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Case Report:

Yunis Varon Syndrome

Sameer I. Dal, Assistant Professor,
Parin Parmar, Resident,
Department of Pediatrics, KT Children Hospital, PDU Medical College, Rajkot – 360 001, Gujarat, India

Address For Correspondence:
Dr. Sameer Dal,
Assistant Professor,
Department of Pediatrics,
P. D. U. Medical College
Rajkot – 360 001, INDIA.
E-mail: dal.sameer@gmail.com

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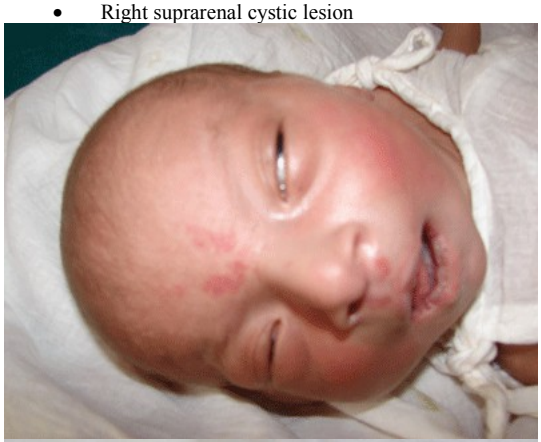
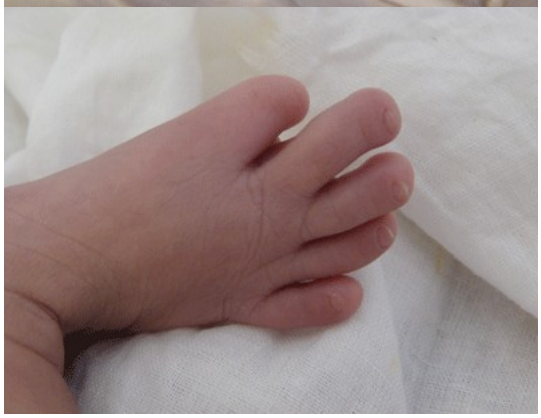
Abstract:
We have reported a case of Yunis-Varon syndrome which is a rare, autosomal recessive syndrome characterized by growth retardation, defective growth of the cranial bones, characteristic facial features, abnormalities of the fingers and/or toes & cleidocranial dysplasia. Additional features in this case were patent ductus arteriosus, CT brain findings suggestive of ischemic changes, CSF examination suggestive of pyogenic meningitis & cystic changes in right adrenal gland.
Key Words: Yunis-Varon syndrome, distal aphalangea

Case Report:
A male neonate weighing 2.9 kg was born at full term by spontaneous vaginal delivery to a 25-year-old gravida 6 mother. There was no history of consanguineous marriage. The mother gave history of having given birth to an earlier 2 female babies with dysmorphic features who had died on day 11 of life. The antenatal period was unremarkable. The neonate did not require resuscitation at birth and developed respiratory distress soon after.

Clinical features seen in the patient were as follows:

- Microcephaly
- Sparse scalp hair, eyebrows & eyelashes
- Short up-slanting palpebral fissures
- Anteverted nares
- Labiogingival retraction
- High arched palate
- Short philtrum
- Thin lips
- Cupid bow like upper lip
- Low-set & dysplastic ears
- Loose nuchal skin
- Micrognathia
- Agenesis/hypoplasia of thumbs & great toes
- Short tapering fingers & toes
- Nail hypoplasia
- Agenesis/hypoplasia of distal phalanges of fingers & toes, 1st metatarsals
- Simian crease (left side)
- External genital abnormality (hydrocele)
- Hazy cornea
- Bilateral nuclear cataract
- Mild ocular hypertelorism
- Patent ductus arteriosus
- Right suprarenal cystic lesion





Discussion:
Yunis-Varon syndrome is an extremely rare inherited multisystem disorder with defects affecting the skeletal, ectodermal tissue and cardio-respiratory systems. Less than sixteen cases have been reported in the world literature. It is characterized by growth retardation prior to and after birth; defective growth of bones of the skull, along with complete or partial absence of the clavicles (cleidocranial dysplasia); charac-

teristic facial features; hypoplasia or absence of thumbs & great toes & distal aphalangea.

The first report of this condition appeared in 1980 when Emilio Yunis and Humbuto Varon described 5 children from 3 families with cleidocranial dysplasia associated with certain other dysmorphic features including micrognathia, pelvic dysplasia, bilateral hip dislocation, and retracted and poorly delineated lips.(1) Two of the three sets of parents were consanguineous, suggesting an autosomal recessive disorder. All died before 10 weeks of age.

Huges and Partington proposed the designation "the syndrome of Yunis and Varon".(2)

Subsequently, there have been isolated case reports numbering a total of approximately sixteen cases with different additional clinical features (3-10), including two cases reported in India. (9,10) Pfeiffer *et al* mentioned aplasia of thumbs & great toes as an outstanding feature of the syndrome.(3) Subsequently other features described were severe hearing impairment, pyloric stenosis, atrophy of left lobe of liver and anomalies of hepatic vessels, congenital heart disease (5), severe osteodysplasty, cardiomyopathy (6), spinal defects, hypertension (7), median pseudocleft.(10) CNS defects that have been associated with Yunis Varon syndrome are absence of corpus callosum, arhinencephaly, hamartomatous lesion of lateral ventricles, cerebellar hypoplasia, hydrocephalus, Dandy Walker malformation.(4,7)

Aedes *et al* have described tetralogy of Fallot (5) associated with this syndrome but our patient had a patent ductus arteriosus.

Adrenal haemorrhage has been not described any where so far in this syndrome. However, CT brain changes of ischemic changes in temporoparietal region, underdeveloped gyri & bilateral lacunar infarcts in middle cerebral artery territory have been described by Kulkarni *et al* for the first time.(10) Our patient also had CT brain suggestive of ischemic changes.

Walch *et al* suggested Yunis Varon syndrome as an evidence for a lysosomal storage disorder.(7) Qualitatively abnormal bands for oligosaccharides and neuraminic acid were seen on urine analysis by thin layer chromatography. Autopsy showed prominent intraneuronal inclusions with vacuolar degeneration mainly in thalamic nuclei, dentate nuclei, cerebellar cortex and inferior olivery nuclei. Severe neurological impairment associated with intraneuronal inclusions and vacuolar degeneration has been taken as evidence for a lysosomal storage disorder. (7)

Dworzak *et al* also suggested the possibility of the syndrome resulting from disordered lysosomal storage based on the finding of vacuolar myopathy on muscle biopsy.(8)

The prognosis of this syndrome is poor. Only three of the 13 patients survived the first year of life (2,5,6), Two of the three survivors developed severe physical and mental retardation(2,5), and one patient showed growth retardation with normal intelligence but this child had only a few characteristics of Yunis Varon syndrome.(6)

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