

# **Noonan Syndrome Information Sheet**

- to be included in the Personal Health Records of people who have been diagnosed as having Noonan Syndrome

(The material in this sheet has been derived from 'Therapeutic Guidelines: Management Guidelines - Developmental Disability' 2005, which can be consulted for more detailed information.)

Noonan Syndrome was first described in 1963. The classical features include a typical facial appearance, valvular pulmonary stenosis, short stature and mild intellectual disability. However there is considerable variation in expression and the phenotype does change with age.

## **CAUSE and PREVALENCE**

The pattern of transmission suggests autosomal dominant inheritance although a significant proportion of cases are sporadic. PTPN11 gene mutations can be found in about 50% of cases and this can be useful in confirming the diagnosis.

It has been estimated that Noonan syndrome with intellectual disability accounts for less than one per 10,000 births. However, Noonan syndrome is associated with an intellectual disability in only one third of cases and the true incidence would be much greater.

## **DIAGNOSIS and CHARACTERISTICS**

Diagnosis is based on clinical findings and may be confirmed by genetic testing. The differential diagnoses include Williams syndrome and foetal alcohol syndrome.

Facial features characteristically include hypertelorism (widely spaced eyes), ptosis, and down-slanting palpebral fissures. Strabismus and refractive errors are common. The ears tend to be low set and posteriorly rotated. There is a high arched palate, flattened nasal bridge and micrognathia. Although wispy in the infant, the hair tends to become more curly or woolly in the older child. The contour of the face becomes more triangular with age. There is a low posterior hairline and excess nuchal skin may be noted in the newborn. Webbing of the neck becomes more obvious in older children.

Growth: In infancy there may be failure to thrive. Short stature is present in 80%. The majority of females are fertile but puberty may be normal or delayed in both sexes. In over half the males one or both testes is undescended and there may be inadequate secondary sexual development associated with deficient spermatogenesis.

## **Medical Problems**

Congenital heart defects occur in about two thirds of cases, with pulmonary valvular stenosis accounting for about half of these. There is a typical ECG pattern.

A chest deformity such as pectus carinatum or pectus excavatum is found in 90% of cases. Cubitus valgus and hand anomalies are also common. Scoliosis and talipes equinovarus may occur.

Abnormalities of clotting factors and platelet dysfunction, lymphoedema and unexplained hepatosplenomegaly occur in a proportion of individuals.

## **Cognitive, Behavioural and Neurological Problems**

About one third of individuals with Noonan syndrome have a mild intellectual disability. The learning disability appears to be associated with specific visual-constructional problems. The language delay may be secondary to perceptual motor disabilities, mild hearing loss or articulation problems. Children with Noonan syndrome tend to be clumsy, stubborn and irritable, and can have psychiatric problems and communication difficulties. Hypotonia is common. Seizures and vision and hearing impairments have also been reported in children.

## **KEY RECOMMENDATIONS FOR MANAGEMENT**

- Referral to geneticist for diagnosis and genetic counselling.
- Management of congenital heart disease and periodic cardiac evaluation.
- Management of undescended testes if present.
- Screening for clotting abnormalities.
- Assessment and management of any learning difficulties
- Behavioural management.
- Monitoring of vision and hearing - ophthalmological examination and audiology.
- Management of seizures, if present.

## **SUPPORT ASSOCIATION**

Information is available from the Noonan Syndrome Association on (02) 9804 6762