

case report

Keratoconus and Turner's Syndrome: A Case Report

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ABSTRACT

We report an unusual combination of keratoconus and Turner's syndrome in a 13-year-old girl.

Key Words: Turner's syndrome, keratoconus, mesoderm

Turner's Syndrome is a well known condition characterized by Karyotype 45XO. Incidence is estimated to be 2 to 3 cases every 1000 newborn¹ but most authors report 1/5000. The clinical features include short stature, a short webbed neck, a low hairline over the back of the neck, a broad short chest with widely spaced hyperplastic nipples, multiple pigmented nevi and short fourth metacarpals.² Ocular involvement is often present³ but no ocular signs are considered characteristic.

CASE REPORT

We present the first report of keratoconus in Turner's Syndrome. A 13-year-old white female with Turner's Syndrome showed complete absence of one X chromosome in all cells. She complained of blurred vision for near and distance and was referred to our Pediatric Ophthal-

mology Service. There was no history of atopic diseases, mitral valve prolapse, osteogenesis imperfecta or other connective diseases. She had never worn contact lenses. Her I.Q. was within normal limits. Keratometric readings disclosed distortion of the mires and the slit lamp examination revealed a cone-shaped thinning of the cornea. Best corrected visual acuity (wearing contact lenses of -1.75 RE and -1.25 LE) was 6/9. No Vogt's striae or Fleischer's ring were seen in the subject's cornea.

DISCUSSION

Keratoconus is a non-inflammatory and usually bilateral disease of the cornea in which progressive thinning, distortion and steepening of the central corneal curvature lead to irregular astigmatism and visual impairment.

The unusual occurrence of keratoconus in a young girl and the coincidence of these two relatively rare conditions led us to some considerations.

Keratoconus is reported to occur frequently in Down Syndrome patients.^{4,5} Eye-rubbing is hypothesized to cause corneal thinning in these patients.⁶ A similar mechanical pathogenesis is proposed for the keratoconus in atopic diseases and in hard contact lenses wearers.^{7,8}

There are no typical eye findings in Turner's syndrome. Single cases reporting various ocular anomalies are listed in Table 1.

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Table 1. Ocular Findings in Turner Syndrome

Pigmented areas on eyelids
Prominent epicanthal folds
Accentuated downslanting of palpebral fissures
Ptosis
Strabismus
Nystagmus
Blue sclerae
Cornea with short horizontal diameter
Pupillary heterotopia
Anterior axial embryonic cataract
Incidence of X-linked color deficiency as in males

We believe that corneal thinning in our patient is an expression of a mesodermal defect, since mesodermal structures are sometimes affected in Turner's Syndrome.^{9,10}

Our case report provides further evidence that Turner's Syndrome is commonly associated with ocular problems, stressing the importance of careful ocular examination in this condition.

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