Treacher Collins Syndrome

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A term female baby weighing 2.6 kg was born by normal vaginal delivery. She had mid face hypoplasia, antimongoloid slanting palpebral fissures, micrognathia, bilateral lower lid colobomas with sparse eyelashes, microtia, cleft palate, projection of lateral hair into lateral cheek and tri-phalangeal thumb (Fig 1 and 2). X ray skull showed hypoplastic maxilla and zygomatic arches on both sides. Ultrasonography of the cranium and abdomen was normal. BERA was suggestive of bilateral conductive hearing loss more on left side. Based on the above features a diagnosis of Treacher Collins Syndrome (TCS) was made.

TCS is a disorder of craniofacial development that is inherited in an autosomal dominant fashion and occurs with an incidence of approximately 1 in 50,000 live births. It is characterized by hypoplasia of the mandible and zygomatic complex, lateral downward sloping of the palpebral fissure, abnormalities of the external ears, conductive hearing loss and cleft palate. Mutation in a gene TCOF 1, which maps to 5q31.3-32, is responsible for this disorder. Chorionic villus samples can be analyzed at around 12th week of pregnancy for prenatal diagnosis. Genetic counseling of families with TCS is complicated by allelic heterogeneity. Hence specific mutation must be identified first within a family before any counseling or prenatal diagnosis can be carried out.

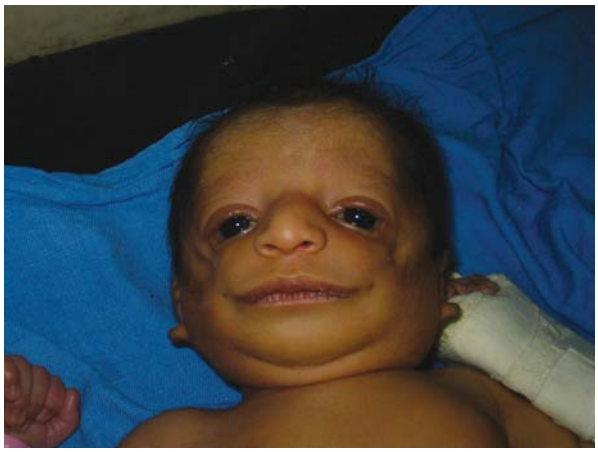

Fig 1: Frontal view showing down slanting palpebral fissure, colobomas of lower eyelids, and paucity of eyelashes. The features show bilateral symmetry.

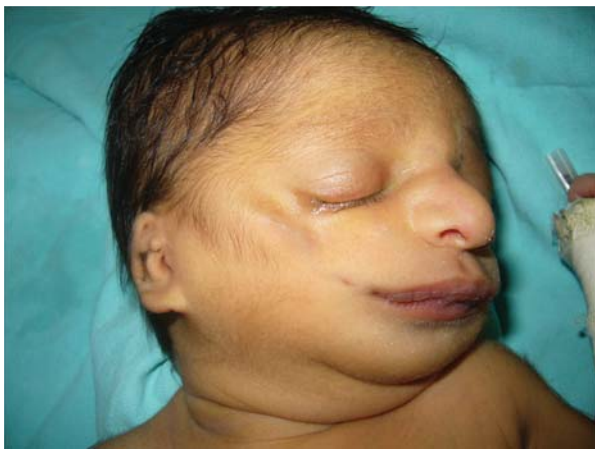

Fig 2: Lateral view showing hypoplasia of mandible and zygomatic complex and severe anomalies of the external ear.

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