

## Meeting Your Needs

People with Noonan syndrome with multiple lentigines and their families have different needs. Our team connects you with providers from a variety of specialty areas. We use a multi-disciplinary approach to meet your needs from head to toe.

### Specialists you may see include:

#### Neurology

Brain, spinal cord & nerves

#### Audiology

Hearing

#### Cardiology

Heart & blood vessels

#### Gastroenterology

Digestive system

#### Physical Therapy

Specialized movement exercises

#### Occupational Therapy

Activities for daily life

#### Genetics

Genes & inheritance

#### Social Work

Support & overall well-being

All,  
for  
your  
one.<sup>SM</sup>



### Resources:

#### RASopathies Network

[rasopathiesnet.org](http://rasopathiesnet.org)

#### Noonan Syndrome Foundation

[teamnoonan.org](http://teamnoonan.org)

#### National Organization for Rare Disorders (NORD)

[rarediseases.org](http://rarediseases.org)

#### Clinical Trials Database

[clinicaltrials.gov](http://clinicaltrials.gov)

### Contact Us:

RASopathy Clinic Appointment

**312.227.6120**

Clinic Coordinator

**312.227.4391**

#### Ann & Robert H. Lurie

#### Children's Hospital of Chicago

225 East Chicago Avenue

Chicago, Illinois 60611

[luriechildrens.org/en/specialties-conditions/rasopathy-program/](http://luriechildrens.org/en/specialties-conditions/rasopathy-program/)

## Noonan Syndrome with Multiple Lentigines

RASopathy Program



# What Is Noonan Syndrome with Multiple Lentiginos?

Noonan syndrome with multiple lentiginos (NSML) is a genetic condition that is part of a larger group of conditions called *RASopathies*. *RASopathies* arise when something goes wrong in a pathway involved in the way the cells of our body communicate the information they need to function properly. Since cells make up our tissues, organs and organ systems, *RASopathies* like NSML can have widespread effects on growth, development and health.



## What Causes NSML?

Genetic changes (sometimes called mutations) in one of the genes listed below cause most cases of NSML:

- *PTPN11* (about 85% cases)
- *RAF1* (about 5 to 10% cases)
- *BRAF* or *MAP2K1* (rarely)

In about 1 to 5% of cases, a specific genetic change is not identified because we are still learning new information about NSML.

## Signs & Symptoms of NSML

**Heart** may have changes in the structure and/or function, such as:

- Narrowing of valve between heart & lungs (Pulmonary valve stenosis)
- Thickened heart muscle (Hypertrophic cardiomyopathy)
- Irregular heartbeat (Arrhythmia)

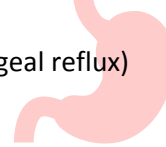


**Unique features** may include:

- Increased distance between the eyes (Orbital hypertelorism)
- Hearing loss
- Dark macules, like birthmarks, appear on the skin over time (Lentiginos)

**Digestive system** issues, including:

- Feeding problems
- Heartburn (Gastroesophageal reflux)
- Bloating and constipation



**Growth and development**

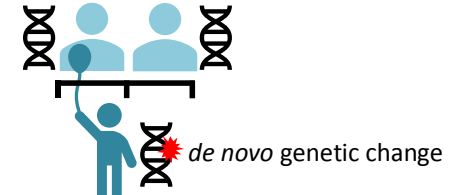
- Delays in meeting milestones such as walking and talking
- Mild learning difficulties
- Slow growth and short height
- Breastbone sunken into the chest (Pectus deformity)



## How Is NSML Inherited?

A gene is a long string of DNA instructions for making proteins that living things need to grow and function. Genes come in pairs. We inherit one copy of a gene from our mother and one from our father.

Since NSML is a genetic disorder, it can be passed down from parent to child. However, in some cases the genetic change that causes NSML is brand new (*de novo*) and occurs for the first time in the child affected by the condition.



## Genetic Counseling

NSML is a very rare condition. It is *autosomal dominant* meaning a change in one copy of a gene pair is enough to cause the condition.

When a genetic change is *de novo*, there is less than a 1% chance for the same biological parents to have more than one affected child.

## Treatment & Management

There is no single or universal treatment for NSML syndrome. Treatment plans are created by a healthcare team with input from the patient and their family. Plans are based on the patient's concerns and accepted guidelines.