Arthrogryposis Multiplex Congenita

Alternative Names
AMC

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
108110

Mode of Inheritance
Isolated cases

Description
Arthrogryposis Multiplex Congenita is a rare disorder that is present at birth (congenital) and it comprises nonprogressive conditions. The major cause of arthrogryposis is fetal akinesia (decreased fetal movements), which could result from fetal abnormalities (e.g., neurogenic, muscle, and connective tissue abnormalities) or maternal disorders (e.g., infection, drugs, and trauma). AMC is characterized by reduced mobility of many joints of the body due to the overgrowth of fibrous tissue in the joints (fibrous ankylosis). The symptoms of arthrogryposis multiplex congenita vary widely among affected individuals depending on the disease type. In the most common form of the disease, the range of motion of the joints in the limbs is limited or fixed. Life span is usually normal, but it may be depend on the disease severity and associated malformations.

Molecular Genetics
Most types of arthrogryposis multiplex congenita are not inherited and do not occur more than once in a family. A genetic cause is only identified in about 30% of the cases.

Epidemiology in the Arab World

Oman
Aithala et al. (1995) studied identical Omani male twins, from first cousin parents, who were suffering from acute spinal muscular atrophy (SMA) in association with arthrogryposis multiplex congenita (AMC). The patients were considered as the first reported cases where both members of identical twins have SMA and AMC. At the age of 3 1/2 months, they showed severe hypotonia, wasting of limb muscles, paradoxical respiratory movements of the chest, symmetrical joints contractures, and mild scoliosis of the spine. A muscle biopsy was taken from one of the twins and histopathology examinations were done. The biopsy revealed an atrophy of the fibers. Joint contractures associated with AMC were thought to be due to decrease fetal movements in the uterus during pregnancy. The twins died at the age of eight months from severe pneumonia.

Rajab et al. (2005) reported six Omani children from two consanguineous families, with a multiple congenital anomaly syndrome defined by arthrogryposis multiplex congenita, typical facial appearance, ophthalmologic anomalies, atrophic calf muscles, and interdigital, neck and axillary pterygia. In addition, the patients had unique features as a furrowed tongue and enlarged corneal nerves, undescribed previously in association with other distal arthrogryposis syndromes (DA). Rajab et al. (2005) suggested that their patients could be classified as having Escobar syndrome type B because of the apparently recessive inheritance, short stature, relative macrocephaly, distinct facial appearance, camptodactyly, congenital foot deformities, and similar skeletal abnormalities such as vertebral defects, scoliosis, and joint dislocations.
Saudi Arabia
Abdullah et al. (2000) reported for the first time in the Arabian Gulf area three patients with arthrogryposis multiplex congenita, cholestasis and renal tubular dysfunction from a Saudi family with two other siblings and three cousins who possibly died with a similar clinical picture. The first patient, the 11th for a first degree Saudi cousin parents, presented jaundice at age of 3 days. Liver biopsy reported by showed intracellular and intracanalicular cholestasis with paucity of intrahepatic bile ducts. Many hepatocytes showed cytoplasmic granular pigment of indeterminate nature and there was no evidence of metabolic or storage disease. She was hypotonic with marked generalized muscle wasting, bilaterally dislocated hips, talipes equinovarus deformity and flexion deformities of the knees. By the fifth month she had severe global developmental delay. MRI brain at the age of 150 days showed hypoplasia of the corpus callosum, with insufficient myelination. Electromyography was consistent with muscular atrophy. The child had polyuria renal tubular acidosis, glucosuria, phosphaturia, hypophosphatemia, hyperchloremia, generalized aminoaciduria and proteinuria. The second case was a female baby, the sister of case 1, and had bilateral hip dislocation, knees flexion and talipes equinovarus. The third case was a female infant, sister of case 1, and was noticed to have flexion deformity of both knees and dislocated right hip at birth. She developed direct hyperbilirubinemia at age of 3 days. Abdullah et al. (2000) pointed that this autosomal recessive disorder is possibly under-diagnosed in the Arabian Gulf region, characterized by a high consanguineous marriage rate.

United Arab Emirates
Sztriha et al. (1998) reported an inbred Arab family with three neonates affected by microlissencephaly syndrome. The index patient was a male infant born by Cesarean section because of fetal distress. The parents were consanguineous of United Arab Emirates origin. Pregnancy was complicated by gestational diabetes requiring insulin treatment. On examination, he had gasping respiration. He died 10 hours after birth. Brain magnetic resonance imaging in the index case revealed very thin brain mantle with agyria-pachygyria, agenesis of the corpus callosum, and hypoplasia of the brainstem and cerebellum. Two male siblings also died soon after birth. All three neonates had microcephaly, arthrogryposis multiplex congenita, and micropenis.

References

Contributors
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