Case Report

Coffin-Siris syndrome in a twelve year old girl

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A 12-year-old girl was admitted to the Colombo South Teaching Hospital with bronchopneumonia. She was a product of a non-consanguineous marriage, born following umcomplicated pregnancy and labour. The perinatal period had been uneventful. However, she has had marked feeding difficulty with recurrent vomiting and regurgitation during infancy.

She was mentally retarded. Both her height and weight were below the 3rd percentile for age. Her head circumference was 48 centimeters, three standard deviations (3SD) below the median for age. Facial features were coarse with partial ptosis of the left eye, epicanthic folds, tall nasal bridge and broad tip of nose, short philtrum, macrostomia and prominent-lips. She had sparse scalp hair in conjunction with bushy eyebrows and long eye lashes. There was nail dysplasia involving all fingers and toes. The terminal phalanges were hypoplastic. The fifth fingernails on both hands was absent. She had a short sternum with pectus excavatum. There were mild scoliosis, hypoplastic patellae and generalised joint laxity with dislocation of radial heads at both elbows.

She had a skeletal maturity of 18 years. The CT scan of the brain was normal. Chromosomal studies revealed normal karyotype of 46, XX. There was evidence of congenital heart disease and the echocardiography studies of the heart revealed multiple defects, such as perimembranous ventricular septal defect, mitral valve prolapse with mitral regurgitation, dilated coronary sinus and a partial anomalous pulmonary venous drainage. A tympanogram showed significant hearing loss in both ears. Her serum calcium and the inorganic phosphate levels were normal. The alkaline phosphatase level was within the normal range for age (396 lU/L).

Discussion

All the above features, apart from the tall nasal bridge and advanced bone age, are typical of Coffin Siris syndrome, first described by Coffin G S and Siris E[1] in 1970 and subsequently by other workers. What have been described originally are flat nasal bridge and the delayed bone age[1]. A few related disorders such as Coffin-Lowry[2] and Coffin-Lowry like syndrome, which share some of the features of Coffin-Siris syndrome, have been described but they are quite distinct from this condition. This is the first time that a case of Coffin-Siris syndrome was described in this country.

This disorder has an autosomal recessive inheritance but no consanguinity was evident in this patient. A small chromosomal deletion described in a few earlier cases was not seen in this patient[3]. Rabe et al 1991 described two sisters with features of this condition who also had hyperphosphatasia[4] but the alkaline phosphatase levels of this girl were within normal limits.

Some of the main features described above are also seen in a few other disorders, such as the fetal hydantoin and other anticonvulsant syndromes, fetal alcohol syndrome, generalised gangliosidosis and mucopolysaccharidosis[5]. However, none of them show all the features of Coffin-Siris syndrome. Therefore, awareness of the existence of this condition is important to the practising paediatrician.

References

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