Case report-Olgu sunumu

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A case of holoprosencephaly with facial anomalies: 11q deletion syndrome

Fasiyal anomalilerin eşlik ettiği bir holoprozensefali olgusu: 11q delesyon sendromu

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Abstract

The term holoprosencephaly defines a group of diseases characterized by separation and differentiation deficiencies of prosencephalon at different stages of development. Craniofacial and extracranial anomalies (polydactly, renal dysplasia, omphalocel, hydrops etc.) may accompany holoprosencephaly. Chromosomal abnormalities are also present in most of these cases. In this report a rare 11q mosaicism holoprosencephaly case with prominent ear and face anomalies is presented.

Keywords: Holoprosencephaly, Jacobsen Distal 11q Deletion Syndrome, craniofacial abnormalities

Özet

Holoprozensefali terimi, prozensefalonun farklı safhalardaki ayrılma ve farklılaşma yetersizlikleri ile karakterize bir grup hastalığı tanımlar. Holoprozensefaliye kraniyofasiyal ve ekstrakraniyal (polidaktili, renal displazi, omfalosel, hidrops vb.) anomaliler eşlik edebilir. Bu olgularda genellikle kromozomal anomaliler de vardır. Bu makalede belirgin kulak ve yüz anomalilerin eşlik ettiği nadir bir 11q mozaisizmli holoprozensefali olgusunun bulguları sunuldu.

Anahtar sözcükler: Holoprosansefali, Jacobsen Distal 11q Delesyon Sendromu, kafa yüz anormallikleri

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Introduction

The term holoprosencephaly defines a group of diseases characterized by separation and differentiation deficiencies of prosencephalon at different stages of development. Craniofacial and extracranial anomalies (polydactyl, renal dysplasia, omphalocel, hydrops etc.) may accompany holoprosencephaly. Chromosomal abnormalities are also present in most of these cases [1-8].

In this article, a rare case of 11q mosaicism holoprosencephaly accompanied by prominent ear and face anomalies is presented.

Case report

One day old preterm male baby evaluated in our hospital for systemic and craniofacial abnormalities after he presented with prominent facial abnormalities (cleft palate, right sided cheek and ear dysmorphism), meconium aspiration, and anoxia. Physical examination revealed cranial trigonocephaly, flat occiput, ptosis, downward located palpebral fissures, posteriorly angled and malformated ears, complete cleft palate, polyport lesion in the cheek, short neck and anteriorly ectopic anus (Figure 1). There was a 2/6 systolic murmur in the heart. Patent ductus arteriosus was found in echocardiography. Laboratory findings were: blood glucose: 38 mg/dL, BUN: 12 mg/dL, Creatinine: 1.5 mg/dL, P: 2.9 mg/dL, Na: 130 mmol/L, K: 5.4 mmol/L, ALT: 95 IU/L, AST: 94 IU/l, CRP: 1.9 mg/L, Hb: 19.8 g/dL, leukocyte: 27900/mL (57% neutrophil), thrombocyte: 81000/mL. Cranial ultrasonography (US) and computed tomography (CT) scan showed dilated and single ventricular system, lacks gyri and sulci altogether, fusion in parenchyma and basal ganglions, and cerebellar aplasia (Figure 2, 3). In addition to similar findings with US and CT, MRI detected absence of corpus callosum, interhemispheric fissure, septum pellucidum and falx cerebri (Figure 4). Chromosomal analysis of the case revealed 46 XY 11q mosaicism.

Figure 1a-b. Cranial trigonocephaly, flat occiput, pitosis, downward located palpebral fissures, posteriorly angled and malformated ears, polyport lesion in right cheek and short neck.

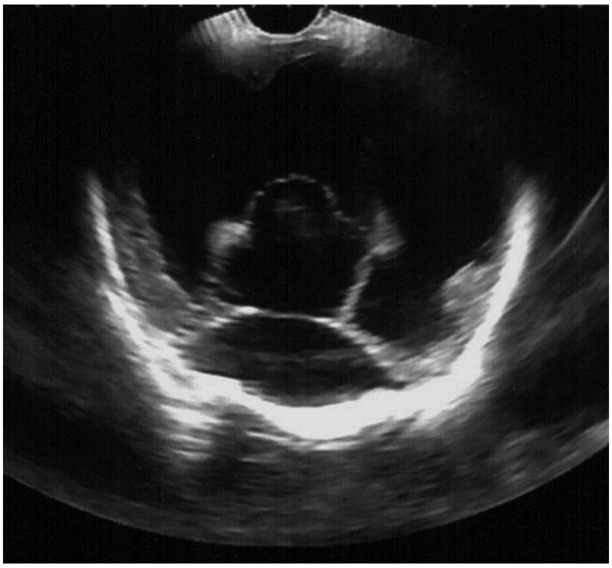

Figure 2. Cranial US show dilated and single ventricular system and cerebellar aplasia.

