Management of Williams Syndrome

A Clinical Guideline

Williams Syndrome Guideline Development Group
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**Acknowledgements**
Introduction...

... to Williams Syndrome (WS)
Williams Syndrome is a rare condition, that occurs in 1 in 20,000 births. The current definition of WS was agreed by the Williams Syndrome Guideline Development Committee at the Williams Syndrome Management Consensus Meeting held in Manchester in May 2009;

“Williams syndrome is a sporadic genetic disorder due to deletion of a small part of chromosome 7. Features may include a distinctive facial appearance, congenital heart defects and high levels of calcium in infancy. Early feeding problems are common and development is delayed. People with WS have sociable personalities, characteristic behavioural traits and variable degrees of learning disability.”

... to the Williams Syndrome Guideline Development Project
The guidelines have been developed using a robust methodology based on the one utilised by the Scottish Intercollegiate Guidelines Network (SIGN). The method has been adapted to suit rare conditions where the evidence base is limited, and where expert consensus plays a greater role. The members of the guideline development group are listed on page 40.

... to the Williams Syndrome Clinical Management Guidelines
What are the aims of the guidelines?
The existing guidelines for the medical supervision of people with WS were published in 2001 by the American Academy of Pediatrics Committee on Genetics (AAPCG). Whilst a valued addition to the available guidance these are not entirely transferable to the UK. Therefore, in order to optimise the medical management of people with WS, the aim of the guidelines is to provide clear evidence-based management recommendations applicable to UK patients.

Who are they aimed at?
As WS is so rare, it is unlikely that the primary care clinicians usually responsible for coordinating the care of people with the condition will have had much prior experience of the syndrome. As it is a multisystem disorder, people with WS require various tests, screenings, assessments, referrals and multidisciplinary interventions at different stages of their lives. These guidelines lay out these requirements in a clear format that are accessible to anybody who is involved in the care of an individual with WS.

How are they used?
The guidelines are divided into recommendations for four age groups: · Infancy: 0—1 year old · Adolescence: 11—18 years old · Childhood: 1—11 years old · Adulthood: 18 years old +

On page 4, recommended baseline investigations are listed, to be consulted alongside the age group-specific recommendations at the time of diagnosis. For each age group, the recommended tests/screenings are listed, and follow-up options depending on the outcome of the test or screening are indicated.

NB. ABNL = Abnormal
## Clinical features and recommended baseline investigations in Williams Syndrome

### Clinical Features of Williams Syndrome

- Congenital heart defects (especially supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis)
- Raised blood/urine calcium levels
- Nephrocalcinosis, bladder and renal tract abnormalities
- Endocrine abnormalities
- Failure to thrive/slow growth rate/feeding problems
- Hypertension
- Scoliosis and other musculoskeletal problems
- Gastrointestinal problems
- Distinctive facial features
- Dental anomalies
- Distinctive behavioural characteristics including irritability, anxiety, overfriendliness
- Hypersensitivity to noise

### Baseline investigations

(Where investigation not indicated for a specific clinical feature, please refer to the relevant age group-specific page for management recommendations)

- Full cardiovascular assessment including scans and BP (blood pressure) measurement in both upper limbs.
- Serum Ca and Urine Ca: Creatinine Ratio
- Renal tract ultrasound to include kidneys and bladder
- Thyroid Function Tests (TFTs)
- Plot growth on appropriate Williams Syndrome growth chart (see pages 14 and 15)

## Confirm diagnosis of Williams Syndrome by testing for microdeletion on chromosome 7 using specialist molecular techniques e.g. FISH test*

*Fluorescence in situ hybridisation (FISH) is the most common, but not the only available test for confirming a diagnosis of Williams Syndrome. Some laboratories may use other DNA-based diagnostic techniques.
Recommendations for the management of Williams Syndrome
~ in neonates & infancy (1) ~

**Recommended Testing/Screening**

- Serum Ca and Urine Ca: creatinine ratio
  - If normal and under 1 year old, repeat test at 12 months.

- Thyroid Function Tests (TFTs)
  - Ensure baseline test undertaken.
  - Measure TSH levels and if elevated, consider thyroid scanning.

- Renal tract screening to include kidneys and bladder
  - If nephrocalcinosis refer to nephrologist for 6 monthly screening.
  - If structural abnormalities, management or referral as necessary.

