PAEDIATRIC DERMATOLOGY

ECTRODACTYLY, ECTODERMAL DYSPLASIA, CLEFT LIP/ CLEFT PALATE (EEC) SYNDROME SANS CLEFT LIP/CLEFT PALATE- A DISTINCT ENTITY?

Samipa Samir Mukherjee [(1)] *- Rohini P Gaikwad* [(2)]

Cloudnine Hospitals, Pediatric Dermatology And Dermatology, Bengaluru, India [(1)] *- Aashay Skin Clinic, Dermatology, Pune, India* [(2)]

Background: The EEC syndrome, consisting of ectrodactyly (E), ectodermal dysplasia (E) and cleft lip (C) with or without cleft palate, is the prototype of syndromes with the presence of heterozygote mutation in the p63 gene in most of the patients. These patients present with the characteristic lobster claw deformity with discoloration of hair and skin. About 30% cases have conductive hearing loss. Although it follows an autosomal dominant inheritance pattern mutations could sometimes be sporadic.

Observation: A seventeen year old male, first child of non consanguineous marriage born of full term normal delivery ,uneventful ante natal history presented with congenital deaf-mutism and deformity of hands and feet. Family history revealed similar complaints in the younger brother, amidst 3 siblings(the sister being normal), however the younger brother was not available for evaluation. Past and personal medical history was non contributory.
Examination revealed ectrodactyly of bilateral hands and split feet or lobster claw deformity of bilateral feet .Conspicuous absence of the dermatoglyphics with atrophy of the thenar eminence was noted in the left hand. The normal skin of the dorsum of the left hand extended on to the palmar aspect. Nails showed two dystrophic nails of the hands and feet. Premature canities and sparse hair on scalp and body was noted. The patient did not have cleft lip or cleft palate. The intelligence quotient as assessed by paediatrician was normal. There were no dental or ocular anomalies. Audiometry revealed 80% bilateral conductive hearing loss.

Key Message: EEC syndrome without cleft lip/palate should be considered as a separate entity.