Patau Syndrome (Trisomy 13): Autopsy Case

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Abstract

Background: Patau syndrome has been estimated to occur in 1 in 12.000-29.000 live births, and the risk is increased with advanced maternal age advanced maternal age.

Case: The ultrasonography was applied to a 40 years old mother and flat nose, holoprosencephaly, cleft palate and cleft lip were seen. Holoprosencephaly, multicystic kidney, cleft palate and cleft lip were found in the autopsy examination of the fetus.

Keywords: Holoprosencephaly, cleft palate, cleft lip multicystic kidney, trisomy 13, prenatal diagnosis.

Patau sendromu (trizomi 13): otopsi olgusu

Amaç: Patau sendromu 12.000-29.000 canlı doğumda bir görülmektedir ve ileri anne yaşı ile risk artmaktadır.

Olgu: 40 yaşındaki annenin yapılan ultrasonografisinde, hipotelorizm, basık burun, holoprozensefali, yarık damak ve yarık dudak tespit edildi. Fetusun otopsi incelemesinde holoprozensefali, multikistik böbrek, yarık damak ve yarık dudak saptanmıştır.

Anahtar Sözcükler: Holoprozensefali, yarık damak, yarık dudak, multikistik böbrek, trizomi 13, prenatal tanı.

Background

Cytogenetics of Patau syndrome is described by Patau et al and clinical phenotype is described by Smith.[1,2] In most of cases, there is nondysjunction-one of morphological chromosomal anomalies. There is approximately 20% translocation and less than 10% mosaicism in cases.[3] Patau syndrome occurs in about 1 out of every 12.000-29.000 live births and the risk is increased with advanced maternal age.[4] Chromosome 13 is larger than chromosome 21 and therefore anomalies are multiple and more severe in trisomy 13. Midline anomalies, facial defects, holoprosencephaly, cardiac defects omphalocele, polycystic kidney disease and polidactyly are prominent in sonography.[5]

Children with Patau Syndrome die in a short time after birth. Seldomly children survive into their first year.[6] Patau Syndrome is presented as a case report because it is seen rarely and the first experience of our clinic.

Case

40-year-old mother with G4, P3, L3, A1, first degree of relationship with her husband and no

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any prenatal follow up hypotelorism, holoprosencephaly (Figure 1), flat nose, cleft lip, cleft palate determined in ultrasonography applied at 24 gestational week. Pregnancy terminated with permission of the family. The birth was via normal vaginal delivery with 1/0 apgar score. In autopsy examination, fetus was 1650 g in weight, 35 cm in length and headcircumference was 20 cm with cleft palate and cleft lip. Plasenta was 350 g in weight with normal appearance (Figure 2). Thoracal and abdominal organs located in their normal anatomic positions. Superior longitudinal fissure was not seen in cranial dissection. Bilateral kidney dimensions were 4x2x1.2 cm. Microscopically holoprosencephaly and bilateral polycystic kidneys determined (Figure 3). Karyotype made by GTG band technique after amniosynthesis was 47, XX+13.

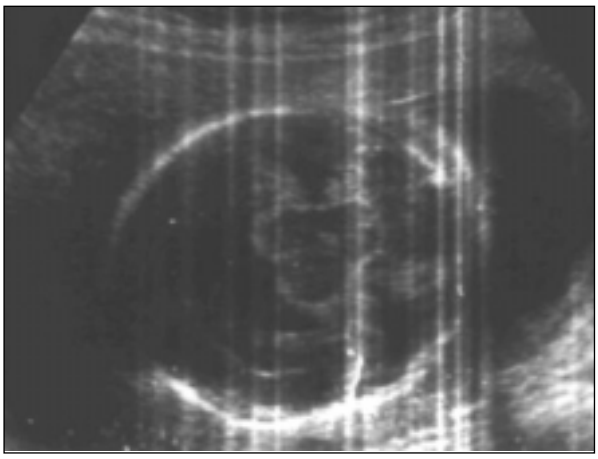

Figure 1. Ultrasonographic appearance of holoprosencephaly.

Discussion

Clinical signs of Patau syndrome (trisomy 13) firstly described at 1967 by Bartolin.[7] Patau et al. described syndrome caused by an extra copy of chromosome 13 in group D. Patau syndrome occurs in about 1 out of every 12.000-29.000 live births but 100 times more in spontaneous abortus.[4,7]

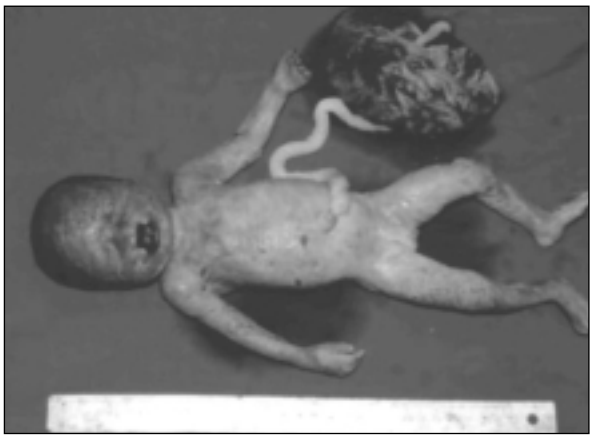

Figure 2. Macroscopic appearance of flat nose, cleft palate and cleft lip malformations.

