

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, Christensen 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*

Case Report

A six-month-old male child was brought to the Orthopedic Out Patient with history of malformed hands and feet since birth. The child was the first issue in the family. There was no history of any drug intake or X-ray exposure in the first trimester of pregnancy or any congenital defects in the family members. Examination revealed defect in the central portion of both the hands with V-shaped cleft tapering proximally and dividing the hand into two parts, one consisting of two ulnar rays with syndactyly and the other consisting of thumb. Both the feet also had a single cleft extending proximally upto tarsometatarsal area and having great toe on its medial aspect and a single toe on the lateral aspect. There was varus of first metatarsal bilaterally with marked hallux valgus (*Fig.*). No other abnormality was detected on detailed examination. The child was able to grasp objects in both the

hands.

X-rays of the hands demonstrated complete absence of index and middle finger bones, fusion of all the carpal bones as a single unit and fusion of the carpal mass with base of metacarpals. X-rays of the feet showed bilateral hallux valgus with varus of first metatarsal making the medial border of cleft. On the lateral side were two metatarsals articulating distally with single phalanx. There were two tarsal bone masses fused separately with two metatarsal base.

Discussion

Barsky(2) described cleft hand of two types. In the typical cleft hand, a deep palmar cleft separates the two central metacarpals. One or more rays are absent, and the existing digits tend to be confluent and of unequal length. In the atypical cleft hand, the central rays are absent and only short radial and ulnar digits remain with a shallow cleft. Treatment of these types vary

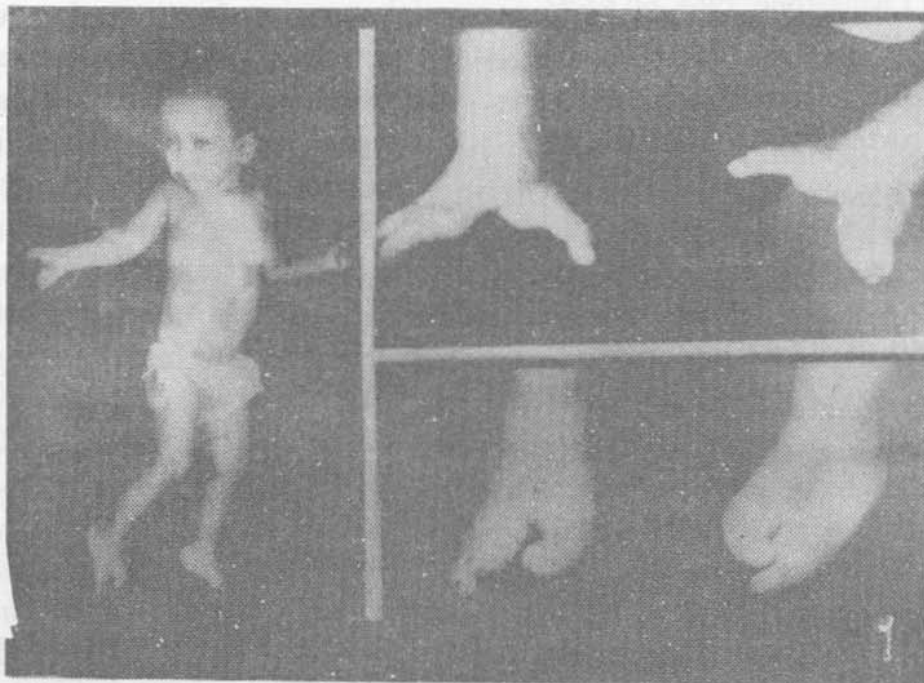


Fig. Clinical photograph of the child and close ups showing bilateral typical cleft hand and cleft foot.

with functional status of the hand. Cleft hand and cleft foot may be found singly or in association with each other. These may be present unilaterally or bilaterally. Miura(3) in a study of 152 congenital anomalies described associated abnormalities of cleft foot, syndactyly, polydactyly, pectoral muscle anomaly and cleft palate. Suzuki *et al.*(4) described 11 cases of typical cleft hand along with bilateral cleft feet in 3 cases, unilateral cleft foot in 2 and syndactyly of fingers and toes in one case each. Miura(5) noted intermediate cases between cleft hand and syndactyly suggesting similarity in the embryological failure in these two types.

The reported case is a rare combination of bilateral typical cleft and with bilateral cleft foot. It also had syndactyly and the classical radiological features. As the clefts were deep and functionally useful, no treatment was offered.

REFERENCES

1. Frantz CH, O'Rahilly. Congenital skeletal limb deficiencies. *J Bone Joint Surg* 1961, 43A: 1202-1224.
2. Barsky AJ. Cleft hand: Classification, incidence and treatment. *J Bone Joint Surg* 1964, 46A: 1707-1720.
3. Miura T. Congenital hand anomalies, and their association with other congenital abnormalities. *The Hand* 1981, 13A: 267-270.
4. Suzuki T, Tsuchiya K, Takazawa H, Matsui I. Dermatoglyphics in typical cleft hand. *The Hand* 1981, 13A: 69-76.
5. Miura T. A clinical study of congenital anomalies of the hand. *The Hand* 1981, 13A: 59-68.

Carbamylphosphate Synthetase-I Deficiency in a Newborn: Survival After Early Diagnosis and Therapy

K. Raghavan
S. Chabra
J. Mondkar
R. Aiyar
L.M. Ambani
A. Fernandez

Carbamylphosphate synthetase-I (CPS) deficiency is a rare, autosomal recessive inborn error of metabolism involving the urea cycle. Deficiency of CPS results in accumulation of ammonia, causing vomiting, lethargy, seizures, coma and death if hyperammonemia is not controlled. Review of the approximately 25 cases in world literature reveals 2 variants of CPS-I deficiency—a group with virtually complete deficiency of the enzyme resulting in death within the first week of life and a group, with a partial deficiency and a variable clinical course(1). Of the previously reported cases with the neonatal form, only 4 have survived with treatment(2,3). We report here a case of the neonatal type of CPS-I deficiency with a typically severe presentation in the first few days of life, but which could be successfully managed. To our knowledge, this is the first report of CPS-I deficiency from India.

From the Department of Pediatrics, L.T.M.G.H. and L.T.M.M.C., Sion, Bombay 400 022.

Reprint requests: Dr. Kalyani Raghavan, 6, Sneh Sadan, Sitaladevi Temple Road, Mahim, Bombay 400 016.

*Received for publication August 17, 1990;
Accepted September 30, 1990*