

## Noonan Syndrome *At a Glance*

Noonan syndrome (NS) may include findings of short stature, heart defects, distinctive facial features, and developmental delays. Other findings may include differences in clotting ability, chest shape, lymph system, and in the eye, etc. Up to 1/3 of individuals with NS have a mild intellectual disability.

This condition is variable even within a family. This can range from subtle physical findings to more extensive changes.

NS is an inherited genetic disorder or can be caused by a spontaneous gene mutation. About 1 in every 1,000 to 2,500 babies is born with NS.

***Learn more about characteristics of NS (not all people with NS will have all of these features):***

### **Congenital heart defect (50%-80%)**

- May have had surgery and/or be on medications
- Most common deficit is pulmonary valve stenosis
- Hypertrophic cardiomyopathy occurs in 20%
- Lifetime cardiac follow-up is important

### **Short stature (50-70%)**

- Final adult height approaches lower limit of normal

### **Developmental delay (variable)**

- Increase in learning disabilities
- Increase in brain differences
  - Wide array of neurologic problems
  - Seizures possible
- Speech articulation difficulties in 72%

### **Physical Differences**

- Broad neck or extra tissue at back of neck
- Unusual shape to the chest
  - Prominence at the top of the breast bone and sunken area at the bottom
  - Nipples wide spaced and low set

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- Rounded shoulders
- Spinal abnormalities
  - Scoliosis (10-15%)
  - Less common spinal differences such as kyphoscoliosis (hunchback), spina bifida, and changes in vertebrae and ribs
- Clubfoot 10-15%
- Ability to hyperextend at the elbow
  - Abnormal forearm angles found in more than >1/2 males and females.
  - May have leg pain

### Facial Features

- Low set ears
- Pale blue or blue green irises
  - Iris is typically lighter in color
- Wide spaced eyes with epicanthal folds (extra fold at the inner part of the eye lid) and thick or droopy eyelids
- Lack of affect or expression

### Skin Differences

- Problems affecting color and texture of the skin
  - Lotions or short courses of topical steroids help dry skin
  - Avoid long hot baths, perfumed soaps, and dry atmosphere
- May have curly, coarse, or sparse hair

### Coagulation (blood clotting) problems

- Specific testing will identify the problem and treatment if necessary.
- May include bruising or nose bleeds, bleeding with surgery
- Avoid aspirin unless documented that they don't have coagulation defects

### Dental Issues

- Poor bite in 50-66%

### Lymphatic dysplasia (difficulty with the lymph system which drains excess fluid from the body and helps fight infection)

- May be in one area or widespread,
- May be before or after birth
- Overall incidence in all age groups is 20%

- Lymphedema in the arms or legs is the most common
  - Typically resolves in first few years of life.
  - Adolescents and adults can also develop peripheral lymphedema

### Ocular (eyes) (95%)

- Strabismus/crossed eyes
- Refractive errors
- Amblyopia
- Nystagmus (rhythmic movement of the eyes from side to side)

### Renal (kidney) abnormalities

- Present in about 11% and are generally minor
- Kidney swelling is most common

### Puberty may be delayed

- Male puberty and fertility may be normal, delayed (about age 13.5-14.5), or inadequate
- Female puberty may be delayed
  - Mean age of first period is 13-14 years
  - Normal fertility typically

## ***Things to Think About***

### **1. Medical/Dietary Needs**

#### **What you need to know**

The list of possible medical problems in NS may be quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely between individuals. Therefore, it is important to ask the parents about the medical issues in their child.

- School age children with NS may have frequent doctor and specialist visits to monitor medical conditions.
- Do not give aspirin as it may cause bleeding problems in children who have the coagulation problems.
- It is important to let parents know if the child has headaches or any other neurological symptom, as this may be indicative of an underlying cause.

- There may be issues affecting attendance due to cardiac involvement, low tone, or leg pain.
- No special diet is required for NS, although a well-balanced diet is important.
- Be aware, or ask parents, if the child wears a medical alert bracelet.

