

Retinal detachment in a child with Frank-ter Haar syndrome

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Background

To present a rare case of ocular involvement in a child with Frank-ter Haar syndrome (FTHS) presenting retinal detachment.

Materials and Methods

Detailed ophthalmological evaluation including examination under general anesthesia, ocular ultrasound, and visual evoked potential testing was completed. Photographic documentation of the physical findings was obtained.

Results

A 3-year-old female patient with FTHS was referred to evaluate for possible ophthalmic involvement. The patient presented with the classical dysmorphic abnormalities of the syndrome. Ophthalmologic evaluation revealed a high, against-the-rule corneal astigmatism in the right eye. In the left eye, the red reflex was absent with a suspicious membrane behind the lens, and a sensory exotropia was present. Ultrasonography confirmed retinal detachment with no history of previous trauma. Due to poor visual evoked potentials, no surgery was planned. Astigmatic refractive error was corrected with routine follow-up.

Conclusions

FTHS is associated with multiple ocular involvement such as megalocornea, congenital glaucoma, or colobomas. This case report is the first to describe a high, against-the-rule astigmatism and retinal detachment in a female child with FTHS and demonstrates that an early and detailed ophthalmological examination is essential for these patients.

KEYWORDS:

- [Frank-ter Haar syndrome](#)
- [genetics](#)

- [retinal detachment](#)
- [SH3PXD2B](#)