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NOONAN SYNDROME

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ABSTRACT

Noonan syndrome (NS) is a genetic disorder that may present with mildly unusual facial features, short height, congenital heart disease, bleeding problems, and skeletal malformations Facial features include widely spaced eyes, light colored eyes, low set ears, a short neck, and a small lower jaw Heart problems may include pulmonary valve stenosis The breast bone may be either protruding or be sunk while the spine may be abnormally curved Intelligence is often normal. Complications of NS may include leukemia

A number of genetic mutations can result in Noonan syndrome The condition may be inherited from a person's parents or occur as a new mutation during early development It is autosomal dominant Noonan syndrome is a type of RASopathy, the underlying mechanism for which involves a problem with a cell signaling pathway. The diagnosis may be suspected based on symptoms, medical imaging, and blood tests Confirmation may occur with genetic testing

KEYWORDS: Autosomal dominant RASOPATHY , HETEROGENUS

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CAUSES

. Mutations in the Ras/mitogen activated protein kinase signaling pathways are known to be responsible for about 70% of NS cases.^[10]

Persons with NS have up to a 50% chance of transmitting it to their offspring. The fact that an affected parent is not always identified for children with NS suggests several possibilities:

1. Manifestations could be so subtle as to go unrecognized (variable expressivity)
2. NS is heterogeneous, comprising more than one similar condition of differing causes, and some of these may not be inherited.
3. A high proportion of cases may represent new, sporadic mutations.

Heart

Up to 85% of people with NS have one of these heart defects:

- Pulmonary valvular stenosis (50–60%)
- Septal defects: atrial (10–25%) or ventricular (5–20%)
- Hypertrophic cardiomyopathy (12–35%)

Lungs

Restrictive lung function has been reported in some patients.

Gastrointestinal tract

- Failure to thrive from infancy to puberty (75%)
- Decreased appetite
- Digestive problems
- Frequent or forceful vomiting
- Swallowing difficulties
- Intestinal malrotation
- Need for a feeding tube
- Low gut motility
- Gastroparesis (delayed gastric emptying)

Genitourinary system

- Cryptorchidism (undescended testicles)

Lymphatic system]

- Posterior cervical hygroma (webbed neck)
- Lymphedema
- Lymphatic anomalies

Blood

- Bleeding disorders
- Easy bruising
- Amegakaryocytic thrombocytopenia (low platelet count)
- Blood clotting disorders
- Von Willebrand disease
- Prolonged activated partial thromboplastin time
- Partial deficiency of Factor VIII:C
- Partial deficiency of Factor XI:C
- Partial deficiency of Factor XII:C
- Platelet dysfunction
- Combined coagulation defects
- Imbalance of plasminogen activator inhibitor type-1 (PAI-1) and tissue plasminogen activator (t-PA) activity^[7]

Musculoskeletal

- Joint pain or muscle pain, especially in adults, which can vary in severity^[8]
- Undifferentiated connective-tissue disorders
- Scoliosis
- Prominence of breast bone (pectus carinatum)
- Depression of breast bone (pectus excavatum)
- Joint contractures (tightness)
- Joint hypermobility (looseness)
- Winging of the scapula
- Hypotonia (low muscle tone)
- Hypermobility syndrome
- Lordosis (increased hollow in the back) due to poor stomach muscle tone

Neurological]

- Arnold-Chiari malformation (type 1), which can lead to hydrocephalus, has been noted in some patients.
- Seizures

Physical appearance**Stature**

For short stature, growth hormone is sometimes combined with IGF-1 (or as an alternative, IGF-1 as a stand-alone) can be used to achieve an increased height/final height quicker.

Head

- Excess skin on the back of the neck
- Low hairline at the nape of the neck
- High hairline at the front of the head
- Large head
- Triangular face shape
- Broad forehead
- Short neck, webbed neck

Eyes

- Hypertelorism (widely set eyes) (95%)
- Epicanthal folds (extra fold of skin at the inner corner of the eye)
- Ptosis (drooping of the eyelids)
- Proptosis (bulging eyes)
- Refractive visual errors
- Strabismus (inward or outward turning of the eyes)
- Nystagmus (jerking movement of the eyes)

Nose

- Small, upturned nose

Ears and hearing

- Low-set ears (in over 90%)
- Backward-rotated ears (over 90%)
- Thick helix (outer rim) of ear (over 90%)
- Incomplete folding of ears

Mouth and speech

- Deeply grooved philtrum (top lip line) (over 90%)
- Micrognathia (undersized lower jaw)
- High arched palate
- Dental problems
- Articulation difficulties
- Poor tongue control

Limbs/extremities

- Bluntly ended fingers
- Extra padding on fingers and toes
- Edema of the back of hands and tops of feet
- Cubitus valgus (Wide carrying angle of the elbows)

Skin

- Lymph edema (lymph swelling of the extremities)
- Keloid formation, excessive scar formation
- Hyperkeratosis (overdevelopment of outer skin layer)
- Pigmented nevi (darkly pigmented skin spots)
- Connective tissue disease

First trimester ultrasound of Noonan syndrome may reveal nuchal edema or cystic hygroma similar to Turner syndrome. Follow up scans may show clinical features as described above. A study shows this disease is also associated with hepatosplenomegaly and with renal anomalies including malrotation and a solitary kidney. A rare incidence of choledochal cysts is also reported.^[20]

Differential diagnosis

- Turner syndrome, a different disorder often confused with NS because of several physical features that they share
- Fetal alcohol syndrome, another disorder sometimes confused with NS because of some common facial features and intellectual disability^[1]
- Other RASopathies, particularly:
 - Costello syndrome
 - Leis syndrome

- Noonan Syndrome with Multiple Lentigines, as known as LEOPARD syndrome, a related disorder caused by mutations in PTPN11 that are catalytically inactivating.
- Cardiofaciocutaneous syndrome, a related disorder which also affects genes encoding elements of the Ras/MAPKina

MANAGEMENT

- Neuropsychological testing is recommended to find strengths and challenges to tailor support needed for school and career.
- Educational customization such as an individualized education program plan is sometimes needed for school-aged children.
- Speech therapy if speech and articulation issues present
- Physical therapy and occupational therapy for gross- and fine-motor delays
- Hypotonia and motor difficulties often impact handwriting. Accommodations for lessening handwriting demands will improve performance and save long-term hand

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