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Editorial inquiries should be addressed to Prof. Dr. Tayyab Afghani, Department of Orbit and Oculoplastics, Al-Shifa Trust Eye Hospital, Jhelum Road Rawalpindi, Pakistan.
Tel: 0092 51 5487821-25, Fax: 0092 51 5487827: Email:aqrcpio@yahoo.com ;
Website: www.asjoalshifaeye.org

CASE REPORT

Ocular features of A Rare Case of Noonan Syndrome in a Pakistani Population

Murtaza Sameen Junejo[1]

Abstract:
A heterogenous congenital disorder characterized as Noonan syndrome (NS), presents with typical features like a triangular face, short stature, and cardiac defects. It typically presents as an autosomal dominant trait. Noonan syndrome is one of the RASopathies due to the involvement of the RAS-MAP-Kinase pathway. Diagnosis is based on clinical features that include, typical facial features (triangular face, hypertelorism, ptosis), skeletal abnormalities(scoliosis), short stature, mild developmental delay, presence of cardiac defects, lymphatic dysplasia, and a family history of NS. Here we report a case of 12 years old boy with bilateral upper eyelid ptosis. On detailed examination, it turned out to be Noonan Syndrome.
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1. *Armed Forces Institute of Ophthalmology.*

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Correspondence to:
Murtaza Sameen Junejo
Armed Forces Institute of Ophthalmology.
drmurtazasameen@gmail.com

Introduction:

Noonan Syndrome (NS)is a congenital disorder with a prevalence of 1:1000 to 1:2500. Mostly it occurs as an autosomal dominant trait. NS is familial in less than 50% of cases. In 2001, the first gene to be connected with Noonan syndrome is PTPN11, while 20 other genes have been discovered, related to this heterogenous clinical condition.[1] Out of 3 RASopathies, Noonan syndrome is considered to be among one of them.

Short stature is one of the main features of this syndrome.[1-2] The syndrome includes several features: Dysmorphic facial features, heart defect, short stature, chest deformity, developmental delay, cryptorchidism, delayed puberty, ptosis, hypertelorism, hand contractures, and hearing problems.[2,3]

The aim of discussing this case is to inform and acquaint ophthalmological community and health care professionals about the signs and symptoms of this rare syndrome.

Case Report:

A 12-year-old boy presented to Armed Forces Institute of Ophthalmology with drooping of both upper eyelids since birth. It was noticed by parents due to his abnormal head posture. There was no history of trauma, redness, photophobia, ocular allergies, or ocular medicines. He has 1 sister of 4 years of age, who also had the same symptoms since birth. While personal history and socioeconomic history were non-contributory.

On general physical examination, a short-stature boy with a lean build and contracted fingers and vertebral problems was standing comfortably and was well-oriented in time, place, and person. Visual acuity was 6/6 OU. An increased intercanthal distance was noticed between two eyes along with drooping of both upper eyelids (hypertelorism and ptosis) and poor levator function (3mm) while rest of anterior and posterior segment examination was within normal limits. He also had triangular face with low set ears and small jaw along with vertebral abnormality like scoliosis. Bilateral hand contractures (Clinodactyly, Brachydactyly, and Blunt fingers) were also present in our patient(Fig 1 A-F, Fig 2). No cardiomyopathy like ASD (atrial septal defect) was noticed in this subject. He was suffering from deafness, for which he was referred to an E.N.T specialist.

Bilateral Upper eyelid ptosis was corrected with a frontalis sling procedure under GA, to prevent amblyopia and correct his head posture (Fig 3).