Recommended screening:

- Annual physical
- Annual eye exam
- Regular developmental assessment of children
- Cardiovascular monitoring in some

For additional information regarding management please see these guidelines:

- “Noonan Syndrome: Clinical Features, Diagnosis, and Management Guidelines” from the American Academy of Pediatrics (*Pediatrics*, Sept. 2010)  
<http://pediatrics.aappublications.org/content/126/4/746.full.pdf>
- “Management of Noonan Syndrome, a Clinical Guideline” from Dyscerne and other partners (with permission of the author, Bronwyn Kerr)  
[http://www.wessland.com/Dyscerne\\_Noonan\\_Syndrome.pdf](http://www.wessland.com/Dyscerne_Noonan_Syndrome.pdf)

## What you can do

- A yearly check-up and studies as needed should occur in the child’s Medical Home (their primary care office).
  - Discuss pain management plan with parents
    - Discuss the use of anti-inflammatories as appropriate
    - Ensure proper storage of ice/gel packs at school
    - Avoid aspirin
  - Individuals may complain of frequent stomach-aches due to reflux, delayed emptying, irritable bowel, etc. A plan should be in place for dealing with these symptoms at home and at school.
  - Important to address injuries quickly
- Notify the parents of any pain.
- Be aware of unusual changes that seem out of line with the situation, and notify parents.
  - Energy level
  - Behavior
  - Mood
  - Academic performance
  - Social

## 2. Education Supports

**It is important to have HIGH LEARNING EXPECTATIONS for children who have Noonan syndrome.** Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

### What you need to know

- Most children with NS have normal intelligence, but 10-40% require special education supports. Even in individuals with NS who have normal intelligence, their IQ is about 10 points lower than unaffected family members.
- Most children will perform well in typical education settings. It is important to identify their strengths and challenges.
- In adolescence, consider vocational needs. This can include: assessing cognitive strengths and weakness, teaching adaptive behavior, and teaching daily living skills as needed.

### Motor development

Motor development may be delayed. This is due to a combination of overly flexible joints and low muscle tone.

- May have a higher rate of clumsiness and coordination
- Physical and occupational therapy may help gross motor and fine motor skills
- Motor issues may lead to poor sitting posture and difficulties with balance
- Short stature may affect positioning
- Activities that require good hand eye control (i.e. writing, drawing and painting) may present difficulties
- Pencil and tool skills may be weak due to poor motor control and difficulty with coordination

Some people with Noonan Syndrome have hyper-elastic skin, joint hypermobility, pain and/or fatigue issues. Tips that have been developed for those with Ehlers-Danlos Syndrome may be useful.

### Verbal / Language Challenges

- Language difficulties in children are common and can be the root of future difficulties in literacy skills, including reading, writing, and spelling.
- Individuals with NS may have signs of developmental disorders such as dyspraxia, ADHD, autism spectrum disorder (ASD).

- Cognitive difficulties including, executive functioning, concentration, attention, impulsivity, short-term memory, receptive language, repetitive behaviors, dislike of change, and visual/spatial abilities may be evident.
- Verbal performance is typically lower than nonverbal performance.
  - May have difficulty with higher order language such as reasoning, problem solving, understanding humor, and perceiving the rhythms and subtle contours of verbal speech.
  - Articulation problems are common. However, most (72%) respond well to speech therapy.
  - Language delay may be related to hearing loss, perceptual motor, or articulation deficiencies.
- If the child with Noonan syndrome experiences dyslexia, the following link may be helpful. Possible signs of dyslexia include difficulty:
  - Moving to the rhythm of music
  - Remembering content of stories
  - Understanding left to right
  - Trouble with visual/spatial concepts
  - Coordinating movement (skipping is hard)
  - Choosing correct words or reversing words
  - Writing – may use reversed letters, words, and/or numbers
  - Proofing their own written work
  - Understanding time
  - Understanding seasons