Major signs of trisomy 13 are motor and mental retardation, microcephaly, microphthalmia, holoprosencephaly, hypotelorism, cleft palate, cleft lip, cardiovascular, genitourinary, ocular malformations and early death.[8]

Chromosomal analysis is necessary for definitive diagnosis. Chromosome 13 is larger than chromosome 21 and therefore anomalies are multiple and more severe in trisomy 13.[9]

Generally Patau syndrome is caused by classical trisomy 47, XX+13 (80%) and also but less frequently (10%) translocation, structural changes and other chromosomal disorders like (5%) mossaicism.[7,9] In this case some characteristical signs of Patau syndrome was determined in ultrasonography applied in perinatology clinic in routine control. In karyotype analysis classical type 47, XX+13 was determined.

Magenis et al announced that 28% of patients with Patau syndrome die in their first week, 44% in their first month and 86% in their first year. Taylor announced the mean life expectancy 89.2+ 29.9 days, but Redheedran et al[12] and Zoll et al.[7] announced that some children with trisomy 13 survive into their teens. Our case died one day after birth. Patients with Patau syndrome are trisomic for chromosome 13, and there is 3 chromosome instead of 2. With extra chromosome in group D, in karyotype there is 47 XX+D total chromosome.

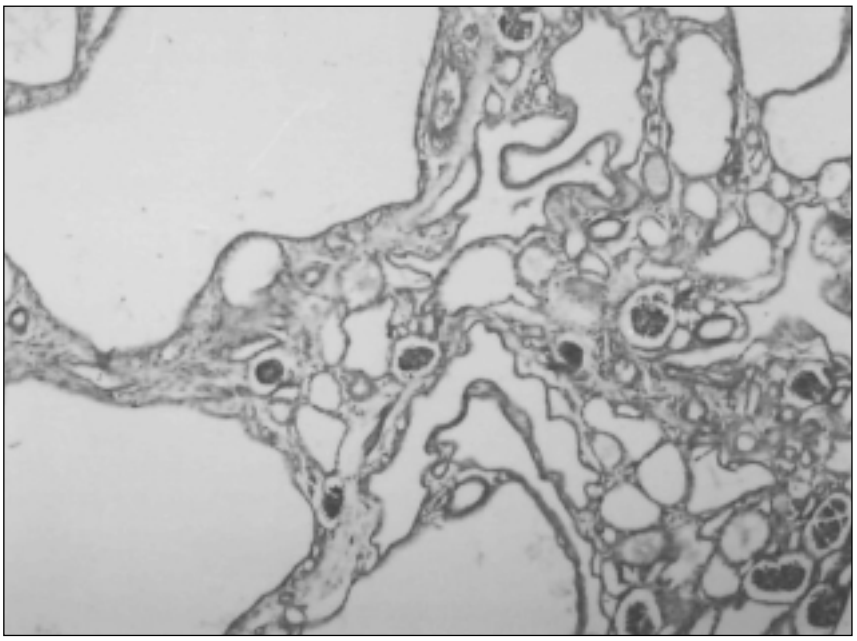

Figure 3. Cystic tubular structures with different dimensions.

20% of cases have 46 chromosome but they have Robertsonian type of translocation between chromosome 13 and 14.[13]

Golstein et al[14] established that mother age is over 35 in their study. Mother age is advanced in cases of chromosomal nondisjunction. Mean mother age is 39 in this group. In our case mother age was 40. In translocation and mosaicism type of trisomic cases, mother or father may be carrier and other children have risk of disease. If mother and father are not carriers, recurrence risk is less than 1%.

Boyd et al[15] reported that mothers who have fetus with trisomy 13 are at risk of preeclampsia more than mothers who have fetus with trisomy 21 and 18 or with no chromosomal anomaly.

As a result, trisomy 13 is known as one of the oldest chromosomal disorder. Autosomal cytogenetic abnormalities should be considered in case of mothers with preeclampsia, mothers older than 35 years old and who have spontaneous abortus and genetic consulting should be recommended. Pregnant woman can be followed up in perinatology clinics for early diagnosis of fetus with abnormality.

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