Blue Eye Delay Syndrome

Other names: Mental retardation, autosomal recessive 38 **Gene involved:** *HERC2*

What is Blue Eye Delay Syndrome?

Blue Eye Delay Syndrome is an inherited condition affecting parts of the brain. Children with Blue Eye Delay syndrome have developmental delay and autism. Sometimes, families will have more than one child with Blue Eye Delay syndrome. Both boys and girls are affected equally. Children are born with Blue Eye Delay syndrome and do not grow out of it.

This condition was identified in 2012 in Anabaptist/Amish communities. Children around the world have condition very similar to Blue Eye Delay syndrome. Changes in brain function and behavior come from spelling changes in a genetic instruction called HERC2.

What are the early signs and symptoms?

- Children often have low muscle tone or "floppiness" from birth
- Some infants may have trouble feeding
- Many don't babble as babies. Speech is delayed
- Sitting, crawling, and walking are delayed
- New situations or crowds can be stressful
- Children may have seizures, especially with fever
- Many, but not all, have beautiful blue eyes

What Happens as Children Get Older?

- Children do best with special education (special ed) in school
- Behavior can be hyperactive
- Children need routines and support when over-stimulated
- The heart, lungs, liver, digestion and kidneys are not affected

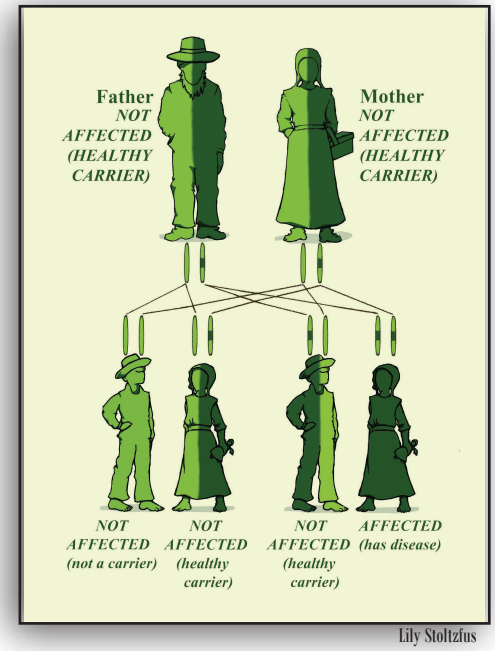
Management

Children with Blue Eye Delay bring good things to their families. Affected children benefit from early physical, speech, and occupational therapy. Eye checkups are recommended because vision problems are treatable. There is no cure for Blue Eye Delay but support from families, special education, and therapists improves communication skills and satisfaction with life.

Many children have difficulty concentrating and find it hard to complete tasks. Medications can be used for attention or behavioral problems if needed. Medications can also be helpful for sleep or seizure problems.

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 Blue Eye Delay Syndrome have a spelling change in BOTH copies of their *HERC2* genes. This means they have no working copy of the *HERC2* instruction.

People with one normal copy of the *HERC2* 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 Blue Eye Delay Syndrome, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *HERC2* 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**