Williams Syndrome Health Surveillance

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.

To: Dr
As you know, your patient has Williams Syndrome/ Infantile Hypercalcaernia 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) Svstematic review of symptoms with appropriate investigation.

Williams Syndrome

1 Eponyms: Idiopathic Infantile Hypercalcaemia Syndrome Williarns- Beuren Syndrome Fanconi's Syndrome Elfin Facies Syndrome

2) Definition: A sporadic congenital syndrome due to a microdeletion of chromosome 7 at the elastin gene focus. There is a typical facies and global developmental delay. There may be abnormalities of calcium metabolism and problems may occur in any of the major systems.

3) Incidence: 1 : 20,000 births (approx.) Males = Females

4) Professional Help: Primary Health Care Team Paediatrician: Developmental General Paediatric Surgeon Nephrologist Cardiologist/Cardiac Surgeon Ophthalmologist Geneticist Clinical Psychologist

5) Typical Diagnostic Features:

a) **Face** (most obvious in childhood) - Broad forehead - Medial eyebrow flare - Periorbital fullness - Strabismus - Stellate iris pattern - Flat nasal bridge - Malar flattening - Full checks and lips - Wide mouth

b) **Cardiac Lesions** (especially) Supravalvular Aortic Stenosis Peripheral Pulmonary Artery stenosis

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.

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 ot 'S.V.A.S. N. B. Antiobiotic cover for dental and surgical procedures. Unexplained fever, think SABE, 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%

6iv) 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 synostenosis: Kyphosils: 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 Hcalth Care Team, many of which may be involved (if available in your area) Communily Developmental Paediatrician

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

Information on claiming benefits is available by dialling 0800 666555 and asking for "FREEPHONE DSY' or writing to the Williams Syndrome Foundation.

Guidelines for parents, teachers and employers & supervisors are available from the registered office of the WS Foundation and are also reproduced online.

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: 9 Park Road New Bamet Herts EN4 3QA Tel. No: 0 181 440 6844

Email: Mikewolfy@talk21.com

Professional Advisory Panel:

Prof. W. Yule MA Dip.Psych. Dr. M. Wolfman MB, ChB Ms. J. Marriage BSc. MSc Prof. J. Atkinson PhD Mike Adlam MBA Dr. N. Martin MD, MRCP Prof. A. Karnriloff-Smith FBA MAE Prof.A.Redington M13,MBBS,MRCP Mrs S Morgan MA(hons) PGCE John Nelson ACIB
