Noonan syndrome with somnambulism: A rare case report

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Noonan syndrome is an autosomal dominant, genetic, multisystem disorder with a prevalence of 1 in 1000–2500 live births. Characteristic features of the condition include distinctive myopathic facial features, hypertelorism, short and broad nose, webbed neck, and low set ears. About 10% of the subjects have auditory defects due to sensorineural hearing loss. The patient also has short stature, chest deformity (superior pectus carinatum and inferior pectus excavatum), widely spaced nipples, and delayed puberty. A rare psychiatric manifestation of somnambulism and somniloquy in a case of Noonan syndrome is reported.

Noonan syndrome with a prevalence of one in 1000–2500 live births is an autosomal dominant, genetically heterogenous multisystem disorder. However, 60% of the cases are reported to be sporadic. Mutations that cause Noonan syndrome alter genes encoding proteins with roles in the RAS-MAPK pathway which is a well-studied, widely important signal transduction pathway through which extracellular ligands — such as some growth factors, cytokines, and hormones — stimulate cell proliferation. The syndrome is characterized by distinctive facial features in the form of hypertelorism, short and broad nose, webbed neck, low set ears, and 10% have auditory defects due to sensorineural hearing loss. There is a delay in puberty; the patient also has short stature, chest deformity (superior pectus carinatum and inferior pectus excavatum), and widely spaced nipples.

Although Noonan (1962) described nine patients with the condition, the initial description was probably by Kobylnski (1883). He reported a 20-year-old male with marked webbing of the neck. Funke (1902) reported a patient with a webbed neck, short stature, micromagnathia, and cubitus valgus, whereas Ullrich (1930) described an 8-year-old girl with similar features.

Noonan syndrome is the second most common syndromic cause of congenital heart disease such as pulmonary stenosis, hypertrophic obstructive cardiomyopathy, atrial septal defect, ventricular septal defect, aortic stenosis, mitral valve abnormalities, aortic coarctation, and coronary artery anomalies. It involves other systems and causes abnormal pigmentation, keratosis pilaris, 80% have unilateral or bilateral cryptorchidism, male gonadal dysfunction, renal anomalies, lymphatic malformations, bleeding problem due to coagulation defects, thrombocytopenia, and myeloproliferative disorder. Many orthopedic abnormalities

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such as cubitus valgus and genu valgum have also been reported.[6,8]

Many children suffering from Noonan syndrome has a significant difference in verbal and nonverbal IQ, they often have impaired social cognition, with difficulty in identifying emotions (alexithymia). They can even have language impairment with difficulty in reading and writing, poor memory, issues with executive functioning, and difficulties with social interaction. Some children can have comorbid autism, mood disturbances, and attention deficit hyperactivity disorder.[9-13] A case of Noonan syndrome with unique psychiatric feature of somnambulism is presented because of its rarity.

CASE REPORT

An 11-year-old boy, class 7th student, was brought to the psychiatry outpatient department by his mother with complaints of sleepwalking from the past 5 months and talking in sleep from the past 1 month. He was apparently alright 5 months back, after which he started getting up at night and would walk out of the door. He would wander aimlessly for few minutes to hours and return subsequently. Parents reported that during his sleepwalk, he would remain unexpressive and uncommunicative and used to become irritable and confused if he was forcibly awakened. The patient had complete amnesia of the event on arousal.

Events were episodic initially and used to occur once or twice in a month. However, from the past 3 months, these episodes are occurring almost every alternate night. The last episode was also associated with nocturnal enuresis. There was no history of any precipitating stressor (parental divorce, interpersonal, sibling issues, no bullying, no change in school environment, no history of sexual assault, or any other major psychological trauma).

There was a past history of tuberculosis of bone at the age of 1 year for which he was treated. There was no history of seizures, head trauma, or psychiatric illness in the past.

Family history was unremarkable. His birth and developmental history was uneventful. There was delayed walking due to weakness in the limb caused by tuberculosis. Secondary sexual characters were not yet developed. His scholastic performance was average.

Physical examination showed a body mass index of 18. The patient had low set ears, hypertelorism, broad and small nose, webbing of the neck was present, the left shoulder was higher set than the right shoulder, low set and widely spaced nipples, and dark patches were present on the forearm. Testes were not palpable [Figures 1 and 2]. Systemic examination revealed murmur on cardiovascular examination. Mental Status Examination showed that patient was well kempt and co-operative, with nonspontaneous speech and decreased content of speech with euthymic affect. He was worried about his health and performance in school. There were no obsessions, delusions or hallucinations. Memory, orientation, insight and judgment were intact.

Investigation

Ultrasoundgram of the abdomen revealed a left ectopic kidney, right undescended testis, and absent seminal vesicles.

Two-dimensional echocardiogram showed branch pulmonary artery stenosis.

Karyotyping showed 46 XY.

EEG and CT scan head were normal.

On Seguin form board test, IQ was 90 (lower range of normal).

On Wisconsin card sorting test, he successfully completed only two categories and made 65 correct responses including 63 errors. He failed to maintain set once, which occurred on the mid-trial of the test. Counting the number of items marked within “p” which denotes perseverative responses, a raw score of 32 was obtained of which 29 were circled, indicating that they were perseverative errors. He made 45 conceptual level responses across 128 trials. His standard scores and “t” scores range from 77 to 84 and 35 to 39, respectively, which indicate mildly impaired range. The normative data reveal that this score is equivalent to a percentile score of 16 under perseverative responses which falls under the below average range.

Toronto Alexithymia Scale-20 Score was 52 which indicated mild alexithymia.
He was treated with tablet mirtazapine 7.5 mg at night with satisfactory response. Parents were advised to take necessary precautions.

**DISCUSSION**

In children, somnambulism is a benign condition which outgrows in later childhood which is suggestive of delayed cerebral maturation. Earlier sleepwalking was even considered a part of hysterical fugue; however, it is apparent now that sleepwalking rests on the abnormality of the sleep mechanism of the brain and represents partial arousal of the deep NREM stages 3 and 4. This is the first case ever reported where the patient primarily presented for sleepwalking and sleep talking on further evaluation and referral a diagnosis of Noonan syndrome was given.

The available literature suggests that the children with Noonan syndrome present with few psychiatric manifestations as mentioned above. Our patient also had mild executive dysfunction, mild alexithymia, and displayed difficulty in having communication and making relations with peers.

As Noonan syndrome is a multisystem disorder and somnambulism a disorder of delayed cerebral maturation, there could be a possible relation among the two which requires further evaluation. Further cases or studies might show some correlation between sleep disorder and Noonan syndrome in the future.

**Declaration of patient consent**

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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**Conflicts of interest**

There are no conflicts of interest.

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