

Atypical association of Duane retraction syndrome and Bardet Biedl syndrome

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Duane's retraction syndrome (DRS) includes changes in palpebral fissure width along with restriction of ocular motility. Bardet Biedl syndrome (BBS) includes presence of retinitis pigmentosa (RP) with obesity, mental retardation, polydactyly and renal abnormalities. We report a case of rare association of DRS with BBS in a seven-year-old child. The ocular motility examination revealed left DRS with esotropia. Fundus examination revealed findings characteristic of an atypical retinitis pigmentosa. The electro-retinogram waveforms were extinguished both for rods and cones. He was diagnosed as a case of BBS on the basis of the ophthalmological findings plus association with the systemic features of obesity, polydactyly, hypogonadism, mental retardation and renal abnormalities. This case gives further evidence of the fact that BBS may be associated with abnormalities of eye movements.

Key words: Bardet Biedl syndrome, Duane's retraction syndrome, retinitis pigmentosa.

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Duane's retraction syndrome (DRS) is characterized by globe retraction and palpebral fissure narrowing on adduction, with restriction of abduction or adduction or both.¹ The proposed mechanism for the impaired ocular motility is congenital miswiring of the lateral and medial rectus muscles.¹ Electromyographic studies show decreased or absent firing of the lateral rectus and medial rectus during attempted adduction causing globe retraction.² It may be associated with other ocular as well as systemic congenital anomalies such as sensorineural deafness, upper limbs defect, facioauriculo-vertebral anomalies, cardiac and genito-urinary anomalies.¹

The cardinal features of Bardet-Biedl syndrome (BBS) are obesity, poly or syndactyly, mental retardation, hypogonadism and retinitis pigmentosa (RP).³ It may be associated with other ocular defects such as nystagmus, strabismus, keratoconus, cataract, ptosis, ophthalmoplegia and constricted visual fields.^{3,4} Herewith, we report a case of unilateral DRS Type I and high myopia in association with BBS.

Case Report

A seven-year-old boy presented with a history of squint since

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birth and progressive night blindness. He was a product of full-term normal delivery, born out of a second degree consanguineous marriage with no family history of strabismus but with a family history of RP. He was one of twins; the other died at birth. He had multiple systemic anomalies including facial asymmetry, ventricular septal defect, polydactyly and hypospadias with mental retardation. The anterior segment examination was normal. The corrected visual acuity of right eye was 7/200 with -7.0 diopters sphere (DS) and left eye 20/80 with -7.0 DS. There was a compensatory face turn to the left [Fig. 1a] along with horizontal jerky nystagmus on all gazes. The ocular motility examination revealed a 30 prism diopters (PD) esotropia with complete inability to abduct the left eye [Figs. 1 a, b]. Palpebral fissure narrowing was present on adduction consistent with DRS Type I (Huber's classification).¹

Indirect ophthalmoscopy revealed myopic discs, attenuated vessels and pigment deposition in the mid-peripheral retina in clumps, characteristic of RP [Figs. 2 a, b]. Electroretinogram (ERG) was performed using bipolar Burian-Allen contact lens electrode (LKC, Gaithersburg); Kurbisfeld stimulation was performed using a photostimulator set at intensity of -25 decibels (db), 0 db after dark-adapting the child for 20 min under general anesthesia. The child was tested both under



Figure 1a: Complete restriction of left eye abduction in levoversion



Figure 1b: The ductions of left eye are full on right gaze. Also the narrowing of left palpebral fissure, characteristic of DRS, is seen

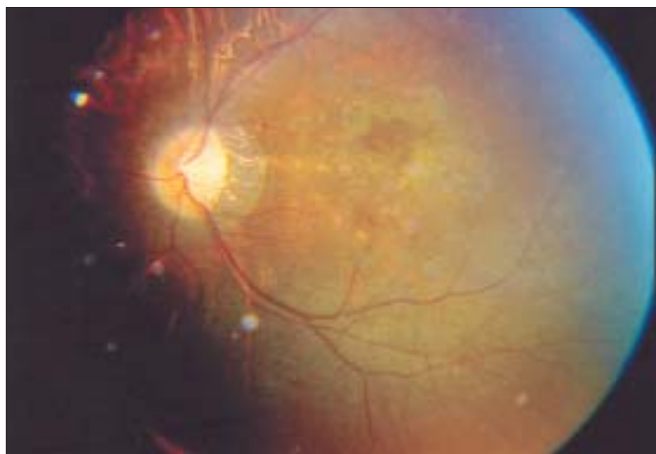


Figure 2a: The myopic disc with crescent in right eye fundus. Arteries are attenuated with pigment mottling and maculopathy

