Hormone Disorders

Noonan’s Syndrome

Patient’s Guide

Average readability
Introduction

The aim of this leaflet is to provide general information about Noonan’s Syndrome. It will give information on how it is diagnosed and treated. It will discuss some of the problems it may cause (specifically looking at endocrine issues).

It has been written in general terms and not all of the information provided will apply to you. Hopefully, this leaflet will help you to understand this condition and give you a basis for discussions with your GP or specialist team.
What is Noonan’s Syndrome?

Noonan’s Syndrome (NS) is a complex condition that can lead to a number of issues including, heart problems (most commonly a narrowing of a heart valve called pulmonary stenosis); some chest shape deformities, growth problems (shorter than average), learning difficulties and unusual facial appearances (broad forehead, drooping eyelids [ptosis] and wide-spaced eyes [hypertelorism]).

What causes Noonan’s Syndrome?

NS is caused by a ‘spelling mistake’ (variant) in certain genes involved in directing cells to grow and divide. It can occur in boys and girls.

Sometimes the variant is inherited from a parent and sometimes it is caused by a new spelling mistake appearing (not inherited) in the child. Over 20 genes have been associated with Noonan’s Syndrome.
How is Noonan’s Syndrome Diagnosed?

Noonan Syndrome should be considered in any child with 2 or more of the following:

- Learning difficulties
- Chest deformity
- Heart problems from birth (congenital heart disease)
- Undescended Testes (Cryptorchidism)
- Short stature
- A family history of NS or any combination of the above features

Nowadays, genetic testing can be done for a number of genes already known to cause Noonan’s Syndrome in a single test (‘gene panel’).
Treatment of Noonan’s Syndrome

There is no way to prevent Noonan’s Syndrome or to cure it. If there is a family history of Noonan’s Syndrome then prenatal tests (testing the fetus during pregnancy) can be considered.

If your child has been diagnosed with Noonan’s Syndrome, it can be helpful to have a doctor co-ordinating the care delivered by different specialists (e.g. a general paediatrician or a community paediatrician).

Treatment and Follow Up
(not an exhaustive list and will depend on the individual needs of your child)

1. Growth and Nutrition with early support from feeding/nutrition specialist if needed and referral to an endocrinologist if concerns regarding growth/undescended testes

2. Assessment of the heart (every 5 years if no pre-existing issues or as per recommendation by cardiac specialist)

3. Hearing assessment

4. Early neuro-developmental or learning assessment if concerns about learning

5. Bleeding disorder assessment prior to surgery or major dental work

6. Monitoring for scoliosis – curvature of the spine (as part of annual assessment)
Specific Endocrine Issues in Noonan’s Syndrome

1. Growth

Children with Noonan’s Syndrome are often shorter than their peers even if they had a normal birthweight. The cause for the short stature is only partially known but children with Noonan’s Syndrome are rarely Growth Hormone deficient.

Growth Hormone treatment is a licensed indication for Noonan’s syndrome although some countries have not approved its use specifically for Noonan’s Syndrome unless additional features (e.g. small for gestational age or Growth Hormone deficiency). Early treatment with GH has been associated with better height outcomes.

2. Puberty

Girls with Noonan’s Syndrome usually have normal puberty and fertility. Boys with Noonan’s Syndrome can have undescended testes which need surgical intervention and often have delayed puberty (even without undescended testes). Treatment options include testosterone (either a daily gel application or monthly injection).
What are other sources of useful information?

The goal of this leaflet was to provide a basic overview of Noonan’s Syndrome.

Educational material can also be found by contacting the following organisations:

- **European Society for Paediatric Endocrinology**
  Starling House
  1600 Bristol Parkway North
  Bristol
  BS34 8YU
  espe@eurospe.org
  Telephone +44 (0) 1454 642246
  www.eurospe.org

- **Child Growth Foundation**
  info@childgrowthfoundation.org
  Telephone +44 (0) 208 995 0257
  www.childgrowthfoundation.org

- **The Endocrine Society**
  www.endo-society.org

- **NHS**
  https://www.nhs.uk/conditions/noonan-syndrome/

- **NORD**
  https://rarediseases.org/rare-diseases/noonan-syndrome/

You can also consult your specialist team for additional information in your local area.
Noonan’s Syndrome

This leaflet is part of the **Hormone Disorders Series**

The following are also available:

- Growth Hormone Deficiency
- Puberty and the Growth Hormone Deficient Child
- Precocious Puberty
- Emergency Information for Children with Cortisol and GH Deficiencies and those Experiencing Recurrent Hypoglycaemia
- Congenital Adrenal Hyperplasia
- Growth Hormone Deficiency in Young Adults
- Constitutional Delay of Growth and Puberty
- Multiple Pituitary Hormone Deficiency
- Diabetes Insipidus
- Craniopharyngioma
- Intrauterine Growth Retardation or Small for Gestational Age
- Hyperthyroidism (overactive thyroid)
- Hypothyroidism
- Type 2 Diabetes and Obesity

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