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CASE REPORT OPEN ACCESS

CONGENITAL ANOMALIES

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ABSTRACT

Congenital anomalies are present at birth and manifest in different parts of the body. But certain anomalies are esthetically compromised which affect the patient psychologically. Here we report four different cases with congenital anomalies. All these anomalies had additional orofacial manifestations. Case reports with review of literature are discussed.

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[I] INTRODUCTION

Congenital anomalies do occur because of a variety of etiologies. It is the prime duty of a clinician to diagnose the patients with congenital anomalies promptly so that proper care could be rendered to the patients. They vary in their clinical presentations. Approximately 2% of infants are born with extrinsically caused deformations that usually arise during late fetal life from intrauterine constraint. Approximately 30% of deformed infants have two or more deformations. Deformed infants tend to show catch-up growth toward their genetic potential during the first few postnatal months after release from the intrauterine constraining environment. Congenital deformations of the head and neck are common, and most resolve spontaneously within the first few days of postnatal life. When they do not, further evaluation may be necessary to plan therapeutic interventions that may prevent long-term consequences [1]. This article would present a series of case reports manifesting with various congenital anomalies.

[II] CASE REPORT

2.1. Case Report-1

A male patient 27 years of age on examination of left side preauricular area there was a sinus opening with no discharge and the patient revealed its presence since childhood. On examination of hands there was syndactly of 2nd and 3rd finger with single palmar crease. Verbal consent was taken from the patient for the photographs **[Figure–1]**.

The opening of the preauricular sinus has also been reported along the posteriosuperior margin of the helix, the tragus or the lobule [2]. The majority of patients with preauricular sinus are asymptomatic [3, 4]. A thorough history of head and neck examination is mandatory in all cases, seeking evidence of associated anomalies. Where it is an isolated, asymptomatic finding no treatment is required.

Syndactyly is a shared feature of more than 28 syndromes, including Poland, Apert, and Holt-Oram syndromes. Syndactyly is a failure of differentiation in which the fingers fail to separate into individual appendages. This separation usually occurs during the sixth and eighth weeks of embryologic development [5].

A simian crease is a single line that runs across the palm of the hand. People normally have three creases in their palms. A single transverse palmar crease formed by fusion of the proximal and distal palmar creases a common but not pathognomonic feature of Down syndrome; also found in 1% of the normal population. The simian crease is the most medically researched marker found on the hands because it is the most noticeable. A very high percentage of Down syndrome children have this marker, however, this doesn't mean everyone with this marker has Down [6].

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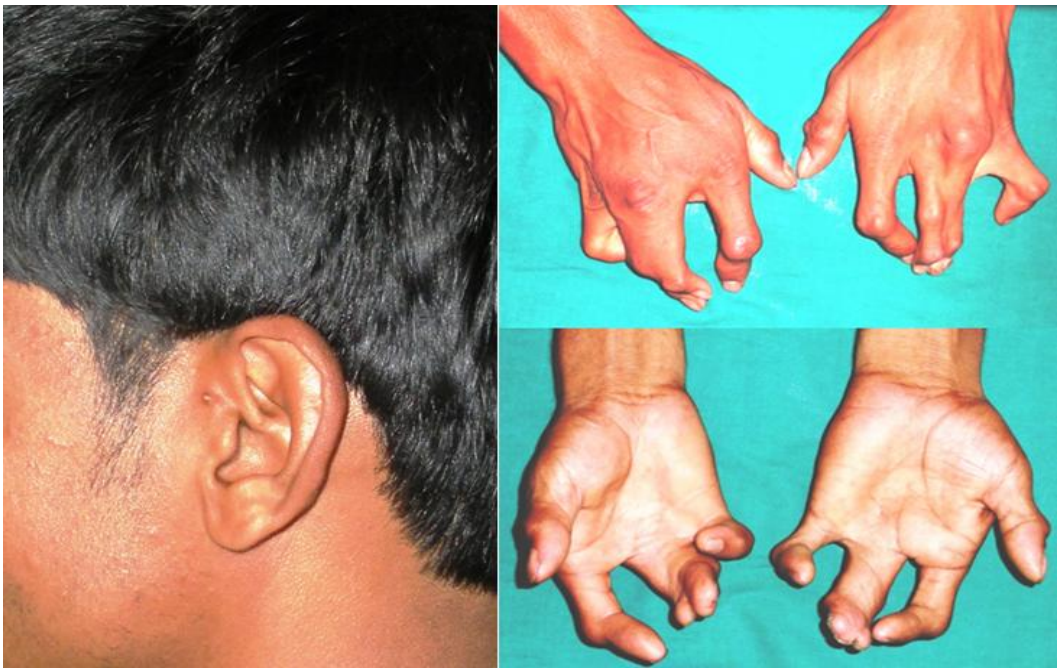

Fig-1: Syndactly of 2nd and 3rd finger with single palmar crease

2.2. Case Report-2

A 13 years old male patient on examination had broad nasal bridge, hypoplastic ala nasi, short first finger, clinodactly of fifth finger, genu valgum, metatarsus adductus. Intraorally he had bifid tongue in the anterior region which continued posteriorly as a fissure in the midline. He also had ankyloglossia along with malocclusion. Verbal consent was taken from the patient for the photographs **[Figure–2]**.

