

## Townes-Brocks Syndrome

A 1½-year-old girl presented with complaint of passage of stools per vaginum since birth. Examination revealed anteriorly placed anus with rectovaginal fistula. There was over folding of the superior helix of right ear (*Fig. 1*). Further examination revealed triphalangeal right thumb (*Fig. 2*). Ultra sonography of abdomen was normal. Skiagram of right hand showed that first metacarpal was not developed and revealed only four metacarpals and one carpal bone. A clinical diagnosis of Townes-Brock syndrome was made.

Townes and Brocks first described this disorder in 1972 and atleast 50 affected individuals have been reported. This single gene disorder (SALLI gene mutation)



*Fig. 1. Overfolding of superior helix of right ear.*



*Fig 2. Triphalangeal right thumb*

encompasses many of the features of both the VATER association and the facio-auriculo-veretebral malformation sequences. It is autosomal dominant with marked variability in the severity of expression for each feature. Thumb, auricular and anal anomalies are the major features of this syndrome. Occasional abnormalities include deafness, mental retardation, microcephaly, microtia, pre-auricular pit, cardiac defect, duodenal atresia, cystic ovary, prominent perineal raphe, hypospadias, 2-3 and 3-4 syndactyly of fingers. Identification of SALLI gene mutation can help differentiate from hemifacial microsomia.

**Priya,  
A.K. Malhotra,**  
*Department of Pediatrics,  
Mahatma Gandhi National Institute of  
Medical Sciences,  
Sitapura, Jaipur 302 022, India.*