- Hypertension screening
  - Annual monitoring of blood pressure in both upper limbs and left leg.
  - If associated with renal artery stenosis (RAS), refer to nephrologist.
  - Surgical treatment where necessary.

- Cardiac screening
  - Full cardiac assessment including scans before one year old if diagnosis made in neonatal period.
  - Annual cardiac examination until 4 years old.
  - Follow up by cardiologist.

**Clinical Management Recommendations**

Management of Hypercalcaemia

- Calcium intake should be equal to or less than half of the recommended nutrient intake (RNI) for the patient's age group.
- Stop use of supplements containing calcium.
- Ensure that infant feeds are prepared using 'soft' water.
- Ensure adequate rehydration.
- Locasol formula milk (SHS Nutrition)
- Steroids (Prenisolone), orally as necessary.
- Monitor blood pressure

- Take sunblock if travelling/in sunny conditions.
- 3 monthly follow up.
- If serum PTH starts to rise, relax calcium intake but monitor blood and urine calcium levels.
- Consider referral to paediatric metabolic bone disorder specialist.
- In rare cases, where hypercalcaemia is refractory to hydration and low-calcium diet, intravenous Pamidronate may be necessary.
### Recommendations for the management of Williams Syndrome

#### ~ in neonates & infancy (2) ~

<table>
<thead>
<tr>
<th>Recommended Testing/Screening</th>
<th>Clinical Management Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Feeding &amp; Gastrointestinal issues</strong></td>
<td>Take feeding history.</td>
</tr>
<tr>
<td></td>
<td>Enquire about bowel habit.</td>
</tr>
<tr>
<td></td>
<td>If problems, refer for appropriate support and treat constipation.</td>
</tr>
<tr>
<td>ABNL</td>
<td></td>
</tr>
<tr>
<td><strong>Growth</strong></td>
<td>Measure height, weight and occipitofrontal circumference (OFC) at birth and 1-3 monthly.</td>
</tr>
<tr>
<td>ABNL</td>
<td>Routine paediatric investigations for failure to thrive and reduced growth velocity.</td>
</tr>
<tr>
<td><strong>Hearing screening</strong></td>
<td>NHS newborn hearing screening programmes throughout UK (NHSP) - screening within the first few weeks.</td>
</tr>
<tr>
<td><strong>Vision screening</strong></td>
<td>Visual screening should take place between 6 and 12 months.</td>
</tr>
<tr>
<td></td>
<td>Parents to report any concerns.</td>
</tr>
<tr>
<td></td>
<td>Refer to community optometric/orthoptic service (via GP) if abnormality found.</td>
</tr>
<tr>
<td>ABNL</td>
<td></td>
</tr>
<tr>
<td><strong>Screening for dental anomalies</strong></td>
<td>Enrol patient in an individualised preventative oral healthcare programme from an early age.</td>
</tr>
<tr>
<td></td>
<td>Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.</td>
</tr>
<tr>
<td>ABNL</td>
<td>Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.</td>
</tr>
<tr>
<td><strong>Multidisciplinary developmental assessment</strong></td>
<td>Between 0-3 years old. Coordinated by hospital or community paediatrician.</td>
</tr>
</tbody>
</table>

**† Anaesthesia**

A paediatric anaesthetist should be involved in the pre-op care of children up to 3-4 years old.

Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.

Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.
**Recommendations for the management of Williams Syndrome ~ in childhood (1) ~**

<table>
<thead>
<tr>
<th>Recommended Testing/Screening</th>
<th>Clinical Management Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Serum creatinine</td>
<td>In all WS children, test serum creatinine every 2—4 years.</td>
</tr>
<tr>
<td>• Serum Ca and Urine Ca: creatinine ratio</td>
<td>Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.</td>
</tr>
<tr>
<td>• Thyroid Function Tests (TFTs)</td>
<td>If normal when under 1 year old, repeat test at 12 months.</td>
</tr>
<tr>
<td>• Renal screening</td>
<td>Management of Hypercalcaemia</td>
</tr>
<tr>
<td>• Hypertension screening</td>
<td>Monitoring for 1—2 years after hypercalcaemia has resolved.</td>
</tr>
</tbody>
</table>

- Calcium intake should be equal to or less than half of the recommended nutrient intake (RNI) for the patient's age group.
- Stop use of supplements containing calcium.
- Ensure that infant feeds are prepared using ‘soft’ water.
- Ensure adequate rehydration.
- Locasol formula milk (SHS Nutrition)
- Steroids (Prenisolone), orally as necessary.
- Monitor blood pressure

- Take sunblock if travelling/in sunny conditions.
- 3 monthly follow up.
- If serum PTH starts to rise, relax calcium intake but monitor blood and urine calcium levels.
- Consider referral to paediatric metabolic bone disorder specialist.
- In rare cases, where hypercalcaemia is refractory to hydration and low-calcium diet, intravenous Pamidronate may be necessary.

