

# Duplication 10q syndrome: A new case

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## SUMMARY

*Partial trisomy of the long arm of chromosome 10 is a well-defined but rare syndrome. Growth retardation, developmental delay and characteristic dysmorphic features are well described in the syndrome. The case had the well characterized phenotype of microcephaly, characteristic dysmorphic facies and limb anomalies. Trisomy in the case involved the 10q25-->qter region.*

**Key words:** Trisomy 10q, characteristic dysmorphic facies, developmental delay

## ÖZET

### 10q duplikasyon sendromu: Yeni bir olgu

*Kromozom 10'un uzun kolunun distal trizomisi iyi tanımlanmıştır fakat nadir bir sendromdur. Büyüme geriliği, gelişme geriliği ve karakteristik yüz görünümü sendromda iyi tasvir edilmiştir. Olgu mikrosefali, karakteristik dismorfik yüz görünümü, ekstremité anomalisi ile iyi karakterize fenotipe sahip idi. Olgudaki trizomi 10q25-->q terminal bölgesinde oluşmuştur.*

**Anahtar kelimeler:** Trizomi 10q, karakteristik dismorfik yüz, gelişme geriliği

Trisomy of more distal 10q is associated with a characteristic syndrome and has been described in many cases which are almost always familial<sup>(1-3)</sup>, but patients with trisomy of the proximal or medial segment of 10q have been less often described<sup>(4)</sup>. Here, we report on a 4-month-old boy with a de novo partial 10q trisomy, karyotype 46,XY,add(10)(q25).

## CASE REPORT

The proband was the second child of a healthy 28-year-old mother and a 30-year-old father. He was born at term after an uneventful pregnancy. His measurements were low but appropriate for a 4-month-old: body weight 3990 g, length 58 cm, and head circumference, 38 cm. The patient had microcephaly, flat face with high forehead, arched eyebrows, blue sclera, depressed nasal bridge, anteverted nares, malformed posteriorly rotated ears, macroglossia, bow-shaped mouth with prominent upper lip and marasmus (Figure 1), hypoplastic liver, hypoplastic spleen, right inguinal hernia, um-

bilical hernia, hypospadias and micropenis, broad chest with widely spaced and long slender limbs. The cardiologist's evaluation showed atrial septal defect. The echocardiogram was normal. Cranial ultrasound examination revealed extensive frontal horns, thin corpus callosum and mild hydrocephalia. Renal ultrasound examination demonstrated medullary nephrocalcinosis, and vesicoureteral reflux. There was no family history of repeated miscarriages, mental retardation, or malformation syndromes.

## CYTOGENETIC ANALYSIS

Cytogenetic analysis was performed on unstimulated 72-hour culture of a peripheral blood specimen. The cells were cultured and processed by conventional methods, and the chromosomes were stained with Giemsa-Trypsin-Giemsa banding (GTG). The karyotype was 46,XY, add(10)(q25) (Figure 2). Parental karyotypes were normal.

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Figure 1. Present case at the age of 4 months.

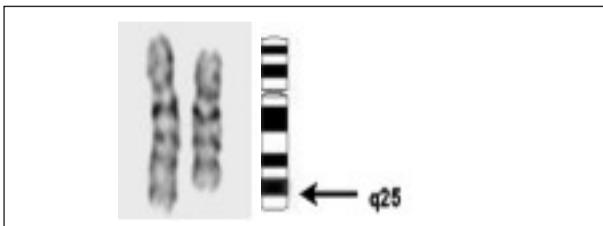


Figure 2. G banded karyotype and ideogram of the present case showing partial trisomy 10q.

## DISCUSSION

Distal trisomy 10q is an extremely rare chromosomal disorder in which the distal portion of the long arm (q) of one chromosome 10(10q) appears trisomy rather than twice in cells of the body. The disorder is characterized by unusually prenatal and postnatal growth retardation, hypotonia, mild to severe mental retardation, and mild to severe delays in the acquisition of skills requiring coordination suggestive of psychomotor retardation. Affected infants and children may also have distinct malformations of the craniofacial area, defects of the hands and/or feet, and/or skeletal, cardiac, renal, and/or respiratory abnormalities. The range and severity of symptoms and physical findings may vary from case to case, depending upon the exact length and location of the duplicated portion of chromosome 10q. In most cases, distal trisomy 10q is due to a chromosomal balanced translocation in one of the parents <sup>(1,2,4)</sup>.

Our patient had the well characterized phenotype of the distal trisomy of 10q in the form of developmental delay, microcephaly, characteristic dysmorphic facies and limb anomalies, and also trisomy if the 10q25-->q ter region was involved. About 25 % of reported patients with distal 10q trisomy die within the first year of life, mostly resulting from internal malformations and respiratory infection, and most survivors suffer from severe mental retardation <sup>(3,4)</sup>. The related outcomes include severe failure to thrive, cardiac involvement and death from respiratory and heart failure.

This study confirmed that unbalanced chromosomal regions of the long arm of chromosome 10 play an important role in the developmental malformations and that a more severe form is associated with involvement of 10q25. It also emphasizes the importance of increasing public awareness regarding these chromosomal rearrangements and the importance of genetic counseling and prenatal diagnosis to avoid recurrences and associated family stress <sup>(5,6)</sup>.

In conclusion, this case implies the importance and worth of prenatal diagnostic procedures for the pregnant with a history of bad obstetric history.

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