

De novo 10q22 interstitial deletion

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Abstract

We describe a 4 month old male with a de novo interstitial deletion of chromosome 10q22. His clinical features included growth deficiency, developmental delay, ocular hypertelorism, posteriorly rotated ears, retrognathia, and fifth finger clinodactyly. He later developed dental lamina cysts of the alveolar ridge. To our knowledge, this is the first reported case of an interstitial deletion of 10q22.

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Deletions of the long arm of chromosome 10 are infrequent. The most commonly reported structural abnormalities are deletions, partial duplications of the short and long arms, and formation of a ring chromosome.¹ An interstitial deletion of only band 22 has not been reported previously to our knowledge. We report a patient with a de novo deletion of 10q22.1 to q22.3 and the clinical history of this patient during the first year of life.

Case report

At the age of 4 months, the proband (FN 89524) was referred for dysmorphic features, developmental delay, and growth deficiency. He was born to non-consanguineous parents

following a pregnancy complicated by first trimester bleeding, upper respiratory infection in the second trimester, and intrauterine growth deficiency diagnosed by ultrasound examination three weeks before delivery. No significant problems were noted after birth with the exception of bilateral undescended testes. Birth weight was 2300 g (<3rd centile) and length was 47.6 cm (5th centile). Developmental delay was noted at 3-4 months of age.

A dysmorphology examination performed at 4 months of age was significant for a height of 54.4 cm (<3rd centile), weight of 5.1 kg (<3rd centile), occipitofrontal circumference 40 cm (-1 SD), large anterior fontanelle, strabismus, mildly downward slanting palpebral fissures, ocular hypertelorism, posteriorly rotated ears (25-30°) with pointed tips, short and upturned nose, smooth philtrum, retrognathia, dimple on the left cheek, diastasis recti, symmetrical puckering of the scrotum, mild fifth finger clinodactyly, and poor head control (fig 1). Dermatoglyphics included: right hand, 2-radial loops, 3 to 5-whorls; left hand, 2 and 3-ular loops, 4 and 5-whorls.

The proband's development was assessed as being 5-9 months in social skills and 2 months in motor skills at a chronological age of 5 months. At 8 months of age he was not yet rolling over or crawling, but had just learned to sit without assistance and was babbling. His general health has been good. He developed several small "gingival cyst"-like lesions on his alveolar ridges at 8 months which were thought to be dental lamina cysts (fig 2).

Materials and methods

GTG banding of peripheral blood lymphocytes showed 46,XY,del(10)(q22.1q22.3) at an average band level of 596 (fig 3). Both parents had normal karyotypes. Fluorescence in situ hybridisation (FISH) analysis was performed to ascertain if the deleted chromosome 10 contained translocated material from another chromosome. Cells, fixed in a 3:1 methanol and acetic acid solution from the original cell

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Figure 1 Proband at 4 months. Note telectanthesis.



Figure 2 Dental lamina cysts of gingiva of maxilla.

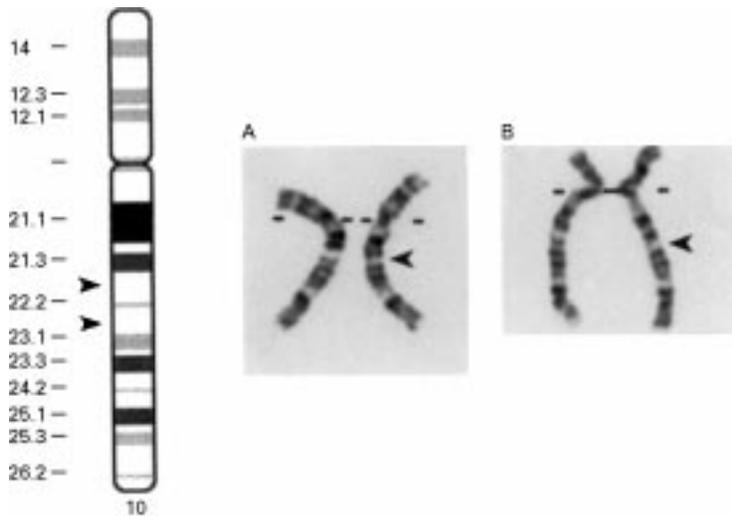


Figure 3 Ideogram and partial deletion of proband's chromosome 10 from a peripheral blood specimen.

pellet, were used for the studies. A digoxigenin labelled coatasome 10 whole chromosome painting probe (Oncor) was hybridised to dividing cells. Twenty cells were analysed and both chromosome 10 homologues showed a uniform signal across the chromosomes, indicating that the derivative (10) did not contain obvious translocated sequences from another chromosome. Additionally, signals from the painting probe were not found on any other chromosome (fig 4).

Discussion

Our patient carries an interstitial deletion in the long arm of chromosome 10 from band q22.1 to q22.3. Because 10q22.2 is lost, the deletion leads to juxtaposition of 22.1 and 22.3, resulting in a broad light band (fig 3). We were not able to find other published cases with this discrete deletion or cases that have similar overlap.²⁻⁴ The case reported by Mori *et al*³ car-

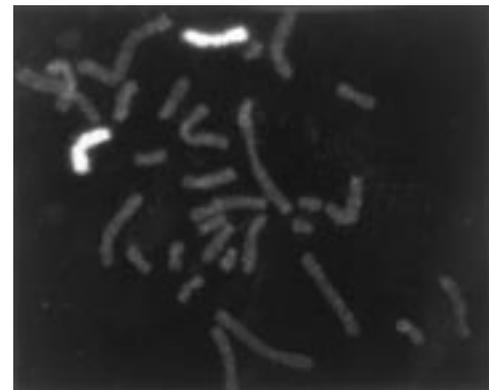


Figure 4 Fluorescence in situ hybridisation (FISH) on partial metaphase peripheral blood lymphocytes with whole chromosome painting probe for chromosome 10. Signal extends across entire chromosome 10 homologues.

ries an interstitial deletion extending from band q22 to band q24. It appears from fig 2 in that article that band q22.2 is retained on the deleted chromosome 10 and therefore the breakpoint is probably located in distal 10q22 at 10q22.3.

The most striking clinical features of this patient are ocular hypertelorism, downward slanting palpebral fissures, posteriorly rotated ears, developmental delay, and growth deficiency. However, his paternal relatives, including his father, also have ocular hypertelorism. Additional cases of 10q interstitial deletions are needed before it can be determined if a specific phenotype is associated with this deletion.

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