

*163950 NOONAN SYNDROME

[Gene Map](#)

CLINICAL SYNOPSIS

Neck :

- Low posterior-hairline
- Webbed neck
- Pterygium colli
- Posterior cervical hygroma

Cardiac :

- Congenital heart defect
- Pulmonic stenosis
- Patent ductus arteriosus
- Coarctation of the aorta
- Pulmonary valvular dysplasia
- Myocardopathy

Thorax :

- Shield chest
- Pectus carinatum superiorly
- Pectus excavatum inferiorly

Growth :

- Short stature

Facies :

- Triangular face
- Deeply-grooved philtrum

Mouth :

- High peaks of upper lip vermilion border
- High arched palate

Eyes :

- Ptosis
- Hypertelorism

Down-slanting palpebral fissures

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Ears :

Low-set ears

Limbs :

Cubitus valgus

Clinodactyly

Brachydactyly

Blunt fingertips

GU :

Occasional hypogonadism

Cryptorchidism

Male infertility

Teeth :

Malocclusion

Neuro :

Articulation difficulties

Oncology :

Malignant schwannoma

Skin :

Lymphedema

Cutaneous lymphangioma

Heme :

Amegakaryocytic thrombocytopenia

Von Willebrand disease

Abnormal bruising

Bleeding tendency

Lab :

Partial deficiency of factor XI:C

Prolonged activated partial thromboplastin time

Partial deficiency of factor XII:C

Partial deficiency of factor VIII:C

Combined coagulation deficiencies

Elevated alkaline phosphatase