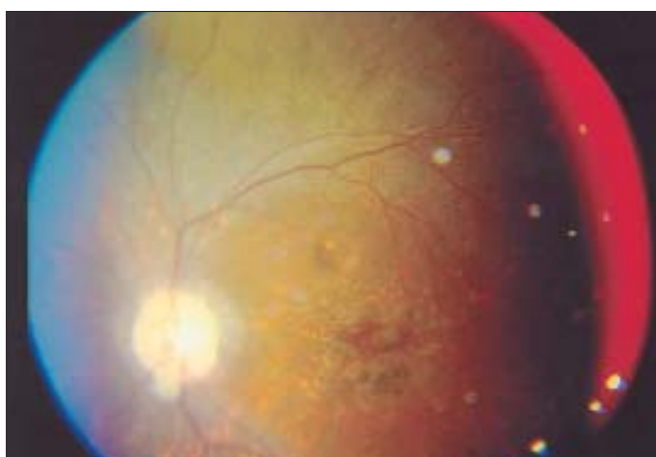


Figure 2b: Pigment mottling with maculopathy in left eye fundus

photopic and scotopic conditions. The ERG waveforms (UTAS 3000, LKC Technologies, Gaithersburg) were extinguished both for rods and cones suggestive of RP [Figs. 3 a, b]. Central visual fields were constricted to 20°.

Discussion

Duane's retraction syndrome is associated approximately 10-20 times more frequently with other congenital systemic anomalies and is thought to be due to an insult during the 10th week of embryogenesis. It has been associated with congenital malformations of the skeletal, auricular, ocular and neural systems.⁴

Guirgis *et al.*⁵ reported a case of DRS with esotropia accompanying cone-rod dystrophy in a patient with achondroplasia. Cone-rod dystrophy has been associated with a number of systemic disorders, including BBS.³ McCullah and Cummings⁶ reported a patient with unilateral DRS with pseudo RP. Pelit *et al.*⁷ reported a case of DRS Type I with RP in a middle-aged woman with ERG showing nondetectable rod and cone responses. The female, however, did not have any features of BBS. The chromosomal study did not reveal any association and the authors postulated it to be a chance occurrence. Lavy *et al.*⁸ reported that 64% of patients with BBS had abnormal eye movements.

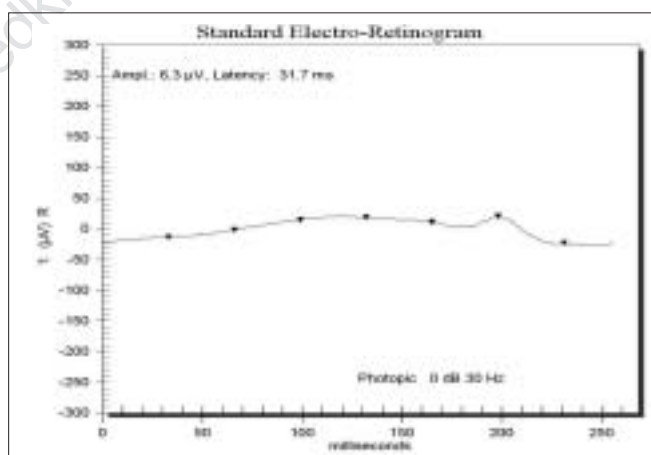
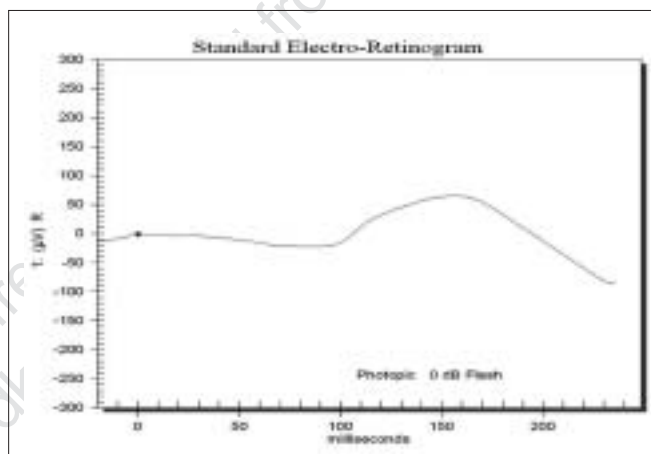
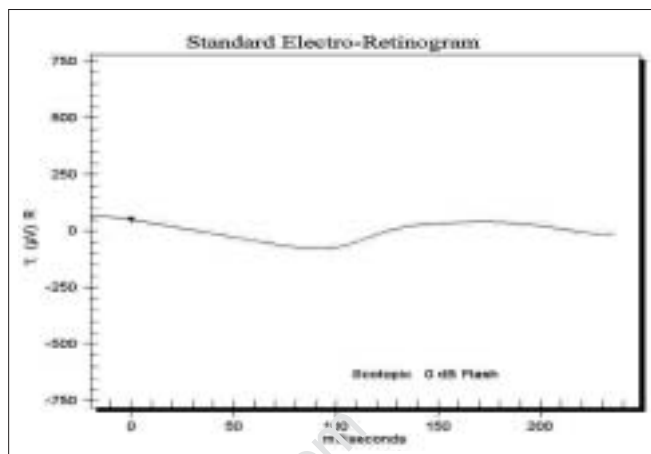


Figure 3a: The waveforms of right eye. Both the rod (scotopic 0dB) and cone responses (photopic 0dB) are extinguished. Photopic 30Hz flicker responses show very small amplitudes and increased latency

BBS may be associated with pigmentary degeneration of the retina and other ocular defects including nystagmus, strabismus, keratoconus, cataract, constricted visual fields, ptosis and ophthalmoplegia.^{3,4} A high rate of consanguinity is found among the parents (25-40%).