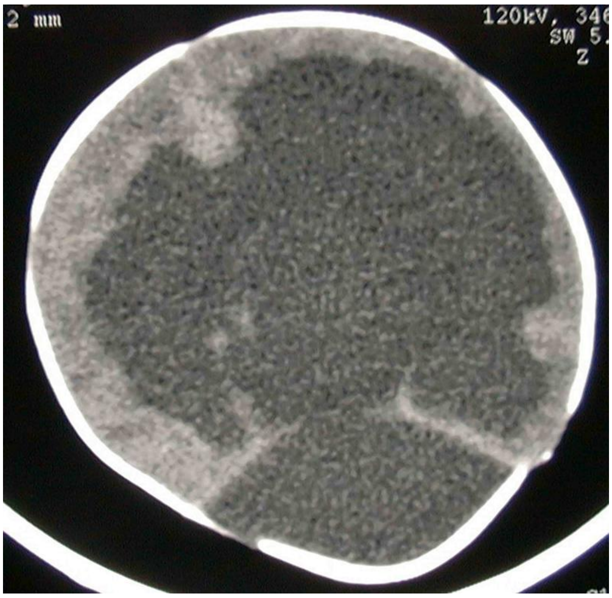

Figure 3. Axial cranial CT shows enlarged ventricular system, disappearance of sulcus and gyrus discrepancy, fusion in parenchyma's and basal ganglia and cerebellar aplasia.

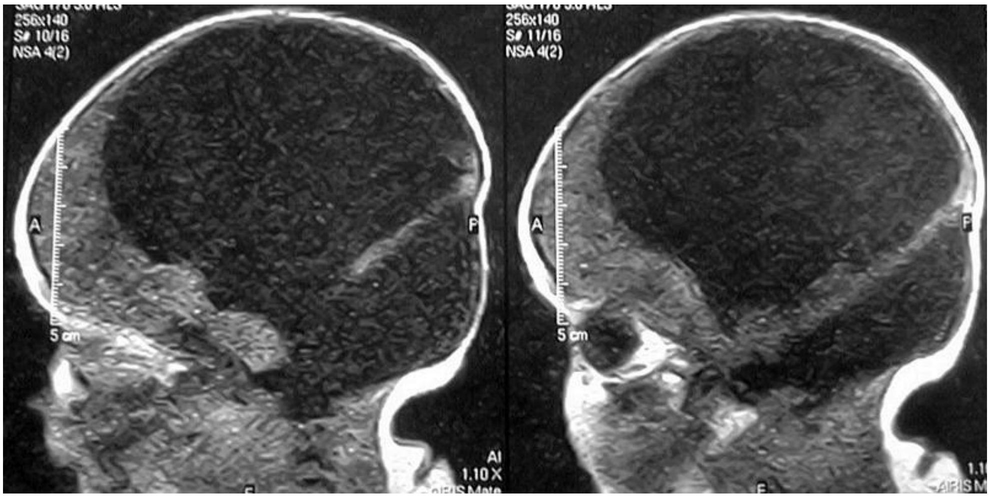

Figure 4. Sagittal cranial MR images show dilated and single ventricular system, deletion of sulci and gyri, fusion in parenchyma and basal ganglia, cerebellar aplasia and absence of corpus callosum, interhemispheric fissure, septum pellucidum and falx cerebri.

Discussion

Holoprosencephaly is a congenital brain abnormality characterized by a single central ventricule with fusioned thalamus. It has three subtypes named as alobar, semilobar and lobar types [1, 2, 4, 7]. Alobar type is the most serious. Third ventricle is usually absent due to fusion of thalami in affected cases. Interhemispheric fissure, falx cerebri and corpus callosum are absent. Semilobar type disease is less severe than the alobar type. Interhemispheric fissure and falx cerebri are partially present at posterior brain area. Callosal splenium may be present without callosal corpus and genu. In other words holoprosencephaly is the only brain anomaly in which corpus callosum develops in the absence of anterior callosal part. Lobar type is the least severe form. Fusion in frontal lobes is observed [1, 4, 7]. Our case has alobar type. Severe hydrocephalus, Dandy-Walker malformation, hydranencephaly and corpus callosum agenesis should be considered in differential diagnosis. There is not fusion in thalami or other basal ganglions in hydrocephalus. Moreover there is an irregular, symmetric and thin cortex [2, 4]. Supratentorial sections are totally normal in Dandy-Walker malformation [7]. Frontal and parietal cortices are most commonly involved in hydranencephaly. Basal ganglia, inferior part of temporal lobe and occipital lob are usually preserved. In corpus callosum agenesis with midline cyst lateral ventricles are not fused. They are separated, dilated or displaced to superolateral site [3, 8].Holoprosencephaly may be accompanied by varying degrees of craniofacial and extracranial (polydactyl, renal dysplasia, omphalocele and hydrops) abnormalities. In these cases chromosomal anomalies such as trisomy 13 or trisomy 18 are usually detected [5-7]. Detection of 11q mosaicism instead of trisomy 13 or trisomy 18 in our case is a very rare incidence.

As a last word, 11q mosaicism should be thought as a rare syndrome in differential diagnosis of holoprosencephaly cases and clinical information important for diagnosis.

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