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A CASE OF CHARGE ASSOCIATION WITH MICROCORNEA

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A 2-year-old Japanese girl presented with esotropia, microcornea and choroidal coloboma in both eyes. There were other abnormalities such as congenital heart disease, retarded growth, and hearing impairment. According to these findings, she was diagnosed as having CHARGE association. To our knowledge, this is the first case of CHARGE association with microcornea.

Key words: CHARGE association, microcornea, choroidal coloboma, esotropia

INTRODUCTION

It is well known that ocular coloboma has been observed in association with other congenital anomalies (1). In 1981, Pagon and co-workers first described and named CHARGE association for six major features: coloboma, heart defects, atresia of the choanae, retarded growth and development, genital hypoplasia, and ear anomalies and/or hearing loss (2). Ocular abnormalities observed with the CHARGE association are microphthalmos, squint, nystagmus, and lacrimal canalicula atresia (3, 4). Additional features are retinal detachment in association with posterior coloboma (5, 6), orbital cyst (7, 8) and optic nerve tumor (9). We examined a rare case of CHARGE association with optic disc coloboma, congenital heart disease, retarded growth, hearing impairment, squint, and microcornea.

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CASE PRESENTATION

A 2-year-old Japanese girl with esotropia was referred to us for further examination. She was the product of a full-term, uncomplicated gestation (birth weight, 2,928 g). No evidence of heritable disease could be reported in her family, and the pregnancy was normal. No known teratogens were identified. She had approximately 50Δ esotropia (Fig. 1). The corneal diameter was 9 mm (normal range, above 10 mm) and axial length was above 21.5 mm (normal range, 21-26 mm) in each eye. Her anterior segments except for the corneas and ocular media were normal. Funduscopy revealed optic disc coloboma and choroidal coloboma at and inferior to the optic nerve head in both eyes (Fig. 2). Echocardiography confirmed the presence of a ventricular septal defect (data not shown). Retarded growth was indicated by a low (14.4) Kaup index (normal range, 15-19) at age one month. Atresia of the choanae, genital hypoplasia, and ear anomalies were not noted, but she reacted poorly to sound. Her chromosomes were normal. Based on these findings, a diagnosis of CHARGE association was made. Three years later on a routine visit, her best corrected visual acuity was light perception OD and 0.3 OS. Ultrasound axial length was 22.6 mm OD and 21.7 mm OS. The corneal diameters were 9 mm in both eyes.

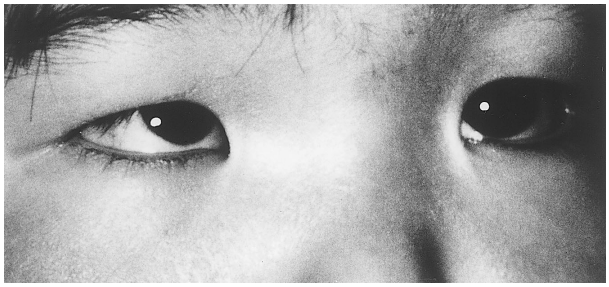

Fig. 1. Esotropia and microcornea are observed in each eye.

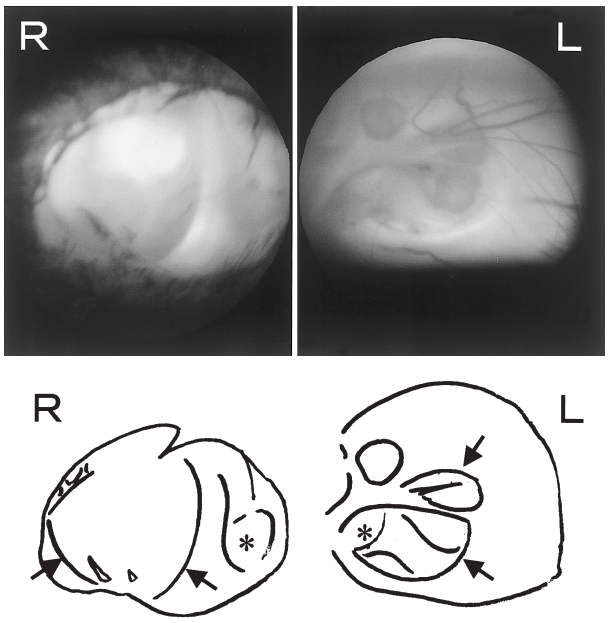

Fig. 2. Fundus photograph shows disc and choroidal colobomas in both eyes. Arrows indicate coloboma and asterisks indicate optic nerve head.

DISCUSSION

We describe a 2-year-old girl with optic disc coloboma, microcornea, congenital heart disease, growth deficiency, and hearing impairment. The CHARGE association is diagnosed on the basis of any four of the six cardinal features of ocular coloboma, congenital heart disease, atresia choanae, retarded growth or development, genitor-urinary anomalies, and ear anomalies or hearing loss. Russell-Eggitt et al. reported 44 cases of CHARGE association with coloboma, and 21 of their patients had microphthalmos (3). Our patient had microcornea (corneal diameter, 9 mm bilaterally), but her axial length was within normal range, as observed by B-scan ultrasonography. We are unaware of previous reports of the microcornea with CHARGE association and could find no reference in a computerized search utilizing Medline. The majority of reported cases of CHARGE association have been sporadic (3, 11). The possibility exists that these malformations are the consequence of unrecognized teratogens (3). Autosomal dominant pedigrees of CHARGE association were also reported (10, 11). Environmental or genetic causes may act similarly. The defects seen in CHARGE association, including microphthalmos, can be attributed to the arrest in various aspects of normal embryologic differentiation during the fifth to sixth weeks after conception (3). On the other hand, embryological ocular impairment after fifth month's gestation can lead to microcornea (12). There appears to be an arrest in the growth of the cornea, a process that begins after the fifth month of gestation, when differentiation is complete (12). Ocular abnormalities in our patient presumably resulted from morphogenetic impairment during this period (fifth weeks to 5 months). In this point of view, our case of CHARCE association (disorder of differentiation) coexisting with microcornea (disorder after differentiation) seemed to be rare and notable.

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