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#### **FUNCTIONAL AND CLINICAL FINDINGS IN THREE MALE CHILDREN WITH STICKLER SYNDROME**

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#### **Introduction**

Stickler syndrome is an autosomal dominant disease with ocular and systemic manifestations. Ocular features are diverse and sometimes vision-threatening. High myopia is frequent, with a high incidence of retinal detachment. Anterior segment anomalies can also be present. Early diagnosis and regular ophthalmic work-up are vital to prevent severe complications. Low-vision aids can dramatically improve the patients' visual performance and life quality, and should be provided from an early age. Here we report on three patients from three unrelated Portuguese families.

#### **Methods**

Diagnosis was made by a multidisciplinary team, based on the rigorous assessment of clinical findings. Detailed characterization of the ocular manifestations was performed through comprehensive ophthalmic examination. Thorough evaluation of retinal phenotype was regularly performed using the RetCam® digital imaging system.

#### **Results**

The first propositus is a 16 year old teenager followed in our clinic because of low visual acuity and high myopia. Objective refraction is -21 diopters (D) bilaterally. BCVA is 20/125 for distance and 20/50 at 10 cm for near vision. Anterior segment anomalies were not present. Fundus observation revealed atrophy in the macular region and a posterior staphyloma. He has mild to severe intellectual and motor disability that impairs the possibility of low-vision aid training.

The second propositus is a 10 year old male. He suffers from high myopia and low visual acuity. Distance BCVA is 20/400 on both eyes, corrected with -16.00(-2.00x90°) D on the right eye and -17.75 D on the left eye. His anterior segment examination is unremarkable. On fundus examination he presented with myopic degeneration, macular atrophy and vitreous liquefaction. No retinal breaks or detachments were ever found. He has been involved in our low-vision consultation since he was 4 years old. With a 10x telescope he achieves a distance BCVA of 9/10. He can read at normal speed using a video magnifier. An effort was made to integrate the school in the visual rehabilitation process, where he uses the same low-vision aids.

The third propositus was seen for the first time in our clinic when he was 5 years old. He had undergone 5 surgeries on his right eye due to a retinal detachment when he was 3 years old. His right eye was left aphakic and with silicone oil tamponade. Anterior segment examination revealed band keratopathy and corectopy on the right eye. Left eye anterior segment was unremarkable. Baseline BCVA was light perception on the right eye and 20/25 on the left eye. He has since been observed twice with the Retcam device, revealing no retinal breaks or detachments.

#### **Conclusion**

Stickler syndrome is a rare disease, highly variable in its ocular presentation and severity. We present three cases that highlight the specific challenges posed by its heterogeneous clinical manifestations and the comprehensive approach needed to help these patients.