

Distal arthrogryposis type 1

Description

Distal arthrogryposis type 1 is a disorder characterized by joint deformities (contractures) that restrict movement in the hands and feet. The term "arthrogryposis" comes from the Greek words for joint (arthro-) and crooked or hooked (gryposis). The characteristic features of this condition include permanently bent fingers and toes (camptodactyly), overlapping fingers, and a hand deformity in which all of the fingers are angled outward toward the fifth finger (ulnar deviation). Clubfoot, which is an inward- and upward-turning foot, is also commonly seen with distal arthrogryposis type 1. The specific hand and foot abnormalities vary among affected individuals. However, this condition typically does not cause any signs and symptoms affecting other parts of the body.

Frequency

Distal arthrogryposis type 1 affects an estimated 1 in 10,000 people worldwide.

Causes

Distal arthrogryposis type 1 can be caused by mutations in at least two genes: *TPM2* and *MYBPC1*. These genes are active (expressed) in muscle cells, where they interact with other muscle proteins to help regulate the tensing of muscle fibers (muscle contraction). It is unclear how mutations in the *TPM2* and *MYBPC1* genes lead to the joint abnormalities characteristic of distal arthrogryposis type 1. However, researchers speculate that contractures may be related to problems with muscle contraction that limit the movement of joints before birth.

In some cases, the genetic cause of distal arthrogryposis type 1 is unknown. Researchers are looking for additional genetic changes that may be responsible for this condition.

[Learn more about the genes associated with Distal arthrogryposis type 1](#)

- MYBPC1
- TPM2

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of an altered gene in each cell is sufficient to cause the disorder. In many cases, a person with distal arthrogryposis type 1 has a parent and other close family members with the condition.

Other Names for This Condition

- AMCD1
- Arthrogryposis, distal, type 1
- DA1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Arthrogryposis, distal, type 1A (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220662/>)

Genetic and Rare Diseases Information Center

- Distal arthrogryposis type 1 (<https://rarediseases.info.nih.gov/diseases/787/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Distal arthrogryposis type 1%22](https://clinicaltrials.gov/search?cond=%22Distal+arthrogryposis+type+1%22))

Catalog of Genes and Diseases from OMIM

- ARTHROGRYPOSIS, DISTAL, TYPE 1A; DA1A (<https://omim.org/entry/108120>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28distal+arthrogryposis%5BTIAB%5D%29+AND+%28type+1%5BTIAB%5D%29%29+OR+%28%28arthrogryposis%5BTIAB%5D%29+AND+%28DA1%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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