Syndrome Specific Medical health check guide - Williams Syndrome (WS)

Introduction

Williams syndrome (WS) is a sporadic genetic disorder caused by the deletion of a small part of chromosome 7. Features include a distinctive facial appearance, congenital heart defects and high levels of calcium in infancy. Early intervention can help with some developmental delays and treatment of heart problems take place as soon as possible.

Early feeding problems are common and development is delayed with small stature. People with WS have sociable personalities, characteristic behavioural traits and variable degrees of intellectual disability (usually mild). WS has an incidence of 1 in 7500 births and presents in childhood with an elfin-like face. The distinct facial features include broad forehead, short nose with a broad tip, full cheeks, wide mouth with full lips, and dental problems.

It is a multisystem disorder that can also include hypertension, supravalvular aortic stenosis (SVAS) and other cardiac and connective tissue related problems and hypercalcaemia.

Many individuals with WS will not achieve normal life expectancy due to complications of the disease.

History

As with all people with LD focus on an assessment of:

- eyesight and hearing
- feeding, bowel and bladder function
- behavioural problems.

Ask about shortness of breath including orthopnoea, chest pain and ankle swelling.

Ask about dental problems and constipation.

Ask about hyperextensible joints and decreased muscle tone.

Advise to wear sunscreen and avoid sunshine to reduce risk of hypercalcaemia.
Examination

1. Sensory
   □ Full assessment by Optician/Optometrist every 2 years.
   □ Check for Stabismus (squint cross-eye)
   People with Williams syndrome may have hearing hypersensitivity.
   □ Assessment of hearing with referral to Audiology if concerns about hearing or hyperacusis.

2. Gastrointestinal
   □ Examine the abdomen for constipation. Screen for coeliac disease and diverticular disease if symptomatic.
   □ Renal tract ultrasound every 5 years for nephrocalcinois.

3. Cardiovascular
   □ Congenital heart defects (especially supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis).
   □ Full cardiovascular assessment with Echocardiogram and Blood pressure measurement in both upper limbs.
   □ Echocardiogram every 5 years throughout life.

4. Musculoskeletal
   □ Height, weight and BMI annually, and avoid excessive weight gain – encourage an ‘active’ lifestyle.
   □ Assessment of spine for scoliosis.
   □ Check for joint contractures and gait problems in older people with WS
   □ Annual blood tests:
     • Creatinine
• Calcium, and Urine Calcium/Creatinine-ratio test if symptomatic of hypercalcaemia.
• TSH
• Screen for diabetes at age 30 years

Resources
Williams Syndrome Foundation: http://www.williams-syndrome.org.uk/wsf-people/i-am-health-professional