

permission to analyse the hospital records and publish this work.

REFERENCES

1. Singh M. Nomenclature. *In: Care of the Newborn*, 3rd edn. New Delhi, Sagar Publications, 1985, pp 1-8.
2. Vaughan VC. The field of Pediatrics. *In: Nelson's Text Book of Pediatrics*, 13th edn. Eds Behrman RE, Vaughan VC, Nelson WE. Philadelphia, WB Saunders Company, 1987, pp 1-5.
3. Sinha VC, Zacharia E. Qualitative aspect of Indian population. *In: Elements of Demography*, 1st edn. New Delhi, Allied Publishers Private Limited, 1984, pp 325-345.
4. Singh M, Paul VK. Strategies to reduce perinatal and neonatal mortality. *Indian Pediatr* 1988, 25: 499-509.
5. Sarna MS, Saili A, Dutta AK, Kumari S. Neonatal mortality patterns in an urban hospital. *Indian pediatr* 1991, 28: 719-724.
6. Bhatia BD, Mathur NB, Chaturvedi P, Dubey AP. Neonatal mortality pattern in rural based medical college hospital. *Indian J Pediatr* 1984, 51: 309-312.

Larsen Syndrome with Cardiac Anomaly

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Larsen's syndrome is a genetic disorder, first reported by Larsen in 1950 characterized by involvement of connective tissue, with varied manifestations. Since the

fully expressed gene results in death of fetus or newborn, the syndrome is very rare. We report here a patient of Larsen's syndrome who in addition to the classical features had mitral valve prolapse (MVP) leading to bacterial endocarditis.

Case Report

A ten-year-old boy was admitted for evaluation of fever, cough and breathlessness. He was born of a second degree consanguinous marriage. At birth he was noted to have hypermobile joints and bilateral clubfeet. The clubfeet were subjected to correction in infancy. His milestones were slightly delayed. All his family members were normal. He was 25th percentile of his height for age, had dolichocephaly, predominant forehead, hypertelorism, low nasal bridge, median cleft of the soft palate, anteriorly directed pinnae, pectus carinatum, hypermobility of the larger joints, bilateral subluxation of the knee joints, long cylindrical fingers with clubbing, short metacarpals, bilateral splayed feet and bilateral retractile testes. He had clinical features of MVP, left ventricular failure, and infective endocarditis. Echocardiogram showed MVP and mild aortic root dilatation. He was put on antibiotics, decongestives and supportive measures. He recovered in 6 weeks.

Discussion

Larsen *et al.*(1) recognized the syndrome consisting of multiple congenital dislocations of the knee and elbows, de-

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Received for publication: June 27, 1991;

Accepted: January 2, 1992

pressed nasal bridge, hypertelorism, prominent forehead, equinovarus or valgus, abnormal segmentation of the spine, and long cylindrical fingers. Three adults also have been described(2) and one of Larsen's patients had survived into adulthood(3). The characteristic facies and dislocation or subluxation of some nature are universal. Our patient had subluxation of both knee joints. Involvement of the knee is commonest followed by the hip. Involvement of the knee alone as seen in two patients(4). Equinovarus is also frequent and the splayed feet in our patient resulted from unsuccessful correction of clubfeet. The proband had long cylindrical fingers with clubbing and short metacarpals. The fingers were described variously as long and cylindrical(1), with short finger-nails and additional finger creases, short metacarpals(5), delta phalanx(6) and tapering or sausage shaped(7).

Our patient did not have vertebral anomalies. Various abnormalities have been described(4,5,6,8) including spinal instability leading to neurological damage and death(2). The bifid calcaneus was seen rarely, because of subsequent fusion as explained by Swensson(9).

Abnormal laxity of cartilage in early infancy, leading to respiratory difficulties and death were also described(8). This indicates abnormalities of the mesenchymal tissues of support. Myxomatous degeneration of the connective tissues supporting the mitral valve could have led to the floppy mitral valve in our patient. This has not been described previously. Swensson *et al.*(9) described a case with dilatation of the aortic root. The occurrence of such a condition was not surprising since the disorder is essentially one of connective tissues. The present case also showed mild aortic root dilatation alongwith MVP. The

implication is that it is a generalized mesenchymal disorder.

Other less common findings are synostosis of the radius and ulna and occipitalisation of the atlas(2), supernumery teeth(4), hypoplasia of the humerus and fibula(5), hydrocephalus(6), pectus carinatum(9) and mental retardation(4). One patient of Curtis and Fisher and abnormal chromosome 45 x 0 mosaicism which according to them was responsible for short stature, cryptorchidism, and mild mental retardation(5). The physical features described by Curtis and Fisher(5) were present in our patient also, but the karyotype was normal.

Occurrence in siblings or other family members have been described(1,3,4,6,7,10). Habermann *et al.*(8) described it in 3 generations of a family and suggested a single gene autosomal dominant transmission. Autosomal recessive mode was also mentioned(3,10). Larsen *et al.*(1) originally concluded that it is not heritable. But one of their patients later had an affected child(3) suggesting an autosomal dominant transmission. Mc-Kusick(7) categorized it as autosomal recessive in 1972. Latta *et al.*(5) suggested autosomal recessive and autosomal dominant single gene disorder with variable expressivity. In our patient, a sporadic mutation like those of Larsen(1) and Habermann *et al.*(8) or an autosomal dominant single gene theory with variable expression had to be thought of. The fully expressed gene results in a lethal effect on the fetus and the newborn. Hence the rarity of the condition. It may be recollected that respiratory distress occurred in several patients in the newborn period(2,5,8). The handicapping nature of the fully expressed syndrome would limit reproduction. Hence, most cases would represent fresh mutations.

REFERENCES

1. Larsen LJ, Schottstoadt ER, Bost FC. Multiple congenital dislocations associated with a characteristic facial abnormality. *J Pediatr* 1950, 37: 574-581.
2. Micheli LJ, Hall JE, Watts HG. Spinal instability in Larsen's Syndrome—report of three cases. *J Bone Joint Surg* 1976, 58A: 562-565.
3. McKusick VA. Mendelian Inheritance in Man. 4th edn. Baltimore, John Hopkins University Press, 1975, pp 84-85.
4. Curtis BH, Fisher RL. Heritable congenital tibiofemoral subluxation. Clinical features and surgical management. *J Bone Joint Surg* 1970, 52A: 1104-1114.
5. Latta RJ, Graham CB, Aase J, *et al.* Larsen's syndrome: A skeletal dysplasia with multiple congenital dislocations and unusual facies. *J Pediatr* 1971, 78: 281-288.
6. Steel RH, Kohl EJ. Multiple congenital dislocations associated with other skeletal anomalies (Larsen's syndrome) in three siblings. *J Bone Joint Surg* 1972, 54A: 75-82.
7. McKusick VA. Heritable Disorders of Connective Tissues, 4th edn. St. Louis, CV Mosby, 1972, pp 154-155.
8. Habermann ET, Sterling A, Dennis RI. Larsen's syndrome. A heritable disorder. *J Bone Joint Surg* 1976, 58A: 558-561.
9. Swensson RE, Linnebur AC, Paster SB. Striking aortic root dilatation in a patient with Larsen's syndrome. *J Pediatr* 1975, 86: 914-915.
10. Gorlin RJ, Pindborg JJ. Syndromes of the Head and Neck. New York, McGraw-Hill Book Co, 1964, pp 126-128.

NOTES AND NEWS

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