

# WILLIAMS SYNDROME FOUNDATION

WILLIAMS SYNDROME FOUNDATION LIMITED

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## WILLIAMS SYNDROME HEALTH SURVEILLANCE

To: Dr .....

As you know, your patient .....  
has Williams Syndrome/Infantile Hypercalcaemia, which was  
diagnosed at the age of ..... months/years.

The Professional Advisory Panel of the WS/IHC Foundation has some  
evidence that the condition is associated with an increased risk from  
cardiovascular disease and renal problems. There is also evidence of  
progressive musculo-skeletal disorders particularly involving the spine.

It is therefore recommended that the persons with WS/IHC should receive  
annual surveillance including:

- i) Blood pressure
- ii) Urinalysis
- iii) Physical examination with special reference to  
bone and joint problems including scoliosis;
- iv) Systematic review of symptoms with appropriate  
investigation.



5/ Typical Diagnostic Features...

- c) **Cognitive Abilities and Behaviour**
  - Mild to severe learning difficulties (more marked in visuo-spatial areas than in language)
  - Global Developmental Delay (especially)
  - Outgoing sociable personality
  - Poor concentration, distractibility
  
- d) **General**
  - Infantile hypercalciuria + hypercalcaemia
  - Failure to thrive
  - Hyperacusis
  - Dental anomalies
  
- 6i) **CLINICAL FEATURES: DEVELOPMENTAL**
  - a) **Intellectual:**
    - Wide variation
    - Mean IQ (ranges from severe learning difficulties to just below average)
    - Speech late to develop "Cocktail Chatter"
    - Visuo-spatial problems
    - Sleep disturbances, often poor sleep pattern
    - Behavioural problems:
      - Overactivity; destructive tendencies
  
  - b) **Motor Skills:**
    - Delayed developmental milestones
    - Hypotonia
    - May not walk until aged 2 to 3 years.
  
  - c) **Growth**
    - Low birth weight
    - Failure to thrive - usually less than 3rd centile for weight, height (particularly with hypercalcaemia), and head circumference
    - Short stature (mean: 5' for girls, 5' 6" for boys)
    - Some catch up growth occurs in middle child-hood but remains below average into adulthood.

6i)/ Clinical Features: Developmental..

**d) Puberty**

Early onset  
Average 9 to 13 years of age for girls, 10 to 14 years  
of age for boys often associated with small genitalia)

6ii) CLINICAL FEATURES: CALCIUM METABOLISM  
Hypercalcaemia ~ hypercalciuria tends to level- off around 12  
to 18 months of age (cause unknown)  
Requires regular biochemical assessment under paediatric  
supervision.

Symptoms

Feeding Difficulties (needs paediatric supervision)  
Vomiting/FTT.  
May need advice from Dietitian/Health Visitor  
Low calcium diet may be indicated  
"Locasol" milk available (Cow & Gate)  
Distilled water  
"Brita Filter- to remove calcium from tap water  
Nasogastric feeding (if severe)

Complications

Nephrocalcinosis; Nephrolithiasis; Osteosclerosis of  
metaphases of long bones; Risk of urinary tract infections.

6iii) CLINICAL FEATURES: CARDIOVASCULAR SYSTEM  
Most have a murmur in the neck due to varying degrees of S.V.A.S.

N. B. Antibiotic cover for dental and surgical procedures.

Unexplained fever, think S.A.B.E, do blood cultures.

Supravalvular Aortic Stenosis 85%o (SVAS)

Most common CHD in Williams Syndrome

Mild form generally does not progress

Moderate/severe does tend to progress with time and may  
require diagnostic cardiac catheterisation prior to surgery.

Peripheral Pulmonary Artery Stenosis -  
may regress; surgery difficult

Other Stenotic Lesions:

Coarctation of the Aorta; Renal Artery Stenosis;  
Hypertension approximately 10%

6i) CLINICAL FEATURES: GASTRO-INTESTINAL TRACT

Feeding Difficulties

Vomiting and refusal to feed  
Often worse in the first year of life  
May be associated with hypercalcaemia and therefore require low calcium diet including special milk ("Locasol" available on prescription).  
May be caused by altered oesophageal motility.  
Oesophageal spasm  
Oesophagitis

Constipation

Possible dietary  
May result in anal fissure  
Treat with dietary advice + laxatives  
Inguinal hernia 30%  
Rectal prolapse 10%

6v) CLINICAL FEATURES: RENAL

A baseline renal and bladder ultrasound examination is recommended because of the high incidence of kidney abnormalities in Williams Syndrome.

