Goldenhar’s Syndrome Associated With Occipital Meningoencephalocele
—Case Report—

Daisuke KITA, Shigeru MUNEMOTO, Yasunao UENO*, and Akiko FUKUDA*

Departments of Neurosurgery and *Pediatrics, Ishikawa Central Prefectural Hospital, Kanazawa, Ishikawa

Abstract

A male neonate presented with Goldenhar’s syndrome (oculoauriculovertebral dysplasia), a combination of facial microsomia and auricular malformation, associated with occipital meningoencephalocele. Three-dimensional computed tomography images clearly showed a suboccipital cranial cleft extending to the foramen magnum and hypogenesis of the left temporal bone. The patient died of heart failure due to ventricular septal defect at age 5 months.

Key words: Goldenhar's syndrome, occipital meningoencephalocele, skeletal anomaly, three-dimensional computed tomography

Introduction

Goldenhar’s syndrome (oculoauriculovertebral dysplasia) is a combination of facial microsomia and auricular malformation,2) and may be caused by dysplasia of the first and second branchial arches.3) The frequency of this syndrome is estimated as one per 5600 to 26550 births, and the male:female ratio is about 3:2.4) We discuss a case of Goldenhar’s syndrome associated with meningoencephalocele and describe the skeletal abnormalities on three-dimensional computed tomography (3D CT) images.

Case Report

The patient was a male infant delivered to a 26-year-old primiparous healthy mother. The pregnancy was uneventful and the family history was unremarkable. Ultrasonography revealed that the fetus had a large mass adhering to the occipital region at the 35th week of gestation. Cesarean section was performed at the 39th week of gestation. Birth weight was 3172 g and height was 49.5 cm. Apgar score was 8 and 10 at 5 and 10 minutes, respectively. An encephalocele of the same size as the neonate’s head was attached to the suboccipital region (Fig. 1). Hemifacial microsomia and deformation of the auricle on the left were also noted. The facial appearance was asymmetrical, indicating left facial nerve paresis.

Cardiac ultrasonography revealed ventricular septal defect. The karyotype was normal. Magnetic resonance imaging revealed that the content of the meningoencephalocele was primarily fluid with a

Fig. 1 Photograph showing the occipital meningoencephalocele and deformation of the left auricle.
small quantity of impacted cerebellar cortex (Fig. 2). 3D CT imaging showed a suboccipital cranial cleft extending to the foramen magnum, hypogenesis of the left temporal bone, and hypoplasia of the left zygomatic arch (Fig. 3).

Resection of the meningoencephalocele was performed on the 5th day after birth. Histological examination of the resected portion of the impacted cerebellum showed complex meninges and dysplastic choroid plexuses. Thirty days after the resection, the patient developed hydrocephalus, which was controlled by ventriculoperitoneal shunting. However, he died of heart failure due to the ventricular septal defect at age 5 months.

**Discussion**

Most cases of Goldenhar's syndrome are sporadic but the reported familial cases are very likely to involve autosomal dominant inheritance. The syndrome is also called oculoauriculovertebral “spectrum” because of the occasional association with skeleton, heart, urinary tract, and other organ anomalies. Cervical vertebral fusion, spina bifida, hemivertebrae, and scoliosis are frequent skeletal abnormalities in this syndrome, and ventricular septal defect or tetralogy of Fallot occurs in about 50% of the heart defects. In addition, pulmonary and renal anomalies are also common major complications of Goldenhar’s syndrome. However, encephalocele has been rarely reported. An autopsy case of Goldenhar’s syndrome with encephalocele (cerebellocele) showed cranium bifidum in the occipital region and defective posterior arch of the C-1 vertebral body. In our case, the occipital meningoencephalocele was possibly caused by dysraphism of the foramen magnum, which is one of the skeletal anomalies seen in the oculoauriculovertebral “spectrum.”

**References**


*Address reprint requests to: D. Kita, M.D., Department of Neurosurgery, Himi Municipal Hospital, 31–9 Saiwai-cho, Himi, Toyama 935–8531, Japan. e-mail: kitad@ns.m.kanazawa-u.ac.jp.*