

Bilateral rhegmatogenous retinal detachment due to unusual retinal degeneration in Down syndrome

A case report

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Abstract

Rationale: The aim of this study was to report a case of Down syndrome (DS) complicated with bilateral retinal detachment (RD) due to unusual retinal degeneration.

Patient concerns: A 9-year-old girl complained of bilateral visual disturbance during a follow-up examination for myopia and strabismus.

Diagnoses: Slit-lamp examination revealed moderate posterior subcapsular cataract in both eyes. B-mode echography showed bilateral bullous RD; however, it was difficult to detect the causal retinal breaks due to poor mydriasis.

Interventions: For treatment, the patient underwent bilateral lensectomy, vitrectomy, and silicone oil tamponade.

Outcomes: Intraoperative findings revealed symmetrical retinal breaks and unusual caterpillar-like retinal degeneration on the upper temporal side of both eyes. Three months later, the patient underwent bilateral silicone oil removal and intraocular lens implantation.

Lessons: In this case, the retinal degeneration was morphologically different from retinal lattice degeneration, thus suggesting that it might be involved in the onset of DS-related bilateral RD.

Abbreviations: DS = Down syndrome, DSCAM = Down syndrome cell adhesion molecule, Dyrk1A = dual specificity tyrosine-phosphorylation-regulated kinase 1A, RD = retinal detachment, VA = visual acuity.

Keywords: Down syndrome (DS), retinal detachment (RD), unusual retinal degeneration, vitrectomy

1. Introduction

Down syndrome (DS) is the trisomy of chromosome 21 of somatic cells, with an incidence of approximately 1 in every 1000 people. Reportedly, its physical abnormalities include characteristic facial features, congenital heart disease, ankyloproctia, and mental retardation.^[1] Ocular complications associated with DS reportedly included refraction abnormality (mainly hyperopia),

strabismus, nystagmus, blepharitis, keratoconus, cataract, and retinal detachment (RD).^[2–7]

Reportedly, the characteristics of DS-related RD include that it is bilateral, that it is deeply related to trauma resulting from self-injurious behavior, and that detection is delayed due to mental retardation, thus tending to increase the severity of the RD.^[8] In regard to the characteristics of causal retinal breaks, numerous studies have reported cases of retinal dialysis due to trauma,^[9] yet there have been a few reports of retinal breaks due to causes unrelated to trauma.^[4]

In this present study, we report a case of DS with bilateral RD that exhibited symmetrical retinal breaks and unusual caterpillar-like retinal degeneration on the upper temporal side, thus leading us to speculate a causal relationship between this type of retinal degeneration and DS.

2. Case presentation

This study involved a 9-year-old girl who complained of bilateral visual disturbance during a follow-up examination for myopia and strabismus. She had previously been diagnosed with DS by genetic testing (21 trisomy), growth hormone secretion failure, and hyperthyroidism at the Department of Pediatrics, Osaka Medical College, Takatsuki-City, Japan, yet she had no previous history of apparent self-injurious behavior. Upon examination, the corrected visual acuity (VA) in her right and left eye was 0.01 (0.02 × S + 2.0D = C-2.5D Ax120°) and 0.02 (0.04 × S + 3.0D = C-2.5D Ax 65°), respectively.

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This case study was approved by the Ethics Committee of Osaka Medical College.

Written informed consent was obtained from the patient.

The authors have no conflicts of interest to disclose.

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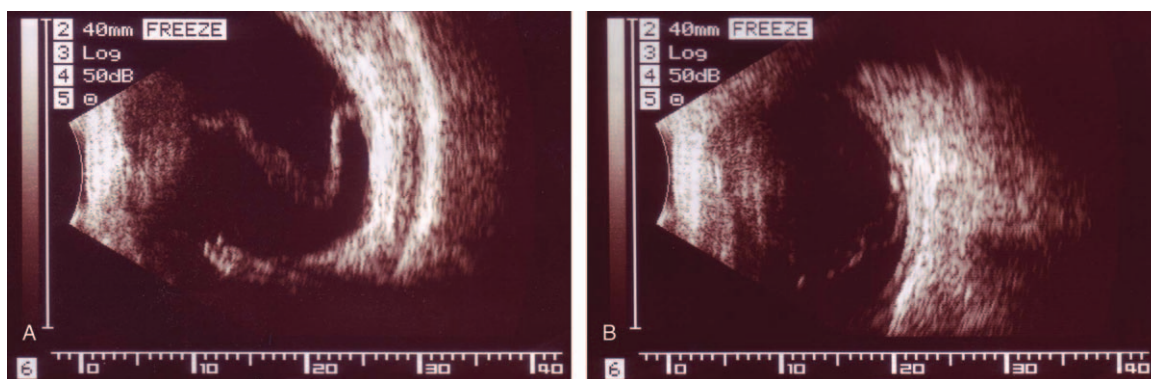


Figure 1. Ultrasound B-mode echography photograph of the patient's right eye (A) and left eye (B) before the initial operation. B-mode echography revealed bilateral bullous retinal detachment.

