UNDERSTANDING NOONAN SYNDROME

A MULTI-SYSTEM GENETIC CONDITION

Noonan syndrome (NS) is a genetic condition that affects multiple systems of the body and may significantly impact health and lifespan. It occurs in approximately 1 in 1,000-2,500 births and is caused by gene variants affecting the RAS-MAPK pathway, which regulates cell growth and development.

Clinical presentation is highly variable, even among members of the same family. Individuals with NS often require care from multiple specialists throughout their lives.

Early diagnosis is important to help identify potential health concerns, guide appropriate monitoring, and connect individuals and families with specialized care and support.

KEY MEDICAL CONSIDERATIONS:

CARDIOVASCULAR

- Present in over 80% of patients
- Pulmonary valve stenosis
- Hypertrophic cardiomyopathy
- Other congenital heart defects

ENDOCRINE

- Short stature
- Delayed puberty
- Thyroid dysfunction

NEURODEVELOPMENT

- Developmental delays
- Learning disabilities
- ADHD and autism spectrum differences

MUSCULOSKELETAL

- Hypotonia (low muscle tone)
- Joint hypermobility
- Chronic pain
- Scoliosis
- Chest wall deformities (pectus excavatum or carinatum)

HEMATOLOGY

- Easy bruising or excessive bleeding
- Transient myeloproliferative disorder
- Increased risk of leukemia (including JMML)

GASTROINTESTINAL

- Feeding difficulties
- Failure-to-thrive
- Gastroparesis
- Feeding tube dependence

ADDITIONAL CONSIDERATIONS

- Hearing loss
- Sleep disorders including obstructiv
- Feeding tube dependence
- Vision problems (ptosis, strabismus, refractive errors)
- Lymphedema
- Gastroparesis
- Sleep apnea
- Chiari Malformation

IMPORTANT FACTS FOR HEALTHCARE PROVIDERS:

- Noonan syndrome is part of a group of conditions known as RASopathies.
- 80-85% of cases can be diagnosed with genetic testing.
- Approximately 50% of cases involve variants in the *PTPN11* gene.
- Diagnosis can be confirmed through genetic testing, including RASopathy panels, exome, or genome sequencing.
- The condition may be inherited or occur de novo (spontaneous genetic change).
- Individuals with NS often have medical concerns affecting multiple organ systems and often require multidisciplinary care.
- Families frequently become highly knowledgeable partners in care due to the complexity and rarity of the condition.

ABOUT THE NOONAN SYNDROME FOUNDATION:

Our mission is to support and empower individuals and families affected by Noonan and Noonan-like syndromes through education, advocacy, and facilitating research to advance knowledge and care. We are a 501(c)(3) non-profit charitable organization.

LEARN MORE

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