Spastic Ataxia with Optic Atrophy

Other names: SPAX4, Spastic ataxia 4, autosomal recessive **Gene involved:** *MTPAP*

What is Spastic Ataxia with Optic Atrophy?

Spastic ataxia with optic atrophy (SPAX4) is an inherited brain disease causing movement difficulties and problems with eyesight. Children are born with SPAX4 and do not grow out of it. Sometimes, a family can have more than one child with SPAX4.

What are the early signs and symptoms?

- Babies look like their siblings. The heart, lungs, liver, and kidneys are healthy.
- Infants may be unsteady when learning to sit or crawl
- Walking may be unsteady or delayed
- Children will be slower in learning to talk

What Happens as Children Get Older?

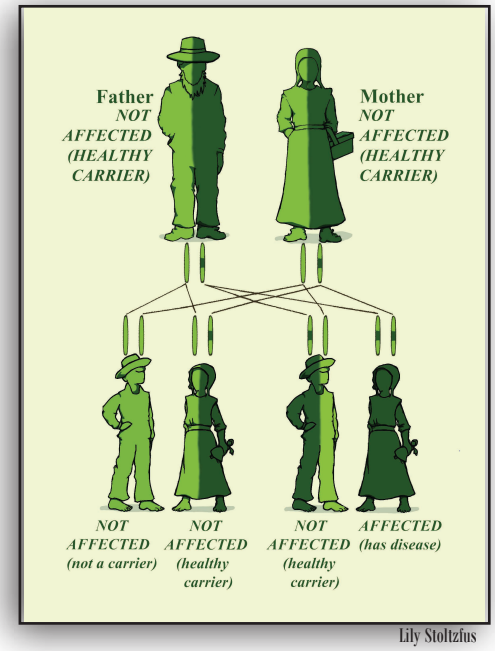
- Children need special support with learning in school
- In early adulthood, problems with speech, swallowing and movement may worsen slightly
- Eyesight may worsen over time

Management

- People with MTPAP bring joy to their families
- Infants and toddlers do best with early therapy. Physical therapy, along with braces and walkers, can help with walking. Speech therapy helps with learning to talk.
- School-aged children benefit from special education and continued speech and physical therapy
- Eyesight should be monitored each year
- Adults need assistance with self-care, but do not lose intelligence

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 SPAX4 have a spelling change in BOTH copies of their *MTPAP* genes. This means they have no working copy of the *MTPAP* instruction.

People with one normal copy of the *MTPAP* 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 SPAX4, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *MTPAP* 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**