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Isolated upper limb phocomelia: A rare limb malformation in a child with chronic adenotonsillitis

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Abstract
Phocomelia is a rare congenital disorder in which the hands and feet are attached close to the trunk, the limbs being grossly underdeveloped or absent. Congenital limb defects occur in approximately 6 per 10,000 live births. It may be inherited as an autosomal dominant or recessive disorder. Although many circumstance can cause phocomelia, this condition was a side effect of the drug thalidomide taken during early pregnancy. Our department report a infrequent case of phocomelia in a 9 year old child with chronic Adeno tonsillitis.

Keywords: phocomelia, tonsillectomy, thalidomide

Introduction
A 9 year old female born of a consanguineous marriage came to our ENT department with the complaints of difficulty in swallowing and throat pain for the past 10 months. Patient also had history of snoring and mouth breathing and recurrent URI for the past 10 months. On general examination child has bilateral non tender enlarged jugulodigastric node, absence of right forearm since birth (phocomelia of right upper limb). Systemic examination was WNL. ENT examination: oral cavity was WNL, Oropharyngeal examination showed bilateral grade 3 tonsillar enlargement and congestion of anterior pillar. Based on symptoms and examination child was detected to have chronic adenotonsillitis. X-ray nasopharynx showed adenoid hypertrophy. Routine blood investigations and ECG was WNL. ECHO was done which showed adequate LV Function, No shunt lesion, No Signific ant PAH, patient was hemodynamically stable, Anaesthetic opinion was obtained and Adenotonsillectomy was performed under GA by Dissection and Snare method. Intra operative and post-operative period was uneventful. Child was treated with appropriate antibiotics and supportive measures.

Fig 1: Pre operative picture of the child showing right upper limb phocomelia

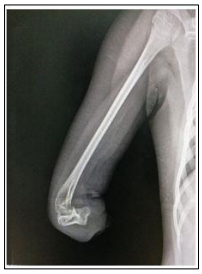

Fig 2: Pre operative X-Ray right upper limb showing absence of radius and ulna

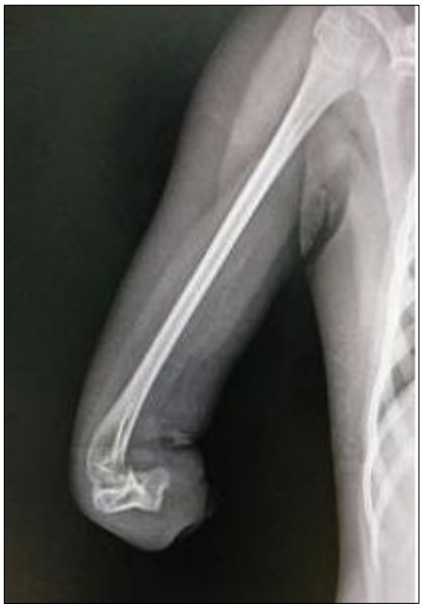

Fig 3: Pre operative X-Ray right upper limb showing absence of radius and ulna

Fig 4: Post operative picture following tonsillectomy

Discussion

Tonsillar enlargement and adenoid enlargement is the most common problem in otorhinolaryngology in children. Chronic Adeno tonsillitis is the main cause of airway blockage in children. Although antibiotic treatment is required in case of acute tonsillitis, tonsillectomy is done in chronic and recurrent tonsillitis.

Phocomelia is defined as the shortening of the long bones of the limb (upper and lower). Phocomelia can occur as an skeletal defect or can occur with other congenital anomalies. The first ever case of phocomelia was described in Germany in 1956 in a baby whose mother had received thalidomide[2] during pregnancy. Although phocomelia is due to maternal exposure to thalidomide during pregnancy, in many cases the underlying cause is poorly understood. The differential diagnosis includes sporadic phocomelia, Holt-Oram syndrome, Thrombocytopenia-absent radius syndrome (TAR syndrome), Roberts Syndrome, and thalidomide induced phocomelia.

Sporadic phocomelia [3] is a genetic disorder inherited as autosomal recessive trait or due to spontaneous mutation. In such scenario, there is 25% possibility for the child to be affected, provided both parents are carriers. Thus, there is more chance of phocomelia when parents have consanguineous marriage.

Thrombocytopenia - absent radius syndrome (TAR syndrome) [4] is characterized by a low platelet count, an absent radius, a hypo plastic thumb, and cardiac anomalies.

Roberts Syndrome [5] is a genetic disorder. It is identified by malformation of bones in face, skull, arms and legs.

Holt-Oram syndrome [6] may be seen as an autosomal dominant disorder Characteristic features include hypo plastic thumb that looks like a finger. Frequently, the radius is missing and the humerous is underdeveloped. The clavicle and the scapula may be affected. About 75% of the patients with Holt-Oram syndrome have cardiac problems the most common being defects in the tissue wall between upper chambers of heart (ASD) or the lower chambers of heart (VSD).

Thalidomide-induced phocomelia [7]: Thalidomide was used to treat nausea and reduce early morning sickness in pregnant mother. The typical clinical presentation includes defects of the arms, defects of long bone of the legs especially of the femur and tibia, absence of auricles, hemangiomata of the nose and upper lip (port wine Stain), atresia of the esophagus, duodenum and anus, cardiac anomalies and aplasia of the gall bladder and appendix.

Conclusion

Our case points to the fact that consanguineous marriage may cause phocomelia. Most of the congenital anomalies can be diagnosed antenatally if an anomaly scan is performed at around 18-20 weeks of gestation. This case had only isolated upper limb phocomelia and no other congenital deformity. With surgical and orthopedic rehabilitation, child will have normal life.

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