

A Rare Case of Down's Syndrome with Ocular Involvement and Hirsutism

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ABSTRACT

Our case highlights a rare presentation in which various associations associated with a particular disease were seen in a single patient.

Key words: Associations, downs, ocular

CASE

A 15-year-old female was brought to us by her parents with a history of severe itching in both the eyes for the past few years. Old records brought by her parents showed that she was a known case of Down's syndrome and had been on topical antihistaminics off and on for the past few years. There was no other significant history. She did not allow refraction nor did cooperate for Snellen visual acuity test. The significant findings on her bilateral eye examination were Horner-Trantas dots with gelatinous limbal membrane plus surrounding triangular conjunctival congestion suggestive of limbal form of vernal keratoconjunctivitis (VKC) and a positive Munson sign and oil droplet reflex (seen on distant direct ophthalmoscopy) suggestive of keratoconus. A central corneal opacity in the right eye was also evident [Figures 1-3]. Keratometry readings could not be taken as the patient had a variable mood pattern. Even on torch examination, the conical cornea could be easily appreciated. Other positive findings were hirsutism as suggested by a visible moustache.

The patient was diagnosed as a case of VKC with keratoconus, a right corneal opacity, Down's syndrome, and hirsutism. The patient was started on topical olopatadine + ketorolac + hydroxypropyl methylcellulose drops twice a day and topical

bepotastine 1.5% twice a day. Her parents were explained in detail the nature of her ocular disease and the need for further follow-ups was required for her ocular treatment. She was also referred to pediatrics and obstetrics-gynecology outpatient department for her other problems and a systemic workup. We did not have any further follow-up from her end.

DISCUSSION

Down's syndrome is typically associated with delay in the physical growth, characteristic facial features, and mild-to-moderate intellectual disability. It is often associated with keratoconus.^[1] Keratoconus is a bilateral non-inflammatory corneal ectasia. The prominent features of VKC include a typical clinical history of severe itching with characteristic signs, including giant papillae on the upper palpebral conjunctiva, limbal infiltrates, and eosinophilic concretions (Horner-Trantas' dots). The association of keratoconus with VKC is known.^[2]

Hirsutism is the presence of terminal (coarse) hairs in females in a male-like pattern. It can be idiopathic or should be considered as a sign of other conditions such as polycystic ovary syndrome, androgen-secreting tumors, non-classic adrenal hyperplasia, or syndromes of severe insulin resistance.^[3]

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Figure 1: Vernal keratoconjunctivitis with conical cornea



Figure 3: Vernal keratoconjunctivitis with conical cornea



Figure 2: Munson sign with hirsutism and right corneal opacity

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