

# Cartilage-Hair Hypoplasia

## Overview

Cartilage-hair hypoplasia, otherwise known as Metaphyseal chondrodysplasia, McKusick type, is a disproportionate form of dwarfism that affects an estimated 1 in 200,000 live births. Despite its name, cartilage-hair hypoplasia (CHH) affects many body systems, particularly the immune system. Common features include fine hair that may be thin or scarce, joint laxity and scoliosis. CHH is the result of mutations in the *RMRP* gene and may be detected using radiological techniques, physical exams and genetic testing. Treatment of symptoms may include monitoring and surgery performed by doctors who specialize in skeletal dysplasia.

## Heredity and genetics

The *RMRP* gene is important for several cell-cycle processes, with over 100 mutations being shown to result in cartilage-hair hypoplasia. Similar mutations can result in related dysplasias like general metaphyseal dysplasias and anauxetic dysplasia.

CHH is inherited in an autosomal recessive manner, meaning a child must get a “bad” copy of the gene from both the mother and the father in order to have the disease. Each time the mother and father conceive, the baby has a 25 percent chance of inheriting CHH.

## Diagnosis and testing

A CHH diagnosis is usually made using a physical exam with radiological images. However, genetic testing may be necessary if a patient does not display the typical radiologic features.

Clinical Features:

- Short stature
  - Average adult height (males): 3-foot-7 to 4-11
  - Average adult height (females): 3-5 to 4-6
- Silky, fine hair that affects the hair, eyelashes, eyebrows and body
- Scoliosis
- Shortened fingers
- Joint hypermobility

Radiological Features:

- Shortening of the long tubular bones with metaphyseal irregularities
- Bowing of the femurs with rounded distal epiphyses
- Shortened ribs
- Disproportionally long fibula

## **Orthopaedic problems and treatment**

### Growth Monitoring

Growth should be monitored using cartilage-hair hypoplasia-specific growth charts. CHH-specific BMI charts are currently unavailable in literature.

- Males: <https://lpamrs.memberclicks.net/assets/documents/CHH%5b1%5d.female.ht.pdf>
- Females: <https://lpamrs.memberclicks.net/assets/documents/CHH%5b1%5d.male.ht.pdf>

### Spine Deformities

Although it is uncommon, people with cartilage-hair hypoplasia can have instability in the cervical spine (upper spine). This can be monitored with cervical spine flexion and extension X-rays, which should take place at the time the patient is diagnosed with CHH, around age 5-6, and then approximately every 5 years until adulthood.

Side-to-side curvature of the spine (scoliosis) affects nearly 20 percent of people with CHH but does not generally require medical treatment (bracing, surgery, etc.). Scoliosis should be monitored yearly until final adult height is achieved via clinical exam and/or radiographs.

### Joint Hypermobility

Hypermobility, or “loose joints,” causes joint instability in the wrists and knees. Severe hypermobility causes difficulty in fine motor activities such as knitting, writing, drawing, etc., and may find the hypermobile joints to fatigue more easily. Joint hypermobility is often noted at a young age then monitored as the child gets older. If symptoms interfere with everyday activities, consider modifying the environment (use of an ergonomic keyboard, modified eating and writing utensils, etc.).

### Leg Alignment Abnormalities

Genu varum, or “leg-bowing,” is common and is most frequently a result of overgrowth of the fibula, a bone in the lower leg. Genu valgum, or “knock-knees,” is another leg alignment issue seen in CHH, though it is less common. A physical exam should be used to monitor leg alignment as well as a series of X-rays called an extremity alignment series, which monitors the mechanical axis of the femoral head, knees and ankles. Orthopaedic treatment may be necessary for genu varum or valgum if the alignment causes pain or a decreased ability to walk distances or participate in physical activity.

## **Other common problems and treatment**

### Immune System Deficiencies

More than 95 percent of people with cartilage-hair hypoplasia have some sort of immune system dysfunction. This can range in severity and type of immune response affected but often results in an increased risk for serious, recurring infections. Children age 2 and younger are especially at risk for infections, particularly bacterial pneumonia.

Because of immune system problems, people with CHH are more likely to have autoimmune disorders, such as inflammation, anemia and thyroid diseases.

See <http://www.lpaonline.org/assets/documents/NH%20Cartilage%20Hair%20Hyoplasia.pdf> for monitoring and treatment recommendations for immune system impairments in CHH.

### Anemia

Macrocytic anemia, where the red blood cells have a larger volume than typical red blood cells and usually a smaller number of cells, is common. This makes it difficult for the blood to have appropriate levels of hemoglobin, the protein that carries oxygen through the body. Anemia can range in severity, with life-threatening cases requiring routine blood transfusions. Macrocytic anemia should be screened with the immune system workup and managed with a hematologist.

### Skin Cancer

People with CHH are at a higher risk of developing skin cancer, particularly non-Hodgkin's lymphoma, basal cell carcinoma and squamous cell carcinoma. Most cases are reported in patients from 15-45 years old. Your primary care provider should begin annual skin checks beginning in adolescence. Make sure to use sunscreen with an SPF of 30 or higher and reapply as necessary. Avoid unnecessary sun exposure and wear proper attire — hats, sleeves, lip balm — when going outside for long durations. If skin cancer is detected, treatment can proceed as with the general population.

### Hair

As mentioned in the name, people with cartilage-hair hypoplasia have distinct changes to their hair. It tends to be very fine or silky feeling and sparse, both on the scalp and the rest of the body. The hair tends to be more lightly colored compared to unaffected members of the family.

### Hirschsprung Disease

Megacolon, or Hirschsprung disease, is very uncommon (affecting about 5-10 percent of people with CHH) and occurs when the nerve cells in a baby's colon are absent. It usually affects the child during infancy and/or childhood and is very rarely diagnosed into adulthood. It can be treated as with others with Hirschsprung disease.

## **Resources**

### **Cartilage Hair Hypoplasia General Information**

Pauli, R.M., Legare, J. Cartilage Hair Hypoplasia Natural History. Rev. 2010.

<http://www.lpaonline.org/natural-histories-of-dwarfism-types>

### **Growth Charts**

[https://lpamrs.memberclicks.net/index.php?option=com\\_content&view=article&id=66](https://lpamrs.memberclicks.net/index.php?option=com_content&view=article&id=66)

**Little People of America**

<http://www.lpaonline.org/>

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