MOPD1

Other names: Microcephalic osteodysplastic primordial dwarfism type 1n
Gene involved: *RNU4ATAC*

What is MOPD1?

MOPD1 is an inherited condition caused by a change in the RNU4ATAC gene (instruction) which is important for growth and brain development.

What are the early signs and symptoms?

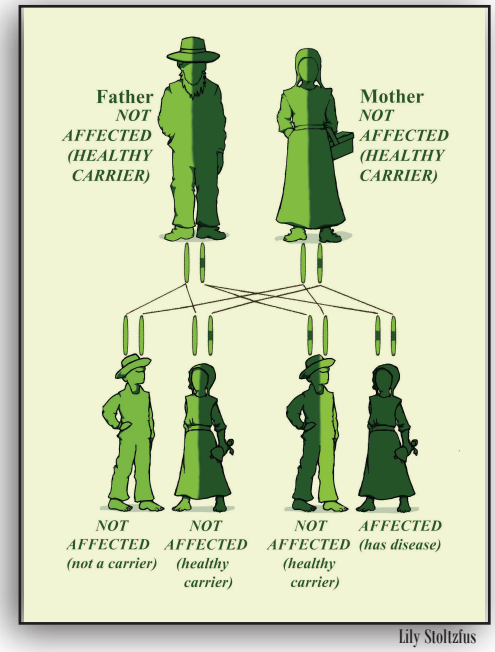
- During pregnancy, babies with MOPD1 grow very slowly and may have an ultrasound because the mother is measuring small for her expected due date. An ultrasound can show a small head, limited growth, and too little fluid.
- Babies are born much smaller than their healthy siblings. They have small heads, thin hair, a unique facial appearance, and bones that do not grow normally.
- Many babies require special support for feeding
- They can have episodes where their breathing stops (apnea)
- Seizures are possible
- There are developmental delays - they can smile and coo, but do not learn to sit or use words
- They usually do not survive longer than their first two years of life

Management

Babies with MOPD1 bring special meaning to their families. There is no cure for MOPD1. Management focuses on the symptoms of MOPD1. Families are encouraged to enroll in hospice care and to focus on keeping their child comfortable. There are prescription medications for seizures and pain that can help babies be comfortable.

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 MOPD1 have a spelling change in BOTH copies of their *RNU4ATAC* genes. This means they have no working copy of the *RNU4ATAC* instruction.

People with one normal copy of the *RNU4ATAC* 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 MOPD1, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *RNU4ATAC* 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**