Case reports

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Deletion (13)(q13q14.3) with retinoblastoma: confirmation and extension of a recognisable pattern of clinical features in retinoblastoma patients with 13q deletion

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SUMMARY A girl with retinoblastoma and a del(13)(q13q14.3) is presented. This case helps to confirm and extend our previous observations¹ regarding a recognisable facial pattern in retinoblastoma patients with 13q deletion involving 13q14 and its adjacent segments.

The proband, a girl (fig 1), was the second child of non-consanguineous parents. Both parents and an

FIG 1 The proband aged three months with a prosthesis in the left orbit.

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older brother were in good health. The mother was 26 and the father 32 years old at the time of her birth. The family history was negative for retinoblastoma or congenital malformations. The mother had had no previous miscarriages. Pregnancy was complicated by hyperemesis requiring infusion therapy and threatened abortion. Delivery was normal and at term. Birth weight was 3742 g and length 55 cm. At one month bilateral leukokorias were noted. At two months enucleation of the left eye was performed after the clinical diagnosis of bilateral retinoblastoma at another eye clinic. Shortly after this surgery, she was referred to our hospital for conservative therapy to the tumour in the right eye. Linear X irradiation (total 3600 rads) and subsequently systemic vincristine and cyclophosphamide were started.

Physical examination showed the following findings: brachycephaly with bilateral parietal bossing, hirsute forehead, upward slanting palpebral fissures, broad nasal bridge, bulbous tip of the nose with anteverted nostrils, long philtrum, thin upper lip, large ears with folded lobule and multiple postauricular pits, and widely spaced nipples. She had normal muscle tone. She began to vocalise with her mother at three months and the developmental quotient at nine months of age was 87. Anthropometric measurements at nine months of age were: weight 8.15 kg (-0.4 SD), height 69.6 cm (-0.3 SD), chest circumference 42.5 cm, and head circumference 44.5 cm.

Fingertip patterns showed UWWWW on the right and all whorls on the left, with a total finger ridge count of 145. The palm showed a vestige in the thenar area, bilaterally. The *atd* angle was 45° bilaterally.

FIG 2 (a) Two pairs of GAG banded chromosomes 13 and (b) two pairs of RBG banded chromosomes 13 derived from the proband. In each pair the deleted chromosome 13 is on the left. (c) Diagram of chromosome 13 showing breakpoints.

CYTOGENETIC STUDIES

For chromosome analysis 72 hour cultured peripheral lymphocytes were used and G and R bands were obtained using GAG and RBG techniques, respectively. All cells examined showed an interstitial deletion of 13q which was evaluated as $\text{del}(13)(\text{q}13 \text{ or } 14.1\text{q}14.3)$ on G banded chromosomes at the 600 band stage (fig 2a) and as $\text{del}(13)(\text{q}13\text{q}14.3)$ on R banded chromosomes at the same band stage (fig 2b). The karyotype was $46,XX,\text{del}(13)(\text{q}13\text{q}14.3)$. Cytogenetic examination of the parents showed normal karyotypes.

Discussion

The present case confirms previous observations

concerning the midface pattern in retinoblastoma patients with 13q deletion, which include a hairy forehead, prominent eyebrows, broad nasal bridge, bulbous tip of the nose, thin upper lip, and a long philtrum.^{1,2} Further comparison of our six retinoblastoma patients with 13q deletion, including five previously published³⁻⁵ and the present one, suggests that brachycephaly with parietal bossing and large ears with folded lobules and postauricular pitting are additional features of 13q deletion with retinoblastoma. Brachycephaly with parietal bossing was observed in three of our six patients and large ears with folded lobules were observed in all six in our study. Postauricular pitting, which Crowder *et al*⁶ described as a unique feature of 13q deletion involving 13q14, was observed in three of our six retinoblastoma patients with 13q deletion. Further accumulation of cases of retinoblastoma with 13q deletion with full description of phenotypes will be required in order to delineate 13q deletion syndrome with retinoblastoma further.

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