

Marshall Syndrome

A seven-year-old boy born of non-consanguineous marriage presented with nasal regurgitation and nasal voice. Examination revealed a large head (head circumference 52 cm) with large eyes and short nose with flat nasal bridge (*Fig. 1*). There was a small submucous cleft palate with bifid uvula (*Fig. 2*). The child weighed 13 kilograms and was 102 cm in height, both parameters below 3rd percentile. Further examination revealed prominent upper incisors and thick lips. Skiagrams of head, dorsolumbar spine, pelvis, both lower limbs and upper limbs revealed calvarial thickening, mild platyspondyly and outward bowing of radius and ulna. A cataract had been noted in infancy but present ophthalmic examination revealed high myopia in both eyes with no evidence of

cataract. A clinical diagnosis of Marshall syndrome was made.

Marshall syndrome is an autosomal dominant chondrodysplasia characterised by mid-facial hypoplasia, sensorineural deafness and ocular defects (cataract, high myopia). Other abnormalities include ectodermal dysplasia, absent frontal sinuses, falx, tentorial and meningeal calcifications, spondyloepiphyseal abnormalities including slightly small and irregular distal femoral and proximal tibial epiphyses and wide tufts of distal phalanges. Occasional abnormalities include mental deficiency, retinal detachment and cleft palate. The cataracts may spontaneously resorb.

Marshall syndrome phenotype often resembles Spondyloepiphyseal Dysplasia Congenita (SED Congenita), Congenital Syphilis, Wagner syndrome and Stickler syndrome. Stickler and Marshall syndromes



Fig. 1. Large head, large eyes and short nose with flat nasal bridge.



Fig. 2. A small submucous cleft palate with bifid uvula.

were considered be the same disorder in the past but are now considered distinct. Patients with Stickler syndrome have flat cheek bones, a small jaw and cleft palate. In most cases of Marshall syndrome genetic studies have demonstrated a splicing mutation of 54-bp exons in c-terminal region of COL11A1 gene.

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