Desiree Vanderstar, O.D.
Resident, Southern College of Optometry
Pediatrics & Vision Therapy/Rehabilitation

Noonan Syndrome – An Autosomal Dominant Genetic Mutation with Ocular Complications; A Case Report

Abstract:
A rare case of Noonan Syndrome: This report details the myriad of potential ocular complications that practitioners should be aware of when this unfamiliar condition presents in your chair.

Case History:
- Patient Demographics: 6-year-old white female
- CC: New patient presents with her mother who requested a visual skills/perceptual evaluation.
  The patients mother reports observation of the patient pulling items of interest towards her left eye predominantly, especially books while learning to read
- Ocular Hx: iris coloboma OD, bilateral upper lid ptosis, hypertelorism, nystagmus OU
- Medical Hx: Noonan Syndrome (NS) with resultant complications including left ventricle non-compaction, hydronephrosis and hydroureter
- Surgeries: uretral repair x 2, strabismus repair OD, bilateral frontalis slings, atrioventricular canal repair, and pulmonary stenosis repair
- Medications: aspirin, norditropan
- Other relevant information: premature birth at 34 weeks

Pertinent findings:
- Unaided VA was 20/100 OD, 20/60 OS with BCDVA is 20/40 OU and BCDVA 20/50 OU; patient uncooperative for monocular BCVA. Testing was done with LEA symbol matching
- Refractive error: OD: +0.50 DS, OS: -0.50 DS +1.50 ADD
- EOMs: Full and unrestricted OU; poor pursuits, saccades and attention during oculomotor entrance testing
- Unable to test for APD due to coloboma OD
- Confrontational Fields: Grossly full
- Visuscopy: target centrally located OS with unsteady fixation. UTT OD secondary to photophobia
- Gross physical assessment: exaggerated philtrum, low set ears, shortened neck with excess skin, microgranthia and a broad forehead lead to a characteristically shaped triangular face

Differential diagnosis:
- Primary/leading: Turner Syndrome
- Other: Fetal Alcohol syndrome, LEOPARD syndrome, Costello syndrome, Legius Syndrome, Cardiofaciocutaneous syndrome

Diagnosis & Discussion:
- This patient has a pre-existing systemic diagnosis of NS. Upon a visual skill set evaluation, she was ultimately diagnosed with intermittent esotropia OD s/p previous strabismus surgery,
amblyopia, oculomotor dysfunction and accommodation insufficiency, all culminating in her difficulties learning to read

- This rare genetic disorder has an incidence rate between 1/1000-1/2500 live births. It is known to affect both sexes equally with no predominance in any race. Primary diagnosis is based on clinical observation. Affected individuals most commonly have inherited a pathologic variant of PTPN11 in an autosomal dominant manner.\(^1\)
- It has been documented to be associated with a great variety of ocular abnormalities including: hypertelorism, downward sloping palpebral apertures, epicanthal folds, bilateral ptosis, NLDO, dermoid cyst, prominent corneal nerves, posterior embryotoxon, anterior stromal dystrophy, cataracts, panuveitis, optic nerve head drusen, optic disc hypoplasia, colobomas, myelinated nerves, amblyopia, refractive error, strabismus, limited ocular motility, anomalous retinal correspondence and nystagmus; at least one of which occurs in 95% of cases.\(^1,2,3\)

**Treatment, management:**

- Treatment of the complications of Noonan syndrome is generally standard and do not differ from treatment in the general population
- Glasses to correct the antimetropic refractive error was released for full time wear. A near add was prescribed to supplement poor accommodative abilities.
- The patient was enrolled in an individualized vision therapy program. Following in-office therapy for 10 weeks, the patient continues to make improvements in steadiness of fixations, visual attentiveness, stereopsis and visual-motor coordination.
- In order to dampen a latent nystagmus present during monocular therapies, a 0.1 Bangerter occlusion foil is used as a patch, which improved her ability to fixate with OD when OS is occluded with this method. When OD occluded, the nystagmus OS persisted. The patient demonstrates moderate opposition to occlusion OS
- With the appropriate interventions at an early age, visual prognosis has been shown to be good.\(^1\)
- As therapy continues, it is expected that her visual function will continue to improve
- It is important that these patients are monitored in a multidisciplinary approach in order to establish the extent of disease in each individual patient. This patient has had a variety of surgical interventions for multisystem involvements and is taking growth hormones to increase growth velocity.

**Bibliography:**


**Conclusion:**

- This patient was plagued by extremely serious cardiac and renal problems early in life, thus amblyogenic factors led to reduced VA. This case study, along with published prospective studies supports the conclusion that ocular symptoms account for a large fraction of the clinical manifestations of NS. It is evident that affected children should have a detailed ocular exam at infancy, even without exhibiting symptoms.