4. Santulli TV, Chen CC, Schullinger JM. Management of congenital atresia of the intestine. Am J Surg 1970, 119: 542-547.

5. Al-Kawas FH. Management of a duodenal web by endoscopic laser therapy. Gastrointest Endosc 1989, 35: 113-115.

De Novo Deletion of Chromosome 9 (9p-) in a Child with Multiple Congenital Anomalies and Psychomotor Retardation

D.S. Krishna Murthy
S.K. Murthy
G.J. Banker
A.J. Patel

Numerical aberrations (trisomy 21, 13, 18) resulting from aneuploidy due to nondysjunction (meiotic or mitotic) are one of the major chromosomal abnormalities in multiple congenital anomalies. Deletion (loss of genetic material) is the most frequently observed structural abnormality in humans. Deletion has been reported for all 22 pairs of autosomes and sex chromosomes, X and Y resulting in varying degree of phenotypic abnormalities and pathogenesis(1,2). Though some manifestations are common in majority of deletion syndromes, some features are characteristic of a particular chromosome region and can be delineated as a definite "clinical entity".

Alfi and coworkers reported the first case of deletion (9p-) syndrome(3). More than 80 cases of this syndrome have been documented in different populations. However, no confirmed case of 9p- has been reported in our population(4). The prevalence of Alfi's syndrome is not well established. However, the prevalence is much lower than trisomy-21. Application of banding techniques, particularly high resolution technique will help in detecting the minor detions like 9p-. An underestimate of the prevalence of this well delineated syndrome cannot be excluded.

From the Department of Zoology (Cell Biology Lab), School of Sciences, Gujarat University, Ahmedabad 380 009.

Reprint requests: Dr. D.S. Krishna Murthy, Department of Zoology (Cell Biology), University School of Sciences, Gujarat University, Ahmedabad-380 009.

Received for publication July 13, 1990; Accepted August 22, 1990

Case Report

The probed, a 8-month-old male child, is the first and only child of healthy, nonconsanguinous parents. Family history was unremarkable. He was suspected to have Down's anomaly by a pediatrician and was referred for chromosome analysis. On examination at 8 months he had marked trigenocephaly, prominent forehead, flat occiput, upslanting palpebral fissures, ocular hyperteleorism, flat nasal bridge, antiverted nostrils, long philtrum, mild micrognathia, high arched palate, low set ears and short neck widely spaced nipples *(Fig 1)*. There was profound hypotonia with reduced reflexes. The genitals were hypoplastic and the gonads were small and undescended. Dermatoglyphics showed simian crease. The child had marked developmental retardation.

Radiological investigations revealed: (*i*) *Skull*: Normal sutures. Antero-posterior diameter was smaller compared to vertical diameter. Pituitary fossa was normal. Petrous temporal bone and orbital margins