"Clinodactyly" or radio-ulnar deviation of the digits is a common congenital anomaly of the hand with a reported incidence of 19.5% in a non-Caucasian population. It is reported in about 1% of normal newborns and 10% of abnormal newborns. The most commonly affected digit is the little finger; the proximal phalanx of the thumb is the second most common site of occurrence. An inherited disorder with a dominant trait, the condition is usually bilateral. It usually results from an angulated short tubular bone - the "delta" phalanx - the middle phalanx commonly as the middle phalanx is the last bone in the hand to ossify [7].

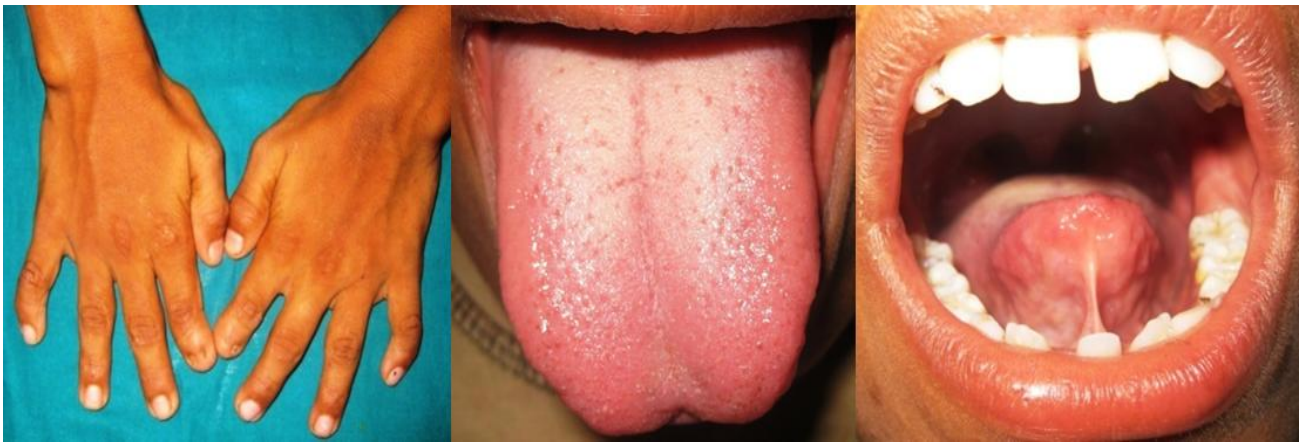

Fig-2: Short first finger, clinodactly of fifth finger, bifid tongue, ankyloglossia

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Ankyloglossia, commonly known as tongue tie, is a congenital oral anomaly which may decrease mobility of the tongue tip and is caused by an unusually short, thick lingual frenulum, a membrane connecting the underside of the tongue to the floor of the mouth. Ankyloglossia varies in degree of severity from mild cases characterized by mucous membrane bands to complete ankyloglossia whereby the tongue is tethered to the floor of the mouth [8, 9].

2.3. Case Report-3

A female patient 7 years of age on examination had triangular shaped forehead, negative canthal tilt, microphthalmia. No other abnormalities were observed. Verbal consent was taken from the patient for the photographs **[Figure–3]**.

Metopic synostosis is a relatively rare form of nonsyndromic synostosis. The incidence of metopic synostosis was reported to be 0.3 per thousand 1.000 live births. Under normal conditions, metopic suture remains patent until 2-3 years of age and so frontal bone development continues. However, premature closure of the metopic suture results in trigonocephaly. In trigonocephaly, a 'keel-shaped' deformity of the forehead is present and this is characterized by a prominent midline ridge, bilateral frontotemporal constriction with compensatory biparietal expansion, supraorbital and lateral orbital retrusion and hypotelorism. Besides that, interorbital distance decreases, orbita is rotated posterolaterally and radiological findings show superomedial harlequin deformity [10]. Triagonocephaly appears as part of the phenotype of some well known syndromes, such as opitz triagonocephaly syndrome, craniotelencephalic dysplasia, Say Meyer syndrome, VSR syndrome, and in autosomal dominant triagonocephaly without associated anomalies [11].

The major causes for this disorder are genetic but environmental factors have also been implicated such as exposure to radiation, chemicals, or viruses. Microphthalmia in newborns is sometimes associated with fetal alcohol syndrome or infections during pregnancy, particularly herpes simplex virus, rubella and cytomegalovirus, but the evidence is inconclusive. Genetic causes of microphthalmia include chromosomal abnormalities Patau syndrome, Triploid Syndrome, and Wolf-Hirschhorn Syndrome or monogenetic Mendelian disorders [12].

