balanced and involved breaks in bands 3q13, 8p22, 8q13, 11p12, and 11q21. Parental karyotypes were normal.

Discussion

Although our patient showed several signs of ectodermal dysplasia we cannot classify him in any of the diagnostic categories proposed by Pinsky¹² and Freire-Maia¹³ because he presented additional features (severe mental retardation and megacephaly). Nevertheless, we think the proband has most of the features of TRP and Langer-Giedion syndromes, including cone shaped epiphyses, short broad middle phalanges, and metaphyseal cupping.

We wish to thank Mr Ernesto Torchia for the photographs, Mr Pablo Ruina for the karyotype, and Miss Alejandra Margiotta for helping us with the translation.

Addendum

Two previous reports of LG syndrome associated with del(8q) have come to our attention since the submission of this manuscript (Zaletaje DV, Marincheva GS. *Hum Genet* 1983;**63**:178–82 and Fryns *et al.* *Hum Genet* 1983;**64**:194–5) which add support to our hypothesis on the possible role of a structural aberration of 8q in the pathogenesis of TRP syndromes.

References

- ¹ Giedion A. Das Tricho-rhino-phalangeal syndrom. *Helv Paediatr Acta* 1966;**21**:475–82.

- ² Giedion A, Burdea M, Fruchter Z, Meloni T, Trosch V. Autosomal dominant transmission of the tricho-rhino-phalangeal syndrome. Report of 4 unrelated families, review of 60 cases. *Helv Paediatr Acta* 1973;**28**:249–59.
- ³ Langer LO. The thoracic-pelvic-phalangeal dystrophy. Clinical delineation of birth defects. Part IV. Skeletal dysplasia. *Birth Defects* 1969;**4**:55–64.
- ⁴ Bühler EM, Bühler UK, Stalder GR, Jani L, Pia Jurik L. Chromosome deletion and multiple cartilaginous exostoses. *Eur J Pediatr* 1980;**133**:163–6.
- ⁵ Pfeiffer RA. Langer-Giedion syndrome and additional congenital malformations with interstitial deletion of the long arm of chromosome 8: 46,XY,del8(q13–22). *Clin Genet* 1980;**18**:142–6.
- ⁶ Zabel BU, Baumann WA. Langer-Giedion syndrome with interstitial 8q– deletion. *Ann J Med Genet* 1982;**11**:353–8.
- ⁷ Beigle C, Karp LE, Hanson JW, Hall JG, Hoehn H. Small structural changes of chromosome 8. Two cases with evidence for deletion. *Hum Genet* 1977;**38**:113–21.
- ⁸ Dallapiccola B, Santoro L, Trabace S, Ramenghi M, Mastroiaco P, Gandini E. Deletion of the long arm of chromosome 8 resulting from a de novo translocation t(4;8) (q13;q21). *Hum Genet* 1977;**38**:125–30.
- ⁹ Taysi K, Noetzel MJ, Strauss AW. Presumptive long arm deletion of chromosome 8: a new syndrome? *Hum Genet* 1979;**51**:49–53.
- ¹⁰ Fryns JP, Logghe N, Van Eygen M, Van Den Berghe H. Interstitial deletion of the long arm of chromosome 8. Karyotype: 46,XY,del(8)(q21). *Hum Genet* 1979;**48**:127.
- ¹¹ Scabright M. A rapid banding technique for human chromosomes. *Lancet* 1971;ii:971–2.
- ¹² Pinsky L. The community of human malformation syndromes that shares ectodermal dysplasia and deformities of the hands and feet. *Teratology* 1975;**11**:227–42.
- ¹³ Freire-Maia N. Ectodermal dysplasias. *Hum Hered* 1971;**21**:309–12.

Correspondence and requests for reprints to Dr J M Sánchez, Fundación de Genética Humana, Salta 661, 1074 Buenos Aires, Argentina.

Interstitial deletion of chromosome 7p detected antenatally

KAREN MARKS, LUCILLE HILL, R G CHITHAM, AND W L WHITEHOUSE*

Regional Cytogenetic Unit, Wandle Valley Hospital, Mitcham Junction, Surrey CR4 4XL; and *Department of Obstetrics and Gynaecology, Queen Mary's Hospital, Roehampton, London SW15.

SUMMARY An interstitial deletion in chromosome 7(p13p15) detected in amniotic fluid cells is presented. After termination, the fetus was noted to have an asymmetrical skull, low set ears, a flattened nose, bifid thumbs and right big toe, pyloric adenomyosis, hypospadias, and simian creases. A brief comparison is made with previously reported cases involving deletions of 7p, including those associated with craniosynostosis.

