Condition: Noonan syndrome

Inheritance:

Autosomal dominant.

Genetic etiology:

Mutation in *PNPN11, BRAF, KRAS, MAP2K1, SOS1, RAF1, or NRAS* genes. One of a group of conditions associated with dysfunctional Ras signaling, also including Costello syndrome, cario-facio-cutaneous syndrome, and neurofibromatosis type 1.

Frequency:

1/1,000 – 1/2,500.

Clinical features:

Short stature, characteristic facial appearance with tall forehead and downslanting palpebral fissures, heart defects (especially pulmonic stenosis and hypertrophic cardiomyopathy), lymphatic anomalies, bleeding diathesis, renal anomalies, developmental delay.

Management:

Surgery for correctable congenital anomalies; supportive care

Genetic counseling:

May occur sporadically or be inherited from a parent; as a dominant genetic testing is available.

Human Genetics and Genomics, Fourth Edition. Bruce R. Korf and Mira B. Irons.
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