



## **NEWBORN SCREENING FACT SHEET**

### **5-Oxoprolinuria (Glutathione Synthetase Deficiency)**

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#### **What is it?**

5-oxoprolinuria, or pyroglutamic aciduria, is an inherited disorder of amino acid metabolism.

#### **What causes it?**

This is caused by an enzymatic defect in glutathione co-enzyme production.

5-oxoprolin, a by-product of glutathione deficiency, accumulates in blood and cerebrospinal fluid, is excreted in urine and is the compound detected by newborn screening.

#### **If 5-oxoprolinuria is not treated, what problems occur?**

Early treatment may prevent:

- 1) Brain damage.
- 2) Seizures.
- 3) Mental retardation.
- 4) Autistic-like disorders.
- 5) Coma, sometimes leading to death.

#### **What is the treatment for 5-oxoprolinuria?**

Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child.

Early supplements with vitamins C and E may improve long-term outcomes for your baby.

#### **Call your doctor at the start of any illness.**

In some children, even minor illnesses such as a cold or the flu can lead to a metabolic crisis. Some of the first symptoms of a metabolic crisis are:

- 1) Poor appetite.
- 2) Extreme sleepiness or lack of energy.
- 3) Behavior changes.
- 4) Irritable mood.
- 5) Muscle weakness.

- 6) Nausea.
- 7) Vomiting.

#### **Is 5-oxoprolinuria inherited?**

5-oxoprolinuria is inherited. It affects both boys and girls equally.

The parents of a child with this condition are unaffected healthy carriers of the condition and have one normal gene and one abnormal gene.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have glutathione synthetase deficiency. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

#### **How many people have 5-oxoprolinuria?**

5-oxoprolinuria deficiency is an extremely rare condition and the actual number of people who have it is unknown.

#### **Does 5-oxoprolinuria go by any other names?**

5-oxoprolinuria is sometimes called:

- 1) Glutathione synthetase deficiency.
- 2) Pyroglutamic academia.
- 3) 5-oxoprolinemia.
- 4) Pyroglutamicaciduria.
- 5) Pyroglutamic aciduria.

#### **Where can I find more information?**

Organic Acidemia Association

[www.oaanews.org](http://www.oaanews.org)

Save Babies Through Screening Foundation

[www.savebabies.org](http://www.savebabies.org)

Children Living with Inherited Metabolic Diseases (CLIMB)  
[www.climb.org.uk](http://www.climb.org.uk)

Genetic Alliance  
[www.geneticalliance.org](http://www.geneticalliance.org)

### Children's Special Health Services (CSHS)

State Capitol Judicial Wing  
600 E. Boulevard Ave., Department 301  
Bismarck, ND 58505-0269  
Toll Free: 800.755.2714  
701.328.2436  
Relay TDD: 701.328.3975  
CSHS website: [www.ndhealth.gov/CSHS](http://www.ndhealth.gov/CSHS)  
North Dakota Department of Health website:  
[www.ndhealth.gov](http://www.ndhealth.gov)

### Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

### *Family Resources*

Family to Family Network  
Center for Rural Health  
University of North Dakota  
School of Medicine and Health Sciences  
P.O. Box 9037  
Grand Forks, ND 58202-9037  
Toll Free: 888.434.7436  
701.777.2359  
Fax: 701.777.2353  
E-mail: [NDF2F@medicine.nodak.edu](mailto:NDF2F@medicine.nodak.edu)  
[www.medicine.nodak.edu/crh](http://www.medicine.nodak.edu/crh)

Pathfinder Services of ND  
Pathfinder Family Center  
1600 2<sup>nd</sup> Ave. SW, Ste. 19  
Minot, ND 58701  
Toll Free: 800.245.5840  
701.837.7500  
Relay TDD: 701.837.7501  
E-mail: [ndpath01@ndak.net](mailto:ndpath01@ndak.net)  
[www.pathfinder.minot.com](http://www.pathfinder.minot.com)

Family Voices of North Dakota, Inc.  
P.O. Box 163  
Edgeley, ND 58433  
Toll Free: 888.522.9654  
701.493.2634  
Fax: 701.493.2635  
[www.geocities.com/ndfv](http://www.geocities.com/ndfv)

\*\*This fact sheet has general information. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

### **Acknowledgement**

The North Dakota Department of Health Newborn Screening Program thanks Star-G Screening, Technology and Research in Genetics for allowing us to utilize its material.

### **Disclaimer**

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