Galloway-Mowat Syndrome

Other names: Nephrocerebellar syndrome, Yoder dystonia, Hershberger syndrome
Gene involved: *WDR73*

What is Galloway-Mowat Syndrome?

Galloway-Mowat syndrome is an inherited condition affecting the brain and kidneys. Sometimes, families will have more than one child with Galloway-Mowat syndrome. Children are born with Galloway-Mowat syndrome and do not grow out of it.

Children around the world have Galloway-Mowat syndrome, but the condition was described in Anabaptist/Amish communities in 2015. Brain function and kidney disease come from spelling changes in a genetic instruction called WDR73.

What are the early signs and symptoms?

- Vision problems or unusual eye movements. Infants do not focus on faces or toys by 6 weeks of age
- Floppy muscle tone and difficulty with head support
- Slow to gain new skills like reaching for toys, rolling over, or sitting up
- Feeding problems and slow weight gain
- Trouble falling asleep or staying asleep
- Fussiness

What Happens as Children Get Older?

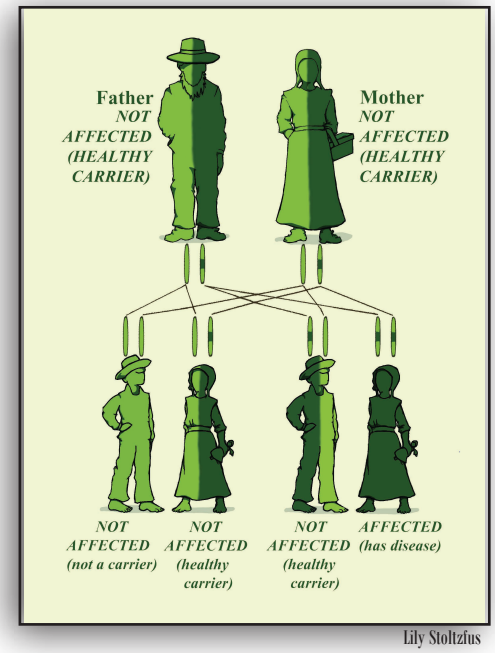
- As they grow, children can learn new skills, but will be delayed compared to their siblings. Some children can only roll over, some can sit, and a few have learned to walk.
- Very few children with Galloway-Mowat learn to speak, but all enjoy being with their families, and a few have learned to speak simple phrases.
- Kidneys fail over time. This is most easy to see when children are sick. Sickness can cause swelling of the eyelids, hands, and feet.
- Eyesight is poor. Most children respond to light and color
- Some children have seizures
- Lifespan is limited and most do not live beyond teenage years. Kidney failure or a lung infection is usually the cause of death.

Management

Although there is no cure for Galloway-Mowat syndrome, treatments are available to help children lead comfortable lives. Children with this condition benefit from early physical, occupational, and speech therapy to strengthen muscles and encourage hand use. Toys and brightly colored objects stimulate visual development. Children with feeding, sleep, or seizure problems can get help for these issues from a medical doctor. The main priority of care is the comfort of the affected child.

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 Galloway-Mowat syndrome have a spelling change in BOTH copies of their *WDR73* genes. This means they have no working copy of the *WDR73* instruction.

People with one normal copy of the *WDR73* 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 Galloway-Mowat syndrome, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *WDR73* 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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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**