MRT70

Other names: Intellectual Development Disorder, Autosomal Recessive 70
Gene involved: *RSRC1*

What is MRT70?

MRT70 is an inherited autosomal recessive disorder. Children with MRT70 are born with special needs. Children who are born with MRT70 will not grow out of it. Sometimes, families will have more than one special child with MRT70.

What causes MRT70?

MRT70 is caused by spelling changes in a genetic instruction called RSRC1. RSRC1 works with other genetic instructions to help the brain develop properly. MRT70 affects children around the world.

What are the early signs and symptoms?

- Children with MRT70 have low muscle tone from birth. Parents describe them as “floppy” or “double-jointed”
- Developmental delay (slower to sit, crawl, talk and walk)
- Slower feeding

What Happens as Children Get Older?

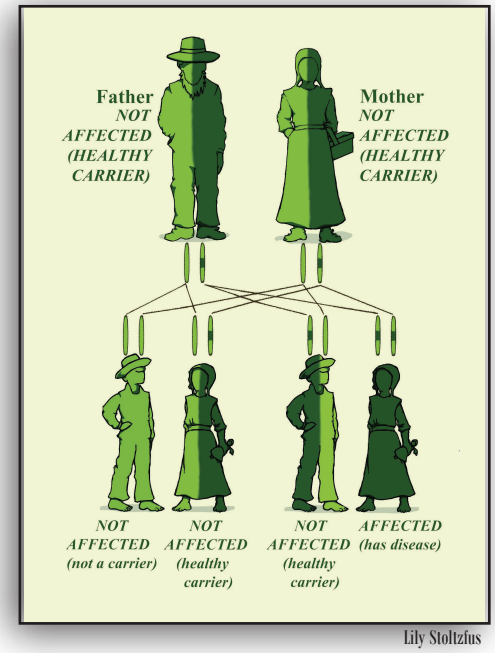
- Some children can be hyperactive
- They may be slower to communicate and need more support in relating to others
- Some children can have seizures
- Many children are less coordinated with walking

Management

Children with MRT70 bring joy to their families. They continue to learn as they get older. They will always require special support and therapy to reach their full potential. There is no cure for MRT70, but support from families, special education, and therapists improves communication skills and the quality of life. Medications can be used for seizures or behavioral problems if needed.

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 MRT70 have a spelling change in BOTH copies of their *RSRC1* genes. This means they have no working copy of the *RSRC1* instruction.

People with one normal copy of the *RSRC1* 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 MRT70, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *RSRC1* 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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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**