### Learning, attention, and memory problems

- Problems with sustaining attention, switching tasks
- Abstract language and concepts can be difficult to grasp
- Metaphors may be rarely used or understood
- Instructions may be heard but not retained long enough for action
- Difficulty with integration skills, working memory, and episodic memory

### Executive Function Challenges

- Executive function issue affect planning, thinking flexibly, and understanding the abstract.
- May struggle to remember, process, and organize information efficiently.
  - This can lead to problems in math and reading.
- Executive function is based on a group of related cognitive and behavioral skills. They are responsible for goal directed activity including:
  - Attention
  - Short term memory

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- Planning and organization
- Behavioral inhibition
- Social interactions

## What you can do

### Interventions for visual-spatial and visual-motor development

- Ensure appropriate seating and positioning
- Make sure desk fits
- Storage and lockers are right size and height
- Occupational therapy may be useful; OT may be needed to make changes due to short stature
- Physical therapy and/or education may help motor skill development
- Modify copying, for example, provide a copy of teacher's or other student's notes
- Provide simple overview or summary before lesson
- Provide clear tests that are as simple as possible with only a few problems on a page. Graph paper may help especially in math problems
- Use lined paper to help place written responses on the sheet
- Allow extra time on work, and limit written homework
- Practice tracing shapes and copying pictures
- Provide feedback as they may not be aware of mistakes
- Use verbal descriptions to reinforce visual information
- May have difficulty matching shapes and sizes
  - Puzzles may be challenging

### Interventions for fine motor development

To help with pencil and implement skills due to poor motor control:

- Use thicker pencils or pencil grips
- Sloping surface may help (easels)
- Use fine motor activities (Legos, play dough)
- Practice folding
- Practice cutting with scissors
- Write on every other line
- Allow tracing

### Strategies to help with Language

- Take learning style into account
- Many strategies that are used with dyslexia can be effective
- May appear to understand but have low comprehension
- Speech therapy for speech and articulation issues

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## Strategies to Help with ADHD

- Seat with limited distractions
- Post schedule and assignments in easy viewing
- Review schedule visually and verbally
- Give reminders of schedule during the day
- Review homework
- Prepare for transitions
  - Provide alerts 15, 10, and 5 minutes before transitions
- Consider visual and verbal reminders and instruction
- Help with daily organization
- Pair student up with well-organized peer
- Break instructions into steps
- Use priority lists for large assignments
- Allow extra time for tests
- Allow use of fidget toy
- Use communication book between teachers and parents
- Develop rules and routines
  - Can be a problem with expressive language delay

## Interventions for communication challenges

- Individuals with speech and motor difficulties often benefit from speech and occupational therapy.
- Promote language understanding by using simple short sentences, visual prompts, and pictures.
- Use a child's experiences and interests to engage in learning.
- Allow extra time
- Repeat directions
- Provide lesson summaries
- Record lesson so child can listen again
- Promote language development
  - Give ample time for responding
  - Boost self-confidence by calling on them when they know answer
  - Encourage child to repeat the questions before responding
  - Allow time to rehearse and respond

## Interventions for attention and memory

- Help with organization
- Present information concretely
- Use manipulative materials to demonstrate concepts
- Simplify verbal and explain concepts clearly

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- Provide visual cues and instructions
- Repeat information and use positive reinforcement.
- Ask child to repeat instructions.
- Help the child find a starting point especially on complex tasks.
  - They often have a hard time with multiple step tasks. They may lose track of what they are doing.
- Select relevant task goals.
- Use a calendar to track important events.
- Organize a means to solve complex problems.
- Monitor and evaluate behavior and emotions.
- Help organize everyday needs at school and at home.
  - Have a place for all things
  - Use different colored notebooks for different subjects

### Strategies for Dyslexia

- Early identification and intervention
- RTI (response to intervention) program
  - <http://www.rti4success.org>

## 3. Behavioral and Sensory Supports

### What you need to know

Individuals with NS may have increased anxiety, panic attacks, social introversion, poor self-awareness, and difficulties identifying and expressing feeling and emotions. They may experience teasing over physical appearance and have poor self-esteem.

