Noonan syndrome is a genetic disorder that is typically evident at birth (congenital). The disorder is 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 posterior hairline; a typical chest deformity and short stature. Characteristic features of the head and facial (craniofacial) area may include widely set eyes (ocular hypertelorism); skin folds that may cover the eyes' inner corners (epicanthal folds); drooping of the upper eyelids (ptosis); a small jaw (micrognathia); a depressed nasal root; a short nose with broad base; and low-set, posteriorly 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) and thickening of the ventricular heart muscle (hypertrophic cardiomyopathy). Additional abnormalities may include malformations of certain blood and lymph vessels, blood clotting and platelet deficiencies, learning difficulties or mild intellectual disability, 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. Noonan syndrome appears to affect more males than females and is thought to affect approximately one in 1,000 to one in 2,500 people. However, other reports indicate that the disorder may affect more than one in 1,000 newborns in the general population. Since Noonan syndrome was originally reported in 1883 (O. Kobylinski) and more thoroughly described in 1963 (J.A. Noonan and D.A. Ehmke), more than 500 patients shave been discussed in the medical literature. Because Noonan syndrome is extremely variable and therefore may be under- or misdiagnosed, it may be difficult to determine the true frequency of the disorder in the general population.