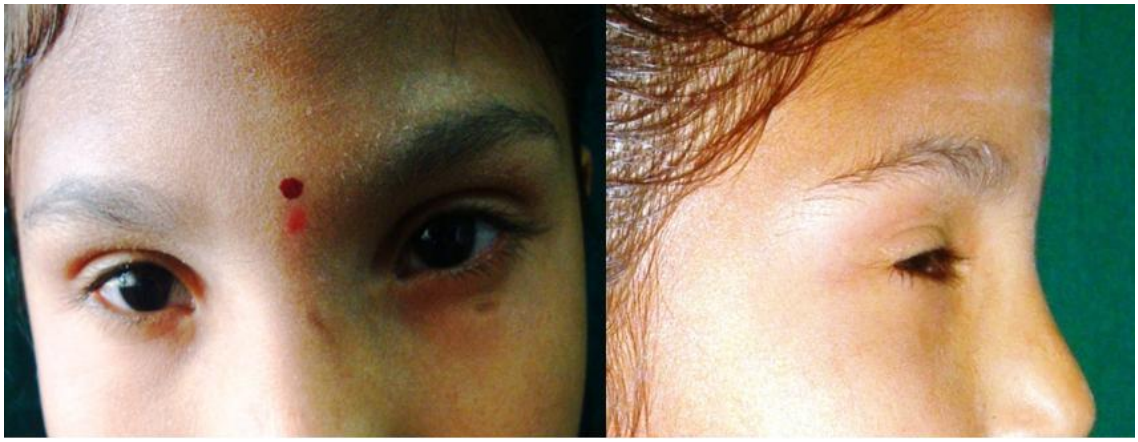

Fig-3: Triangular shaped forehead, negative canthal tilt, microphthalmia

2.4. Case Report-4

A female patient 16 year of age came to our department for regular check up. She gave history of consanguineous marriage and frequent fracture of bones on any severity of trauma. On examination she had bluish discoloration of sclera of eyes. On forehead she had scar resembling cigarette foil appearance. Her oral examination revealed no abnormality except flourosis. Radiological investigation revealed normal thickness of enamel and dentin. So a provisional diagnosis of osteogenesis imperfect was given. Verbal consent was taken from the patient for the photographs **[Figure–4]**.

Osteogenesis imperfecta (OI) is a heterogeneous group of genetic disorders that affect connective tissue integrity [13].

Tissues in which the principal matrix protein is type I collagen (mainly bone, dentin, sclerae, and ligaments) can be affected. The resultant abnormalities include blue sclera, rigidity of the osseous tissue, hearing loss, dentinogenesis imperfecta (DI), growth deficiency, laxity of the joints, and any combination of these characteristics [14]. The incidence of OI in infancy is about 1 per 20,000-30,000 in an Australian study [15].

There are extreme phenotypic variations within the OI

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population. Four types of OI including mild, perinatal lethal, progressive deforming, and moderately severe were classified according to clinical, genetic, and radiographic criteria [16]. Each of the 4 types of OI is further subdivided on the basis of the absence or presence of D [17].

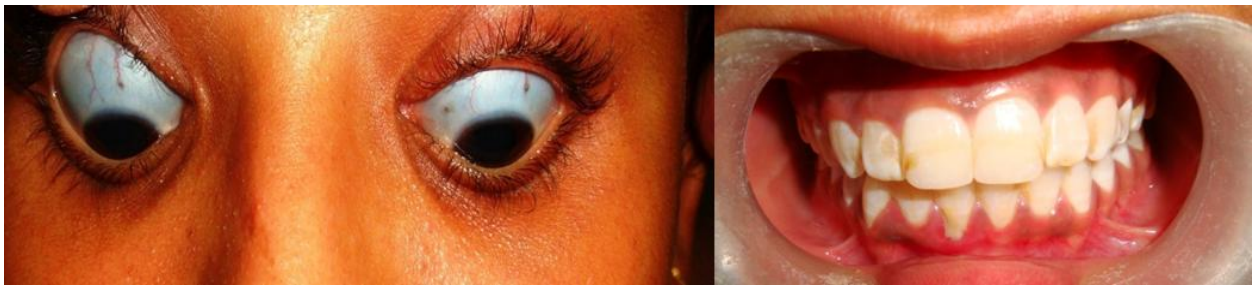

Fig-4: Bluish discolouration of sclera of eyes, flourosis

[III] CONCLUSION

A thorough knowledge about the developmental anomalies is essential so as to diagnose the ailments of the patients and intervene at an earlier stage. Hence, as a futuristic approach; proper genetic counseling is essential to prevent such anomalies in the progeny of the affected family.

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CONFLICT OF INTERESTS

All authors declare that they have no conflict of interest.

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