Rare disease

Surgical management of unilateral rhegmatogenous retinal detachment associated with ocular coloboma in a 7-year-old child with Noonan syndrome

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Summary

The authors would like to present an unusual case of unilateral retinal detachment, phacodonesis, dense cataract and ocular coloboma in a 7-year-old Indian Muslim boy with Noonan syndrome. He underwent lensectomy, 23G pars planar vitrectomy, air fluid exchange, endolaser and silicone oil injection which successfully reattached the retina. His best corrected visual acuity was 6/24 6 months after the surgery and oil removal.

BACKGROUND

This is an unusual case of ocular coloboma which developed rhegmatogenous retinal detachment (RRD) secondary to an occult break within the optic disc itself in a child with Noonan syndrome (NS). We would like to share with our fellow colleagues a technique of barricade laser to the coloboma with short-term silicone oil tamponade which successfully reattached the retina for up to 6 months postoperatively.

CASE PRESENTATION

A 7-year-old Indian Muslim boy presented to the eye clinic with poor vision, noted by his parents, in the left eye for 1 year. He had been previously diagnosed to have refractive error bilaterally at another centre. He had been wearing spectacles since the age of 4. There was no history of eye pain, redness or trauma.

He had been diagnosed with NS since the third month of gestation by amniocentesis. He was delivered by spontaneous vaginal delivery postdates. Systemic history included intestinal obstruction since birth for which surgical repair was performed successfully in infancy, undescended testis (operated) and spontaneous closure of patent ductus arteriosus with now normal cardiac function.

There was no history of parental consanguinity with three otherwise normal older siblings.

On examination, he was a cheerful and lovable child with dysmorphic features (figure 1) with global developmental delay and short stature. He had bilateral partial ptosis with small-angle left convergent squint. Extraocular movements were full bilaterally. Visual acuity was 6/36 *oculus dextrum* (OD) and hand motions *oculus sinistrum* (OS) with good light projection. There was bilateral microcornea with right cornea 1–2 mm smaller than left. Other ocular abnormalities included bilateral iris colobomata, cataracts (more dense in the left, precluding fundal view) with phacodonesis bilaterally (figure 2) and bilateral optic disc, retina and

choroidal coloboma (figure 3). AB-scan ultrasonography of the left eye showed a mobile RRD (figure 4).

DIFFERENTIAL DIAGNOSIS

Exudative RD associated with optic disc coloboma
Baraitser-Winter syndrome (BWS).

TREATMENT

Urgent lens aspiration, 23G pars planar vitrectomy, superior drainage retinotomy, heavy liquid, endolaser, cryotherapy, peripheral iridotomy, heavy liquid-air exchange and air-silicone oil exchange was performed under general anaesthesia. Small slit-like breaks were noted within the optic disc coloboma, which could not be lasered. There was a RRD involving the macula. Following meticulous release of vitreous traction at the disc and coloboma, barricade endolaser was applied to the edge of coloboma including the papillomacular (PM) bundle on flattened retina under heavy liquid.

Postoperatively, his intraocular pressure was elevated and attributed to overfill of silicone oil which was released and the pressure was treated medically. His retina remained attached. He received routine tapering dose of topical and oral steroids and topical antibiotics. Apart from the first 24 h postoperatively face down, posture was suggested but not enforced. Instead the patient was given toys to play which would encourage a sit up face tilt downwards position. Silicone oil was removed 12 days after the primary procedure due to elevated intraocular pressure. The eye was left aphakic.

OUTCOME AND FOLLOW-UP

Following removal of silicone oil, the intraocular pressure returned to normal and best corrected visual acuity was 6/18 OD and 6/24 OS at 6 months postoperatively. He has been fitted with contact lenses while awaiting placement of an intraocular lens.

Figure 1 Physical features of our patient consistent with Noonan syndrome.

DISCUSSION

NS is a rare condition that occurs at an incidence of 1:1000 to 1:2500 live births.¹ The phenotype is similar to Turner syndrome. However, there is no sex chromosomal abnormality and it is usually diagnosed clinically. It is due in half of cases to a missense mutation in the *PTPN11* gene on chromosome 12.¹ It has a sporadic or autosomal dominant inheritance with a predominance of maternal transmission² although male-to-male transmission has been recorded.³

Characteristic facial features include hypertelorism, epicanthal folds, downsloping palpebral fissures, expressive triangular-shaped eyebrows and ptosis and strabismus similar to our patient¹ (figure 1) although the phenotype is extremely variable. Other common ocular features include

refractive errors, amblyopia and nystagmus in up to 95% affected individuals.^{3–4} Fundus changes such as optic disc hypoplasia, coloboma and myelinated nerve fibres occur in 20%.^{1–4} Iris and retinal colobomas are a rare feature of NS.⁵ Cataract is even less common at 5%.⁴

Another rare syndrome, the BWS is characterised by iris coloboma, ptosis, hypertelorism and mental retardation.⁶ However, our patient had antenatal genetic diagnosis of NS and is not mentally retarded. This alternative diagnosis remains a possibility. Therefore, to our best knowledge, this is the first reported case of NS/BWS presenting with RRD in association with iris and choroidal coloboma in the presence of cataract.

Of causes of RRD in paediatric population of East Asians, high myopia was the most common while 17%

Figure 2 Anterior segment photograph showing left eye mature cataract and iris coloboma.

Figure 3 Fundus photograph of the left eye postoperatively following oil removal showing flat retina and laser photocoagulation marks involving the edge of coloboma including the papillomacular bundle.

had structural abnormalities such as coloboma.⁷ Poor surgical outcome was significantly correlated with congenital anomaly, macula off and total RD.⁷ The presence of bilateral microcornea in this patient is also interesting. There was a case of a RRD associated with microphthalmos, microcornea and choroidal coloboma successfully treated

with vitrectomy and silicone oil in a 28-year-old Chinese patient.⁸

Conventional surgical management of RRD in coloboma is vitrectomy, endolaser and silicone oil tamponade.^{9–10} However, barricade laser involving the PM bundle is somewhat controversial.⁹ In our experience, this was

Figure 4 B scan ultrasonography of the left eye showing a retinal detachment.

necessary to seal off the coloboma from the flat retina and did not severely compromise vision. Furthermore, persistent detachments are associated with persistent intercalary membrane connections which should be lasered.¹¹ This also supports the aetiology for RRD in disc coloboma as being within the colobomatous disc itself as no peripheral breaks or tears were detected in our patient.

Ocular hypertension appears to be a common complication occurring in 47% of cases and should be treated routinely.¹⁰

The best corrected vision of 6/24 6 months after surgery we believe, is a reflection of the underlying amblyopia secondary to vision deprivation as a result of cataract and ocular misalignment present in this patient. However, with dedicated correction of the refractive error and amblyopic therapy as appropriate in the presence of retinal reattachment, we are optimistic of his ability to function into adulthood with this level of visual acuity.

Learning points

- ▶ RRD may occur in cases of coloboma of the lens and choroid in NS
- ▶ Breaks in such situation are located in the coloboma itself
- ▶ Reattachment of the retina requires drainage retinotomy and barricade laser photocoagulation to the normal retina at edge of coloboma with judicious usage of intraocular tamponade.

Competing interests None.

Patient consent Obtained.

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