CEP55 Disorder

Other names: Hydranencephaly and Cystic Kidney Disease **Gene involved:** *CEP55*

What is CEP55 Disorder?

CEP55 disorder is an inherited condition that causes severe problems with brain, kidney, and lung development.

What are the early signs and symptoms?

Signs of CEP55 disorder can be seen in a baby on ultrasound during pregnancy. The ultrasound might show a very small brain that is filled with fluid, kidneys that have pockets of fluid (cysts), poor growth, and little amniotic fluid.

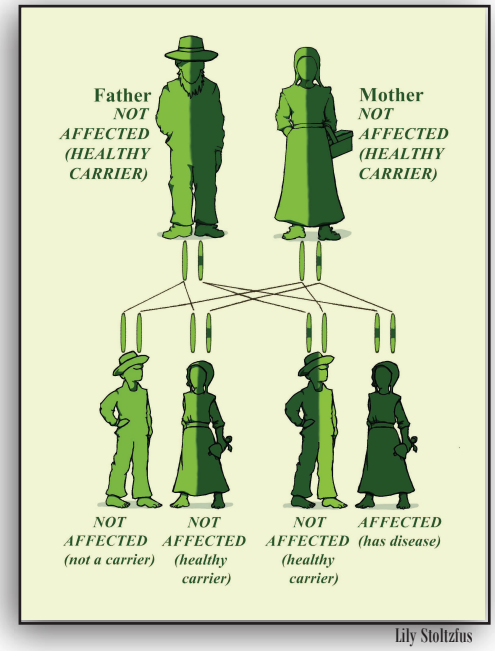
At birth babies often have a flattened nose, small chin, unique ears, and stiff joints in their arms and legs. Babies with CEP55 have lungs that cannot breathe well on their own and are too small. Unfortunately, babies with CEP55 die around the time of birth.

Management

There is no cure for CEP55 disorder. If a baby is suspected of having CEP55 we can work with the mid-wife or OB/GYN doctor to help them understand that we do not expect babies with CEP55 to live long past the time of birth. We can focus on keeping the baby comfortable and the family supported.

Diagnosis

A gene test is required to make the diagnosis. It is possible to test a pregnancy at 11 weeks to see if a baby is affected by the CEP55 disorder. At 16 weeks an ultrasound can be used to look for the signs of the CEP55 disorder.



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. eww with CEP55 disorder have a spelling change in BOTH copies of their *CEP55* genes. This means they have no working copy of the *CEP55* instruction.

People with one normal copy of the *CEP55* 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 CEP55 disorder, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *CEP55* 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**