The usefulness of repeat ultrasound examinations in the absence of documented renal disease or hypercalciuria is not established.

In children, a urinalysis should be performed every 1-2 years as part of routine paediatric care.

Blood creatinine levels should be followed periodically (frequency not specified) in adults with Williams Syndrome.

Specific urinary tract investigations should be performed only for specific indications such as chronic urinary frequency and bedwetting which persist beyond 6-8 years of age, recurrent urinary tract infectious, urine retention, and or chronic difficulties or discomfort in urinating.

6vi) CLINICAL FEATURES: LOCOMOTOR SYSTEM

Poorly developed pectoral/pelvic girdle with associated increased thigh muscle bulk, sloping shoulders (more noticeable with age).  
Poor gross and fine motor skills.

Musculoskeletal anomalies Radio-ulnar synostosis: Kyphosis:  
Flexion contractures; Recurrent joint dislocation

6vii) CLINICAL FEATURES: DENTAL

High dental caries rate both in deciduous and permanent dentition.

Poor response to dental treatment:

Partly due to anxiety; Poor tolerance to the noise of the dental drill (hyperacusis)

Advise: Fluoride drops,  
Oral hygiene instructions  
Fissure sealants on permanent teeth

NB Antibiotic cover re: congenital heart disease.

Anomalies include:

Small teeth  
Abnormal shape (pointed incisors, bulbous molars)  
Malocclusion  
Invagination of incisors  
Late eruption

6viii) CLINICAL FEATURES: AUDIOLOGICAL

Hyperacusis in 80 to 90% of cases

6ix) CLINICAL FEATURES: EYES

Squints 50% Myopia: Lenticular opacities (rare)

7) PROGNOSIS:

The natural history in adult life is not well documented, but there may be increased risk from heart disease and renal failure.

8) MANAGEMENT:

**Primary Health Care Team**

Support to parents (particularly at time of diagnosis)  
Parental involvement in decisions of treatment and referral  
Parental access to medical correspondence.  
Co-ordination of, and liaison between, different specialists.  
Liaison between members within Primary Health Care Team, many of which may be involved (if available in your area)  
Community Developmental Paediatrician,

8 Management: Primary Health Care Team...

Community Physiotherapist.  
Paediatric Occupational Therapist.  
Speech Therapist.  
Health Visitor.  
Pre-school teacher/Portage scheme  
Clinical Psychologist.

9) SCHOOLING:

In order to address their special needs, all Williams children should be statemented.

Pre-School teacher (may be available from time of diagnosis).

Portage workers

Special needs teachers

Seek advice from education officer for children with special needs.

Children may cope with ordinary school at infant and primary levels, but are likely to require special school at secondary level.

10) REFERRAL:

As necessary to:

Paediatrician

Nephrologist

Cardiologist

Orthopaedic Surgeon

Dentist (Dental Surgeon)

Gastroenterologist

Geneticist

Local Child Development (if available)

11) GENERAL ADVICE:

May claim for:

Disability Living Allowance

Invalid Care Allowance

Free Prescriptions (depending on disability)

Severe Disablement Allowance

11/ GENERAL ADVICE...

Information on claiming benefits is available by dialling 0800 666555 and asking for 'FREEPHONE DSY' or writing to the Williams Syndrome Foundation at the address on the front of this booklet.

Guidelines for parents, teachers and employers & supervisors are available from the registered office of the WS Foundation and are also reproduced on our website: <<http://www.williams-syndrome.org.uk>>

Tertiary referrals may be made to members of the Professional Advisory Panel of the WS Foundation. Addresses of the panel can be obtained from the Williams Syndrome Foundation or:

Dr Mike Wolfman, at:  
Burnham Medical Centre  
Love Lane  
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**Professional Advisory Panel:**

Dr. N. Martin MD, MRCP	Prof. D. Donnai, MB BS FRCP
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Mike Adlam MBA	John Nelson ACIB
Ms E Hurst	Nurse Wendy Smith RN (Child) DipHE
Prof. P. Howlin MSc PhD Cpsychol PBPSS	

*This leaflet has been produced by the Williams Syndrome Foundation for family doctors: it is very comprehensive and therefore covers all possible features of Williams Syndrome. Each person with Williams Syndrome will demonstrate some of these features but not all of them. Even the rarest possibilities have been included to enable your doctor to manage your child's health efficiently.*