Acute onset esotropia in a child with Turner Syndrome
Author: Kelsey A. Thomson, OD – The New England College of Optometry
Co-Author: Mitchell Strominger, MD – Tufts/New England Medical Center

Abstract:
This case report discusses the ocular manifestations of a pediatric patient with Turner syndrome and a multidisciplinary approach to the diagnosis and management of this condition.

Key Words: chromosomal abnormalities, ocular abnormalities, strabismus, amblyopia, Turner syndrome

Case History:
A 6 year old Caucasian female presents for annual follow-up for amblyopia and partially accommodative esotropia. Significant ocular history includes isolated acute onset of right esotropia without signs of infection or trauma at age 3, full time wear of bifocal eyeglasses, and previous patching and atropine therapy. Significant surgical history includes a bilateral medial rectus recession (BMR) of 5.5mm. Her medical history is significant for Turner syndrome (diagnosed at age 5), developmental delay, and hypotonia. She was born full term weighing 5 pounds, 14 ounces with no complications during the pregnancy or birth. She receives Norditropin growth hormone injections nightly.

Pertinent findings:
Initial presenting acuity at age 3 was 20/50 OU uncorrected. Cover testing showed a 30pd AltET at distance and 40-50pd AltET at near. There were no signs of motility issues at that visit suggesting cranial nerve involvement. Cycloplegic retinoscopy revealed +3.00sph OU. Due to the acute onset of the esotropia, an MRI was ordered which came back within normal limits. Glasses were prescribed for full time wear and she was followed every 3 months. There was little improvement in her visual acuity and mild improvement in her ET throughout consecutive visits. A BMR was performed to improve alignment. After the surgery the alignment improved significantly and her vision started to improve with her bifocal eyeglasses.

The patient required multidisciplinary care after the strabismus surgery as she was diagnosed with global developental disorder, hypotonia, and sensory integration dysfunction. On physical exam, her height, weight, and head circumference were in the 3rd percentile. Referral to genetics revealed a diagnosis of Turner syndrome.

The most recent examination at age 6 showed corrected visual acuity of 20/20 OD and 20/25 OS at distance and near with habitual bifocal correction of +4.00+1.00x090 OD and +4.50+0.50x078 OS with +3.00 add OU. Uncorrected cover test showed 25pd ET at distance and 10pd E(T) at near. With the glasses, cover testing revealed 6pd EP at distance and Ortho at near when looking through the bifocal lens. Ocular motility testing revealed mild inferior oblique over action of the left eye. Stereopsis was reduced to 100 seconds of arc using Randot testing. All other tests, anterior and posterior segment examinations were unremarkable.

Diagnosis and discussion:
Turner syndrome affects about 1 in 2000 females and is defined as a chromosomal anomaly affecting the X chromosome\(^1,2\). It can be an alteration or a deletion of the X chromosome. Diagnosis can be made in the womb via amniocentesis, or after birth with a karyotype blood test. Short stature is a universally recognized feature of this syndrome, along with low birth weight, and other systemic findings\(^3\). Ocular sequelae are common in this syndrome and often overlooked. Studies have found that strabismus, amblyopia, and hyperopia are among the most common of these abnormalities\(^1\) and affect individuals with Turner syndrome more than the general population\(^1\). The patient reported in this case had many of the classic features that define Turner syndrome as listed above.
**Treatment and management:**

The patient discussed in this case was treated for her amblyopia and strabismus with eye muscle surgery, full time wear of bifocal eyeglasses, and previous patching and atropine therapy. She is followed every 6 months for her eyes. She will continue to be monitored for any further ocular sequelae secondary to her Turner syndrome. She also continues to be followed by a neurologist, her pediatrician, an endocrinologist, and other members of a multidisciplinary team who work together to ensure the best prognosis for this patient.

Turner syndrome is a lifelong condition. Patients are prone to many chronic systemic conditions and are typically managed by a multidisciplinary team of doctors. In children, growth hormone therapy is used to prevent short stature in adulthood. Typically these patients have fertility issues and impaired sexual development. These patients require estrogen replacement therapy. For ocular manifestations of Turner syndrome, early diagnosis and treatment provides an excellent long term visual prognosis for these patients. Ocular treatment indications and options will be discussed.

**Conclusions:**

Turner syndrome is a relatively prevalent genetic condition in which ocular sequelae are a common finding. The diagnosis of Turner syndrome is often delayed which results in a later onset of treatments. The patient discussed in this case was not diagnosed with Turner syndrome 3 years after her initial presentation with an acute onset esotropia. With pediatric patients, strabismus or amblyopia are not the only condition a patient may have. It is important to consider systemic conditions or genetic syndromes that may coexist with these ocular diagnoses. Also, it is important to understand genetic syndromes and the associated ocular manifestations in order to properly manage these patients. If Turner syndrome is suspected or has been previously diagnosed, co-management with a multidisciplinary team is imperative for successful treatment and management.

**References:**