In our case, the child was diagnosed as a case of BBS with progressive night blindness, visual field constriction noted to be progressive. ERG showed diminution of both photopic and

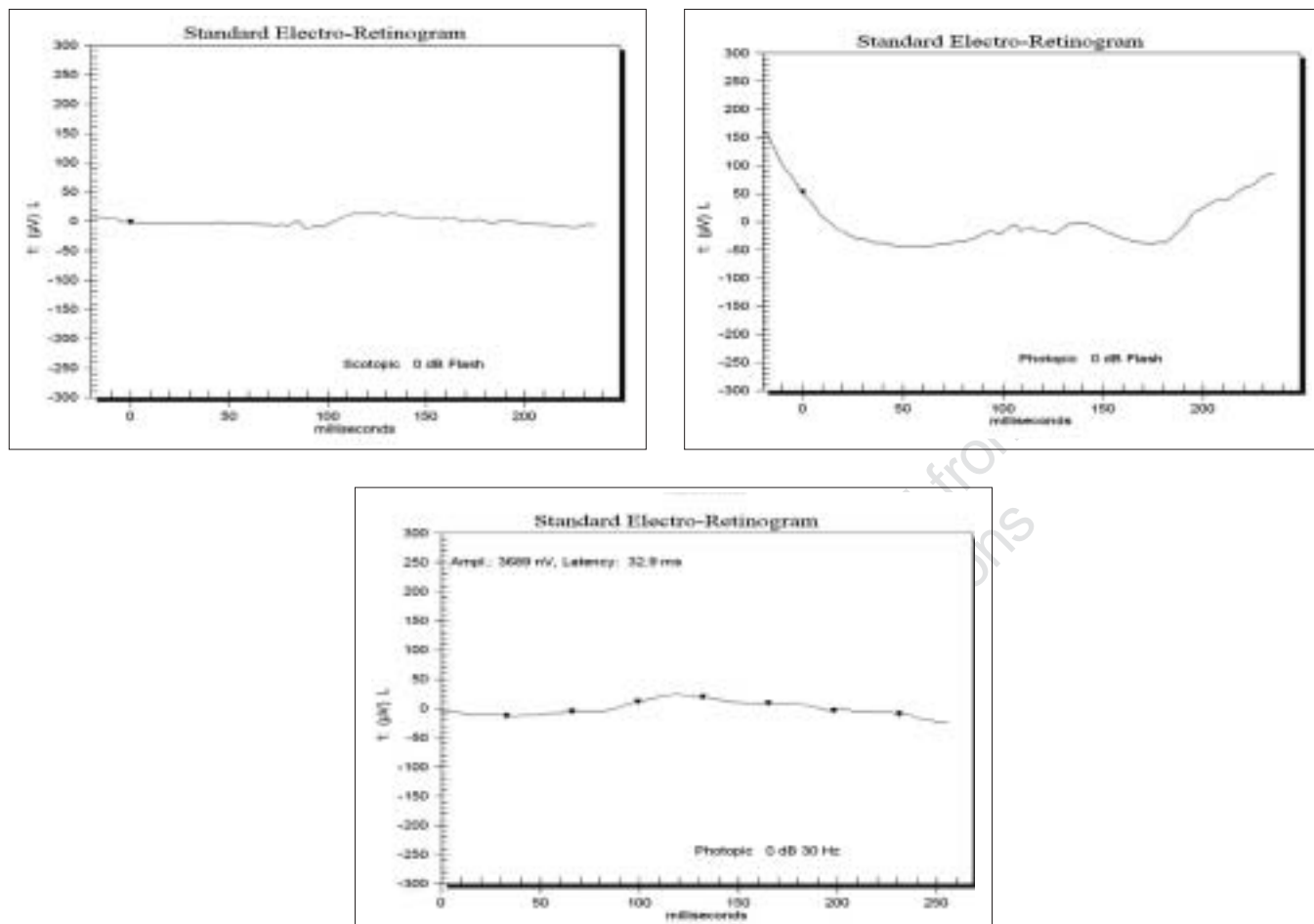


Figure 3b: The waveforms of left eye. Both the rod (scotopic 0dB) and cone responses (photopic 0dB) are extinguished. Photopic 30Hz flicker responses show very small amplitudes and increased latency

scotopic responses. There is a definite family history of RP but not of strabismus. Pelit *et al.*⁶ reported a similar case and found that this was a chance occurrence and the chromosomal analysis was normal. We did not do a detailed chromosomal analysis since it was not possible in our setup. This rare case of DRS in a patient of BBS gives evidence of the variability of the features of BBS and also demonstrates that this condition can be associated with abnormalities of eye movements.

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