Disability Awareness Begins With You:

Noonan Syndrome

Noonan syndrome is a rare genetic disorder that is typically evident at birth (congenital). The disorder may be characterized by a wide spectrum of symptoms and physical features that vary greatly in range and severity.

In many affected individuals, associated abnormalities include a distinctive facial appearance; a broad or webbed neck; a low hairline in the back of the head; and short stature.

Characteristic abnormalities of the head and facial (craniofacial) area may include widely set eyes (ocular hypertelorism); vertical skin folds that may cover the eyes' inner corners (epicanthal folds); drooping of the upper eyelids (ptosis); a small jaw (micrognathia); a low nasal bridge; and low-set, prominent, abnormally rotated ears (pinnae). Distinctive skeletal malformations are also typically present, such as abnormalities of the breastbone (sternum), curvature of the spine (kyphosis and/or scoliosis), and outward deviation of the elbows (cubitus valgus).

Many infants with Noonan syndrome also have heart (cardiac) defects, such as obstruction of proper blood flow from the lower right chamber of the heart to the lungs (pulmonary valvular stenosis).

Additional abnormalities may include malformations of certain blood and lymph vessels, blood clotting and platelet deficiencies, mild mental retardation, failure of the testes to descend into the scrotum (cryptorchidism) by the first year of life in affected males, and/or other symptoms and findings.

In some affected individuals, Noonan syndrome appears to result from spontaneous (sporadic) genetic changes (mutations). In others, the disorder may be transmitted as an autosomal dominant trait. Genetic analysis of one affected multigenerational family (kindred) suggests that the disorder may result from mutations of a gene located on the long arm (q) of chromosome 12 (12q24). However, many investigators indicate that Noonan syndrome may be caused by mutations of different genes (genetic heterogeneity).

<u>Resources</u>

Human Growth Foundation
800-451-6434

MAGIC Foundation for Children's Growth
800-362-4423

March of Dimes
888-663-4637

The ARC
800-433-5255

American Heart Association
800-242-8721

Congenital Heart Anomalies, Support, Education, & Resources
419-825-5575

Big Hearts for Little Hearts
516-741-5522

National Heart, Lung, & Blood Institute
301-592-8573

Noonan Syndrome Support Group
410-374-5245

Children's Cardiomyopathy Foundation
201-227-8852

http://my.webmd.com/hw/raising_a_family/nord412.asp?lastselectedguid={5FE84E90-BC77-4056-A91C-9531713CA348}