

cleft and the x-rays of her hands and feet showed normal bony components. The common parent could be considered to be a non-expressing heterozygote and an obligate carrier. Another explanation would be to assume that the mother had a balanced chromosomal translocation and that, in the children, an unbalanced form of the translocation is present, resulting in the anomalies. In the absence of any detectable changes in the karyotype of either child, this interpretation becomes hypothetical. A fourth explanation would be that the mother had a gonadal mosaicism which could result in repeated births of children with similar congenital malformations not present in the maternal phenotype, but this assumption is also speculative and cannot be demonstrated.

If these two sibs had been the children of the same parents, the cases would have been quoted as 'suggesting' that these anomalies were attributable to homozygosity of an autosomal recessive gene, an explanation that is unlikely in view of the different fathers. In the case of a common mother and different fathers, X linked recessive inheritance can be involved if the affected children are males. An exception to this rule is the rare possibility that a female has an X linked recessive disorder (Lyon hypothesis).

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References

- Walker JC, Clodius L. The syndrome of cleft lip and palate and lobster claw deformities of hands and feet. *Plast Reconstr Surg* 1963;**32**:627-36.
- Wiedermann HR. Missbildungs-retardierungs-syndrom mit fehlen des 5 strahl an handen und fussen: Gaumins-plate dyplastischen ohren und augenlide und radioulnerne synotosis. *Klin Paediatr* 1973;**185**:181-6.
- Pashayan HM, Pruzansky S, Solomon L. The EEC syndrome. *Birth Defects* 1974;**10**(7):105-27.
- Temtamy S, McKusick VA. Synopsis of hand malformations with particular emphasis on genetic factors. *Birth Defects* 1969;**5**:125-84.
- Brill CB, Hsu LYF, Hirschhorn K. The syndrome of ectrodactyly, ectodermal dysplasia and cleft lip and palate: report of a family demonstrating a dominant inheritance pattern. *Clin Genet* 1972;**3**:295-302.
- Robinson GC, Wildervanck LS, Chiang TP. Ectrodactyly, ectodermal dysplasia and cleft lip and palate syndrome. *J Pediatr* 1973;**82**:107-9.
- Preus M, Fraser FC. Lobster claw defect with ectodermal defects, cleft lip and palate, tear duct anomaly and renal anomalies. *Clin Genet* 1973;**4**:369-75.
- Critchley M. Observations on essential (heredofamilial) tremor. *Brain* 1949;**72**:113-39.

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18p- syndrome with a single central maxillary incisor

SUMMARY A child with a single central maxillary incisor and a deletion of the short arm of chromosome 18 (18p-) is presented. He is the first patient in whom this association has been found.

A syndrome of short stature and single central maxillary incisor has been described by Rappaport *et al.*¹ Five of the seven patients in the original report had cytogenetic studies, all of which were normal. Four of these five patients were growth hormone deficient.

We are reporting a child with short stature and a single central maxillary incisor who was found to be missing the entire short arm of chromosome 18 (18p-). To our knowledge, he is the first patient in whom this dental anomaly has been associated with an abnormal karyotype.

Case report

A 28-month-old male was referred for evaluation of short stature, dysmorphic facies, and developmental delay. He was born after an uncomplicated 37 week pregnancy. The birthweight (2.4 kg) and length (45 cm) were at the 25th and 50th centiles, respectively, for this gestational age. Overlapping of the second and third toes was noted in the newborn period.

His early linear growth rate, weight gain, and cranial growth were normal, but have fallen off since the age of 6 months. At 9 months of age his third tooth, a large single central maxillary incisor, appeared.

The parents report that his early developmental milestones were normal; he rolled over at 4 months and sat at 6 months. His language development has been delayed. Although he spoke his first word at one year, he does not combine words at 28 months.

The mother (aged 24 years), father (aged 25 years), and a sister (aged 7 years) are of normal stature and dentition. There is no consanguinity or history of similarly affected relatives. The home environment is stable.

The patient has had no other admissions to hospital for serious illnesses. His diet is appropriate for age.

At 28 months, his height (79 cm), weight (8.8 kg), and head circumference (44 cm) were below the 3rd centile for age. Pertinent craniofacial features included prominent simply-formed ears, downward slanting palpebral fissures, mild epicanthal folds

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FIG 1 Patient aged 28 months. Note the prominent ears, downward slanting palpebral fissures, epicanthal folds, unusual nose, and single central maxillary incisor.

