

CASE REPORT

NOONAN SYNDROME: AN EARLY DIAGNOSIS

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ABSTRACT: Noonan syndrome is a genetic multisystem disorder characterised by facial dysmorphism, learning difficulties, developmental delay, short stature, cardiac defects, bleeding manifestations and lymphatic malformation affecting 1 in 1000-2500 children. This is a case report of a twenty six month old boy diagnosed to have Noonan syndrome with sporadic inheritance secondary to advanced paternal age. Multidisciplinary approach is the key to success in managing these children, who are diagnosed at an early age.

KEYWORDS: Genetic, dysmorphism, de novo mutagenesis, multidisciplinary approach.

CASE REPORT: A twenty six month old male child born of a non-consanguineous marriage was brought to the paediatric outpatient clinic by his parents with the complaints of delayed development of speech. General examination of the child revealed that he was short statured [<fifth percentile], underweight [<third percentile], had facial dysmorphism with low set ears, inverted triangular face, ptosis, bulbous nasal tip, tall forehead, webbed neck and high arched palate. However, the motor development and systemic examination was unremarkable.



Figure 1

The maternal and birth history were apparently normal and the family history revealed no similar complaints. There was no history of bleeding manifestation or feeding problems. In addition to this child, the parents have a healthy fourteen year old daughter who is attending school.

In view of typical facial features and speech delay, the child was admitted to our inpatient ward and investigated thoroughly. The biochemical and haematological investigations showed no abnormality. Ultrasonography of abdomen and thyroid profile was normal. 2D Echocardiography showed a congenital heart defect [mild valvular pulmonary stenosis]. Audiological evaluation [BERA] was done in view of delayed speech, which revealed profound hearing loss in both the ears. Karyotyping was normal [46XY].

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Mutational analysis of PTPN11 gene showed a heterozygous pathogenic missense mutation in exon 13 was detected [c.1510A>G (p.M504V)], confirming the diagnosis of Noonan's syndrome in this child.

DISCUSSION: Noonan syndrome is relatively common congenital genetic disorder in the developed countries compared to the developing countries, with an estimated prevalence of 1 in 1000-2500 live births.^{1,2}

In India, there are no available statistics and what makes this disorder uncommon here is:

- Early marriages (Some of the cases with sporadic inheritance are linked to advanced paternal age).
- Unreported (Due to lack of availability/insufficient resources).
- Overlooked (Typical facial features recede with age).
- Missed (Due to wide variability, the presentation may be subtle and patients appear as normal variants to remain undiagnosed).

Noonan syndrome is an autosomal dominant disorder with complete penetrance and variable expressivity, but a significant percentage of cases represent *de novo* mutagenesis (Sporadic inheritance).³ Approximately one half of the known mutations are in the protein tyrosine phosphatase non receptor type 11 (PTPN 11) genes. The *de novo* PTP11 mutation in sporadic cases is predominantly of paternal origin.⁴

Early diagnosis is extremely essential as the developmental disabilities can be addressed by early intervention enabling these children to grow up and function normally in the adult world. The ongoing and comprehensive care can also prevent or lessen some of its complications.

However, there is no cure for Noonan syndrome. These children have a wide array of health issues making it important for the doctors to be aware of the child's special care needs. Multidisciplinary approach with regular follow up care and genetic counselling to apprise the family of new development and recommendation is the key for successful management of children with Noonan syndrome.⁵

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