PIERRE ROBIN SYNDROME

**Includes:** Pierre Robin sequence, Glossoptosis, Micrognathia and Cleft Palate

**FACTS**

Pierre Robin syndrome is a condition in which the jaw bone or mandible is small. The downward position of the tongue may lead to breathing difficulties and there may be a high arched or cleft palate resulting in feeding problems. The jaw continues to grow after birth and the condition improves with age.

Other features include cardiovascular and lung conditions, such as benign heart murmurs, high blood pressure in the arteries of the lungs (pulmonary hypertension), and narrowing of the opening between the lung artery and the right ventricle of the heart (pulmonary stenosis). Abnormalities of the musculoskeletal system, including those in the arms, legs, feet, and vertebral column, are also common. Inflammation of the middle ear (otitis media) occurs in about 80% of individuals who are affected. Eye (ocular) defects occur in about 10% to 30% of cases.

Males and females are affected in equal numbers. Pierre Robin may occur alone, or in conjunction with other syndromes. When it occurs isolation, it is nearly always a sporadic (spontaneous or random) event, although there may be an inherited basis for Pierre Robin syndrome in some instances. When it occurs in isolation, Pierre Robin syndrome may follow an autosomal recessive inheritance pattern.

Support Groups can provide individuals and families with information about genetic conditions, community resources and an understanding and empathic ear.

If you are not able to contact the services listed below, please call the Association of Genetic Support of Australasia (AGSA) on (02) 9211 1462 or visit www.agsa-geneticsupport.org.au for support and assistance.

Further information and details of your local genetic counselling service can be found at www.genetics.edu.au, or please call The Centre for Genetics Education on (02) 9462 9599.