

MULTIPLE PTERYGIUM SYNDROME: ESCHOBAR SYNDROME. A RARE DISORDER OBSERVED IN PAKISTANI COUPLE



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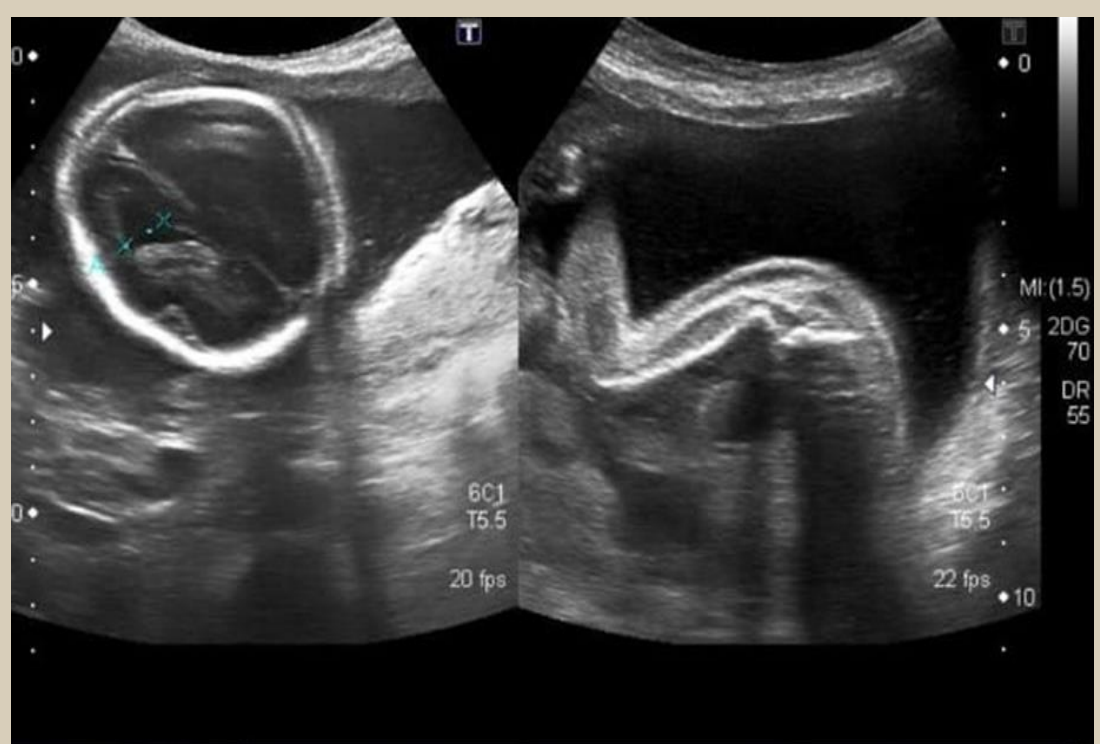
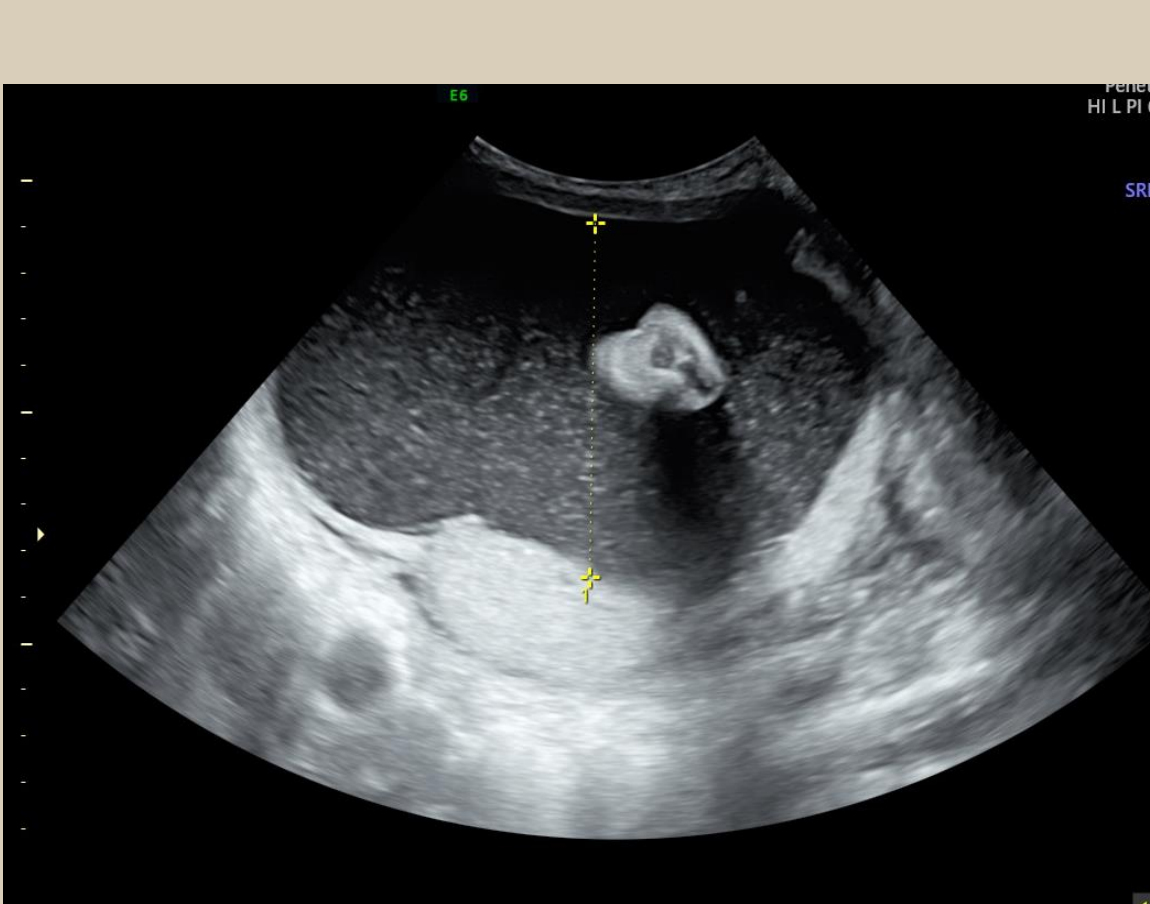
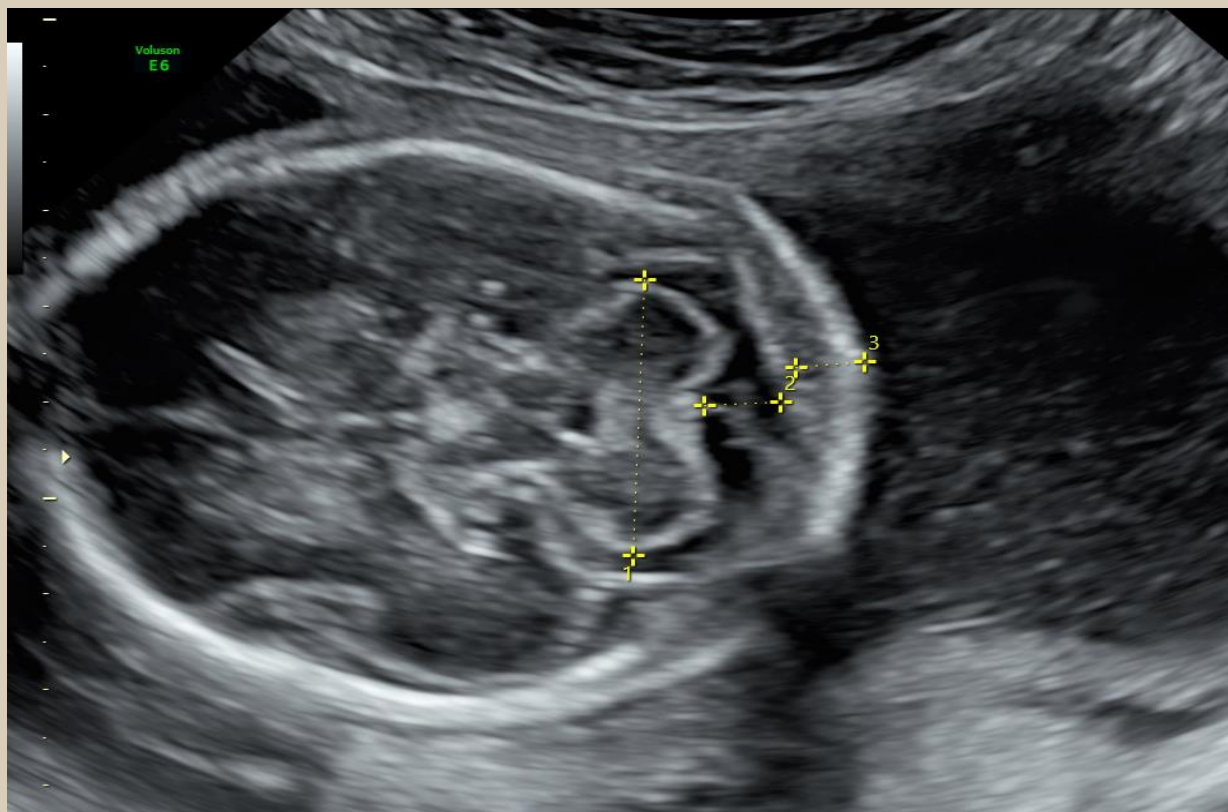
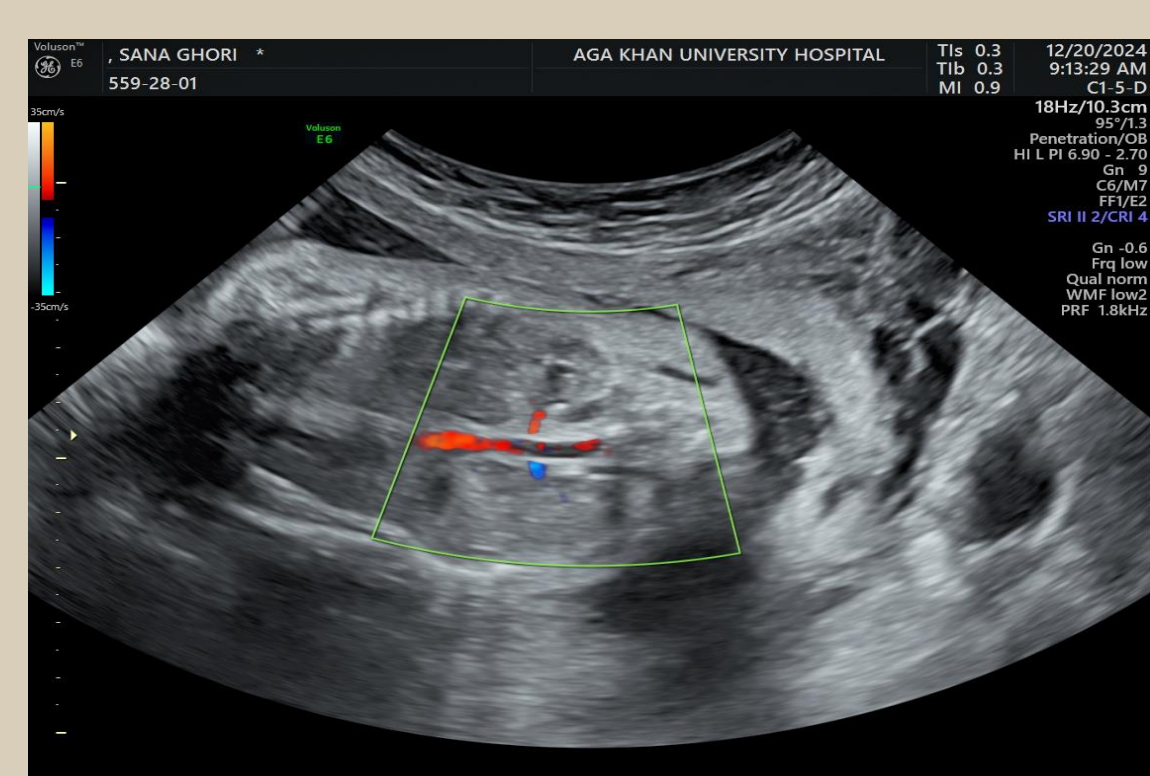
CASE

