Nanophthalmos and situs inversus totalis

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Nanophthalmos is characterized by short axial length, high hypermetropia, thick sclera and a normal-sized crystalline lens. Situs inversus totalis is the mirror image of the normal morphology of the thoracic and abdominal viscera. To the best of our knowledge this is the first report of a nanophthalmic patient with situs inversus totalis. Therefore, we would like to invite the attention of our colleagues to our case and underline the importance of the systemic examination of the nanophthalmic patients to detect systemic malformations and visceral transpositions.

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Nanophthalmos represents an arrested development of the globe in all directions after the closure of the embryonic fissure. It is a rare clinical disorder characterized by a small eye with short axial length, high hypermetropia, thick sclera and a normal-sized crystalline lens.[1,2] The axial length in nanophthalmos measures 14-20 mm.[3] Nanophthalmic patients have a high risk for developing glaucoma and choroidal detachment. Moreover, in children, high hyperopia with resultant amblyopia and strabismus are among the more important ophthalmic considerations.[4] To the best of our knowledge, this is the first report describing the association of nanophthalmos and situs inversus totalis.

Case Report

A five-year-old boy presented with complaint of esodeviation on his right eye. The best-corrected visual acuities in the right eye and in the left eye were 20/60 with +14.00 diopter (D) (+0.25 D cylinder α 140°) and +14.00D (+0.50 D cylinder α 50°), respectively. He had a comitant 25-D esotropia at distance and near in his right eye. Both eyes had Grade 2 overaction of inferior oblique muscles. Patient had suppression of the right eye. Ductions and versions were normal in both eyes.

Horizontal and vertical corneal diameters were 11.5 mm and 10.0 mm, respectively in each eye. Results of a biomicroscopic examination of the anterior and posterior segment of the eyes were unremarkable. His intraocular pressures were 13–14 mm Hg in both eyes. The axial length measured 16.24 mm in the right eye and 15.64 mm in the left eye [Fig. 1].

The patient was consulted by a pediatrician due to growth retardation. No pathologic finding, but situs inversus totalis was found by physical examination, computed tomography and echocardiography. Computed tomography showed mirror image transposition of the abdominal and thoracic viscerae [Fig. 2]. Dextrocardia was diagnosed but anatomic appearance and the M mode measurements of the heart were normal [Fig. 3]. Family history and maternal history during pregnancy revealed no abnormal finding. Intrauterine growth retardation (IUGR) was not stated by the past medical history of the patient. Postnatal history of the case revealed that the child had been born as 2700 g in weight and 48 cm in length, which excluded IUGR. Chromosomal analysis gave no abnormality and no chronic respiratory infection episodes were reported. The study of his pedigree showed sporadic pattern. The growth retardation of the patient was suggested as related to insufficient dietary intake of nutrients.

Discussion

Nanophthalmos is characterized by short axial length, thick sclera, narrow palpebral fissure, small orbit, shallow anterior chamber, normal-sized lens, and hyperopia of up to 20 D.[1,2] The small eye without ocular malformations is considered to be nanophthalmic.[5] Complications such as glaucoma, macular hypoplasia, papillomacular folds, and pigmentary retinal dystrophy have been reported.[3] But these complications were not found in our case.

The etiology of nanophthalmos is still not known. The majority of the cases are sporadic, but autosomal recessive and autosomal dominant inheritance have been reported. Glycogen-like deposits, together with the abnormal fibrils, are thought to lead to scleral rigidity that causes a small intraocular volume, choroidal congestion, detachment, and exudative retinal detachment.[6,7] Karyotype analysis of the patient revealed no chromosomal anomalies. Both orbital fissure closure and visceral organ rotation occur between 5th-10th gestational weeks.[8] Thus we suggest that our patient might have been affected during this time period.

It was shown by histological examination that the nanophthalmic sclerae were much thicker, and the collagen fibrils showed a less orderly arrangement than the sclerae of the normal controls.[7] Nanophthalmic eyes are at risk for angle-closure glaucoma, uveal effusion, and exudative retinal detachment.[9] Angle-closure glaucoma occurs in nanophthalmic eyes because of the large lens in the small eye. Uveal effusion is the result of choroidal congestion secondary to impaired vortex venous drainage through the thick sclera.[6]