Research suggests that adults with NS may have mild problems in

- Attention
- Organizational skills
- Psychosocial maturity
- Difficulty expressing emotions and feelings

Adults with NS may also have body image problems with poor self-esteem, depression, and social inadequacy. It is important to note that the majority of adults with NS will finish high school and have paying jobs, and many will attend college.

### What you can do

- Early identification and behavioral interventions can be helpful for anxiety and self-esteem issues.
- Provide a peer buddy or social skills group.
- Unstructured situations may be difficult for individuals with mood and anxiety disorder (e.g., school assemblies, in-between classes, on bus, and on field trips).
  - Watch for overload
  - Prepare them ahead of time
  - Offer supports
  - Redirect behaviors

## 4. Physical Activity, Trips & Events

### What you need to know

No special accommodations are needed for individuals with NS syndrome, unless required due to the child's learning, medical, sensory and/or behavioral difficulties.

- Individuals may need extra preparation about what to expect during the activity.

Individuals who have NS are not usually limited in their activities. The only exception is for those children with a specific complication.

- A medical doctor will point out if there are any restrictions (i.e., heart, bleeding, etc.).

If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming ([www.nepassage.org](http://www.nepassage.org)).

## 5. School Absences and Fatigue

### What you need to know

- School age children who have NS may have increased or frequent absences due to medical issues.
- Individuals with NS may have fatigue which may or may not be related to cardiac involvement. Leg pain may also occur.

### What you can do

- Contact parents if changes are noticed.

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- Provide assignments and notes

## 6. Emergency Planning

### What you need to know

- Emergency plans will be very individually determined, based on child's behaviors and medical issues.

### What you can do

- It is important to mention any new signs or symptoms and/or pain to the child's parents.

## 7. Resources

### Noonan Syndrome Foundation

The Noonan Syndrome Foundation, a branch of the RASopathies Foundation, offers help to get answers, find support, and stay connected. NS: "the most common syndrome you've never heard of."

<http://www.teamnoonan.org/>

### 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).

*"The guidelines aim to provide clear, and whenever possible, evidenced-based recommendations for the management of patients with Noonan syndrome."*

[http://www.wessland.com/Dyscerne\\_Noonan\\_Syndrome.pdf](http://www.wessland.com/Dyscerne_Noonan_Syndrome.pdf)

### Noonan Syndrome Association (UK)

The Noonan Syndrome Association, in the United Kingdom, provides comprehensive support through research and raising awareness. See their document *Education in Noonan Syndrome*

<http://www.noonansyndrome.org.uk/>

[http://www.noonansyndrome.co.uk/docs/pdf\\_docs/Education\\_in\\_NS\\_2004\\_Website.pdf](http://www.noonansyndrome.co.uk/docs/pdf_docs/Education_in_NS_2004_Website.pdf)

### GeneReviews

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Learn more about the genetics of Noonan syndrome.

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

### **PubMed Health**

Learn more about Noonan Syndrome from the U.S. National Library of Medicine.

<http://www.ncbi.nlm.nih.gov/pubmedhealth/PMH0002621/>

### **Noonan Syndrome: Clinical Features, Diagnosis, and Management Guidelines**

This article was featured in an issue of Pediatrics, the Official Journal of the American Academy of Pediatrics in September 2010. The article includes information about the diagnosis of Noonan syndrome and how to manage it.

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

### **Marla Wessland's Noonan Syndrome Information webpage**

This comprehensive list of information and resources is maintained by Marla Wessland, whose daughter, Sara, is featured in GEMSS – “Meet a Child with Noonan Syndrome”.

<http://www.wessland.com/noonansyndrome.htm>

### **Rene Pierpont, PhD – Selected Peer-Reviewed Publications**

Dr. Pierpont's is looking into neuropsychological features of Noonan syndrome, which may interest those involved in education.

