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Floppy eyelid syndrome in stickler syndrome

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ABSTRACT

Purpose: To report the possible connection between Stickler syndrome and floppy eyelid syndrome.
Observations: A 36-year-old man with genetically confirmed Stickler syndrome presented with prominent bilateral eyelid laxity consistent with floppy eyelid syndrome. He had a high-arched palate and reported years of apneic episodes during sleep consistent with obstructive sleep apnea (OSA).
Conclusions and importance: To our knowledge, there have been no prior reported cases of floppy eyelid syndrome in Stickler syndrome patients. However, many patients with Stickler syndrome have palate abnormalities, which are associated with a higher risk of OSA. Given the known association between floppy eyelid syndrome and OSA, this case highlights the potential role for the ophthalmologist in identifying these patients and referring them for a sleep study if floppy eyelid syndrome is identified.

1. Introduction

Autosomal-dominant Stickler syndrome results from mutations in *COL2A1*, *COL11A1*, and *COL11A2* genes, which encode various collagen proteins.[1] These mutations manifest in ocular, orofacial, auditory, and skeletal tissues.[2] To our knowledge, floppy eyelid syndrome (FES) has not been reported with Stickler syndrome despite the plausible connection between abnormal eyelid laxity and collagen abnormalities. We present a case of bilateral FES in a patient with Stickler syndrome.

2. Case report

A 36-year-old man with IVS4A+ 1G > A transition *Col2A1* mutation Stickler syndrome was examined in follow-up of a prior retinal detachment and found to have prominent bilateral eyelid laxity consistent with FES (Fig. 1). The patient had a high-arched palate, retromicrognathia, BMI of 35, and reported years of snoring with witnessed apneic episodes consistent with obstructive sleep apnea (OSA). The patient was treated with a sleep apnea oral appliance with significant improvement in his OSA.

3. Discussion

FES is well known to coexist with OSA,[3] which is common in Stickler syndrome and reported in 14% of patients because of the associated palate abnormalities, such as cleft palate and high-arched palate.[2,4–7] Although there appears to be a relationship between collagen subtype mutations in Ehlers Danlos syndrome and floppy eyelids,[8] a similar association has not been reported between Stickler syndrome and FES. Histologically, collagen appears normal in FES eyelids, while elastin is decreased and disrupted.[9] It seems possible that the collagen mutation contributed to the development of FES in our patient, though there are data that suggests that OSA might cause FES by its effect on metalloproteinases and development of a proinflammatory response.[10] Even if OSA is not the cause of FES, there is certainly an association between the two. Therefore, if FES is present, OSA is more likely to be present as well. Lastly, it is possible that our patient's OSA was from obesity alone, but given the patient's high-arched palate and literature which support coexistence of OSA and palate abnormalities, its presence was likely multifactorial.

4. Conclusions

Because Stickler syndrome has numerous ophthalmic manifestations, it is important for ophthalmologists to recognize the increased prevalence of OSA in this condition and to refer patients with FES for sleep medicine studies.

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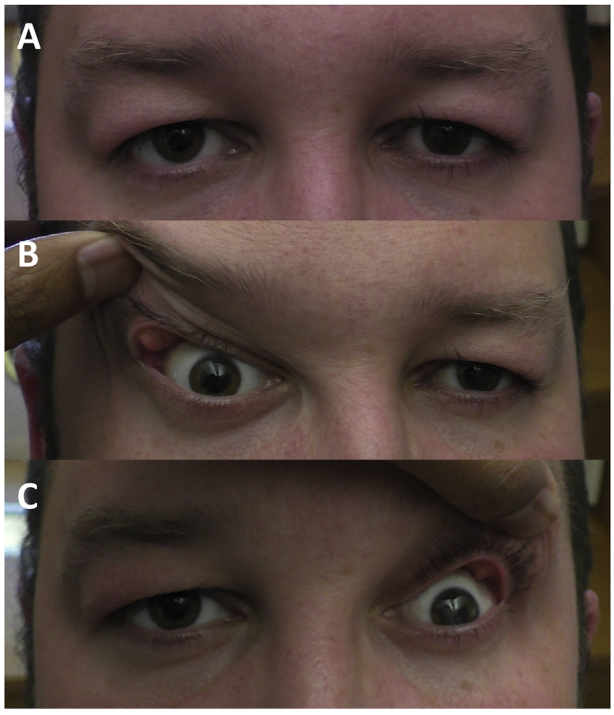

Fig. 1. Patient with floppy eyelid syndrome (A) with laxity of the right upper eyelid (B) and left upper eyelid (C).

Patient consent

The patient consented to publication of the case in writing.

Conflicts of interest

None of the authors have financial disclosures.

Authorship

All authors attest that they meet the current ICMJE criteria for authorship.

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