Hirschberg-method testing of the anterior segment revealed an esotropia of approximately 15°, and slit-lamp examination revealed moderate posterior subcapsular cataract in both eyes. B-mode echography showed bilateral bullous RD (Fig. 1 A, B); however, it was difficult to detect the causal retinal breaks due to poor mydriasis.

Under general anesthesia, vitreous surgery was performed on both of the patient's eyes. Following plana lensectomy, the vitreous gel was removed and an artificial posterior vitreous detachment was created from the posterior pole toward the periphery. Intraoperative findings revealed that caterpillar-like unusual retinal degeneration had been spreading in a circumferential direction on the upper temporal side in both eyes (Fig. 2 A, B). We detected an oval retinal break in the posterior edge of the retinal degeneration in her left eye and small atrophic holes in the thin retinal degeneration in her right eye. Multiple white cross-linked tissues traversing in the meridian direction were observed in the retinal degeneration, yet we found no retinal blood vessels in that area. Those cross-linked tissues were slightly raised and thickened; however, the retina between those tissues had become extremely thin. Thus, these findings were obviously different from those usually observed in retinal lattice degeneration. Moreover, in a somewhat extensive area around the degenera-

tion, firm vitreoretinal adhesions were observed. After the vitreous shaving, we performed pneumatic retinal replacement, endophotocoagulation, encircling by use of a #240 silicon band, and silicone oil tamponade. Post-surgery, the retina was restored, and at 3 months after the initial surgery, silicone oil removal and intraocular lens implantation were performed. At 3 years after the final surgery, the retina was successfully restored in both eyes and the patient's corrected VA improved to (0.6) in her right eye and (0.4) in her left eye.

3. Discussion

Previous reports have pointed out that RD related to DS is sometimes bilateral,^[8] so it is possible that the cases in those studies already had an existing vulnerability apart from a traumatic factor that likely led to RD. In this present case, retinal degeneration spreading in a circumferential direction was observed in a symmetrical position of both eyes, and retinal breaks were observed in and around these degenerations. Lattice degeneration related to the onset of RD had also spread in a circumferential direction; however, the retinal degeneration in this present case was obviously different from the morphology of traditional lattice degeneration.

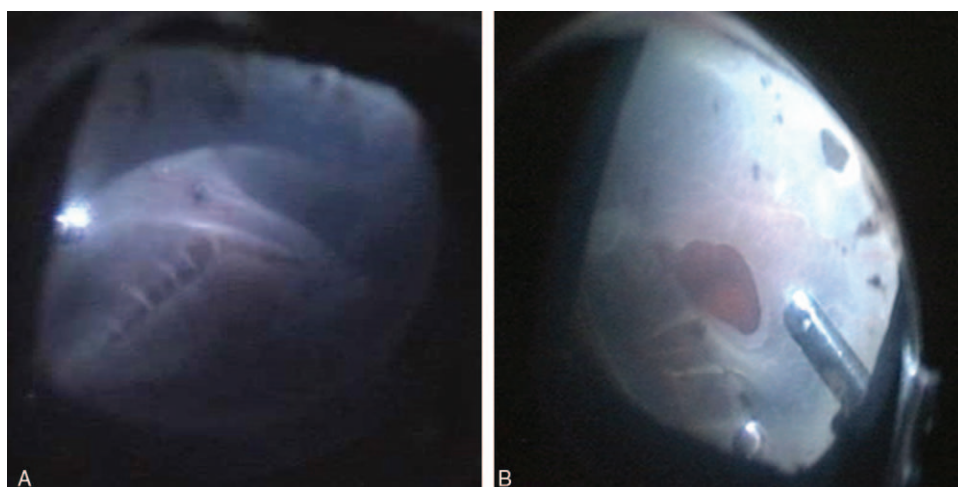


Figure 2. Intraoperative findings of the patient's right eye (A) and left eye (B). The images show unusual caterpillar-like retinal degeneration spreading in a circumferential direction on the upper temporal side in both eyes.

In the degenerations in this present case, we observed multiple, white cross-linked tissues traveling in the direction of the meridian. Usually, hyalinized retinal blood vessels are found in cases of traditional lattice degeneration. However, the cross-linked tissues in this present case were unrelated to retinal blood vessels. In addition, an oval hole was observed in the posterior site of this degeneration in the patient's left eye. In general, a horseshoe tear is formed along the edge of lattice degeneration, yet the oval hole in this case was somewhat distant from the edge of the degeneration. The retinal breaks in our patient's right eye occurred in the thinned area between the cross-linked tissues, which also differs from the atrophic hole in traditional lattice degeneration. To the best of our knowledge, there have been no previous reports of the unique findings on the causal retinal breaks of DS that were observed in this present case.