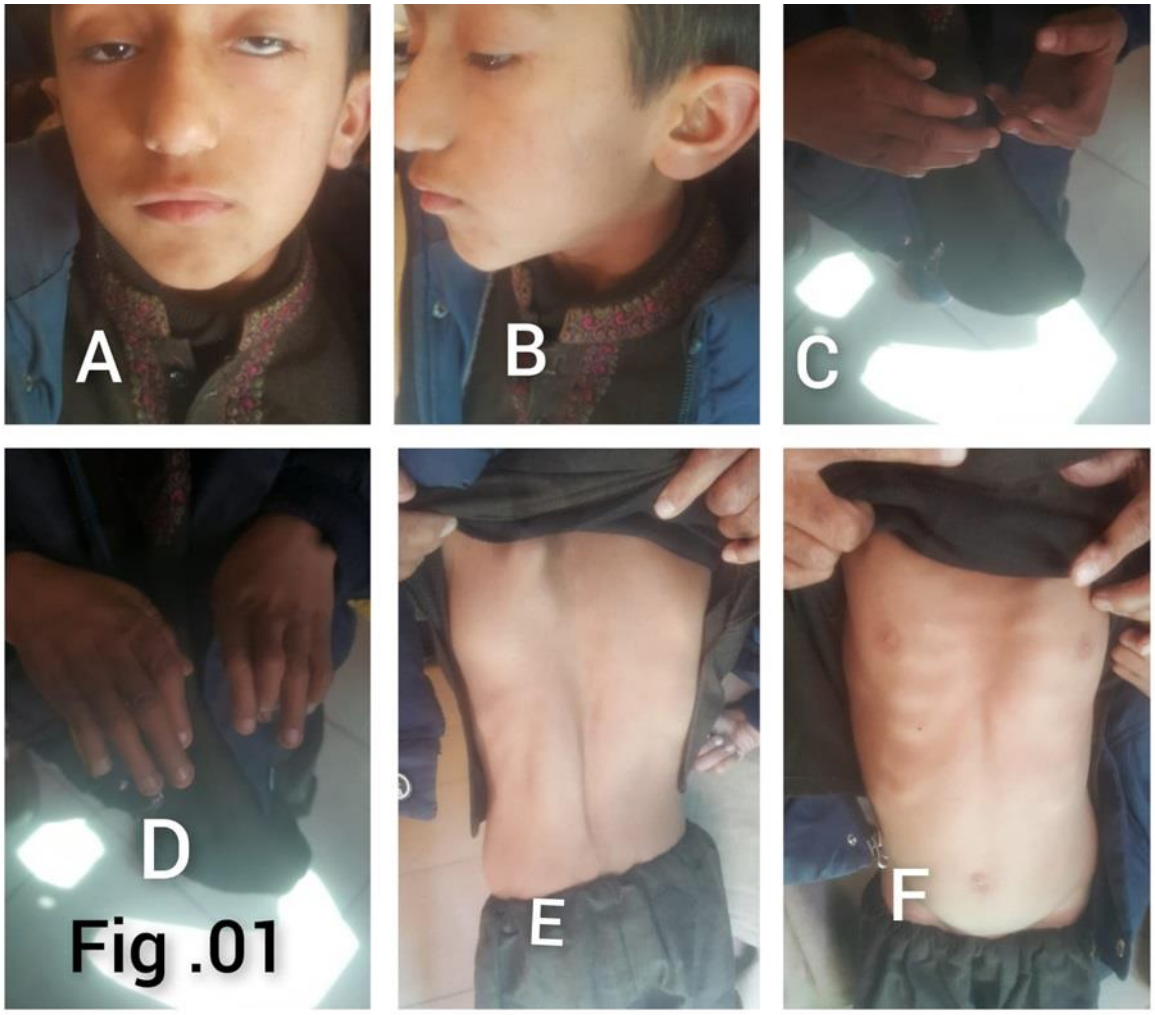

Fig: 1 A) Bilateral Ptosis with hypertelorism B) Low set ears C & D) Hand Contractures (Clinodactyly, Brachydactyly and Blunt fingers) E) Scoliosis F) Pectus Excavatum

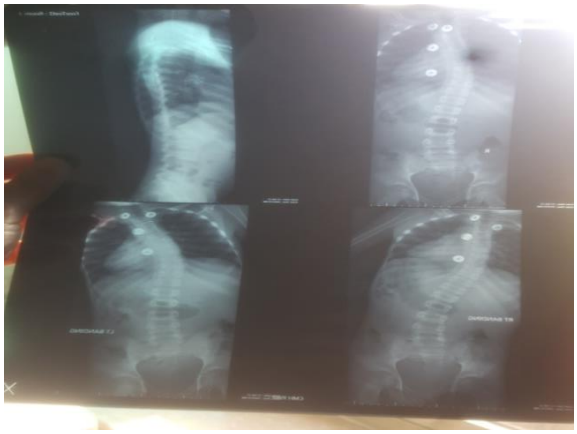

Figure 2: Xray Chest showing Scoliosis

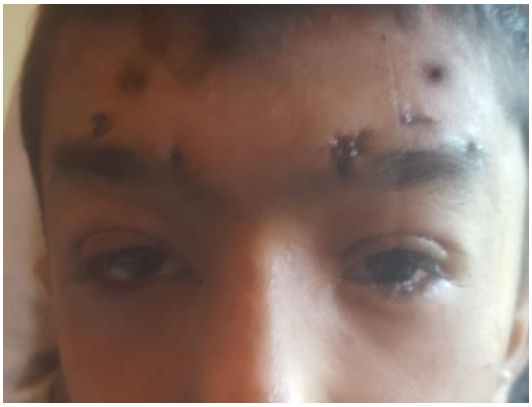

Figure 3: Frontalis Sling Procedure

Discussion:

In 1963, Noonan described many features that were also common in Lentigines syndrome, so the name Noonan was labeled. The same pleiotropic gene has been observed in both syndromes (Noonan and Lentigines).[4]

It has been observed that a patient with Noonan syndrome requires a multidisciplinary team approach to treat and manage this rare syndrome. We also sent our patient to a cardiologist, dermatologist, Endocrinologist, E.N.T specialist, orthopedic surgeon, and pediatrician.

The patient was operated on for bilateral ptosis correction with a frontalis sling procedure under GA in our case report to make his chin-up posture a more comfortable posture and prevent him from developing amblyopia.

Mendez and Optiz in their study confirmed that ocular manifestations are the commonest and consistent features in almost 95%, occurring in Noonan Syndrome. [5]

Marin et al, in their study, also suggest that ocular features account for larger clinical features in Noonan syndrome patients.[6]

The patient we reported here was a young male with ocular and systemic features of Noonan Syndrome.

In summary, NS is a rare disorder with multiple ocular features that should be diagnosed and treated early to prevent vision-threatening complications, therefore long-term follow-up and a multidisciplinary team approach are required.

Increased awareness of Noonan syndrome among ophthalmologists and other health care professionals could help parents/ guardian to seek specialist advice and proper management.

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Authors Contribution

Concept and Design: Murtaza Sameen Junejo
Data Collection / Assembly: Murtaza Sameen Junejo
Drafting: Murtaza Sameen Junejo
Statistical expertise: Murtaza Sameen Junejo
Critical Revision: Murtaza Sameen Junejo