

## *Images in Clinical Practice*

### **Delleman Syndrome**

A one-month-old male child presented with abnormal looking left eye surrounded by skin tags, and focal alopecia. On examination left eye showed upper eyelid coloboma, epibulbar dermoid and surrounded by skin tags. Facial cleft was present on left (*Fig. 1*). He had left frontal and parietal focal alopecia. Overlying skin on these regions showed patches of focal dermal hypoplasia (*Fig. 2*). Neurological examination was unremarkable. On follow-up at age 2 years, he had developed hydrocephalus with right-sided hemiparesis and mild psychomotor retardation.

MRI brain at age one month revealed left intracerebral cyst communicating with dilated left lateral ventricle (*Fig. 3*). Brain surface overlying cyst showed pachygyria (posterolateral and medial aspect of parieto-occipital region). In addition MRI showed maldevelopment of corpus callosum, lipoma in left basitemporal region and arachnoid cyst in the left temporal lobe. Follow up MRI brain at age 2 years showed increase in the size of the cyst.



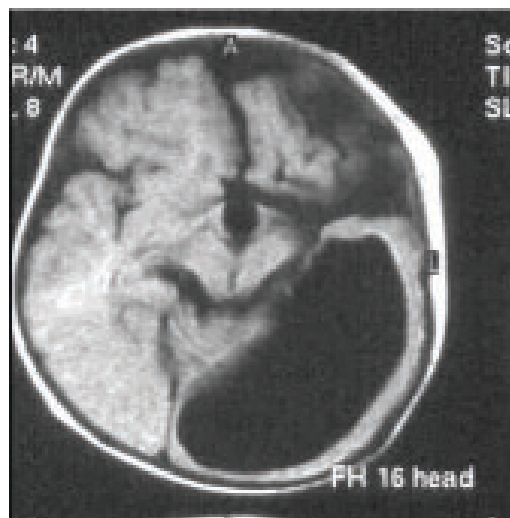
*Fig. 1. Left eye upper eyelid coloboma, epibulbar dermoid, skin tags.*

Chromosomal analysis revealed normal male karyotype with no detectable abnormality.

Delleman syndrome (syn: occulo-cerebrocutaneous syndrome) is a rare sporadic



*Fig. 2. Focal alopecia with focal dermal hypoplasia.*



*Fig. 3. MRI showing left intracerebral cyst.*

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.

**S.J. Patil,**  
**Shubha R. Phadke,**  
*Department of Medical Genetics,*  
*SGPGIMS, Raebareli Road,*  
*Lucknow 226 014 (UP), India.*  
*E-mail: shubha@spggi.ac.in*

## 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.*