LETTER TO EDITOR (VIEWERS CHOICE)

CARDIOFACIOCUTANEOUS SYNDROME - A RARE ENTITY

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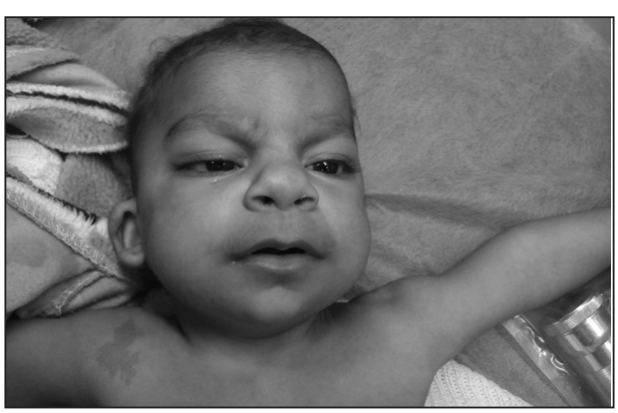
A six-month old male infant born out of non-consanguineous marriage presented with developmental delay, one episode of focal seizure two days prior to admission and abnormal facial appearance. On examination, weight was 5.5 kg (-2 to -3 SD), length was 63.5 cm (0 to -2 SD) and head circumference was 38 cm (< -3SD). He had microcephaly with large anterior fontanel and bitemporal narrowing. There were thin, curly, dry and sparse hairs along with down slanting palpebral fissures and nystagmus. Midfacial profile showed depressed nasal bridge with wide ala nasi, long and prominent philtrum, low set dysplastic ears, cupid bow lip and short neck. (Figure 1). Cutaneous manifestations in form of palmoplantar hyperkeratosis, follicular keratosis, and hyperkeratotic patch over right shoulder and hyperhydrosis with peculiar smell were noted. On cardiovascular examination there was early systolic murmur which on echocardiography was suggestive of moderate atrial septal defect (ASD) with mild pulmonary stenosis. On systemic examination, he had generalized hypotonia, cryptorchidism and mild hepatosplenomegaly. On the basis of above features possibilities of cardiofaciocutaneous syndrome (CFCS), Noonan syndrome, Costello syndrome and mucopolysacchridosis (MPS) were considered. Concentration of glycosaminoglycans (GAGs) by electrophoresis was normal and skeletal survey was not suggestive of MPS. To differentiate CFCS from related phenotypically similar conditions, CFC index proposed by Kavamura et al (1) was used. CFC index was calculated by adding the allocated value for each of the 82 characteristics in our index case. In our case calculated CFC index was 14.667 which strongly favored diagnosis of CFC syndrome. Genetic test could not be done due to unaffordability.

Figure 1: Facial features of the patient



Cardiofaciocutaneous syndrome is a condition of sporadic occurrence in which patients have multiple congenital anomalies, peculiar facial profile and mental retardation. (2) It is characterized by failure to thrive, distinctive facial features in form of prominent forehead, bitemporal narrowing, sparse/ absent eyebrows, hypertelorism, epicanthic folds, downward-slanting palpebral fissures, depressed nasal root and a bulbous tip of the nose and relative macrocephaly (3). Many of the patients have a congenital heart defect in form of atrial septal defect, pulmonary stenosis or hypertrophic cardiomyopathy. The cutaneous manifestations consist of hyperelastic dry and scaly skin, palmoplantar hyperkeratosis, non seborrhoeic eczema, pigmented nevi/ generalized hyper pigmentation; sparse, slow growing curly hair, and cutaneous hemangioma. (2-5) Moderate to severe developmental delay (most commonly in motor and language) is quite common. (5) CFC index was proposed by Kavamura et al (1) to differentiate this entity from Noonan syndrome and Costello syndrome, which have overlapping phenotypic features. The normal distribution of CFC index curve (mean ±2SD) ranges between 9.5 and 19.9. Management of a patient with CFC syndrome requires a multidisciplinary team approach involving pediatrician, cardiologist and developmental supportive care team with active surveillance for possible secondary complications. With adequate medical care, social and emotional support along with special education they are able to live an acceptable quality of life. (3)

Funding : None

Conflict of Interest : None

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DOI No. 110.7199/ped.oncall.2016.32