

X-linked chondrodysplasia punctata 1

Description

X-linked chondrodysplasia punctata 1 is a disorder of cartilage and bone development that occurs almost exclusively in males. Chondrodysplasia punctata is an abnormality that appears on x-rays as spots (stippling) near the ends of bones and in cartilage. In most infants with X-linked chondrodysplasia punctata 1, this stippling is seen in bones of the ankles, toes, and fingers; however, it can also appear in other bones. The stippling generally disappears in early childhood.

Other characteristic features of X-linked chondrodysplasia punctata 1 include short stature and unusually short fingertips and ends of the toes. This condition is also associated with distinctive facial features, particularly a flattened-appearing nose with crescent-shaped nostrils and a flat nasal bridge.

People with X-linked chondrodysplasia punctata 1 typically have normal intelligence and a normal life expectancy. However, some affected individuals have had serious or life-threatening complications including abnormal thickening (stenosis) of the cartilage that makes up the airways, which restricts breathing. Also, abnormalities of spinal bones in the neck can lead to pinching (compression) of the spinal cord, which can cause pain, numbness, and weakness. Other, less common features of X-linked chondrodysplasia punctata 1 include delayed development, hearing loss, vision abnormalities, and heart defects.

Frequency

The prevalence of X-linked chondrodysplasia punctata 1 is unknown. Several dozen affected males have been reported in the scientific literature.

Causes

X-linked chondrodysplasia punctata 1 is caused by genetic changes involving the *ARSL* gene. This gene provides instructions for making an enzyme called arylsulfatase L. The function of this enzyme is unknown, although it appears to be important for normal skeletal development and is thought to participate in a chemical pathway involving vitamin K. Evidence suggests that vitamin K normally plays a role in bone growth and maintenance of bone density.

Between 60 and 75 percent of males with the characteristic features of X-linked

chondrodysplasia punctata 1 have a mutation in the *ARSL* gene. These mutations reduce or eliminate the function of arylsulfatase L. Another 25 percent of affected males have a small deletion of genetic material from the region of the X chromosome that contains the *ARSL* gene. These individuals are missing the entire gene, so their cells produce no functional arylsulfatase L. Researchers are working to determine how a shortage of arylsulfatase L disrupts the development of bones and cartilage and leads to the characteristic features of X-linked chondrodysplasia punctata 1.

Some people with the features of X-linked chondrodysplasia punctata 1 do not have an identified mutation in the *ARSE* gene or a deletion involving the gene. Other, as-yet-unidentified genetic and environmental factors may also be involved in causing this disorder.

[Learn more about the gene associated with X-linked chondrodysplasia punctata 1](#)

- *ARSL*

Inheritance

This condition is inherited in an X-linked recessive pattern. The gene associated with this condition is located on the X chromosome, which is one of the two sex chromosomes. In males (who have only one X chromosome), one altered copy of the *ARSL* gene in each cell is sufficient to cause the condition. In females (who have two X chromosomes), a mutation would have to occur in both copies of the gene to cause the disorder. Because it is unlikely that females will have two altered copies of this gene, males are affected by X-linked recessive disorders much more frequently than females. A characteristic of X-linked inheritance is that fathers cannot pass X-linked traits to their sons.

Other Names for This Condition

- Arylsulfatase E deficiency
- CDPX1
- Chondrodysplasia punctata 1, X-linked
- X-linked recessive chondrodysplasia punctata 1

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Chondrodysplasia punctata, brachytelephalangi, autosomal (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1844853/>)

Genetic and Rare Diseases Information Center

- Brachytelephalangic chondrodysplasia punctata (<https://rarediseases.info.nih.gov/diseases/1296/index>)

Patient Support and Advocacy Resources

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

Catalog of Genes and Diseases from OMIM

- CHONDRODYSPLASIA PUNCTATA 1, X-LINKED RECESSIVE; CDPX1 (<https://omim.org/entry/302950>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28cdpx1%5BTIAB%5D%29+OR+%28%28chondrodysplasia+punctata%5BTIAB%5D%29+AND+%28recessive+tiab+OR+brachytelephalangic+tiab%29%29+OR+%28%28chondrodysplasia+punctata%5BTIAB%5D%29+AND+%28ARSE%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D>)

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