Aicardi-Goutières Syndrome

Other names: SAMS Association **Gene involved:** *SAMHD1*

What is Aicardi-Goutières Syndrome?

Aicardi-Goutières syndrome (AGS) is a rare inherited condition that affects the brain, immune system, skin, joints, and blood vessels. The symptoms of AGS are caused by the body having too much inflammation. AGS varies widely in its initial symptoms and in how the disease progresses, even between members of the same family. Both boys and girls can be affected. Some children will have severe developmental problems whereas others may develop normally and only be diagnosed because of the skin or joint problems. Children are born with Aicardi-Goutières and they do not grow out of it.

What are the early signs and symptoms?

AGS can present immediately after birth with signs of brain damage that can look like a viral infection. It can also present in infancy after a period of apparently normal development. Children can have a small head, seizures, irritability, intellectual and physical disabilities. Infants can have low platelets and red blood cells (anemia). Feeding may be slow and difficult. Weight gain is often slow.

What Happens as Children Get Older?

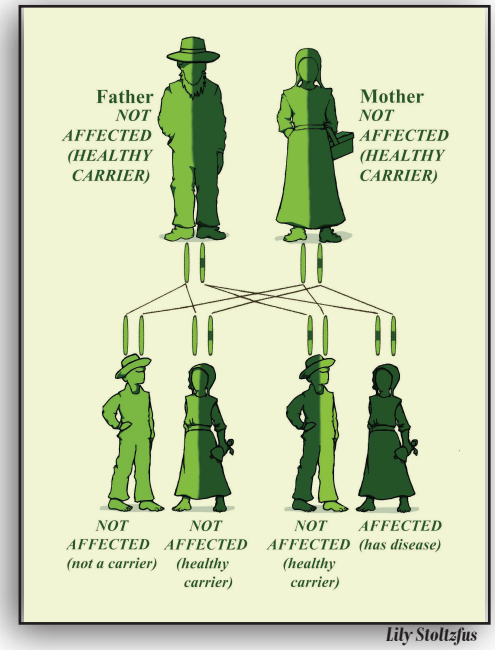
Most children, as they get older, will make limited developmental progress. They can continue to have difficulties with feeding and seizures. Poor circulation in the hands and feet are common. People with AGS can develop a painful, itchy rash on the fingers, toes, nose, and ears that worsens in cold weather. Additionally, they can have recurrent fevers, vision problems, joint and muscle stiffness. People with AGS are at an increased risk for strokes.

Management

There is currently no cure for AGS, but there are new treatments that doctors are learning about to help patients with AGS. These new medications work by decreasing inflammation which can be one of the main causes of brain damage in patients with AGS. Children often benefit with physical and occupational therapy. A feeding tube may be helpful for growth, especially in children who have difficulty swallowing. Medications can be used to treat seizures and help manage irritability.

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 AGS have a spelling change in BOTH copies of their *SAMHD1* genes. This means they have no working copy of the *SAMHD1* instruction.

People with one normal copy of the *SAMHD1* 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 AGS, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *SAMHD1* 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**