Distal Arthrogryposis Syndrome

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CASE REPORT

ABSTRACT

Arthrogryposis multiplex congenital (AMC) is characterized by contractions of multiple joints present at birth. The involved muscles are partially or totally replaced by fat or fibrous tissue. AMC is a distinct entity and distinction with the distal forms of arthrogryposis can be difficult, since there is a considerable clinical and genetic heterogeneity. A comprehensive musculoskeletal evaluation and genetic consultation is necessary.

Keywords: Distal arthrogryposis, Whistling face, Bowing of legs.

INTRODUCTION

The term arthrogryposis or (AMC) is often used to describe multiple congenital contractures that affect two or more different areas of the body. Arthrogryposis is not a specific diagnosis, but rather a clinical finding, and it is a characteristic of more than 300 different disorders.1

Distal arthrogryposis is a group of autosomal dominant disorders that mainly involve the distal parts of the limbs. It is characterized by congenital contractures of two or more different body areas without a primary neurological and/or muscle disorder. Features shared among all distal arthrogryposis include a consistent pattern of hand and foot involvement, limited involvement of proximal joints and variable expressivity.1

CASE REPORT

A 3-year-old female patient reported to the department of oral medicine and radiology with the complaint of multiple decayed teeth and recurrent swelling and discharge in upper anterior region since 1 year.

Patient’s medical history revealed that the child was premature baby with hydrocephalus and multiple joint deformities of wrist, ankle, knee and was diagnosed as distal arthrogryposis syndrome. The patient is undergoing physiotherapy since last 2 years.

The clinical examination revealed whistling face appearance with a small mouth and puckered lips. The patient also exhibited pinched nose, squint, hypertelorism, the skin over face showed tightening. Stiffness of metacarpophalangeal and interphalangeal joint of thumb and all four fingers in both hands, forefoot adduction and mild varus of heel was present bilaterally with bowing of legs (Figs 1 and 2). Rest of the joints and spine was normal.

Intraoral examination showed normal oral mucosa. There was generalized microdontia of all deciduous teeth with multiple remaining root rest of teeth carious.

RADIOGRAPH

Lateral skull view showed slight frontal bossing with prominent marking (silver-beaten appearance), micrognathia and hypoplastic mandible and suture are closed (Fig. 3).

Chest view showed hypoplastic second, third and fourth ribs, bowing of humerus, rest of the finding were normal and lungs were clear (Fig. 4).

Fig. 1: Facial feature of syndrome, varus of heel, bowing of legs
AMC is nonprogressive congenital syndrome complex characterized by contracture of several joints in different parts of the body. The genesis of AMC involves varying degrees of fibrosis of the affected muscles, thickening and shortening of periarticular capsular and ligamentous tissue with or without primary neurological or muscle disease that affect limb function.2

Ten different types of distal arthrogryposis have been described to date. The prototypic distal arthrogryposis is distal arthrogryposis type 1 (DA1, OMIM 108120) (OMIM—online Mendelian inheritance in man) and is characterized by camptodactyly (permanent flexion of finger or toes) and clubfoot. Hypoplasia and/or absence of some interphalangeal creases is common, with or without fifth digit, to severely clenched fists and ulnar deviation of the wrist. The shoulders and hips are less frequently affected.

DA1 is phenotypically similar to a condition called Freeman-Sheldon syndrome (FSS or DA2A, OMIM 193700). In addition to contractures of the hands and feet, FSS is
characterized by oropharyngeal abnormalities, scoliosis and a distinctive face that includes a very small oral orifice (often only a few millimeters in diameter at birth), puckered lips, and an H-shaped dimple of the chin; hence, FSS has also been called ‘whistling-face syndrome’. Individuals with DA1 and FSS have similar limb phenotypes and they can only be distinguished by the differences in their facial morphology.

The finding of our case is similar to type DA2A which showed all signs, such as stiffness of metacarpophalangeal and interphalangeal joint of thumb and all four fingers in both hands, forefoot adduction and mild varus of heel present bilaterally and clubfoot. Small mouth and whistling-face appearance, puckered lips except scoliosis was absent in our case. Our patient also has silver-beaten appearance on skull radiograph which probably may be attributed to hydrocephalus during early phase of life. As the patient is undergoing physiotherapy treatment her condition has improved.

DA2B or Sheldon-Hall syndrome (SHS, OMIM 601680)\(^3,4\) is probably the most common of the distal arthrogryposis disorders and shows more prominent nasolabial folds, high arched palate, attached earlobes, mild cervical webbing, short stature, severe camptodactyly, ulnar deviation and vertical talus and/or talipes equinovarus.

Distal arthrogryposis types 3, 4 and 6 (DA3, DA4 and DA6): DA3, DA4 and DA6 are very rare. DA3 or Gordon syndrome(OMIM 114300) is distinguished from other distal arthrogryposis by short stature and cleft palate. DA4 is a form with severe scoliosis.

DA5 (OMIM 108145) includes ptosis, restricted movement of the extraocular muscles and/or strabismus.

DA7 or trismus-pseudocamptodactyly syndrome (TPS, OMIM 158300) is an uncommon distal arthrogryposis characterized by an inability to fully open the mouth (trismus) and pseudocamptodactyly.

DA8 or distal arthrogryposis type 8 (OMIM 178110) autosomal dominant multiple pterygium syndrome.

DA9 or distal arthrogryposis type 9 (OMIM 121050) congenital contractual arachnodactyly.

DA10 or distal arthrogryposis type 10 (OMIM 187370) congenital plantar contractures and have short tendo calcaneus. The molecular basis of distal arthrogryposis syndromes mutations in at least five genes (TNNI2, TNNT3, TPM2, MYH3 and MYH8) that encode components of the contractile apparatus of fast-twitch myofibers can cause distal arthrogryposis 30 to 32. FSS and SHS are caused by mutations in MYH3, a gene that encodes embryonic myosin. Mutations in MYH3 explain approximately 90% of the cases of FSS and approximately 40% of the cases of SHS, making MYH3 mutation the most common known cause of distal arthrogryposis. No mutations overlap between those that cause FSS and SHS, suggesting an unambiguous genotype-phenotype relationship.

CONCLUSION

AMC is commonly seen by orthopedic surgeon because it is associated with multiple fixed deformities of both upper and lower limbs, however its association with dental problem has not been studied in great details in the past. The present case highlights the associated dental problem in patient with AMC and needs proper consideration in patient management.

REFERENCES