

syndrome characterised by orbital cysts, microphthalmia/anophthalmia, focal skin hypoplasia and skin tags, and cerebral malformations. Delleman syndrome show overlapping clinical features with other syndromes like Goldenhar syndrome, encephalo-cranio-cutaneous lipomatosis and Goltz syndrome. All the reported cases are sporadic with no risk of recurrence in the siblings. No etiological cause has been found yet. Proposed hypothetical molecular pathology are autosomal dominant gene-lethal gene survival

by somatic mosaicism, and twin spotting-coexistence of focal dermal hypo-plastic/hyperplastic lesion. In this case surgery for hydrocephalus is indicated to prevent further neurological damage and poor outcome.

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Langer-Giedion Syndrome

A 12-year-old girl presented with bony swellings that were seen over anterior chest at birth and at ends of long bones appearing sequentially over last 2 years. Her facial features included fine sparse scalp hair, eyebrows and eyelashes, bulbous nose, elongated philtrum, thin upper lip, hypoplastic mandible, irregular teeth, small head and large laterally protruding ears (*Fig. 1*). Exostoses were seen over ends of almost all long bones of limbs (*Fig. 2*) and left 5th and 9th rib



Fig. 1. Facial features characteristics of TRP II.



Fig.2. Exostoses of long bones of lower limbs.

anteriorly. Her scholastic performance was average and sexual maturity rating was stage III. There was no affected family member in 3 previous generations.

Langer-Giedion Syndrome (LGS) or type II trichrhinophalangeal syndrome (TRP) is usually sporadic and features may overlap with type I TRP, which is usually autosomal dominant. Theoretically, deletion of long arm of chromosome 8 in the region q24.11-q24.13 seen in approximately 50% of patients can be detected antenatal. But there is no previous attempt at antenatal diagnosis in literature possibly because only a few cases of vertical transmission have been reported so far. In addition to aforementioned features, other symptoms like hearing loss, submucous cleft palate, skin and joint laxity, melanocytic nevi, myopia, growth delay, mental deficiency, epilepsy, psychological disturbances, sarcomatous changes in exostoses, uretrohydro-nephrosis, hematometra, endocrine problems

like diabetes mellitus and hypothyroidism, epiphyseal dysplasia, avascular necrosis of femoral head and decreased reproductive fitness may be associated and require a multidisciplinary therapeutic approach. Bony exostoses require excision only if neurovascular or sarcomatous complications occur. Despite these numerous associated problems, the prognosis for general health is good. Differential diagnoses include Ehlers-Danlos syndrome (because of excessive redundancy and looseness of skin especially at birth and during early infancy) and Osteochondromatosis syndrome or Ollier's disease (if striking facial features of LGS are absent).

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