



distal arthrogryposis type 1

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.

Genetic Changes

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.

Inheritance Pattern

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

Diagnosis & Management

These resources address the diagnosis or management of distal arthrogryposis type 1:

- Genetic Testing Registry: Arthrogryposis multiplex congenita distal type 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220662/>
- Merck Manual for Health Care Professionals
<http://www.merckmanuals.com/professional/pediatrics/congenital-craniofacial-and-musculoskeletal-abnormalities/arthrogryposis-multiplex-congenita>
- New York University Langone Medical Center
<http://nyulangone.org/conditions/arthrogryposis-in-children>

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy
<https://medlineplus.gov/drugtherapy.html>
- Surgery and Rehabilitation
<https://medlineplus.gov/surgeryandrehabilitation.html>
- Genetic Counseling
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care
<https://medlineplus.gov/palliativecare.html>

Additional Information & Resources

MedlinePlus

- Encyclopedia: Clubfoot
<https://medlineplus.gov/ency/article/001228.htm>
- Encyclopedia: Contracture Deformity
<https://medlineplus.gov/ency/article/003185.htm>
- Health Topic: Joint Disorders
<https://medlineplus.gov/jointdisorders.html>

Genetic and Rare Diseases Information Center

- Distal arthrogyriposis type 1
<https://rarediseases.info.nih.gov/diseases/787/distal-arthrogryposis-type-1>

Educational Resources

- Boston Children's Hospital
<http://www.childrenshospital.org/conditions-and-treatments/conditions/arthrogryposis>
- Disease InfoSearch: Distal arthrogyriposis type 1B
<http://www.diseaseinfosearch.org/Distal+arthrogryposis+type+1B/8272>
- MalaCards: arthrogyriposis, distal, type 1b
http://www.malacards.org/card/arthrogryposis_distal_type_1b
- My46 Trait Profile
<https://www.my46.org/trait-document?trait=Distal%20arthrogryposis%20type%201%20and%20type%202B&type=profile>
- Orphanet: Digitolar dysmorphism
http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1146

Patient Support and Advocacy Resources

- Resource list from the University of Kansas Medical Center
<http://www.kumc.edu/gec/support/arthrogr.html>

Genetic Testing Registry

- Arthrogyriposis multiplex congenita distal type 1
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220662/>

ClinicalTrials.gov

- ClinicalTrials.gov
<https://clinicaltrials.gov/ct2/results?cond=%22distal+arthrogryposis+type+1%22+OR+%22Arthrogyriposis%22>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?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>

OMIM

- ARTHROGRYPOSIS, DISTAL, TYPE 1A
<http://omim.org/entry/108120>

Sources for This Summary

- Bamshad M, Bohnsack JF, Jorde LB, Carey JC. Distal arthrogryposis type 1: clinical analysis of a large kindred. *Am J Med Genet.* 1996 Nov 11;65(4):282-5.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8923936>
- Bamshad M, Jorde LB, Carey JC. A revised and extended classification of the distal arthrogryposes. *Am J Med Genet.* 1996 Nov 11;65(4):277-81. Review.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/8923935>
- Gurnett CA, Desruisseau DM, McCall K, Choi R, Meyer ZI, Talerico M, Miller SE, Ju JS, Pestronk A, Connolly AM, Druley TE, Weihl CC, Dobbs MB. Myosin binding protein C1: a novel gene for autosomal dominant distal arthrogryposis type 1. *Hum Mol Genet.* 2010 Apr 1;19(7):1165-73. doi: 10.1093/hmg/ddp587. Epub 2010 Jan 2.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/20045868>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2838534/>
- Sung SS, Brassington AM, Grannatt K, Rutherford A, Whitby FG, Krakowiak PA, Jorde LB, Carey JC, Bamshad M. Mutations in genes encoding fast-twitch contractile proteins cause distal arthrogryposis syndromes. *Am J Hum Genet.* 2003 Mar;72(3):681-90.
Citation on PubMed: <https://www.ncbi.nlm.nih.gov/pubmed/12592607>
Free article on PubMed Central: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1180243/>

Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/distal-arthrogryposis-type-1>

Reviewed: January 2011

Published: February 14, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services