(fig 1), normal interpupillary distance, and normal ocular fundi. The nose was unusual in appearance with a short columella, deficient tip, hypoplasia of the nasal alae, and a fleshy prominence at the base of the nasion. Examination of the oral cavity revealed a single central maxillary incisor and torus palatinus. The dentition was otherwise unremarkable except for the absence of the two-year molars. The chest, abdomen, and genitalia were normal. Examination of the extremities revealed long third toes which deviated laterally. The palmar creases and digital dermal ridge patterns were normal. There was a left distal palmar triradius. An arch tibial pattern was present in the right hallux area.

The neurological examination was unremarkable with the exception of delayed speech. Audiological evaluation indicated a mild conductive hearing loss bilaterally. His performance on the Bayley Scale of Infant Development was at the 20-month level.

Laboratory data included normal serum electrolytes, BUN, creatinine, and a normal haemogram. The urine analysis revealed a specific gravity of 1.029 and was otherwise unremarkable. Endocrine studies included T_4 , TSH, LH, FSH, and morning serum cortisol, which were normal for age. A random serum growth hormone determination was 7.4 ng/ml. The serum growth hormone rose from a baseline of 8.4 ng/ml to 13.9 ng/ml after exercise, indicating a normal pituitary response to this stimulus. A random somatomedin C was 0.18 U/ml (RIA Nichols Laboratory). His bone age was 1 year 6 months (\pm SD 4.5 months). A lateral skull radiograph was normal.

Lymphocyte karyotype was evaluated using Giemsa staining and quinacrine mustard banding.

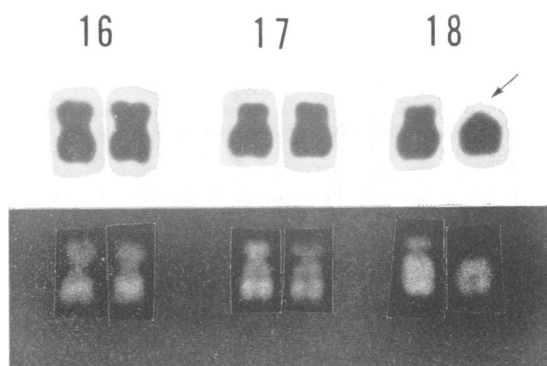


FIG 2 Partial karyotype of E group chromosomes from the patient. Top line shows chromosome 18 with short arm deletion (arrow) stained with Giemsa; bottom line shows corresponding E group from 2nd cell stained with quinacrine.

The karyotype revealed a complete deletion of the short arm of chromosome 18 (46,XY,18p-) (fig 2). Karyotypes of both parents were normal.

Discussion

Dental caries and absence of the lateral incisors have been described in the 18p- syndrome, as well as a variety of midline cranial defects such as cebocephalus, agenesis of the pituitary gland, and agenesis of the corpus callosum.² Short stature is also a common feature,^{2,3} although only one patient with 18p- syndrome has been documented as being growth hormone deficient.⁴

Our patient had an acceptable growth hormone response to exercise, but his somatomedin C level was in the range seen in children older than 4 years of age who are growth hormone deficient. Normal somatomedin C levels in younger children have not been established. We are unable to state whether the short stature in our patient is a manifestation of his cytogenetic abnormality or is the result of inadequate generation of somatomedin.

Of interest is the similarity in nasal configuration between our patient and patient 7 described by Rappaport *et al.*¹ That patient had a normal karyotype, but details of the cytogenetic methods used were not published. The newer high resolution cytogenetic techniques may be useful in searching for minor chromosomal abnormalities in patients with the single central maxillary incisor-short stature syndrome.

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References

- ¹ Rappaport EB, Ulstrom RA, Gorlin RJ, Lucky AW, Colle E, Miser J. Solitary maxillary central incisor and short stature. *J Pediatr* 1977; **91**: 924-8.
- ² de Grouchy J, Turleau C. *Clinical atlas of human chromosomes*. New York: Wiley, 1977: 164-9.
- ³ de Grouchy J. The 18p-, 18q- and 18r- syndromes. *Birth Defects* 1969; **5**(5): 74-87.
- ⁴ Leisti J, Leisti S, Perheentupa J, Savilahti E, Aula P. Absence of IgA and growth hormone deficiency associated with short arm deletion of chromosome 18. *Arch Dis Child* 1973; **48**: 320-2.

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