Monitor for 1—2 years after hypercalcaemia has resolved.

Test if patient is symptomatic.

Measure TSH levels and if elevated, consider thyroid scanning, consider referral to endocrinologist for treatment with L-Thyroxine if patient has overt hypothyroidism, or progressive deterioration of thyroid function.

Renal tract ultrasound to include kidneys and bladder if symptomatic.

If nephrocalcinosis refer to nephrologist for 6 monthly screening.

Annual monitoring of blood pressure in both upper limbs and left leg.

If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary.

**NB.** If RAS is present, angioplasty is not recommended due to elastinopathy.

If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out).
## Recommendations for the management of Williams Syndrome

### in childhood (2)

<table>
<thead>
<tr>
<th>Recommended Testing/Screening</th>
<th>Clinical Management Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Cardiac screening</strong></td>
<td>Annual cardiac examination until 4 years old, and once between 5-13 years old. Full cardiac assessment including scans every 5 years.</td>
</tr>
<tr>
<td><strong>Feeding &amp; Gastrointestinal issues</strong></td>
<td>Enquire about feeding problems annually. Enquire about bowel habit annually. Treat constipation.</td>
</tr>
<tr>
<td><strong>Screen for coeliac disease</strong></td>
<td>Once, after 3 years of age, with low threshold to repeat if suggestive symptoms. Height, weight and OFC measurements 1-3 monthly until 2 years of age. Annualy thereafter (use WS growth charts). Mid parental height centile should be estimated.</td>
</tr>
<tr>
<td><strong>Growth &amp; Puberty</strong></td>
<td>Check spine clinically for kypho/scoliosis at puberty and x-ray/refer to orthopaedic team as indicated. Routine paediatric investigations for abnormal growth velocity and precocious puberty (&lt; 8 years). Where necessary, consider gonadotropin releasing hormone (GnRH) therapy.</td>
</tr>
<tr>
<td><strong>Hearing screening</strong></td>
<td>18 months: screen for otitis media with effusion (OME) &amp; hyperacusis. 3 years: screen for OME &amp; language development. 5-10 years: screen for hyperacusis &amp; hearing loss. 11-18 years: screen for hyperacusis &amp; high frequency hearing loss. If hyperacusis, implement a programme of desensitisation (plus maskers if necessary).</td>
</tr>
<tr>
<td><strong>Vision screening</strong></td>
<td>Visual screening should take place between 6 and 12 months. Parents to report any concerns. Refer to community optometric/orthoptic service (via GP) if abnormality found.</td>
</tr>
<tr>
<td><strong>Screening for dental anomalies</strong></td>
<td>Enrol patient in an individualised preventative oral healthcare programme from an early age. Routine follow up and regular dental examinations by a family dentist or local community dental services are essential. Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.</td>
</tr>
</tbody>
</table>
## Recommendations for the management of Williams Syndrome

### in childhood (3) ~

<table>
<thead>
<tr>
<th>Recommended Testing/Screening</th>
<th>Clinical Management Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Multidisciplinary developmental assessment</td>
<td></td>
</tr>
</tbody>
</table>
Between 0-3 years old. Should involve local Child Development/ Learning Difficulties (LD) Teams.  
Involve Child and Adolescent Mental Health Services (CAMHS) if necessary. |
| Behavioural & Mental Health issues |  
Ongoing review and support of learning and development with further assessment of special educational needs as required.  
Behavioural management advice and support to family as required.  
Refer for psychological intervention for anxiety, and when major life events. |

### Anaesthesia

A paediatric anaesthetist should be involved in the pre-op care of children up to 3-4 years old.  
Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.  
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.
## Recommendations for the management of Williams Syndrome