<https://sites.google.com/site/renepierpontphd/publications-1>

### **Classroom Accommodations for Students with Visual Issues**

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

### **Noonan Syndrome Awareness Association (Australia)**

The [Noonan Syndrome Awareness Association](http://www.noonansyndrome.com.au/) (NSAA) works to increase awareness of Noonan Syndrome among health professionals and the general public, as well as to help families find and access services. They are located in Australia.

<https://noonansyndrome.com.au/>

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## Genetics Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

<https://ghr.nlm.nih.gov/condition/noonan-syndrome>

**Note:** This printable version does not include the information found under the green button marked "Transitions" on the website. Those general pages may be printed separately.

### **Meet a Child with Noonan Syndrome: Spunky Sara!**



Sara was introduced to our GEMSS website by her geneticist as "Spunky Sara!" Sara's mother Marla echoes the sentiment with her description of Sara as an extremely energetic little girl who is enthusiastic about almost everything and is "a lot of fun!" Sara is 9 years old and lives near Boston, MA. She is in third grade and is in all typical classes at her local community school.

"Many children with Ns have learning disabilities but Sara does not have any that are significant enough to be noticed at school," says Marla. She describes Sara as a quirky kid who excels in her academics but "marches to the beat of her own drummer." At one time, Sara did have an IEP to get physical, occupational and speech/language therapies in school, especially for articulation. However, she now has a 504 plan which is helpful for her vision and some GI issues. She has preferred seating so she can see well in the classroom and has access to the bathroom whenever she needs it.

She achieved her early gross motor milestones later than many children, but she now participates in kung fu and running programs with her peers.

Sara was diagnosed at about 6 months of age. A cardiologist noticed a heart murmur during her first few days of life and, although she did not have any of the more significant cardiac issues that many with Ns have, he later referred her family to a geneticist because he recognized other signs of Ns. Sara tested positive for a genetic mutation for Noonan that is present in about 50% of the people diagnosed with this syndrome. Prior to that, Sara had surgery when she was five days old to correct her severe laryngomalacia (floppy larynx tissue) that was causing breathing problems.

Over the years, she has had quite a few specialists in and out of her life. She is currently on growth hormone and is now about even height with some of the children in her class and she is now on the CDC growth chart. She sees an endocrinologist, ophthalmologist, Gastroenterologist, orthopedist, and geneticist. Although some children with NS may have a G-

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tube, especially during their first few years, Sara did not need one. She had supports to help her gain weight from nutritionists and speech/language pathologists, as well as her gastroenterologist.

Sara is very social and loves birthday parties, neighborhood block parties, and family events. She loves being in groups of kids and “loves life!”

Her mother Marla is active in supporting other families as part of an international Facebook group. She has also organized a regional support group to help local families connect with one another.

*Marla, a former preschool special education teacher, has some advice to other parents whose children have NS:*

- *Get all the early intervention you can! They really can catch up with this extra help and the feeding support can be so helpful.*
- *Because there is so much variation in each child, if you give any information (articles, etc.) about Ns to the school team, highlight the items that affect your child.*
- *Work with the school system for any IEP/504 accommodations. There are many things that can easily be done. For example, a small step stool at the sink for the shorter children may be needed, or an OT to help adjust the seating so feet are on a firm surface and the children have a good position for handwriting.*
- *Teach your child to advocate for her/himself. If there is a substitute teacher who isn't aware of their needs, they may need to advocate for themselves.*



*Marla also passed on some ideas for teachers, school nurses, and others at school:*

- *Talk to parents about the specific issues for their child. The list of possible issues is so long, and no one will have all of the possible issues. Get parents to highlight the issues that affect their child.*
- *Sometimes, you would never even know a child has Noonan syndrome because they are so mildly affected.*
- *Fatigue and pain is very real for SOME children. They may need to be excused from gym or allowed to rest if they are fatigued.*
- *They may have some extra absences due to doctor's appointments or fatigue.*

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