Corneal-scleral contact lens fitting for an atypical Noonan syndrome patient: A case report

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Background

To report an atypical case of irregular cornea in a young Noonan syndrome patient who was successfully fitted with corneal-scleral rigid gas permeable (RGP) contact lenses.

Case report

A 20-year-old male, MCT, was presented to our optometry clinic for routine eye examination. The patient was suffering from Noonan syndrome (NS), and his chief complaints included blurry distant vision and monocular diplopia in both eyes. The patient reported that cardiovascular surgery, strabismus surgery, ptosis surgery, and amblyopic treatment had been performed to him before the age of six. His general health condition was currently monitored by genetic and cardiovascular specialists. Patient’s aided visual acuity (VA) were 6/18 (OD) and 6/38 (OS). Biomicroscopy revealed nystagmus, microcornea, conjunctival tissue proliferation, and upward displacement of the pupils in both eyes (see Fig. 1).

Regular aspheric RGP lens with different diameters and bi-toric RGP lens were fitted to the patient. The parameters of different trial fittings were summarized as follows:

1) Inferior decentration
2) Heavy inferior pool with air bubbles trapped in both eyes
3) Mild monocular diplopia

As optimal visual acuity and lens fitting could not be obtained by using normal RGP lenses, he was re-fitted with corneal-scleral RGP lenses. Due to the patient’s small and irregular cornea with different diameters at different meridians, instead of standard 14.0mm diameter lenses, 13.5mm lenses with the BC on flat-K and peripheral curve (PC) 1.5D flatter than standard were fitted to the patient.

Visual performances were good with both lenses. However, heavy conjunctival jerk (especially at the area of conjunctival tissue proliferation) and central bearing were noted (see Fig. 3). Moderate diffused punctuate staining and lens binding appeared after wearing the lenses for a few days.

Thus, lenses with smaller diameter, flatter PC and steeper BC were re-fitted for the patient to reduce the conjunctival jerk on the proliferation tissue and the central bearing.

Finally, the new lenses were fitted successfully to the patient (see Fig. 4). Visual performance was good and monocular diplopia symptom disappeared. The corneal health, lens fitting, and subjective feeling were optimal during subsequent aftercare consultations.

Discussion

The Noonan syndrome is a multifaceted genetic disorder characterized by a series of congenital systemic malformations, including one in the visual system. It has no racial predilection and may be sporadic, autosomal dominant or autosomal recessive. The incidence of NS is reported to be between 1 in 1000 and 1 in 2500 live births.

Frequent ocular abnormalities include hypertelorism (74%), ptosis (48%), epicanthal folds (39%), antimongoloid slant (38%), downward-sloping palpebral fissures (38%), strabismus (48%-63%), refractive errors (61%), amblyopia (33%), anterior segment changes (63%), fndal abnormalities (20%), and nystagmus (9%). In addition, some other ocular abnormalities such as nasolacrimal duct obstruction, dermoid cyst, increased cup-to-disc ratio, and optic nerve pit may also be associated with Noonan syndrome.

Most of the above ocular abnormalities can be managed by muscle surgery, vision therapy and spectacle. However, for patient complicated by microcornea, conjunctival tissue proliferation, upward displacement of pupils and irregular cornea, visual performance cannot be improved by spectacle. In this case, RGP lenses, especially corneal-scleral RGP lenses may be useful in providing optimal visual performance and maintaining good ocular health for the patients.

Conclusion

The above case demonstrated that patients with Noonan syndrome, complicated by the presence of irregular cornea, can be successfully managed by using corneal-scleral RGP lenses.

Reference
