

Noonan Syndrome *At a Glance*

Noonan syndrome (NS) includes 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. 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

- Estimated between 50-80%
- May have had surgery and/or be on medications
- 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
 - Rounded shoulders
- Spinal abnormalities

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- 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.

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%)

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- 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 in the Classroom

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 annual 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.

Recommended screening:

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

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No special diet is required for NS, although a well-balanced diet is important.

For additional information regarding management please see these guideline:

- “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
http://www.dyscerne.org/dysc/digitalAssets/0/265_Noonan_Guidelines.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).
- Notify the parents of any pain or changes in energy level.
- Be aware of unusual changes in behavior or mood. Notify parents.
- Be aware of any academic, social, behavioral changes. Contact parents when differences are noticed.

2. Education Supports

It is important to have HIGH EXPECTATIONS for learning for children who have Noonan syndrome.

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

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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

Verbal / Language Challenges

- Language difficulties are common and may lead to reading and spelling difficulties.
- Individuals with NS may have signs of developmental disorders such as dyspraxia, dyslexia, ADHD, autism spectrum disorder (ASD).
- Executive function, 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 cognitively with reasoning.
 - 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.

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

Dyslexia

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

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- 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

Executive Function Challenges

- Executive function issues 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
 - 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

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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

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
- 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/whatisrti>

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. Considerations for Field Trips and Special Functions

No special accommodations are needed for individuals with NS syndrome, unless required due to the child's learning 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.

5. Absences and Fatigue

- School age children who have NS may have increased school absences due to medical issues
- Individuals with NS may suffer fatigue related to cardiac involvement.

6. Emergency Plan Considerations

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. Noonan Syndrome: "the most common syndrome you've never heard of."

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

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

Management of Noonan Syndrome Clinical Guidelines

These guidelines were developed by the Noonan Syndrome Guideline Development Group.

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

http://www.dyscerne.org/dysc/digitalAssets/0/265_Noonan_Guidelines.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

National Center for Biotechnology Information (NCBI) Bookshelf – Noonan Syndrome

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/>

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