Nance Horan Syndrome:
Contact Lens Considerations for a Rare Genetic Condition

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Background

Nance Horan Syndrome (NHS) is a rare X-linked disorder caused by a mutation in the *NHS* gene.[1,2] This condition, also known as cataracts-oto-dental syndrome, is characterized by developmental problems, dental anomalies, facial dysmorphism, and congenital cataracts.[1,2]

Ocular Manifestations

- Bilateral congenital cataracts
- Microphthalmia
- Microcornea
- Nystagmus
- Strabismus
- Glaucoma

Case History

A twelve-year-old male was referred to the NSU Health Eye Care Institute for a contact lens (CL) fitting to optimize visual acuity.

Previous Medical History

- Nance Horan Syndrome, diagnosed at birth

Previous Ocular History

- Congenital cataracts, post extraction w/ aphakia OU
- Glaucoma OD
- Polycoria OD
- Strabismus OD
- Nystagmus

Ocular medications

- Dorzolomide/ Timolol BID OD
- Prednisolone Acetate 1% qhs OD

Pertinent Findings

Manifest Rx: OD +5.50 -2.00 x 075, DVA 20/300
OS +8.50 -2.50 x 155, DVA 20/100
Pupils: irregular, non-reactive OD and OS
Nystagmus: Pendular
Strabismus: Constant Right Esotropia
Slit lamp examination: see Figure 1

Clinical Presentation

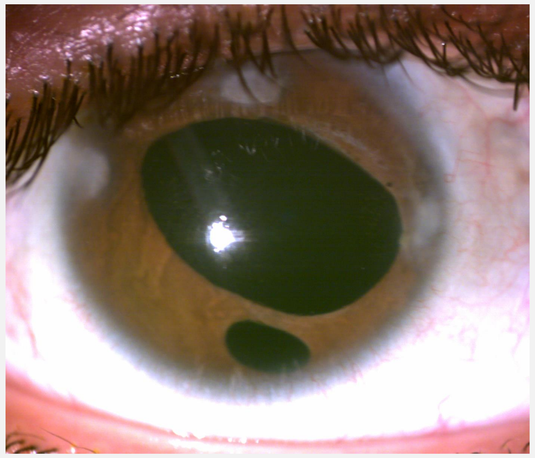

Figure 1: Anterior segment photo, OD. Polycoria with superior, temporal, and nasal stromal haze were observed during slit lamp biomicroscopy. The patient was newly diagnosed with presumed iridocorneal endothelial syndrome (ICE), most likely the progressive iris atrophy subtype.

Contact Lens Fitting

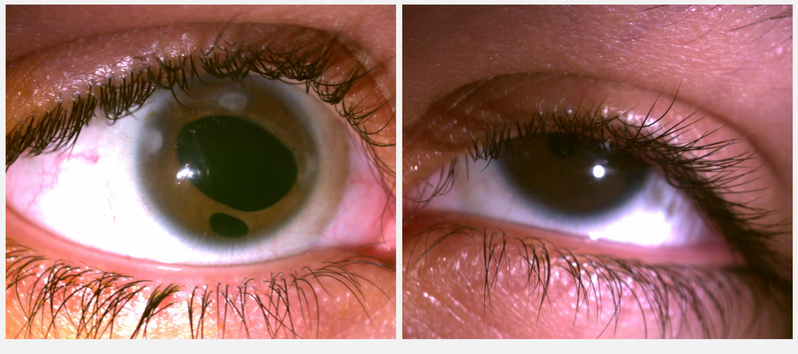

Figure 2: CL Fitting, OU. The patient was successfully fit with Cooper Vision Biofinity® XR Toric soft contact lenses.

Final CL Rx:

Biofinity® XR Toric // 8.70 Base Curve // 14.50 diameter
OD: +8.50 -2.75 x 065 DVA: 20/70
OS: +10.00 -3.75 x 160 DVA: 20/60

Discussion

Although cataract surgery is necessary in these patients, it may initiate glaucoma.[2] The mechanism that causes the elevated glaucoma risk in these patients is still unknown but may be secondary to a poorly developed aqueous humor drainage system.[2]

After reviewing the literature and considering this patient's findings, it appears there may be a link between NHS and progressive iris atrophy. In one case report, endothelial cell attenuation with a poorly developed thin iris was observed during pathological examination of an NSH patient's enucleated eye.[2] More studies must be conducted to confirm or refute this potential link.

Future Considerations

Due to the patient's polycoria, a prosthetic contact lens with an opaque iris and underprint should be considered if the patient develops symptoms of diplopia and/or light sensitivity.

Conclusion

NHS is associated with ocular manifestations including congenital cataracts, microcornea, microphthalmia, nystagmus, strabismus, and glaucoma.[1,2] Young NHS patients are usually left aphakic after early cataract extraction, with the majority suffering from significant visual impairment even after early surgical intervention has been established.[2] Toric soft contact lenses are a great option for young patients who cannot adapt to other contact lens modalities. When considering this case and others reviewed, the suspicion of an association between NHS and iridocorneal endothelial syndromes, including progressive iris atrophy, is heightened.

References

1. Coccia M, Brooks SP, Webb TR, Christodoulou K, Wozniak IO, Murday V, Balicki M, Yee HA, Wangensteen T, Riise R, Saggar AK, Park SM, Kanuga N, Francis PJ, Maher ER, Moore AT, Russell-Eggitt IM, Hardcastle AJ. X-linked cataract and Nance-Horan syndrome are allelic disorders. Hum Mol Genet. 2009 Jul 15;18(14):2643-55. doi: 10.1093/hmg/ddp206. Epub 2009 May 4. PMID: 19414485; PMCID: PMC2701339.

2. Ding X, Patel M, Herzlich AA, Sieving PC, Chan CC. Ophthalmic pathology of Nance Horan syndrome: case report and review of literature. Ophthalmic Genet. 2009. Sep: 30 (3):127-35.