PMRED

Other names: SNIP1, Psychomotor Retardation, Epilepsy, and Craniofacial Dysmorphism
Gene involved: *SNIP1*

What is PMRED?

PMRED is an inherited disorder caused by changes in the SNIP1 gene (instruction) which is important for brain development. Children with PMRED have developmental delays (sitting, talking, and learning), seizures, and a unique skull shape. These problems can start shortly after birth. Children are born with PMRED and do not grow out of it.

What are the early signs and symptoms?

- Babies with PMRED can have seizures, pauses in breathing (apnea)
- Feeding difficulties
- Some children have heart defects that require surgery within the first year of life
- Babies are often floppy (hypotonia), and struggle to meet their early milestones
- They can have low blood sugar and low thyroid

What Happens as Children Get Older?

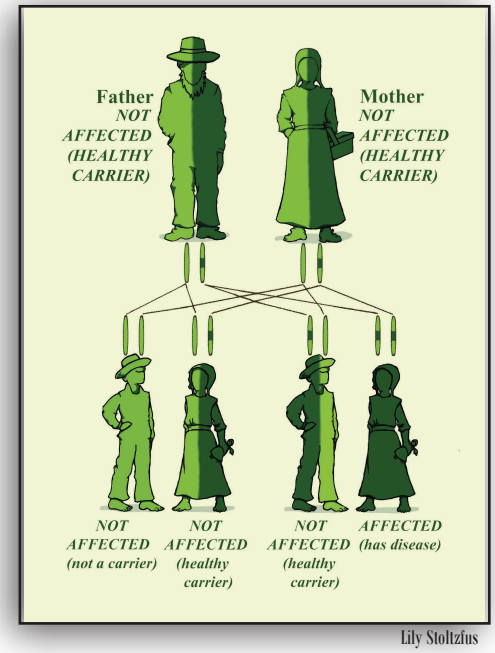
- Children with PMRED will continue to have developmental delays and need support from their families
- Some children can walk by mid childhood, and learn basic communication skills (pointing, signing)
- Can have frequent infections
- Some might have difficulty with feeding
- Seizures can be difficult to control and may require several medications

Management

Currently, there is no cure for PMRED. Management includes treating the symptoms and complications of PMRED such as respiratory distress, feeding difficulties, low blood sugar, low thyroid, and seizures. A feeding tube (g-tube) may be recommended. Medications for seizures can reduce the risk of hard seizures which can cause loss of milestones.

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 PMRED have a spelling change in BOTH copies of their *SNIP1* genes. This means they have no working copy of the *SNIP1* instruction.

People with one normal copy of the *SNIP1* 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 PMRED, a
- 1 in 2 chance they will be a healthy carrier
- 1 in 4 chance they will inherit two normal copies of the *SNIP1* 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**