Cartilage-Hair Hypoplasia

Other names: McKusick Type Metaphyseal Chondrodysplasia, Dwarfism
Gene involved: *RMRP*

What is Cartilage-Hair Hypoplasia?

Cartilage-Hair hypoplasia (CHH) is an inherited condition that is the most common cause of short stature in the Amish community. People with CHH can have frequent and severe infections, low levels of red blood cells (anemia), increased risk of cancer, lung problems (bronchiectasis), and short brittle hair. Children are born with CHH and do not grow out of it.

What are the early signs and symptoms?

- Short stature is the hallmark of CHH. Patients with CHH can also have curving in their spine (scoliosis).
- Many children have bowing of their legs, sometimes this causes pain that is severe enough that children need surgery.
- Children usually have fine, sparse, light hair.
- Some children with CHH are identified on the newborn screen to have a poor immune system. Children with CHH have a higher risk of severe infection, with about 35-60% having recurrent infections.
- Anemia, or low red blood cells occurs in about 80% of patients with CHH. This can cause fatigue.
- Digestion problems; some newborns with CHH can have a bowel problem called Hirschsprung's, which causes severe constipation that might require surgery.

What Happens as Children Get Older?

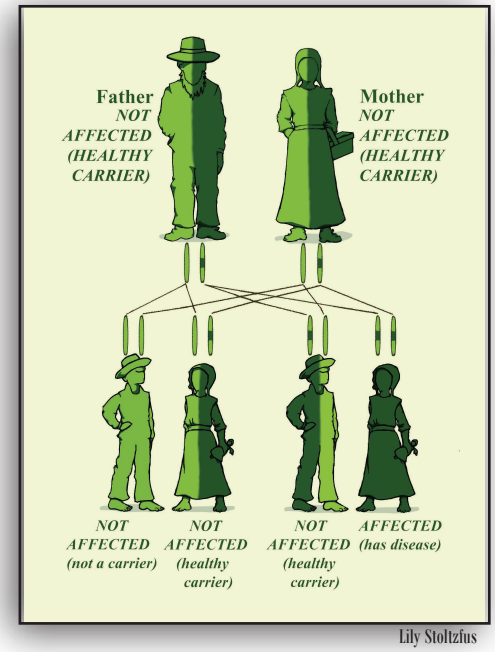
- Infections can continue to happen more frequently in adults with CHH, even if they did not have infections as children.
- About 30% of patients with CHH develop bronchiectasis, which is a lung disease that can cause problems with breathing and lead to recurrent lung infections.
- There is an increased rate of cancer in patients with CHH. Cancers tend to develop beginning in the teenage and young adult years.

Management

Although there currently is no cure for CHH, there are many ways to keep people with CHH healthy. It is important that children and adults have regular check-ups to monitor for infections, bowing in their legs, breathing problems, anemia, and cancer. It is important to get vaccines to help protect children from infections. To better understand a child's immune system, sometimes blood work is needed. Chil-dren may need to see a bone surgeon for the bowing in their legs or curve in their spine. Children with very poor immune systems may need a bone marrow transplant (BMT).

Diagnosis

A gene test is required to make the diagnosis. To arrange this, please see the contact information below.



Autosomal Recessive Inheritance

Genes are instructions for growth and development. We have two copies of each gene. One copy is inherited from our mother, and one copy is inherited from our father. Children with CHH have a spelling change in BOTH copies of their *RMRP* genes. This means they have no working copy of the *RMRP* instruction.

People with one normal copy of the *RMRP* gene and one misspelled copy are called "healthy carriers".

When two healthy carrier parents have a child, there is a:

- 1 in 4 chance the child will have CHH, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *RMRP* gene and be neither affected nor be a carrier

This informational pamphlet is a project of New Leaf Center - a member of the Plain Community Health Consortium (PCHC)

NEW LEAF CENTER
CLINIC FOR SPECIAL CHILDREN
WINDOWS OF HOPE PROJECT

16014 E. Chestnut St., PO Box 336
Mt. Eaton, OH 44659

330-359-9888
330-359-9890 (F)
info@newleafclinic.org
www.newleafclinic.org

Additional PCHC Members Include:

Center for Special Children (La Farge, WI) **608-625-4039**
Central Pennsylvania Clinic (Belleville, PA) **717-935-2065**
Clinic for Special Children (Strasburg, PA) **717-687-9407**
DDC Clinic (Middlefield, OH) **440-632-1668**
Midwest Genetics Network (Okemos, MI) **517-324-8300**
The Community Health Clinic (Topeka, IN) **260-593-0108**
UPMC Children's Hospital of Pittsburgh (Pittsburgh, PA) **412-692-5070**
WeCare Clinic - Medical Care for Special Needs (Pembroke, KY) **270-962-7383**