### in adolescence (1)

<table>
<thead>
<tr>
<th>Recommended Testing/Screening</th>
<th>Clinical Management Recommendations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Serum creatinine</td>
<td>In all WS adolescents, test serum creatinine every 2—4 years. ABNL Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.</td>
</tr>
<tr>
<td>Serum Ca and Urine Ca: creatinine ratio</td>
<td>Test if symptomatic of hypercalcaemia. ABNL If abnormal, investigate and manage as appropriate. ABNL Test if patient is symptomatic.</td>
</tr>
<tr>
<td>Thyroid Function Tests (TFTs)</td>
<td>ABNL Measure TSH levels and if elevated, consider thyroid scanning, Consider referral to endocrinologist for treatment with L-Thyroxine if patient has overt hypothyroidism, or progressive deterioration of thyroid function.</td>
</tr>
<tr>
<td>Renal screening</td>
<td>Renal tract ultrasound at puberty, before leaving paediatric care, and 5 yearly thereafter, or if symptomatic. ABNL If nephrocalcinosis refer to nephrologist for 6 monthly screening.</td>
</tr>
<tr>
<td>Hypertension screening</td>
<td>Annual monitoring of blood pressure. ABNL If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary. NB. If RAS is present, angioplasty is not recommended due to elastinopathy.</td>
</tr>
<tr>
<td>Cardiac screening</td>
<td>Cardiac examination once between 5-13 years old and 13-21 years old (follow up if symptomatic). Full cardiac assessment including scans every 5 years.</td>
</tr>
<tr>
<td>Gastrointestinal issues</td>
<td>ABNL Enquire about bowel habit annually. ABNL Treat constipation and consider investigating for diverticular disease.</td>
</tr>
</tbody>
</table>
Recommendations for the management of Williams Syndrome
~ in adolescence (2) ~

**Recommended Testing/Screening**

- **Screen for coeliac disease**
  - If patient symptomatic.

- **Growth & Sexual Health**
  - Check spine clinically for kypho/scoliosis at puberty and x-ray/refer to orthopaedic team as indicated.
  - Weigh annually, and avoid excessive weight gain.
  - Offer contraceptive advice/contact details of organisations who can advise on contraception for people with learning disabilities.
  - Consider GnRH therapy for precocious puberty.

- **Hearing screening**
  - 11-18 years: screen for hyperacusis & high frequency hearing loss
  - If hyperacusis, implement a programme of desensitisation (plus maskers if necessary).

- **Screening for dental anomalies**
  - Ensure patient enrolled in an individualised preventative oral healthcare programme.
  - Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
  - Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.

- **Multidisciplinary developmental assessment**
  - Should involve local Child Development/ Learning Difficulties (LD) Teams.
  - Involve Child and Adolescent Mental Health Services (CAMHS) if necessary.
  - Ongoing review and support of learning and development with further assessment of special educational needs as required.
  - Referral to local Connexions service may be appropriate/helpful.

- **Behavioural & Mental Health issues**
  - Behavioural management advice/support for family as required.
  - Access to social skills training, and programmes to teach basic self help and daily living skills.
  - Refer for psychological intervention for anxiety, and when major life events.
  - **NB.** Apparent friendliness and sociability can mask depression and anxiety.

**High Anaesthesia**

*Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.*

*Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.*
### Recommendations for the management of Williams Syndrome

#### in adulthood (1)

<table>
<thead>
<tr>
<th><strong>Recommended Testing/Screening</strong></th>
<th><strong>Clinical Management Recommendations</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>• Serum creatinine</td>
<td>In all WS adults, test serum creatinine every 2—4 years.</td>
</tr>
<tr>
<td></td>
<td>Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.</td>
</tr>
<tr>
<td>• Serum Ca and Urine Ca: creatinine ratio</td>
<td>Test if symptomatic of hypercalcaemia.</td>
</tr>
<tr>
<td>• Thyroid Function Tests (TFTs)</td>
<td>If abnormal, investigate and manage as appropriate.</td>
</tr>
<tr>
<td></td>
<td>Measure TSH levels and if elevated, consider thyroid scanning, if compensated hypothyroidism present, refer to endocrinologist and monitor TFT and TSH annually.</td>
</tr>
<tr>
<td></td>
<td>If TSH level significantly low, consider thyroid replacement therapy.</td>
</tr>
<tr>
<td>• Renal screening</td>
<td>Bladder &amp; kidney ultrasonography every 5 years and if/when symptomatic.</td>
</tr>
<tr>
<td></td>
<td>If nephrocalcinosis refer to nephrologist for 6 monthly screening.</td>
</tr>
<tr>
<td>• Hypertension screening</td>
<td>Annual monitoring of blood pressure.</td>
</tr>
<tr>
<td></td>
<td>If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary.</td>
</tr>
<tr>
<td></td>
<td>NB. If RAS is present, angioplasty is not recommended due to elastinopathy.</td>
</tr>
<tr>
<td></td>
<td>If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out). Consider referral to renal specialist for care of adults with hypertension.</td>
</tr>
<tr>
<td>• Cardiac screening</td>
<td>Full assessment including scans, every 5 years throughout life.</td>
</tr>
</tbody>
</table>
Recommendations for the management of Williams Syndrome
~ in adulthood (2) ~

