

Noonan Syndrome (NS) *For Healthcare Providers*

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Noonan syndrome will have all of these features.

- Congenital heart defect
- Short stature
- Developmental delay (variable)
- Characteristic facial features
- Coagulation defects
- Dental Issues
- Ocular abnormalities
- Renal abnormalities
- Physical Differences
 - Broad neck or webbed neck
 - Unusual shape to the chest with pectus carniatum and pectus excavatum
 - Spinal abnormalities
 - Clubfoot

Recommended Routine Surveillance

- Monitor anomalies found in any syndromes; especially cardiovascular abnormalities
- Ophthalmologic evaluation as needed
- Routine dental care

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
 - Monitor for cardiac abnormalities
- Dermatologist
 - Keratosis Pilaris
 - Ulorythema
- Developmental evaluation:
 - Speech therapy
 - Physical therapy
 - Occupational therapy
- Endocrinologist
 - Growth monitoring
 - Delayed puberty
- Geneticist / Genetic Counselor:
 - Diagnosis
 - Coordination of care
 - Genetic risk for family
 - Clinical trials
- Genitourinary
 - Renal abnormalities
- Hematologist/Oncologist
 - Treat specific coagulation defects
 - Treat specific childhood cancers
 - Varied lymphatic abnormalities
- Ophthalmology surveillance:
 - Strabismus
 - Refractive errors
 - Amblyopia
 - Nystagmus
- Neurologist
 - If Arnold-Chiari malformation present

Sample Forms

- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient _____ has been diagnosed with Noonan syndrome (NS). NS is characterized by characteristic facies, congenital heart defects, short stature, and

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developmental delays. Medical complications with NS syndrome include management of heart and renal, and coagulation defects. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

“[Noonan Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



Resources

Gene Reviews Noonan syndrome

<https://www.ncbi.nlm.nih.gov/books/NBK1124/>

Management of Noonan Syndrome Clinical Guidelines

These clinical guidelines were developed by the “Dyscerne” Noonan Syndrome Guideline Development Group, and are provided here with permission of the key author, Bronwyn Kerr (UK).

http://www.wessland.com/Dyscerne_Noonan_Syndrome.pdf

Noonan Syndrome: Clinical Features, Diagnosis, and Management Guidelines

<http://pediatrics.aappublications.org/content/126/4/746.full.pdf%20html>