**Comments**

Heterozygous mutations of the gene encoding ribosomal protein S19 (RPS19) on chromosome 19q13.2 are detected in 25% of familial and sporadic cases of DBA. This case met the diagnostic criteria of DBA, but we have not been able to confirm the mutations. These mutations are disorders of ribosomal biogenesis frequently associated with congenital anomalies of midline and craniofacial malformations. The frequent disturbance of extraocular muscles might also be found in craniofacial malformations because the disturbances form orbital cavities. This case showed both strabismus and craniofacial malformation as hypertelorism. We hypothesize that the simultaneous occurrence of dysmorphism and erythroid agenesis in this case may have been the consequence of an insult to the fetus at a critical stage of development of the craniofacial structure and of primitive erythroid cell migration from the yolk sac to the fetal liver and bone marrow.

It is noteworthy that the abnormalities associated with DBA are observed in the head, face, bone, and cartilage, which means they are easily recognized in external examinations. Similarly, ocular manifestations, such as strabismus and ocular hypertelorism, are not difficult to detect by a standard ophthalmological examination. In contrast, pediatric bone marrow failure syndromes such as DBA and Fanconi’s anemia are not easily detected because pediatric patients show only anemia as a symptom.

Ophthalmologists, as the first medical examiners of patients with DBA, have a unique opportunity to make an early diagnosis of a rare disorder with a possible mortal cause. We recommend that patients who present with strabismus, ocular hypertelorism, and a pale complexion should be referred to a pediatric hematologist for a hematological work-up. This case raises the awareness of ocular hypertelorism and exotropia as presenting signs of Diamond-Blackfan anemia.

**Key Words:** congenital hypoplastic anemia, Diamond-Blackfan anemia, exotropia, ocular hypertelorism

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**References**


**End-Stage Glaucoma in Stevens-Johnson Syndrome**

Stevens-Johnson syndrome is an acute, self-limiting disease of the skin and mucous membranes that predisposes patients to ocular complications ranging from minimal (e.g., mild conjunctival hyperemia) to very severe (e.g., corneal melting and perforation). Visual impairment and ocular discomfort continue throughout life. Although secondary glaucoma is a severe complication of ocular surface disease, the thinning and unevenness of corneal thickness in Stevens-Johnson syndrome, as well as the rough ocular surface, may lead to delayed diagnosis of secondary glaucoma as a result of underestimation or misreading of the true intraocular pressure (IOP).

**Case Report**

A 25-year-old Japanese woman with Stevens-Johnson syndrome had been treated since March 2003 with topical dexamethasone three times per day for conjunctival inflammation. The ocular surface inflammation worsened, and from February 2007 topical dexamethasone was increased to six times per day. While her visual acuity decreased from 0.07 to light perception, her ocular inflammation increased. She was referred to the corneal clinic of Keio University Hospital in March 2007 complaining of visual loss and ocular surface irritation. Vision in both eyes was hand motion. IOP in both eyes was 15 mmHg, measured by Goldmann applanation tonometry (GAT), but when measured with Tono-Pen XL (Medtronic Solan, Jacksonville, FL, USA), the IOP was 45 mmHg in the right eye and 30 mmHg in the left eye. Slit-lamp examination revealed conjunctivalization, corneal keratinization, and corneal opacification, typical manifestations of Stevens-Johnson syndrome (Figs. 1 and 2). Central corneal thickness measured by ultrasound pachymetry was 235 μm in the right eye and 240 μm in the left eye. Optic disc evaluation showed glaucomatous optic neuropathy with total cupping. Visual field measured by Goldmann kinetic perimetry showed only central vision. Angles evaluated by ultrasound biomicroscopy were open.

We diagnosed her condition as end-stage secondary glaucoma. Dexamethasone was stopped and latanoprost once daily, timolol twice daily, and brinzolamide twice daily.
Secondary glaucoma is a severe complication of ocular surface disease. Chronic inflammation induced by Stevens-Johnson syndrome decreases the central corneal thickness, increases unevenness of the central corneal thickness, and roughens the ocular surface, which makes measuring true IOP difficult. The true IOP of the present patient was underestimated because of the reduction in corneal thickness, or the corneal edema, when measured by GAT. One case of misreading of the IOP, after which laser in situ keratomileusis was performed, has been reported; this case resulted in end-stage glaucoma with thinning of the central corneal thickness.

A characteristic ocular surface finding in elevated IOP is corneal edema that mimics the ocular inflammation of Stevens-Johnson’s syndrome. IOP elevation itself causes visual acuity loss and corneal edema that appear as a worsening ocular surface inflammation. The glaucoma remained undetected in this patient because the IOP measured by GAT remained within normal limits because of the corneal thinning, the rough ocular surface, and the corneal edema. IOP measured by Tono-Pen was also affected by central corneal thickness, but Tono-Pen has proved accurate when used with scarred corneas. Therefore, the IOP of severe Stevens-Johnson syndrome patients should be measured not only by GAT but also by Tono-Pen and other available methods.

Key Words: glaucoma, Stevens-Johnson syndrome, tonometry
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in both eyes were started for 1 week. However, since IOP, as measured by Tono-Pen, remained at 30 mmHg in both eyes, a trabeculotomy was performed in both eyes. Three months after surgery, the IOP in both eyes, without anti-glaucoma medication or topical dexamethasone, decreased to 12 mmHg when measured by Tono-Pen and to 10 mmHg when measured by GAT; her vision in both eyes, however, remained light perception. IOP was measured at least three times with Tono-Pen and with GAT, always by the same investigator (K.Y.), who has a subspecialty in glaucoma. We used the final mean IOP value measured by Tono-Pen with a coefficient of variation ≤5%.

Figure 1. Slit-lamp examination of the right eye of a 25-year-old Japanese woman with Stevens-Johnson syndrome. Typical manifestations of Stevens-Johnson syndrome such as conjunctivalization, corneal keratinization, and corneal opacification can be observed.

Figure 2. A Pentacam rotating Scheimpflug camera revealed a significant thinning of corneal thickness.