What is Noonan syndrome?

Noonan syndrome (NS) is a genetic syndrome that affects around 1 in every 1000 - 2500 individuals. It is equally common among males and females, and can occur in all ethnicities. Genetic changes leading to Noonan syndrome, or related conditions such as Noonan syndrome with multiple lentigines (NSML), cause individuals to develop unique characteristics and behaviors. Common features include:

- Short stature
- Congenital heart disease
- Distinctive facial characteristics: widely spaced eyes, low set ears, low hairline, webbed neck
- Feeding and gastrointestinal problems
- Skeletal anomalies
- Bleeding disorders
- Varying degrees of developmental delay

What causes Noonan syndrome?

Noonan syndrome is caused by changes in a person's genes. Genes are like maps within our cells that tell a body how to develop and function in a particular way. Just like people follow directions on a map to reach a specific place, the body follows the directions given by its genes to grow and develop. When changes occur in the body's maps, it causes the body and brain to develop in a way that is different from what is typical.

In Noonan syndrome, this genetic difference may be found in one of many different genes. Noonan syndrome can be inherited from an affected parent. If a person with Noonan syndrome has a child, their baby will typically have a 50% chance of inheriting the genetic change and also having Noonan syndrome. In some families, the genetic change happens randomly and the child will be the first person in their family to have Noonan syndrome.

Resources

Anna's Big Week: A Story About Living with Noonan syndrome

This children's picture book tells a story about the everyday life of a child with Noonan syndrome. It is available on Amazon.com. Proceeds from the sale of this book support research on Noonan syndrome.

Noonan Syndrome Foundation: teamnoonan.org

RASopathies Network: rasopathiesnet.org

Genetics Education Materials for School Success (GEMSS): gemssforschools.org

RASopathies Family Facebook Group: facebook.com/groups/noonanfamily

GeneReviews: ncbi.nlm.nih.gov/books/NBK1124/

NS Resources Complied by Wessland Family: wessland.com/noonansyndrome.htm

Global Genes: globalgenes.org

National Organization for Rare Disorders: rarediseases.org

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TALKING WITH YOUR CHILD ABOUT NOONAN SYNDROME: A GUIDE FOR CAREGIVERS

How and when should I tell my child about their Noonan syndrome diagnosis?

Given the many different ways in which Noonan syndrome can affect people, the best approach to discussing the diagnosis depends on each individual's needs. Here are some suggestions that you may find helpful:

1. Become informed about Noonan syndrome

Seek answers to your own questions from doctors, researchers, and trusted books and websites. Your physicians and genetic counselors can help you prepare to answer questions that your child may ask in the process of learning about their condition.

2. Start conversations about Noonan syndrome with your child early and be willing to have further discussions

You may find it less overwhelming to provide information about Noonan syndrome to your child gradually over time rather than trying to disclose a lot of information in a big conversation with them. This will also prepare your child to bring up questions regarding their diagnosis as they grow older.

A natural place to start the conversation about Noonan syndrome with your child might be to explain to them why they have certain medical or therapy appointments, or are experiencing learning, social, or physical challenges.

Research shows that most people prefer to learn information surrounding their health care from someone they feel comfortable with. Learning about Noonan syndrome from a family member or trusted doctor will be better than learning about it by chance when older. Resentment can occur when a person learns that information about their identity has been withheld from them for a long period of time.

3. Provide age-appropriate information

If your child is in *preschool/kindergarten,* it might be helpful to explain to them that their body is made just a little differently from other children. Tell them that every child has certain strengths, and faces some challenges, and that having Noonan syndrome is something special and unique about them.

If your child is *school-aged*, talking to them about the genetics behind Noonan syndrome might be beneficial. This will reassure them that nothing they did caused them to have the condition. If they are the first person in the family to have NS, you can explain that it happened by chance. If they have relatives who have Noonan syndrome, you can tell them that it can be passed down from parents to children.

During *adolescence/adulthood*, encourage your child to talk about their feelings about Noonan syndrome. Help them understand and learn the ways in which they should manage their own health care.

4. Be open and honest

Do not mislead your child surrounding their condition, and try not to avoid difficult questions. Children are often able to sense when information is held back from them. If they don't receive a response to questions, they may feel anxious or assume that something is worse than it actually is.

5. Connect with other families affected by Noonan syndrome

If possible, try to attend conferences, advocacy group meetings, or fundraising events where families gather to support people with Noonan syndrome. You will learn more about the condition, and meet other people who might be facing similar challenges, which will help you build a support system. If appropriate, you could bring along your child affected by Noonan syndrome so that they could make friends with other children who have the same condition. It may also be encouraging for them to meet adults with Noonan syndrome. If attending family events is not feasible, there are other ways to connect with families via telephone, email, and social media. Sharing stories and advice with other families is a valuable tool!

Will telling my child about having Noonan syndrome cause them to feel badly about themselves?

Research suggests that many children feel relieved when they learn about the reasons for the challenges or struggles that they face. Because of learning disabilities, health issues and/or distinct physical features, some children with Noonan syndrome feel different from their friends. Learning that there are other children like them may be reassuring.

If your child responds negatively to learning about their condition, encourage them to talk about what they feel and identify what is upsetting to them. It is often necessary to repeat information and check your child's understanding, in case they are experiencing confusion. Be patient in allowing your child to process and accept all the information surrounding Noonan syndrome. If you are a parent with Noonan syndrome and feel nervous or embarrassed about disclosing the diagnosis with your child, know that your own experiences of coping with challenges could be very helpful in supporting them through life.