Since this paper was submitted, Mr W L Whitehouse has died.
Received for publication 30 August 1984.
Accepted for publication 2 October 1984.

A review of published reports showed five previously reported cases involving the deletion of band 7p14^{1–4} and nine other cases with associated short arm deletions and translocations.² Previous cases with deletion 7p14 exhibited varying associated clinical features. To our knowledge, this is the first case of an interstitial deletion of band 7p14 encountered during antenatal screening. Common anomalies include developmental delay, flattened nose, low set ears, and abnormalities of the extremities including simian creases. Craniosynostosis was not present in our case, nor in the five others involving band 7p14 where the breakpoints were proximal to band 7p21.

FIG 1 (a) Profile of fetus. (b) Bifid thumbs.

Case report

The mother, aged 30, para 1, was offered amniocentesis because of two consecutive high serum AFP levels. The amniotic fluid level of AFP was found to be within normal limits.

However, cytogenetic analysis revealed a chromosomal abnormality in the fetus. In view of this, and after discussion with reference to the previously reported cases, the parents elected for a termination. Necropsy showed a male fetus weighing 578 g with a head circumference of 19 cm, crown-rump length of 20 cm, and crown-heel length of 30.8 cm. There was some asymmetry of the skull, abnormal distribution of hair, oedema around the left ear, which like the right ear was low set, flattened nose, slight epicanthic folds, very prominent protruding upper lip, and a receding chin. The thumbs and right big toe showed distal flattening with spoon-shaped nails. X-rays revealed that both thumbs and the right big toe had two sets of phalanges. Transverse creases were noted on both hands. Hypospadias was present but internal examination showed no gross abnormalities apart from pyloric adenomyosis.

CYTOGENETIC STUDIES

Chromosomal analysis was initially performed on amniotic fluid cells from four independent cultures. G, Q, and R banding revealed a deletion of band 7p14 in all cells, with breakpoints at bands p13 and p15.1, involving a partial loss of material from these two bands, although it was not possible to state the precise limits of the deletion. The karyotype was 46,XY,del(7)(pter→p15.1:p13→qter). Both parents had normal karyotypes. Fetal blood lymphocytes and fibroblasts from fetal muscle, skin, and eye confirmed the presence of the deletion.

FIG 2 Partial karyotype of the fetus and diagrammatic illustration of the interstitial deletion 7p14. Assumed breakpoints are arrowed.

Discussion

Four cases (including ours) had an apparently identical interstitial deletion of bands 7p13–7p15,^{1,4} while two other cases had a larger deletion involving bands 7p13–7p21.^{2,3} Common features include developmental delay, low set ears, a broad nasal bridge, hypertelorism, and malformation of the digits. It is interesting to note the similarity in the duplication of the phalanges as seen in this case and the one reported by Bianchi *et al.*¹ In two out of the four cases with the deletion 7p13–7p15 both parents had apparently normal karyotypes, indicating a de novo origin, while in the other two cases the father was not available for testing and so the possibility that the deletion may have arisen from the crossing over between an inverted 7 and its normal homologue cannot be excluded. The wide variety of clinical symptoms seen in apparently identical rearrangements may be due to the limitation of present

day techniques to enable a precise identification of the breakpoints. The difference in symptoms is probably the result of submicroscopic variation in the amount of deleted material. It is not surprising, therefore, that no clear cut clinical syndrome emerges.

References

- ¹ Bianchi DW, Cirillo-Silengo M, Luzzatti L, Greenstein RM. Interstitial deletion of the short arm of chromosome 7 without craniosynostosis. *Clin Genet* 1981;**19**:456–61.
- ² Muller U, Staudt F, Hameister M. A patient with interstitial deletion 7(p13–p21). *Ann Genet (Paris)* 1981;**24**:239–41.
- ³ Crawford Md'A, Kessel I, Liberman M, McKeown JA, Mandalia PY, Ridler MAC. Partial monosomy 7 with interstitial deletions in two infants with differing congenital abnormalities. *J Med Genet* 1979;**16**:453–60.
- ⁴ Moedjono SJ, Funderburk SJ, Sparkes RS. Chromosome 7 short-arm interstitial deletion (p14). *Hum Genet* 1978;**44**:51–7.

Correspondence and requests for reprints to Dr R G Chitham, Regional Cytogenetic Unit, Wandle Valley Hospital, Mitcham Junction, Surrey CR4 4XL.