Cockayne Syndrome

Other names: CS **Gene involved:** *ERCC6 (CSB)*

What is Cockayne Syndrome?

Cockayne syndrome is an inherited autosomal recessive disorder. Children with Cockayne syndrome have abnormally small heads and do not gain weight or grow at the expected rate. Some signs of Cockayne syndrome may be apparent at birth and will continue to worsen over time. Children are born with Cockayne syndrome and do not grow out of it.

What causes Cockayne Syndrome?

Cockayne syndrome is caused by spelling changes in the ERCC6 gene. This gene helps with repair of damaged genetic instructions. Usually, our body can repair damage before it becomes problematic. However, in Cockayne syndrome, damage is not repaired as it should be. This causes poor growth, slow development, and eventual death. It is not completely understood how the spelling changes in the ERCC6 gene cause the various features of this condition.

What are the early signs and symptoms?

Early signs and symptoms that are present early in infancy include those mentioned earlier, as well as:

- Hearing loss
- Vision loss
- Severe tooth decay
- Bone abnormalities
- Fragile skin
- Hands and feet that are cold all the time
- Changes to the brain
- Increased sensitivity to sunlight
- Seizures

What Happens as Children Get Older?

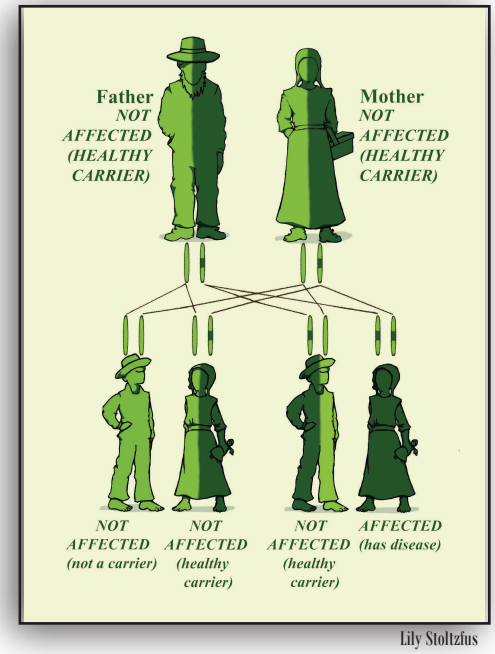
Children with Cockayne syndrome are special. They bring joy to their families and communities. They may not speak with words but enjoy the attention of everyone they meet. They have favorite toys and games. They need help with eating, diapering, and movement throughout their lives. As the disease progresses, joint stiffness may increase, and children do not move as easily. Feeding can become more difficult as well.

Management

While there is no cure for Cockayne syndrome, the various symptoms of the condition can be helped. Early physical and feeding therapy helps with developmental, movement, and feeding concerns. A feeding tube can be placed as needed. Regular dental care can help painful tooth problems. Limiting exposure to the sun protects the skin. Many families are glad for the chance to talk with other parents with similar children. Doctors need to support families in giving quality of life through all stages of Cockayne. Hospice care is available that allows children to be home at the end of life.

Diagnosis

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



Lily Stoltzfus

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 Cockayne Syndrome have a spelling change in BOTH copies of their *ERCC6* genes. This means they have no working copy of the *ERCC6* instruction.

People with one normal copy of the *ERCC6* 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 Cockayne Syndrome, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *ERCC6* 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)

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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**