Original Research Paper

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Paediatrics

ARTHROGRYPOSIS MULTIPLES CONGENITA-A CASE REPORT.

Dr. S Imtyaz Ahammad — 3rd Year Post Graduate, M.D. Paediatrics.

Dr. Shaik Neha Amreen — Fathima Institute Of Medical Sciences, Ramaraju Palli, Pulivendula Road, Kadapa-516003

KEYWORDS :

INTRODUCTION:

Arthrogryposis Multiplex Congenita is a rare sporadic nonprogressive congenital disorder characterised by multiplejoint contractures and can incorporate muscle weakness and fibrosis.

CASE REPORT:

A preterm 36 week , Male ,1.75kgs born to a 20 year G2P1L1 mother by Emergency Lscs through non consanguineous marriage.

- Antenatal no H/O maternal fever, no H/O medical ailments, there are decreased fetal movements ,TIFFA not done.
- Natal history : Breech presentation ,severe oligohydramnios
- Clinical examination:severe IUGR , Left parietal bossing , multiple joint contractures elongated philtrium ,
- SHOULDER: internally rotated and adducted,

ELBOWS :extended,

FOREARM :pronated with ulnar deviation,

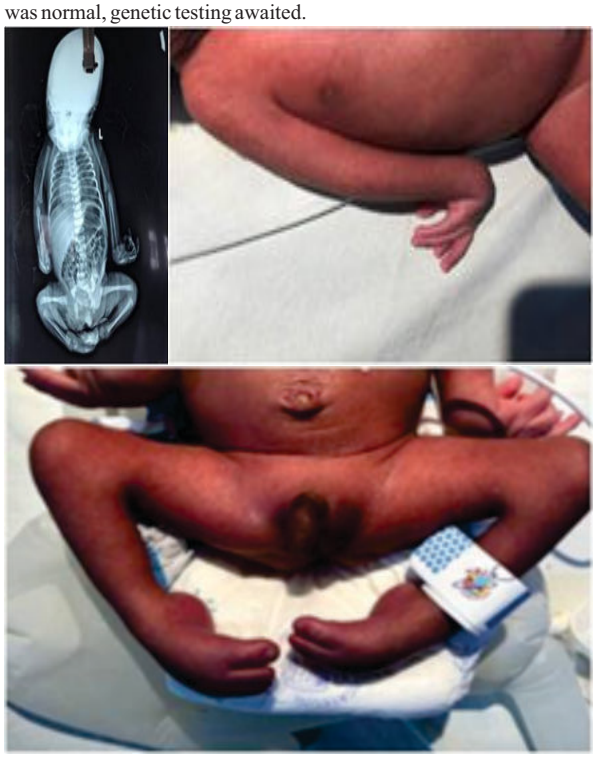
WRIST : flexed with flexed fingers

KNEE: flexed,

FEET: with severe equinovarus deformity and B/L undescendend testes

INVESTIGATIONS:

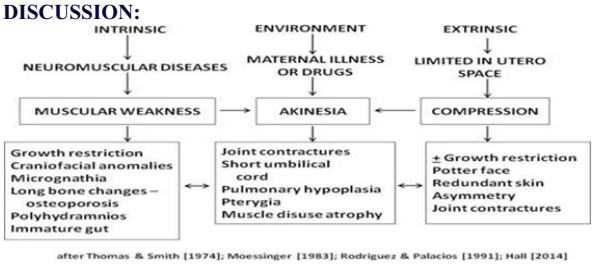
2D ECHO, USG joints,USG abdomen chest xray were normal, NSG was normal, genetic testing awaited.



Differential Diagnosis:

Fetal akinesia deformation sequence, Pena shokeir syndrome

DISCUSSION:



after Thomas & Smith [1974]; Moessinger [1983]; Rodriguez & Palacios [1991]; Hall [2014]

Fetal Akinesia Deformation Sequence:

Homozygous mutation in MUSK gene on 9q31 inherited in autosomal recessive manner .May sometimes be caused by mutations in RAPSN or DOK7 genes . About 30% of affected individuals are stillborn many live born infants survive only a short time due to complications of pulmonary hypoplasia

CONCLUSION:

this case report highlights the clinical presentation of Arthrogryposis Multiplex Congenita in a preterm infant, emphasizing the importance of early diagnosis and management. Despite the challenges posed by the condition, including multiple joint contractures and associated anomalies, appropriate interventions and supportive care can significantly improve the quality of life for affected individuals. Further genetic testing is awaited to confirm the diagnosis and guide long-term management strategies. This report underscores the need for increased awareness, early detection, and comprehensive multidisciplinary care to optimize outcomes for patients with Arthrogryposis Multiplex Congenita.

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