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SHORT COMMUNICATION

Case report

Ocular findings in cerebro-oculo-facial-skeletal syndrome (Pena-Shokeir-II syndrome)

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PURPOSE. *To report the ocular findings in cerebro-oculo-facial-skeletal syndrome or Pena-Shokeir-II syndrome.*
METHODS. *Case report.*
RESULTS. *A five-month-old male infant presented with bilateral posterior polar cataract, microphthalmos, nystagmus, and marked non-glaucomatous optic nerve atrophy. Systemic abnormalities such as microcephaly, micrognathia, flexion contractures of the elbows and knees, hypotonic musculature, and failure to thrive, with pronounced statomotor retardation, led to the diagnosis of cerebro-oculo-facial-skeletal syndrome or Pena-Shokeir-II syndrome. Cataract surgery did not improve the poor visual performance.*
CONCLUSIONS. *Cerebro-oculo-facial-skeletal syndrome (Pena-Shokeir-II syndrome) should be included in the differential diagnosis of bilateral microphthalmos, congenital cataract, nystagmus, and pronounced optic nerve atrophy, and cataract surgery does not markedly improve vision. (Eur J Ophthalmol 2003; 13: 209-11)*

KEY WORDS. *Cerebro-oculo-facial-skeletal syndrome, Pena-Shokeir-II syndrome, Bilateral congenital cataract, Microcephaly, Neurogenic arthrogryposis*

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INTRODUCTION

The cerebro-oculo-facial-skeletal syndrome, originally described by Pena and Shokeir in 1974 (1), is a rapidly progressive neurological autosomal recessive inherited disorder of neurogenic arthrogryposis, failure to thrive, microencephaly, mild ventriculomegaly, delayed and abnormal cerebral myelination, severe degenerative changes of the cerebellum in older children involving the internal granular layer and Purkinje cell layer, infantile spasms, characteristic facial changes such as large ears, prominent nose, micrognathia, and a long philtrum, wide-set nipples, camptodactyly, and flexion contractures of the elbows and knees (2-10). Ophthalmic features include microphthalmos, cataract, microcornea, and optic nerve atrophy.

The cerebro-oculo-facial-skeletal syndrome must be distinguished from Cockayne´s syndrome which is a recessively inherited neurodegenerative disorder involving low-to-normal birth weight, growth failure, brain dysmyelination with calcium deposits, cutaneous photosensitivity, retinitis pigmentosa, cataracts, and sensorineural hearing loss. Cells of patients with Cockayne syndrome are hypersensitive to ultraviolet radiation because of their impaired nucleotide excision repair of UV-induced damage in actively transcribed DNA. The abnormalities in Cockayne's syndrome are associated with mutations in the Cockayne syndrome group A or B gene. It has now been reported that children suffering from the cerebro-oculo-facial-skeletal syndrome have the same mutation in the Cockayne syndrome group B gene, suggesting that the two syndromes share a common pathogenesis (10).

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Case report

Described here are the ocular findings in a child with Pena-Shokeir-II syndrome. A five-month-old male infant presented with marked failure to thrive, and pronounced statomotor retardation. This was the first child of his 19-year-old father and 19-year-old mother who were first cousins with an unremarkable family history. Pregnancy had been normal, birth occurred spontaneously in the 39th week of gestation, and birth weight was 2640 g. Pediatric examination revealed microcephaly, micrognathia, flexion contractures of the right knee, muscle hypotony, and general statomotor retardation. There was low-frequency nystagmus, probably mainly due to the inability to take up fixation. According to the parents, the nystagmus had started around the third month. No other neurologic deficits were detected. Cranial computed tomography showed general atrophy of the brain including the cerebellum, with enlargement of the ventricular spaces. All these features led to the diagnosis of cerebro-oculo-facial-skeletal syndrome or Pena-Shokeir-II syndrome. Cockayne's syndrome, as differential diagnosis, was not considered to be the main diagnosis mainly on account of the child's general appearance and since there was no cutaneous photosensitivity or pigmentary retinopathy.

Ophthalmologic examination under general anesthesia revealed anterior microphthalmos, with a corneal diameter measuring 10.0 mm horizontally and 9.5 mm vertically in both eyes. The radius of the anterior corneal curvature was 6.95 mm and 6.98 mm. There were bilateral posterior polar cataracts, slightly decentered in the nasal direction. No persisting anterior or posterior hyaloid artery was detected.

The visibility of the macular region by direct or indirect ophthalmoscopy, and by Goldmann contact lens examination of the fundus, was reduced, so that in the eye that was eventually operated the details of the macula were first seen only after cataract surgery. The depth of the anterior chamber, measured sonographically, was 1.90 mm for the right eye and 2.10 mm for the left, and the lens thickness was 4.30 mm and 3.90 mm. Axial length measured sonographically during a follow-up examination at the age of 14 months was 14.3 mm for the right eye and 14.7 mm for the left eye, i.e. much shorter than normal values for the age. Retinoscopy revealed bilateral hyperopia of +7 diopters in the right eye and +3.5 diopters in the left.

Ophthalmoscopy indicated the optic disc area was approximately 1.5 mm^2. The configuration of the neuroretinal rim was normal with its smallest part in the temporal region of the optic disc, the rim was pale, and visibility of the retinal nerve fiber layer was markedly reduced. The macula appeared mostly unremarkable except for a lack of the wall reflex. Intraocular pressures in the right and left eyes were 12 and 13 mm Hg. In the bright-flash Ganzfeld electro-retinogram, photopic and scotopic responses were normal for both eyes.

At the age of 20 months, body weight had increased to 7300 g. Head circumference was 38.5 cm. The child reacted only slightly to calling, touching or other external stimuli. His muscles were hypotonic, and he was unable to sit or stand. There was a flexion contracture of his knees of about 20 degrees. Cataract surgery was performed in the right eye with implantation of a posterior chamber lens combined with planned posterior capsulotomy without vitreous prolapse. The dioptric power of the intraocular lens implanted was +35 diopters, the highest power available.

To deal with a secondary cataract growing on the intact anterior vitreous surface, Nd-Yag laser capsulotomy was done six months after cataract surgery. Despite cataract surgery and capsulotomy, visual acuity as determined from the child's behavior did not improve. The child did not take up fixation depending on the stimulus presented. Fixation behavior and nystagmus were unchanged. Measurement of visual acuity by preferential looking tests was not possible because of the child's lack of cerebral development.

DISCUSSION

The present report on the association of cerebro-oculo-facial-skeletal syndrome or Pena-Shokeir-II syndrome with bilateral cataract suggests that this should be added to the panoply of syndromes such as Cockayne`s and Seckel's syndromes, and Potter's anomaly, which can be associated with congenital bilateral cataract in microphthalmic eyes. Because of the microcephaly, including involvement of the optic nerve, cataract surgery may not markedly improve visual performance in infants suffering from Pena-Shokeir-II syndrome.

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