A case report of distal arthrogryposis

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Objective

To present a case of distal arthrogryposis

Method

A 37-years-old, G8P0 lady presented in her 8th pregnancy after 3 spontaneous miscarriage. The partner was 42-years-old. affected by distal arthrogryposis with flexion and ulnar deviation of the metacarpophalangeal joints and calcaneovalgus at birth. He underwent corrective surgery in childhood and was functioning normally. His mother was also affected. He had a normal daughter with his previous partner. Morphology scan performed at 19 weeks showed mild polyhydramnios and retronagthia, bilateral talipes equinovarus, left clenched fist with adducted thumb and flexed right. Active movements at the wrists and other joints were observed.

Result

The couple decided to continue with the pregnancy. An emergency Caesarean section was performed at 40 weeks gestation for non-reassuring cardiotopography. The baby had extended metacarpophalangeal joints on the right and flexed on the left. The interphalangeal joints were flexed on the right and extended on the left. Both thumbs were adducted at rest.



Figure 1. Findings on antenatal ultrasound include bilateral talipes equinovarus, clenched left wrist, flexed metacarpal phalangeal joints, adducted thumb and retronagthia.







Figure 2. The ultrasound findings were confirmed postnatally.

Active movements could be observed in the thumbs and fingers. There was bilateral talipes equinovarus and mild retronagthia. The proximal joints showed normal movements. Genetic study was not performed. The baby had physiotherapy and occupational therapy after birth.

Discussion

The distal arthrogryposes (DA) are a group of autosomal dominant disorders that mainly involve the distal parts of the limbs. It belongs to the group of arthrogryposis multiplex congenita (AMC), where there are congenital contractures in more than two joints in multiple body areas. The clinical findings and the severity may vary with the type of DA, and within the affected family. The affected individual usually has normal mental development.

Conclusion

Relatively good outcome could be expected in some types of distal arthrogryposis.