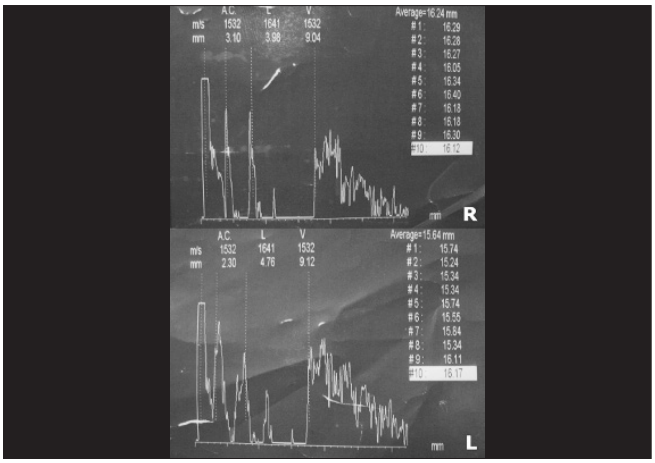

Figure 1: The A-scan measurements of the eyes

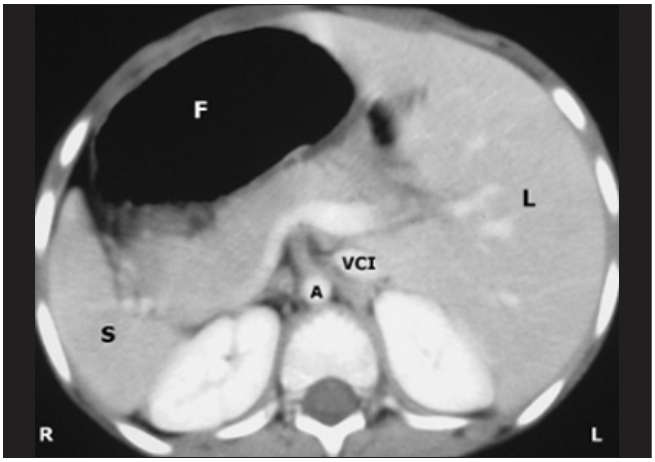

Figure 2: The computed tomography image of the abdomen. Note the situs inversus anatomy of the abdominal organs. A: Aorta F: Fundus of the stomach L: Liver S: Spleen VCI: Vena Cava Inferior

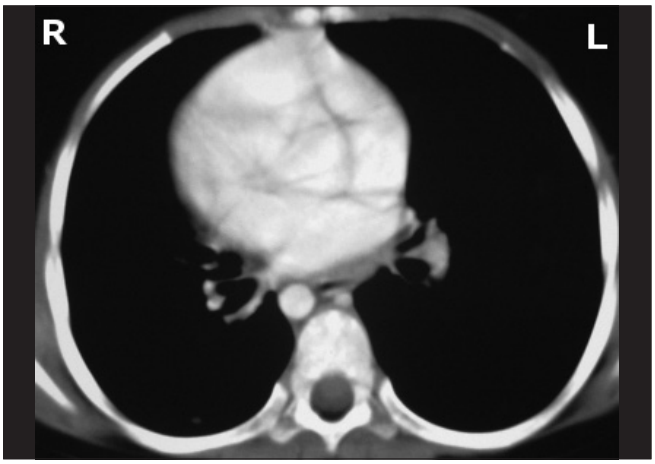

Figure 3: Thoracic computed tomography scan shows the dextrocardia

The high hyperopia leading to ametropic amblyopia, associated strabismus leading to strabismic amblyopia are the main complications in children.[4] Strabismus usually presents with esotropia such as in our case. The short axial length with high hyperopia is the most likely factor in the development of esotropias in nanophthalmic eyes.[4]

Situs inversus totalis is a condition with left-to-right reversal of the viscera combined with dextrocardia. In addition to their abnormal position, individual organs present a symmetrical mirror image morphology. Situs inversus totalis associated with primary ciliary dyskinesia is known as Kartagener syndrome. Patients with primary ciliary dyskinesia have recurrent sinus and pulmonary infections due to immotile cilia that line their upper airways. No recurrent infections were reported in the medical history of our patient. So we did not think immotile cilia syndrome in the case.

The prevalence of situs inversus totalis seems to range between 1 in 8000 and 1 in 25000.[10] No racial predilection exists and the male-to-female incidence is 1:1. Situs inversus totalis is usually discovered incidentally as in our case. Dextrocardia is easily diagnosed by clinical examination. Although chest roentgenogram, ultrasound, scintigraphy and angiography are used in the diagnosis of visceral organ transpositions, computed tomography and magnetic resonance imaging are the preferred examinations for the definitive diagnosis of situs inversus totalis. The recognition of situs inversus totalis may help avoid mishaps at surgery or other interventions that result from the failure to recognize reversed anatomy or an atypical history.

Both nanophthalmos and situs inversus totalis are rare clinical conditions. To the best of our knowledge, this is the first report of a nanophthalmic patient with situs inversus totalis. The clinicians should keep in mind that nanophthalmos may accompany situs inversus totalis.

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