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Uveitis In Turner's Syndrome – A Case Report

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Abstract
Background*: Turner's syndrome is chromosomal abnormality were where phenotypic females have either missing X chromosome. In turner syndrome several ocular diseases are associated in the past, including one case only of proven iridocyclitis.*
Methods: *In this case study we report the clinical finding of single female with turner's syndrome and uveitis for a mean period of 12 month.*
Result*: This patient has recurrent iridocyclitis and amenorrhea. The iridocyclitis in turner syndrome was chronic and may be due to contraindication to use of systemic steroids. Although the final visual outcome is 6/6.*
Conclusions*: Iridocyclitis should be included in the list of ocular manifestation in turner syndrome.*
Keywords: *Turner's syndrome, Chromosome, Iridocyclitis, Amenorrhea, Steroid.*

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I. Introduction

Turner's syndrome is a chromosomal disorder where phenotypic females have either a missing chromosome (45X,0) [1] or a structural aberration of one of the X chromosomes such as an isochromosome of the longarm, ring chromosome or small deletions [2].

Approximately 80% of females with Turner's syndrome have 45 chromosomes with a single X, while the remaining 20% have 45, X/46, XX mosaicism and remainder have structural abnormality of the X chromosome such as X fragments, isochromosome or rings.

The clinical feature of turner syndrome results from haploinsufficiency of multiple X chromosomal genes (e.g. short stature homeobox, SHOX). However, imprinted genes also may be affected when the inherited X has different parental origin.

The incidence of Turner's syndrome is estimated to be 1 in 2,000–5,000 live births [3, 4].

Short stature, ovarian dysgenesis with primary amenorrhea, sexual infantilism with small uterus, sterility, webbed neck, broad shield chest with widely spaced nipples, cubitus valgus, left-sided congenital heart defects and multiple pigmented nevi are main systemic features [2].

Autoimmune disorder such as Hashimoto thyroiditis, diabetes, juvenile rheumatoid arthritis, and Crohn's disease, have been associated with the syndrome.[2,5,8,9]

Ocular abnormalities including Strabismus (18.9–37.5%) [1,3,4], Ptosis (16–29.1%) [1,3,4], Hypertelorism in 10% [3], Epicanthus in (10–45.8%) [1,3,4], Color deficiency (2.7–10%), Blue sclerae (29%), and congenital cataract (4.2%–8.1%).

Additionally, several case reports addressed other ocular diseases, such as glaucoma [6], retinal detachment [7], anterior segment dysgenesis, choroidal and retinal neovascularization, Coat's disease, and keratoconus.

Only one patient with anterior uveitis and Turner's syndrome has been described in the literature [5].

We here report one case of iridocyclitis in turner's syndrome.

II. Case Report

A 16-year-old girl had visited to our OPD in 2020 with complain of Redness, Pain, Photophobia and Blurring of vision in left eye since 10 days.

Her clinical history revealed Recurrent episode of similar event occur previously, first episode was occurred when she was in class 5th. After that she had 4-5 more episode.

She had been diagnosed as having Turner's syndrome at the age of 13 (45X,0 karyotype) with amenorrhea and short stature.

On General examination, Patient has Short stature, webbed neck and has average weight.

She has also taken treatment for Amenorrhea from gynecology department.

At our first Ophthalmic examination, visual acuity of patient was recorded by Snellen's chart, best corrected Vision in OD is 6/6 and in OS 6/18.
On slit lamp examination of Right eye, conjunctiva is normal, cornea clear and no cells and flare in Anterior chamber, Pupil is normal in size and normal in reaction to light.
Left eye, conjunctiva has ciliary congestion, hazy cornea with fine KP's on endothelium. Anterior chamber cells 4+, Flare 3+ (by SUN Classification of uveitis) and fibrin exudate of approximately 3mm in size present, Pupil is irregular and sluggish in reaction to light, Posterior synechiae present, Iris pigment present over lens, no cells are present in vitreous.
On dilated fundoscopy by indirect ophthalmoscope fundus is within normal limit.
A complete work for iridocyclitis including Complete blood count, ESR Rheumatoid factor, Immunoglobulins (IgG, IgA, IgM), Autoantibodies (ANA), p-ANCA and c-ANCA, complement factors, Urinalysis, Mantoux test, VRDL, Angiotensin-converting enzyme, Serum calcium test, Chest X-ray, HLA typing, HRCT was unremarkable.
She was given a course of Topical steroids (Dexamethasone 0.1% six times daily), Mydriatics (Atropine 1% TDS) and Topical NSAID (Flurbiprofen 0.03%).
In follow up after 5 days the Anterior Chamber Cells, Flare and Fibrin Exudate doesn't decrease so doses of Topical steroid (Dexamethasone 0.1% given 2 hourly daily).
In next follow up after 10 days anterior chamber cells decrease to 2+, Flare 1+ and exudate height decreases, dexamethasone dose than gradually started tapering. Best corrected visual acuity improves after 15 days of treatment that is 6/6 in left eye. Iridocyclitis subsided after a 2 -month course of therapy.

III. Result

Our patients developed iridocyclitis in childhood approximately at the age of 10 year and was unilateral. Systemic steroids were avoided in the patient because of the endocrinological disease. The patient was treated with topical steroids and mydriatics and systemic NSAIDs, but the uveitis tended to be-come chronic with time. She was also having amenorrhoea as endocrinological abnormality. Patient was follow up for 9 months.

IV. Discussion

Many ocular abnormalities have been described in patients with Turner's syndrome [1,4] but in only one reported case was the patient affected by uveitis [5]. In this study we report a case of anterior uveitis in patients with Turner's syndrome which was recurrent as well as chronic. The trend toward the chronicity of uveitis may be partially due to the difficulty of using steroids either peribulbar or systemically, since they might affect the endocrinological abnormalities related to the chromosomal alterations. The association between autoimmune diseases and chromosomal anomalies such as Down's syndrome and Turner's syndrome has been already reported [5,8, 10, 11,12, 13, 14], although the pathogenetic role of the chromosomal abnormalities in the development of auto-immune disorders has been yet not defined. Experimental studies on animal models have demonstrated that polymorphic genes on the X chromosome may modulate susceptibility to collagen-induced arthritis, a systemic polyarthritis resembling human rheumatoid arthritis [15]. In patients with Turner's syndrome many studies have demonstrated that the risk of developing autoimmune thyroid disease is particularly high in women with an X isochromosome [16], suggesting that a gene on the long arm of the X chromosome plays an important pathogenetic role in the development of autoimmune disease [16]. Alternatively, an imbalance between loci on the short and long arms of the X chromosome may increase the risk of immune dysfunction. Nevertheless, whether the Turner's syndrome by itself might be a cause of uveitis remains unclear. A longer follow-up of this case and further studies may clarify the relationship between X-chromosome abnormalities and the possible development of uveitis.

V. Conclusion

Uveitis should be added to the list of ocular lesions in Turner's syndrome and a full ophthalmologic follow-up is strongly indicated in these patients. In order to identify any possible systemic or genetic disease associated with uveal inflammation, careful systemic investigation of young patients with uveitis is always recommended.

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