

Williams Syndrome Information Sheet

(Optional sheets to be included where recipient of PHR has Williams syndrome)

The material in this sheet has been adapted from the Therapeutic Guidelines book 'Management Guidelines for People with Developmental and Intellectual Disabilities' and updated from the 2005 version 'Management Guidelines – Developmental Disability' which can be consulted for more detailed information.

INTRODUCTION

Williams and Barret-Boyes first described this syndrome in New Zealand in 1961. The classical features include a typical facial appearance, supravalvular aortic stenosis and variable intellectual disability. Children and adults with this disorder usually have a friendly, outgoing personality.

AETIOLOGY and INCIDENCE

- It has recently been discovered that the great majority of Williams syndrome cases involve a microdeletion in the gene for the extracellular-matrix protein elastin (ELN). This gene is situated on the long arm of chromosome 7. Several other genes have also been implicated. As the gene deletion is usually de novo and parents have normal chromosomes, the recurrence risk in siblings is extremely low.

A defect in elastin may account for the abnormalities found in connective tissue in the aorta and in the larynx but not for the cognitive and behavioural problems. The latter may be explained by a deletion in adjacent genes.

Limited studies indicate that Williams syndrome occurs in less than 1 per 20,000 births.

DIAGNOSIS

- Irritability and failure to thrive, often with constipation, are the most consistent early features.
- The characteristic facial features become more evident after the age of one year.
- Fluorescence in situ hybridisation (FISH) will identify a submicroscopic deletion at 7q11.23 in 95% of cases.
- Investigation and assessment includes use of Williams syndrome growth charts, checking calcium levels and thyroid function, cardiology, urinary tract and eye checks and genetic review.

CHARACTERISTICS

Commonly seen **facial features** include periorbital fullness, stellate irides, malar hypoplasia, a broad nasal tip with anteverted nares and a full lower lip often with an open mouth appearance. In infancy and childhood the cheeks are sagging but become thin in adolescence or in early adulthood. Strabismus is common. Characteristic dental anomalies may also occur.

Williams syndrome may be associated with intrauterine **growth retardation**. Both children and adults may be short in stature and microcephaly is often present. Menarche may be early.

Associated medical conditions are common. Cardiac abnormalities occur in about 75% of cases, the commonest being supra-valvular aortic stenosis and peripheral pulmonary artery stenosis. Hypertension occurs frequently in adults. Myocardial ischaemia associated with stenosis of the left coronary artery and strokes and chronic hemiparesis associated with stenosis of the cerebral arteries can occur.

Renal abnormalities, including bladder diverticulae, renal hypoplasia and duplicated kidneys, were found in 18% of cases. Urinary tract infections, chronic constipation and diverticulosis may also occur and require management.

Other conditions include a hoarse voice, inguinal hernia, orthopaedic problems such as joint contractures and scoliosis and infantile hypercalcaemia which may present as constipation.

Cognitive, Behavioural and Neurological Problems

Infantile behaviour is often described as difficult with irritability and poor feeding. Older children tend to be friendly and talkative, and may be socially uninhibited. Their intellectual ability may be overestimated because of their apparently good verbal skills.

Cognitive deficits vary from mild to moderate intellectual disability, with perceptual and motor function more reduced than verbal and memory performance. Emotional and behavioural disturbances including anxiety attention deficit problems may improve in adolescence and adulthood. Hyperacusis has also been reported. Hypotonia is frequent in the younger years progressing to hypertonia with age.

KEY RECOMMENDATIONS FOR MANAGEMENT

- Screening for and management of congenital heart disease
- Screening for and management of hypertension in adults
- Management of urinary and gastro-intestinal and feeding problems
- Regular dental care
- Ophthalmological review
- Physiotherapy to aid management of musculoskeletal and neurological problems
- Management of behaviour
- Ultrasound screening of the renal system

SUPPORT ASSOCIATIONS

Information about state support associations can be obtained by contacting :

Williams Syndrome Association of Australia.

C/- Association of Genetic Support of Australiasia

66 Albion St, Surry Hills, NSW 2010

Ph: (wk) (02) 9211 1462, (hm) (02) 9332 1361 Fax: (02) 9211 8077

Internet: <http://www.agsa-geneticsupport.org.au/Index.html>