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Case Report

An Unusual Case of Ptosis in a Child

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ABSTRACT

Noonan syndrome is a developmental disorder characterized by facial dysmorphia, short stature, cardiac defects and skeletal malformations. Noonan syndrome (NS) is one of the most common genetic syndrome and is also an important differential diagnosis in children presenting with syndromic facies. This syndrome is characterized by facial dysmorphism, congenital heart defects, short stature and also a wide phenotypicvariations. We report a eight year old boy, who had Noonans syndrome and presented with low set ears, widely spaced nipples, had ocular features of ptosis and hypertelorism, mild pulmonary valve stenosis and delayed mile stones of development. His father also had ocular and facial findings. Multidisciplinary treatment is the key to success in managing children with Noonan syndrome and the pediatric ophthalmologists play an important position to lead the health team.

Keywords: Noonan Syndrome, Ptosis.

INTRODUCTION

Noonan syndrome (NS) is a congenital condition characterised by short stature, facial dysmorphism, skeletal anomalies, congenital heart defects, bleeding diathesis and reproductive anomalies [1] It may be sporadic or inherited as an autosomal dominant or recessive trait. [2] It occurs in 1/1000 to 1/2,500 births. [3] Ocular abnormalities in 95% of these patients occur as ptosis with hypertelorism, refractive errors and strabismus. Out of these anomalies ptosis and nystagmus are most common ocular features, however visible corneal nerve, cataract and uveitis occur less frequently. We describe here a patient with NS who presented with right eye ptosis.

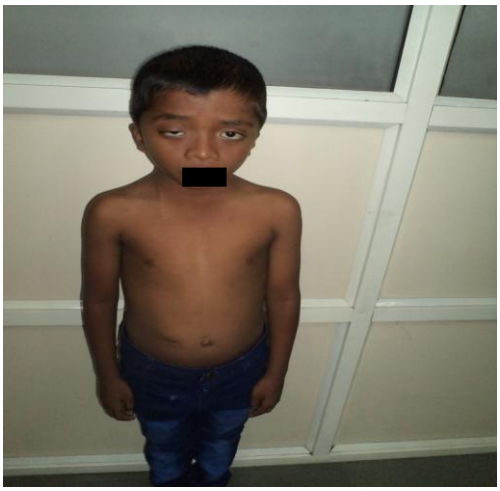
CASE REPORT

An eight year old, first born male child, reported to our outpatient department with main complaint of drooping of right eye lid since birth. The child was born out of non consanguineous marriage and was delivered vaginally after 38 weeks of gestation. His birth weight was 2500 g and length was 50 cm.The boy's father had bilateral ptosis but his younger brother had no such problem. [fig 1] The child had a history of surgery done for hydrocephalous by shunt surgery, at 1 year of age, elsewhere

in the city hospital. His blood investigations revealed Hemoglobin of 12 g/dl, Total Leukocyte Count of 6600/mm^3 with differential count of P 41%, L58%, E 01 % and peripheral smear showed a picture of normocytic normochromic, with reticulocyte count of 1 % and Platelets were adequate. Smear was negative for any parasites. His ESR was 29 mm at end of hour and blood urea nitrogen was 17 mg/dl, Serum Creatinine was 0.7 mg/dl, Serum Glutamate Oxaloacetate Transaminase (SGOT) was 26 IU/l, Serum Glutamate Pyruvate Transaminase (SGPT) was 28 IU/l. Serum Total Proteins was 5.8 gm/dl and Serum Albumin was 4.3 gm/dl and all his blood chemistry parameters were within normal limits. No genetic testing of his parents or of the child could be done due to financial constraints. Ultra Sonography, ECG, Xray chest and blood gases estimation showed no abnormality. However clinical cardiological assessment and echocardiography had established the presence of mild pulmonary valve stenosis and MRI brain showed features of communicating hydrocephalous. Ocular examination showed the best corrected visual acuity in both eyes was 6/9 and there was no improvement .Auto refraction finding was Rt Eye -1.25 Dsph/-0.74Dcyl 62^0 and Lt Eye -1.00 Dcyl 160^0 Anterior segment examination showed ptosis in right eye with poor function of levator palpebrae superioris (2 mm) and marked hypertelorism. Bell's phenomenon was present. There was no nystagmus or strabismus. There were no visible corneal nerves and ocular mobility was full & normal. Posterior segment examination was unremarkable [fig 2].

On systemic examination the child showed two surgical scars (each about 2.5 cm in size) on the right side of his scalp. There was facial dysmorphism with low set ears. He had short stature, short neck, widely spaced nipples on chest and had undescended testis on right side . The child had delayed mile stones of development.

[Fig 1] Family photo showing right ptosis in child and father, younger child is unaffected.



[Fig 2]. Child presented with right ptosis, facial dysmorphism with low set ears, short stature, short neck, widely spaced nipples on chest and undescended testis on right side.

DISCUSSION

Noonan Syndrome (NS) is a eponymous for a disorder described in 1963 by two paediatric cardiologists, Noonan & Ehmke, in conference and later published by Noonan alone. They based their description of this syndrome on observations made over nine subjects with pulmonary valve stenosis, low set ears, webbed neck, chest deformities and a distinctive dysmorphic facial appearance with hypertelorism and ptosis.

Opitz suggested that this disorder should be called Noonan Syndrome, a terminology that was subsequently adopted. [4] Noonan syndrome may occur in pattern consistent with autosomal dominant inheritance with almost complete penetrance. [4-7] In recent years germline mutations in the RAS-MAPK (mitogen activated protein kinase) pathway have been shown to be involved in the pathogenesis of NS. Mutations in PTPN11, KRAS, SOS1, NRAS, RAFI, BRAF, SHCO2, CBL e MEK1 can explain 60 to 70% of the molecular cause Genotype-phenotype correlations have been documented (Romano et al 2010; Tataglia et al 2011). [8,9]

Differential diagnosis is to be done from a number of conditions with phenotype strikingly similar to NS. First, Turner's Syndrome, a well known chromosomal abnormality occurring in girls. Then, there is a group of distinct syndromes with potentially overlapping phenotypes in which causative mutations are found in genes of RAS-MAPK pathway. These include cardiofacial - cutaneous syndrome, Costello syndrome, neurofibromatosis type 1, generalised woolly hair and LEOPARD syndrome. [5,10]

The diagnosis is based on a clinical score system proposed by van- der- Bugrt. [3] A dysmorphic face is mandatory, associated with other major criteria. The major criteria are: Pulmonary valve stenosis, hypertrophic cardiomyopathy, short stature (below third percentile), pectus carinatum and/or pectus excavatum and a first degree relative with diagnosis of NS. The minor criteria are: other cardiac defects, short stature (below tenth percentile), other thoracic abnormalities, first degree relative suggestive of NS and other findings (bad dental occlusion, micrognathism, mental retardation, developmental delay, deafness). [4,9,11] Our patient presented with typical facies (ptosis & hypertelorism) and cardiac defect (pulmonary valve stenosis), short stature, developmental delay and a degree of learning disability. So this patient had diagnostic criteria of Noonan syndrome. The genetic abnormalities were not investigated in this case.Treatment for patients with Noonan syndrome is symptomatic and directed to the specific associated anomalies or clinical findings. [7]

CONCLUSION

Though a rare disorder, it is important to recognise Noonan syndrome and do workup with multisystemic approach and rule out any genetic defect in a child who presents with ptosis, pulmonary valve stenosis and delayed milestones. There is no way to prevent Noonans syndrome. However, early diagnosis can help to control the disease manifestations once a person has it.

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