Case Report:
Yunis Varon Syndrome
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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 & clidocranial 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

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 (clidocranial 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 dys-
plasia, bilateral hip dislocation, and retracted and poorly delin-
eeated lips.(1) Two of the three sets of parents were consan-
guineous, 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 osteodys-
plasty, cardiomypathy (6), spinal defects, hypertension (7),
median pseudocleft.(10) CNS defects that have been associ-
dated with Yunis Varon syndrome are absence of corpus cal-
losum, arhinencephaly, hamartomatous lesion of lateral vent-
ricles, cerebellar hypoplasia, hydrocephalus, Dandy Walker
malformation.(4,7)

Aades et al have described tetralogy of Fallot (5) associated
with this syndrome but our patient had a patent ductus ar-
teriosus.

Adrenal haemorrhage has been not described anywhere so far
in this syndrome. However, CT brain changes of ischemic
changes in temporoparietal region, underdeveloped gyri & bi-
lateral lacunar infarcts in middle cerebral artery territory have
been described by Kulkarni et al for the first time.(10) Our pa-
tient 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 associ-
ated 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 find-
ing 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 charac-
teristics of Yunis Varon syndrome.(6)

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