A Rare Case of Escobar Syndrome

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Background
A 34 year old woman gravida 5 para 3+1 booked at eight weeks gestation. She was serology negative, varicella negative, rubella immune and rhesus positive. She has a past medical history of migraine, no alcohol or illicit drugs use during pregnancy. She has a complicated past obstetric history including a 36 week pre labour caesarean section for non reassuring CTG and delivered a male infant 2460g diagnosed with Escobar syndrome, now 6 years old requiring significant support including tracheostomy. Following this she had a 33 week intra uterine demise of a female infant weighing 1950g with suspected Escobar syndrome. Subsequently she had an unaffected monochorionic diamniotic twin pregnancy.

Current Pregnancy
A scan at 18 weeks gestation revealed a large cystic hygroma, scoliosis and permenantly flexed limbs. Repeat ultrasound at 20 weeks revealed micrognathia, pleural effusion, contractures and polyhydramnios. She went on to have serial growth scans and developed significant polyhydramnios with an AFI of 70 before delivery, amnioreduction was offered but declined.

At 35 weeks she was delivered by elective repeat caeseran section. A male infant was delivered weighing 3290g. The decision was made for comfort care only and he died one hour post delivery. On clinical examination there was found to be a cystic hygroma present at the neck, multiple contractures at neck, upper and lower limbs. Small thorax was noted and equinovalgus deformity of both feet. Parents declined a post mortem.

Discussion
Escobar syndrome is a form of arthrogryposis multiplex congenital. It is a rare autosomal recessive condition characterized by congenital contractures, excessive webbing (pterygia), and scoliosis(1). Escobar syndrome is a non lethal variant of multiple pterygium syndrome produced by a mutation in the gamma unit of the acetylcholine receptor gene held on chromosome 2q. The defect produced in the fetal cholinergic receptor from the mutated CHRNG gene (MIM100730) cause abnormal development of the neuromuscular junction with the resulting limitation in movement producing abnormal fetal morphogenesis(2)(3). The lethal multiple pterygium syndrome is also autosomal recessive with features including severe contractures, hypertelorism, cervical pterygia, narrow chest, and hypoplastic lungs(4).

References

Fig 1: 3D Ultrasound at 35weeks showing fixed flexion of fetal upper limbs.