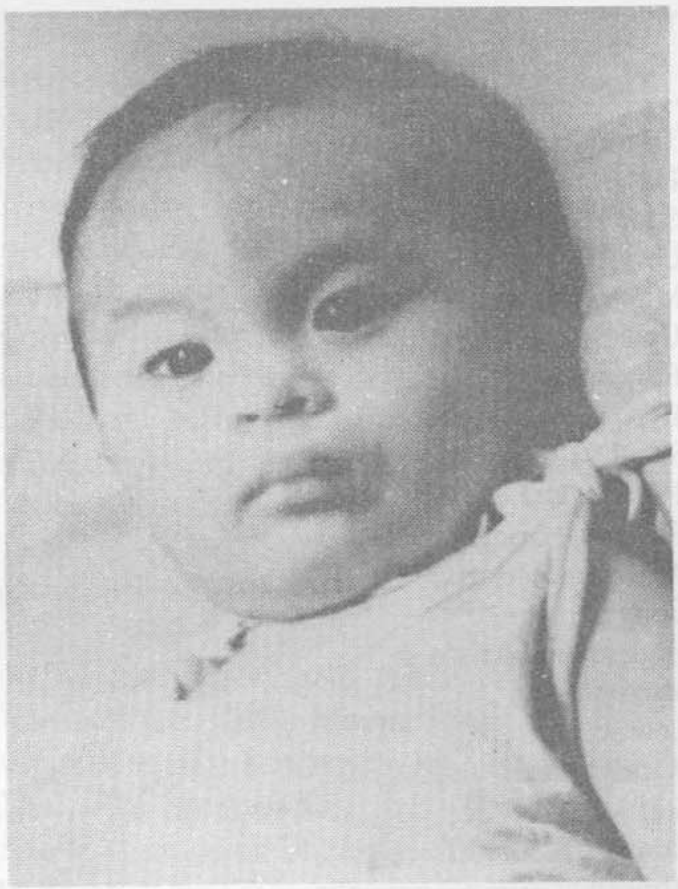

Fig. 1. Phenotype of the proband

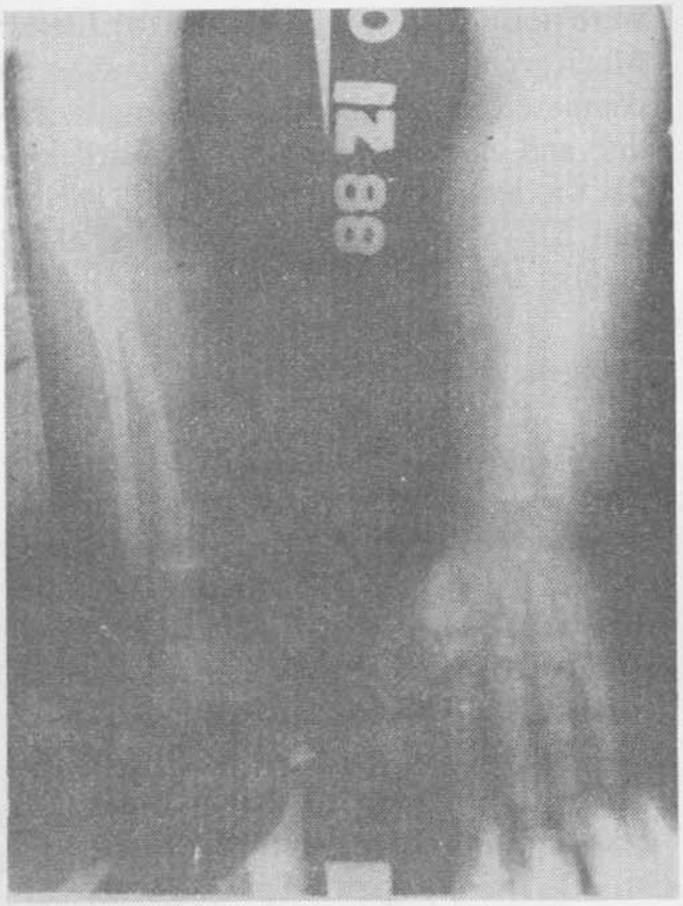

Fig. 2. X-ray of hand.

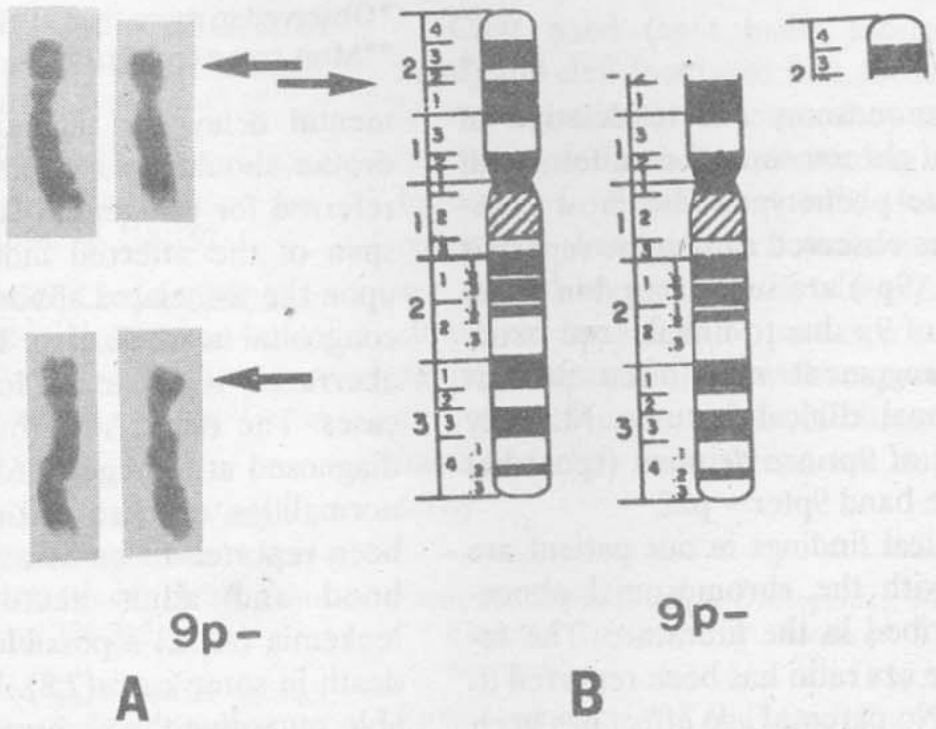

Fig. 3. A. Partial karyotype (G-banding) showing deletion (Arrow—break point).
B. Diagramatic representation of the deletion.

were normal on both sides; and (*ii*) *Limbs:* Metacarpel bones were relatively short as compared to phalanges of both hands. Radius and ulna were normal. Metacarpel index was less compared to normal (3.8, Normal 5.3 ± 0.41) for the average of 12 months *(Fig. 2).*

Cytogenetic techniques

Metaphase chromosomes of the proband and parents were prepared from peripheral blood culture as per standard procedure and stained for conventional staining and Giemsatrypsin banding. A minimum of 30 metaphase spread were analyzed in each case. The karyotype of the proband showed apparent deletion of the short arm of chromosome 9 in all the cells analyzed. The karyotype is interpreted as, 46, XY, del (9) (pter → p22). Partial karyotype of the proband is shown in *Fig. 3.* Karyotypes of parents were normal.