DS-related ocular findings, such as tigroid fundus and polytomous retinal blood vessels around the optic disc, are observed in many cases, yet they are not usually considered to have no effect on visual function.^[10] However, it should be noted that there have been reports on a few complications that have serious effects on visual function, such as optic atrophy, morning glory syndrome, macular hypoplasia, and macular coloboma.^[11-14] Such findings gave rise to the speculation that DS was associated with the congenital abnormality of the optic disc or retina.

There have been several reports on the genetic abnormalities related to DS and retinal abnormalities. Laguna et al^[15] reported that the dual specificity tyrosine-phosphorylation-regulated kinase 1A (*Dyrk1A*) gene, a serine/threonine kinase essential for brain development and function, was overactivated in DS. In that study, which involved experiments using a DS mouse model, the authors reported that the inner granular layer and the inner reticular layer of the mice whose *Dyrk1A* had tripled were histologically thickened. Although its relationship with the cross-linked tissue observed in this present case is unknown, it may be related to the fact that the retina of this portion is slightly raised and thickened. In addition, the genes present in chromosome 21, such as the DS cell adhesion molecule (*DSCAM*), play an important role in the formation of a network structure in the retina, and it is widely manifested in the central nervous system. Fuerst et al^[16] reported that *DSCAM* was excessively manifested in patients with DS and that it was responsible for retinal structural changes. Having found that the caterpillar-like retinal degeneration with the cross-linked tissues in this present case was very unusual, we speculated the possibility that such a gene abnormality was involved in the retinal structure change.

In conclusion, we believe that the caterpillar-like retinal degeneration in this case can provide new findings on the pathogenesis of RD related to DS.

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References

- [1] Patterson D. Molecular genetic analysis of Down syndrome. *Hum Genet* 2009;126:195–214.
- [2] da Cunha RP, Moreira JB. Ocular findings in Down's syndrome. *Am J Ophthalmol* 1996;122:236–44.
- [3] Creavin AL, Brown RD. Ophthalmic abnormalities in children with Down syndrome. *J Pediatr Ophthalmol Strabismus* 2009;46:76–82.
- [4] Liza-Sharmini AT, Azlan ZN, Zilfalil BA. Ocular findings in Malaysian children with Down syndrome. *Singapore Med J* 2006;47:14–9.
- [5] Berk AT, Saatci AO, Erçal MD, et al. Ocular findings in 55 patients with Down's syndrome. *Ophthalmic Genet* 1996;17:15–9.
- [6] Traboulsi EI, Levine E, Mets MB, et al. Infantile glaucoma in Down's syndrome (trisomy 21). *Am J Ophthalmol* 1988;105:389–94.
- [7] Scherbenske JM, Benson PM, Rotchford JP, et al. Cutaneous and ocular manifestations of Down syndrome. *J Am Acad Dermatol* 1990;22:933–8.
- [8] AlAhmadi BO, Alsulaiman SM, Arevalo JF. Retinal detachment in Down syndrome: characteristics and surgical outcomes. *J Ophthalmol* 2016;2016:6971591.
- [9] Ahmad A, Pruett RC. The fundus in mongolism. *Arch Ophthalmol* 1976;94:772–6.
- [10] Sherk MC, Williams TD. Disc vascularity in Down's syndrome. *Am J Optom Physiol Opt* 1979;56:509–11.
- [11] Altun A, Altun G, Kurna SA, et al. Unilateral morning glory optic disc anomaly in a case with Down syndrome. *BMC Ophthalmol* 2014;14:48.
- [12] Aziz HA, Ruggeri M, Berrocal AM. Intraoperative OCT of bilateral macular coloboma in a child with Down syndrome. *J Pediatr Ophthalmol Strabismus* 2011;48:e37–9. Online.
- [13] Parsa CF, Almer Z. Supranumerary optic disc vessels may indicate reduced systemic angiogenesis in Down syndrome. *Br J Ophthalmol* 2008;92:432–3.
- [14] Hayasaka Y, Hayasaka S. Bilateral congenital macular coloboma in a boy with Down syndrome. *Eur J Ophthalmol* 2004;14:565–7.
- [15] Laguna A, Barallobre MJ, Marchena MÁ, et al. Triplication of *DYRK1A* causes retinal structural and functional alterations in Down syndrome. *Hum Mol Genet* 2013;22:2775–84.
- [16] Fuerst PG, Koizumi A, Masland RH, et al. Neurite arborization and mosaic spacing in the mouse retina require *DSCAM*. *Nature* 2008;451:470–4.