# CHONDRODYSPLASIA PUNCTATA

**Definition**

Chondrodysplasia Punctata is a group of hereditary disorder that affects infants and young children. It usually affects the skeletal system, skin, eyes and mental functioning. It is characterized by punctate calcification of the cartilage of the epiphyses, larynx and trachea or an inherited form of dwarfism.

**Symptoms**

- Growth retardation
- Bilateral Cataracts
- Dry and scaly skin (Icthyosis)
- Large Skin pores
- Dry Hair or Patchy alopecia
- Mildly Retarded
- Abnormal facial features ( depressed nasal bridge)
- Curvature of the spine
- Cartilage Abnormalities
- Microcephaly

**Diagnostic Tests**

For parents who know that they are carriers of the X-linked type of chondrodysplasia punctata, there is a prenatal procedure and test called preimplantation genetic diagnosis (PGD). After in vitro fertilization (IVF), PGD can test for genetic abnormalities, as well as gender before an embryo is implanted.

"Preimplantation genetic diagnosis is essentially an alternative to prenatal diagnosis, in which genetic testing is performed on embryos before a clinical pregnancy is established."

Prenatal ultrasound may be helpful in diagnosing chondrodysplasia punctata in the fetus. A second trimester ultrasound may detect the characteristic punctated calcifications of the spine and feet. Combined with evidence of shortened limbs, a diagnosis may be made. However, in milder cases of the disorder, the defects may be too subtle for detection by a routine prenatal ultrasound.

A physical examination may diagnose the external features of this disorder, including the facial abnormalities, shortened limbs, curvature of the spine, and ichthyosis.

A definitive diagnosis may be made by x ray of the limbs and spine. In children of one year or younger, punctated calcifications may be seen in the long bone and the feet in the areas of cartilage at the ends of growing bones. This cartilage disappears after the first year of age and is replaced with growth plates. These plates appear normal on x ray. In adults and older children, the diagnosis is based on shortened bones in the arms and legs and the presence of other physical characteristics of the disorder.

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**Treatment**

The treatment and management of chondrodysplasia punctata is primarily orthopedic and dermatologic. The characteristic stippling or dotted cartilage will disappear as the child ages; however, shortened arms and legs and curvature of the spine require orthopedic treatment. In some cases, surgery may be necessary to help patients whose legs are different lengths.

In some individuals, bone growth may be induced by a surgical bone-lengthening procedure. This procedure involves several surgeries and an extensive recovery period. The bone to be lengthened is cut. Leaving a narrow gap between the two pieces of bone, metal pins are inserted into the bone and the skin is closed. An external frame is attached to the pins. Gradually, the bone is pulled apart just enough to provide a small gap for the bone to grow into. As the bone grows, the space is widened and more bone grows. After the bone has healed, the pins are surgically removed.

Spinal abnormalities, such as spinal cord compression and **scoliosis**, may be treated surgically. A spinal column fusion can relieve the stress on the spinal cord caused by malformations of the spinal column. In a spinal fusion, two or more vertebrae are fused together using bone grafts or metal rods.

Ichthyosis is often most severe at birth and can resolve completely as the child ages. However, in some individuals, the skin lesions may be extensive and long lasting, leading to recurrent skin infections. Management of ichthyosis involves topical treatment and, in severe cases, bandaging to help prevent infection.

**Prognosis**

Prognosis of chondrodysplasia punctata depends on the type. The rhizomelic form of this disorder has a very poor prognosis. Most individuals with this type of chondrodysplasia punctata do not survive the fetal period or die shortly after birth. Of those that do survive, life expectancy is 10 years or less. Along with the skeletal anomalies, profound mental retardation is common as well.

The non-rhizomelic type, also known as Conradi Hunermann disease, can have a better prognosis. Though the condition is extremely rare, a range of outcomes has been reported from death to mildly affected adults. The X-linked dominant type, or Happle's type, is usually lethal to males, and they generally do not survive past the second trimester of pregnancy. However, females with this type usually survive and may have normal intelligence.

**Complications**

- Breathing Difficulty
- May die shortly after birth

**Prevention**

Genetic counseling may be helpful for prospective parents when one or both have chondrodysplasia punctata.