A 36 yrs old G5P3+1, at 30 weeks of gestation presented at MFU.
In her past obstetrics history, first born was well while the second born was a male baby with suspected pterygium syndrome who is alive and has normal cognition. Third baby had similar phenotypic features and died of Respiratory distress in early neonatal period. Fourth baby's anomaly scan showed multiple anomalies and couple decided for interruption of pregnancy.

In current pregnancy NT scan showed increased nuchal thickness. In genomic testing whole exome sequencing was performed and while waiting for results anomaly scan done.

ULTRASOUND FINDINGS

Anomaly scan at 24 weeks showed fetal growth at 4.2 centile polyhydramnios and short long bones. Raised nuchal fold, scoliosis of fetal spine at thoracolumbar region. Kidneys seem echogenic, hypoplastic nasal bone, micrognathia, mild ventriculomegaly. Fetal extremities have fixed flexion at knee, ankles, elbows and wrist joint (arthrogryposis) and rocker bottom feet.



Family screening Genetic testing report of affected child showed ESCHOBAR SYNDROME (which is autosomal recessive gene with mutation in gene variant of CHRNA3).
Parents whole exome sequence shows heterozygosity for the same gene.

KEY LEARNING POINTS

Escobar syndrome is a rare autosomal recessive disorder that affects the Musculo skeletal system and the skin. It is characterized by pterygia or webs of skin, joint flexion and contractures, cleft palate, micrognathia and spine deformity

. The mutations in the gamma subunit of nicotine acetylcholine receptors of CHRNA3 gene results in this disorder.

This recessive disorder that can present in utero with multiple congenital anomalies affecting skin and musculoskeletal systems. Prenatal findings suggestive of Escobar should prompt prenatal genetic diagnosis and appropriate counselling. .