Clinical Brief

Yunis Varon Syndrome

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Abstract. In this communication is reported a neonate with Yunis Varon syndrome, a rare autosomal recessive disorder, born to a consanguineously married couple who had microcephaly, wide cranial sutures, prominent eyes, hypertelorism, dysplastic ears, sparse hairs, cupid bow like upper lip with median pseudocleft and labio-gingival retraction. Bilateral hypoplasia of thumbs, absent great toes, short phalanges were other features. Additional features in this case included median pseudocleft unreported earlier and C.T. findings of underdeveloped gyri, ischemic changes in temporoparietal region and bilateral lacunar infarcts in middle cerebral artery territory. [Indian J Pediatr 2006; 73 (4): 353-355] E-mail : drvani_hn@yahoo.com

Key words : Cleidocranial dysplasia; Distal aphalangea; Yunis-Varon syndrome (YVS).

Yunis Varon syndrome is an extremely rare autosomal recessive inherited multisystem disorder with defects affecting the skeletal, ectodermal and cardiorespiratory system. The striking characteristics are prenatal and postnatal growth deficiency, defective growth of bones of the skull, along with complete or partial absence of clavicles (cleidocranial dysplasia), characteristic facial features, hypoplasia or absence of thumbs and big toes and distal aphalangea. The first case was reported relatively recently in 1980 by Yunis et al. Since then less than 15 cases have been reported from different parts of the world.

Here is reported a 3-day-old child with typical phenotypic features and radiologic features of YVS, along with additional features like median pseudo cleft of upper lip and CT findings of underdeveloped gyri, ischemic changes in temporoparietal region and bilateral lacunar infarcts in MCA territory.

CASE REPORT

A 3 day old female child with dysmorphic features was brought to the hospital by anxious grandparents for genetic counselling as there was recurrence in the family. The child was the second child born to a healthy consanguineously married couple. The mother was 22 year old gravida 2, with no antenatal history of polyhydramnios or history suggestive of intrauterine infection like fever, rash or 'flue like' illness. Birth events were normal. The parents confirmed that the previous sibling had features similar to the index (propositus) and expired on ninth day of life.

On examination the baby was short stunted and dysmorphic (Fig 1), her weight was 2.5 Kg and length 45 cm and a head circumference was 28.5 cm, US to LS ratio 1.9:1.

The child had microcephaly, soft skull, wide anterior and posterior fontanelle. Eyes were prominent, and other

![Fig 1. Characteristic facial features, bilateral hypoplastic thumbs and absent great toes.](image-url)
features noticed were hypertelorism, dysplastic ears, sparse hairs, absent eyelashes and eyebrows, thin lower lip, narrow high arched palate, glossoptosis, cupid bow like upper lip with median pseudocleft and labiogingival retraction (Fig.2).

There was bilateral hypoplasia of thumb. Fingers were short and tapering. In the lower limbs great toes were absent and nails were hypoplastic. The skin over the neck was loose and joints were lax with generalized hypotonia and genitals were normal. Systemic examination was essentially normal.

Radiological examination revealed absence of first metacarpal bone and distal aphasis of both hands and feet (Fig.3). Clavicles were hypoplastic typically on left side. The long bones were slender. With these typical features, the diagnosis of YVS was made. Echocardiogram was normal. CT scan of head revealed microcephaly, wide separated sutures, features of bilateral MCA lacunar infaracts, ischemic changes in temporo parietal region and underdeveloped gyri.

**DISCUSSION**

Yunis Varon syndrome is a relatively recently described syndrome. It was first described in 1980 by Emilio Yunis and Humbuto Varon in five children from three Colombian families. The patients had multiple malformations, including cleidocranial dysostosis, micrognathia, absent thumbs and distal phalanges of fingers, hypoplasia of proximal phalanx and absence of distal phalanx of the big toes, pelvic dysplasia, bilateral hip dislocation and retracted and poorly deliniated lips. All died before the age of 10 weeks. Because of parental consanguinity and affection of both sexes the author suggested autosomal recessive inheritance. The facial features of the present case were strikingly similar to these cases but had additional feature of median pseudocleft, not described earlier.

Hughes and Partington proposed the designation “the syndrome of Yunis and Varon”. Subsequently only few case reports numbering approximately fifteen have been reported. Pfeiffer et al (1988) described a patient with consanguineous parents and emphasized aplasia of the thumbs and great toes as an outstanding feature. Rabe and colleagues reported two affected sisters from a German family, one sister had severe hearing impairment and pyloric stenosis, features not previously described in the syndrome. In some patients additional abnormalities have been described such as atrophy of left lobe of liver and anomalies of hepatic vessels, congenital heart disease, hypodontia, cardiomyopathy, osteodysplasty, spinal defects, CNS defects (Aplasia of corpus calosum, arhinencephaly, hamartomatous lesion of the lateral ventricles, cerebellar hypoplasia, hydrocephalus). In the present case CT scan showed features of bilateral MCA lacunar infaracts, ischemic changes in temporo parietal region and underdeveloped gyri.

Walch et al reported a 15 week female infant with YVS with microcephaly, hydrocephalus and Dandy-Walker syndrome.