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A CASE OF YUNIS-VARON SYNDROME

Sudhakar Ajmera[1] and Nagesh Adla*[2]

[1]Department of Paediatrics, Kakatiya Medical College, Mahatma Gandhi Memorial Hospital, Warangal, Telangana, India.
[2]Department of Clinical Pharmacy, Vaagdevi College of Pharmacy, Warangal, Telangana, India.

***Author for Correspondence: Nagesh Adla**
Department of Clinical Pharmacy, Vaagdevi College of Pharmacy, Warangal, Telangana, India

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ABSTRACT
A rare autosomal recessive disorder, Yunis Varon syndrome is reported. A male child had microcephaly, large fontanella with wide sutures, protruding eyes, absent /sparse scalp hair, eyebrows and eye lashes, bilateral absent great toes, and rudimentary thumbs (no phalanges) hypoplasia of bilateral first metatarsal and metacarpals, absent distal phalanx of bilateral toes and fingers. Additional features are micropenis, single palmar crease, with hypo densities involving grey and white matter of bilateral temporo occipital lobes and right partial lobe, and congenital heart disease Atrial septal defect (ostium secundum).

KEY WORDS: Microcephaly, absent great toes and rudimentary thumbs, atrial septal defect, Yunis Varon Syndrome.

INTRODUCTION
Yunis Varon Syndrome is an extremely rare autosomal recessive disorder. It was first reported by Yunis and Varon in 1980. The striking characteristics are prenatal and postnatal growth deficiency.[1] Till now 22 cases were reported in the literature.[2] It appears to be a generalized disorder effecting growth and development of the skeletal, ectodermal, central nervous and cardiovascular systems.[3] We report a male child with phenotypic features of Yunis Varon Syndrome with additional findings of micropenis and atrial septal defect.

CASE REPORT
A 2 days old male child, product of consanguineous marriage, born to a 19 years old primi para came to the hospital with history of shortness of breath. The antenatal history was uneventful except history of fever with cough for 3 days in the last month of pregnancy. There is no history of polyhydramnias, rash or unknown drug exposure. The child was delivered by Cesarean section and no resuscitation done. No similar complaints in the family or near relatives.

At admission, the child was dull and respiratory distress was present with respiratory rate of 78/min. There were sub costal retractions, intercostals retractions and nasal flaring. The heart rate was 128/min; SpO2 was 95% with oxygen supplementation. Child developed neonatal Jaundice on 5th day and was treated accordingly. On examination the birth weight was 1.9 kgs (< 5th percentiles), and gestational age was 37 weeks (full term small for gestation) with new Ballards score. The length of the child was 46 cms (5th percentiles) and the head circumference was 30 cms (< 5th percentiles). The child had microcephaly, enlarged anterior fontanella (4.5cm x 4.5cm), posterior fontanella (1cm x 1cm), and widely placed sutural lines. The eyes were protruding, absent supra orbital ridges, spares hair on the scalp and eye lashes, absent eyebrows, low set and dysplastic ears, thin lips, short philtrum, labiogingival retractions, cupid bow like upper lip, high arched palate and micrognathia were noted [Fig: 1].

There were rudimentary thumbs and absent bilateral great toes. The fingers and toes are short and tapering, absent nails in both the fingers and toes were noted. There was Single palmar crease bilaterally. Stretched penile length measured from penile base to pubic symphisis was 1.7 cms, with both the testis in scrotum. Radiologically, the child showed hypoplasia of the medial end of right clavicle, periosteal reactions at shaft of femur, bilaterally (with left more than right). There was hypoplasia of middle phalanx of index finger and the 1st metacarpal bone. In the foot there was absent great toe with hypoplasia of 1st metatarsal bones bilaterally. There was bilateral absence of distal phalanx of all the fingers and toes [Fig: 2].

CT scan of the head and brain showed hypo densities involving grey and white matter of bilateral temporo occipital lobes and right parietal lob. The 2D ECHO showed ostium secundum Atrial septal defect with right

to left shunt. Neurosonogram and ultrasound abdomen were normal. There was incomplete ossification of skull bones (bilateral partial, temporal, and occipital).

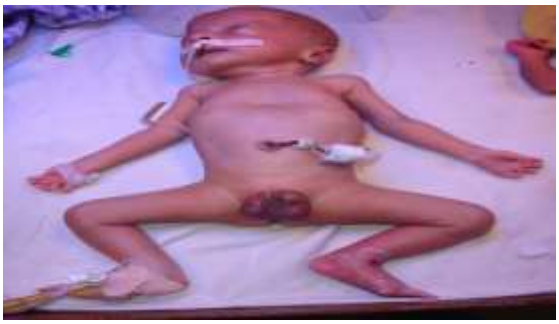

Figure: 1 general physical

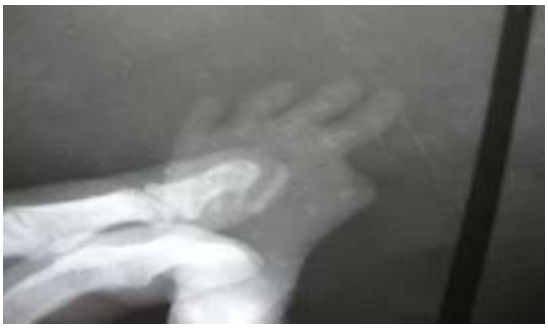

Figure: 2 X-ray of hand examination

DISCUSSION

Yunis Varon syndrome is relatively new and rare autosomal recessive disorder. In 1980 Yunis and Varon described 5 children from 3 families. They had multiple malformations like micrognathia, absent thumbs, distal phalanges of fingers and big toes, pelvic dysplasia and poorly delineated lips.[1] Occurrence of the syndrome in both the sexes, absence of reports of the syndrome in more than one generation and consanguinity points to the autosomal recessive inheritance. But the exact mechanism and gene responsible are not yet identified. It is characterized by pre and post natal growth deficiency, craniofacial disproportion, agenesis and hypoplasia of the clavicles, digital anomalies, with extreme hypoplasia of the first rays.[4] Very few cases were reported from India.[5]

Since then further features were reported like osteodysplasty[6], atrophy of left lobe of liver and anomalies of hepatic vessels[7], severe hearing impairment and pyloric stenosis[8], Walch *etal* reported Dandy walker malformation, cerebellar hypoplasia, hydrocephalus and intra neuronal inclusions with vacuolar degeneration in central nervous system grey matter.[9]

Cardiovascular features such as tetrology of Fallot[3], cardiomyopathy[10] and primary pulmonary hypertension[2] were reported. Our patient had Atrial septal defect (ostium secundum type) which was not reported earlier. Based on the abnormal bands of unidentified oligosaccharides in urine chromatography and vacuolar myopathy in muscle biopsy Dworzak *etal* proposed the possibility of the syndrome due to lysosomal storage disorder. Though similar vacuoles were found in heart, cartilage, central nervous system and cultured fibroblast but no biochemical defect was identified.[11]

Our patient showed micropenis and hypo densities of bilateral temporo occipital lobes and right parietal lobe in the CT scan and micropenis. Kulkarni *etal*[5] reported CT scan showing features of bilateral MCA lacunar infarcts, ischemic changes in temporo parietal region and under developed gyri. The prognosis is poor and death occurs mostly in the neonatal period attributed to cardiorespiratory failure. Only 3 of the 13 patients from the literature reviewed by Ades[3] *etal* survived the first year of life. And the survivors developed severe physical and mental retardation. They have severe failure to thrive and marked psychomotor delay.

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