than 90% of children with bipolar disorders will have comorbid ADHD [5,6]. Moreover, both bipolar disorder and ADHD were more likely to be diagnosed in boys than in girls. ADHD was more common in children and adolescents with childhood-onset mania than in patients with adolescent-onset mania [7]. Third hypothesis is that presence of mania could be coincidental with Aarskog-Scott Syndrome.

This case is reported because of its unique occurrence along with Mania.

Contributors: RBN: diagnosed the case and prepared the manuscript; AL: dental opinion, collecting the literature and revising the manuscript; GSB: case workup; AP: managing the case, follow up of the case and will act as the guarantor. The final manuscript was approved by all authors.

Funding: None; Competing interests: None stated.

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