TABLE I– *Features in Del (9p) Syndrome (Huret et al. 1988)*

Trigonocephaly/prominent head**
Flat occiput
Short/broad neck/webbing
Low hair line
Mild facial dysplasia
Upward slanting of eyes**
Epicanthus**
Hypertelorism**
Small palpebral fissures
High arched eye brows
Flat nasal bridge**
Antiverted nostrils**
Long philtrum**
High arched palate
Micrognathia
Low set ears
Widely spaced nipples
Stiff joints/scoliosis
Congenital heart disease
Abnormal external genitalia
Hyper convex, square nails
Delayed development/mental retardation

*Observed in more than 50% of cases.
**Most common features.

Discussion

Partial monosomy due to deletion of short arm of chromosome 9 manifests with characteristic phenotype. The most common features observed among the reported cases of del (9p-) are summarized in *Table I.* Deletion of 9p due to unbalanced structural rearrangement may often present with additional clinical features. Majority of the cases of 9p- are *de novo* (sporadic) involving the band 9pter→ p22.

The clinical findings in our patient are consistent with the chromosomal abnormality described in the literature. The female to male sex ratio has been reported to be 1.6 : 1.0. No parental age effect has been observed unlike Down syndrome. In the presence of trigonocephaly/prominent head, long philtrum and marked developmental delay the diagnosis of Alfi syndrome should be suspected at birth and referred for cytogenetic investigation. Life span of the affected individuals depends upon the associated abnormalities, such as congenital heart disease. The chromosome aberration is not lethal in majority of the cases. The oldest surviving case has been diagnosed at the age of 61 years(5,6). Abnormalities of chromosome 9 have also been reported to be associated with childhood and adult acute lymphoblastic leukemia (ALL) a possible cause for early death in some cases(7,8). No data is available regarding the recurrence risks among the *de novo* deletion (9p-) cases. However, the prenatal diagnosis can be made by amniotic fluid culture or chorionic villi biopsy.

Counselling of families having a child with *de novo* deletion syndrome is difficult as no accurate risk figures are available, though chances of recurrence are extremely rare. Chromosome analysis helps in the precise and early diagnosis of the pathogenesis as well as to exclude other causes. No specific etiological factors for del (9p-) has been established. The presence of a fragile site at 9p22 might be one of the predisposing factors for the origin of *de novo* deletion of the short arm, and needs to be confirmed.

Acknowledgements

The authors wish to thank Prof. A.B. Desai and Dr. (Mrs) Meena Shah. This investigation was supported by the Indian Council of Medical Research, New Delhi and the Council of Scientific and Industrial Research, New Delhi.

REFERENCES

1. Grouchy de J, Turleau C. Clinical Atlas of Human Chromosomes. Edinburgh, Churchill Livingstone, 1984, pp 87-92.
2. Schinzel A. Catalogue of Unbalanced Chromosome Aberrations in man. Berlin, De Gruyter, 1984, pp 350-357.
3. Alfi OS, Donnell GN, Crandall BF, Derencsenyi A, Mennon R. Deletion of the short arm of chromosome 9 (46, 9p-): A new deletion syndrome. Ann Genet (Paris) 1973, 16: 17-22.
4. Borgoankar DS. Chromosomal Variation in Man. A catalog of Chromosomal Variants and Anomalies, 4th edn. USA, Alan R. Liss Inc, 1984, pp 276-325.
5. Huret JL, Leonard C, Forestier B, Rethore MO, Lejeune J. Eleven new cases of del (9p) and features from 80 cases. J Med Genet 1988; 25: 741-749.
6. Nielsen J, Homma A, Christiensen F. The deletion 9p- syndrome: A 61-year-old man with deletion of short arm of 9. Clin Genet 1977, 12: 80-84.
7. Lai JL, Fenaux P, Poldet JP, *et al.* Acute lymphocytic leukemia with 9p anomalies: A report of 24 additional cases and review of literature. Cancer Genet Cytogenet 1988, 33: 99-109.
8. Hecht F, Hecht BK. Chromosome 9 in ALL: Breaks in 3 cases of AMLL. Cancer Genet Cytogenet 1986, 21: 1-5.

Congenital Cleft Hand and Cleft Foot

K.R. Patond
N. Kumar
S.K. Srivastava

Cleft hand (split hand, lobster claw hand) and cleft foot (split foot, lobster foot, lobster claw foot) are intercalary congenital bone deficiencies. According to the Frantz and O'Rahilly classification(1) of congenital skeletal limb deficiencies, these are named as partial adactylia, which includes absence of all or part of a metacarpal or metatarsal along with respective finger or toe. Herein, a case of bilateral cleft hand and cleft foot is reported due to its rarity.

From the Department of Orthopedics, Mahatma Gandhi Institute of Medical Sciences, Sevagram.

Reprint requests: Dr. K.R. Patond, Professor and Head, Department of Orthopedics, MGIMS, Sevagram 442 102, Wardha.

Received for publication July 19, 1990; Accepted September 3, 1990