Recommended Testing/Screening

- Gastrointestinal issues
  - Enquire about bowel habit annually.
  - Treat constipation and consider investigating for diverticular disease.
- Screen for coeliac disease
  - If patient symptomatic.
- Screening for diabetes
  - At 30 years old: Oral Glucose Tolerance Test (OGTT), (or fasting insulin if considered more appropriate).
  - Repeat OGTT if rapid weight gain.
  - NB/ Do not use haemoglobin A1C as a screening tool.
  - Control impaired glucose tolerance with exercise & diet.
  - Avoid large glucose loads over a short time period.
  - Avoid diabetogenic drugs.
  - Manage clinical diabetes in WS in the same way as in general population.

- Growth & Sexual Health
  - Weigh annually, and avoid excessive weight gain—encourage an ‘active’ lifestyle.
  - Offer contraceptive advice/contact details of organisations who can advise on contraception for people with learning disabilities.
- Hearing screening
  - Every 10 years (for hearing loss and wax build-up).
- Screening for dental anomalies
  - Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
  - Missing teeth/malocclusion/other dental anomalies: refer to a consultant in Adult Restorative Dentistry or Special Care for multidisciplinary management.
- Behavioural & Mental Health issues
  - Access to support for employment, self help and independent living.
  - Social skills intervention as needed.
  - Refer for psychological intervention/support for anxiety, and when major life events.
  - NB. Apparent friendliness and sociability can mask depression and anxiety.

† Anaesthesia
Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.
Williams Syndrome Growth Charts

For Girls

... WEIGHT: 0–1 years old
... WEIGHT: 1–5 years old
... WEIGHT: 5–18 years old
... LENGTH: 0–1 years old
... HEIGHT: 1–5 years old
... HEIGHT: 5–18 years old
... OFC: 0–1 years old
... OFC: 1–5 years old

For Boys

... WEIGHT: 0–1 years old
... WEIGHT: 1–5 years old
... WEIGHT: 5–18 years old
... LENGTH: 0–1 years old
... HEIGHT: 1–5 years old
... HEIGHT: 5–18 years old
... OFC: 0–1 years old
... OFC: 1–5 years old

All growth charts are reproduced with the kind permissions of Harlow Printing Limited and Dr Neil Martin.

GIRL'S WEIGHT: (kg)
BIRTH - 1yr

NAME: ................................................
D.O.B. ........../........../ .........
**GIRL’S WEIGHT: (kg)**

1 - 5yrs

NAME: ________________________________
GIRL’S WEIGHT: (kg)
5 - 18yrs
With provision for school reception class
NAME: ........................................
D.O.B. ........../......./........

Williams Syndrome Clinical Management Guidelines
GIRL’S LENGTH: (cm)
BIRTH - 1yr

NAME: .................................................................
D.O.B. ........../........../.........

weeks/months

GIRLS 0–1 LENGTH
GIRL’S Height: (cm)
5 - 18yrs
With provision for school reception class
NAME: ........................................
D.O.B. ......../......../........
GIRL’S HEAD CIRC: (cm)
BIRTH - 1yr

NAME: ..................................................
D.O.B. ........../........./.........
GIRL’S HEAD CIRC: (cm)
1 - 5yrs
NAME:______________________________
BOY'S WEIGHT: (kg)
BIRTH - 1yr

NAME: .................................................................

D.O.B. ........../........../..........
BOY’S HEIGHT: (cm)
1 - 5yrs
NAME: _______________________________
BOY’S HEIGHT: (cm)
5 - 18yrs
With provision for school reception class
NAME: ........................................
D.O.B. ........../......./.......
BOY'S HEAD CIRC: (cm)
BIRTH - 1yr
NAME: ..................................................
D.O.B. ........../........../.........
Bibliography

**General papers & Guidelines**


**Anaesthesia**


**Calcium metabolism**

Bibliography continued...

Calcium metabolism continued...

Cardiovascular
Bibliography continued...

Cardiovascular continued...


Dental

- Fearne, J. "Dental Advice for Children with Williams Syndrome." UNPUBLISHED - from Williams Syndrome Foundation website: www.williams-syndrome.org.uk
Bibliography continued...

Dental continued...

Development, Behaviour and Mental Health

Endocrine & Thyroid
### Bibliography continued...

#### Endocrine & Thyroid continued...

#### Gastrointestinal & Feeding

#### Growth, Puberty and Sexual Health
Bibliography continued...

**Hearing**

**Orthopaedic**

**Renal & Hypertension**
Bibliography continued...

Renal & Hypertension continued...


Vision

Bibliography continued...

Other Resources

- Williams Syndrome Review Checklist
  This is aimed at carers involved in the care of adults with Williams Syndrome.
  It clearly states which tests and what screening should be undertaken at the GP surgery, and how often.
  It also lists symptoms which may present between GP appointments, and what to do should they occur.
  The checklist is currently being finalised, and will be available from Kay Metcalfe, Consultant Geneticist at St Mary’s Hospital in Manchester (kay.metcalfe@cmft.nhs.uk) when it is completed.

- Orphanet (www.orpha.net)
Orphanet is an online database of rare diseases and related services provided throughout Europe. It contains information on over 5,000 conditions, including Williams Syndrome, and lists specialised clinics, diagnostic tests, patient organisations, research projects, clinical trials and patient registries relating specifically to Williams Syndrome.

Resources for Patients

- Personal Health Record for Williams Syndrome (Blue Book)
  All babies in the UK are issued with a red book to record their health, growth and development. We have previously designed similar Blue Books for several rare conditions requiring multi-disciplinary management, including 22q11 Deletion Syndrome, Achondroplasia and Neurofibromatosis Type 1.
  As part of this project a Blue Book has been designed for people with Williams Syndrome.
  The primary aim of the Blue Book is to empower patients and their families, giving them more information about and ultimately more control over their health. It will also benefit the healthcare professionals involved in managing these patients, by facilitating inter-speciality communication, educating non-specialists and allied healthcare professionals, providing a readily accessible summary ‘snapshot’ of a patient’s condition, and they can also be used as a tool for clinical audit and research.
  They are available from the Williams Syndrome Foundation (see below)

- The Williams Syndrome Foundation UK (www.williams-syndrome.org.uk)
The Williams Syndrome Foundation is run for parents by parents. They aim to be the first point of contact for individuals with Williams Syndrome, their families, and professionals needing support and information regarding the Syndrome.
  The Foundation actively supports research into the educational, behavioural, social, scientific and medical aspects of the Syndrome, and seeks to organise their financial and personnel resources so as to achieve their mission on a sustainable basis.
Acknowledgements

- The Williams Syndrome Guideline Development Group
  Dr Jane Ashworth, Dr Susmito Biswas, Professor Bruno Dallapiccola, Dr Mark Dalzell, Dr Jane Deal, Professor Dian Donnai, Pam Griffiths, Dr Kay Hood, Professor Pat Howlin, Dr Ed Ladusans, Dr Ralph MacKinnon, Dr Josephine Marriage, Dr Neil Martin, Dr Kay Metcalfe, Dr Zulf Mughal, Dr Ramanlal Patel, Dr Alison Pike, Dr Christopher Stinton, Kate Strong, Dr Rajat Verma, Dr Mike Wolfman

- DYSCERNE: A Network of Centres of Expertise in Dysmorphology (www.dyscerne.org)

- The Williams Syndrome Foundation (www.williams-syndrome.org.uk)
  Regional Coordinators
  Professional Advisory Panel

- Nowgen—A Centre for Genetics in Healthcare (www.nowgen.org.uk)

These guidelines were produced thanks to funding from DYSCERNE: A Network of Centre of Expertise for Dysmorphology (funded by the European Commission Public Health Executive Agency (DG Sanco) Project: 2006122), and the Williams Syndrome Foundation UK.