

## Prader-Willi Syndrome (PWS): Multi-Disciplinary Paediatric Health Oversight

<b>Contents</b>	<b>Page</b>
Characteristics and need for multidisciplinary approach .....	2
Birth .....	3
0 - 12 months .....	4
1 - 5 years .....	5
5 - 11 years .....	6
11 - 17 years .....	7
17 years plus - adult services .....	8



Prader-Willi Syndrome is a rare genetic condition which is caused by an abnormality of chromosome 15.

**Characteristics of the syndrome include:**

- Hypotonia and major feeding difficulties in infancy which may require tube feeding
- Hyperphagia starting in pre-school years and leading to a lifelong tendency to excessive or extreme weight gain. However severe obesity is preventable through consistent management from a young age
- Hypogonadism and pubertal dysfunction; occasional precocious puberty
- Reduced height. Responsive to Growth Hormone which is licenced for PWS
- Delayed gross motor development, ongoing hypotonia and reduced exercise endurance
- Delay and disorder in social and emotional development and characteristic behaviours
- Variable cognitive development: Usually mild or moderate general learning disabilities; may have mainstream learning abilities or a more severe learning disability.
- Specific learning disabilities: eg slow processing speed; specific or general aspects of language ; executive functions; switching focus
- Complex behaviours: reduced flexibility; emotional lability; skin picking
- An increased incidence of Autistic Spectrum Disorder
- Increased incidence of scoliosis

**Other medical implications:** A high pain threshold, poor temperature regulation and lack of vomiting can mean that an injury or a serious medical problem may present in an unusual way and so could be overlooked if carers and professionals are not aware of this.

**Children and young people with PWS therefore require multi-professional advice and oversight from infancy onwards**

This should be provided by local services working in conjunction with either tertiary endocrine advice and reviews or with a specialist PWS clinic - although these are still limited in number. The current specialist Paediatric PWS clinics in the UK are: Brighton, Birmingham, Chelsea and Westminster, Glasgow and Dublin and there may be more.

The health oversight required from secondary and tertiary care professionals working jointly or in close liaison is summarised in age bands on the following pages..

**Age: Birth**

<b>Hospital Paediatrician</b>	<b>Paediatric Dietician</b>	<b>Child Development Team</b>	<b>Other Services</b>
<ul style="list-style-type: none"> <li>• Refer to genetics consultant</li> <li>• Refer to paediatric dietician</li> <li>• Consider specialist SALT re feeding</li> <li>• Refer to paediatric physiotherapy re hypotonia and motor delay</li> <li>• Review 3-4 months by paediatrician with endocrine and PWS expertise</li> <li>• <b>Information about PWS for families</b></li> <li>• <b>New Diagnosis Video</b></li> </ul>	<ul style="list-style-type: none"> <li>• Anticipate feeding difficulties</li> <li>• Anticipate faltering growth</li> <li>• <b>More dietary information</b></li> </ul>	<ul style="list-style-type: none"> <li>• Physiotherapist: hypotonia and motor delay</li> <li>• <b>More physiotherapy information</b></li> </ul>	

**Age: 0 - 12 months**

<b>Hospital Paediatrician</b>	<b>Paediatric Dietician</b>	<b>Child Development Team</b>	<b>Other Services</b>
<ul style="list-style-type: none"> <li>• Regular reviews by a local paediatrician with endocrine expertise linked to a tertiary endocrine PWS service</li> <li>• Assess growth and feeding</li> <li>• Genetics: subtype; any genetic questions</li> <li>• <b>Discussion about growth hormone therapy</b></li> <li>• Investigations prior to starting GH</li> <li>• Assess for respiratory symptoms and sleep apnoea</li> <li>• Undescended testes - consider referral to paediatric surgery</li> <li>• Assess for squint; scoliosis</li> <li>• Information about PWS features - feeding issues; may not vomit; high pain threshold; temperature regulation; refer to community development/child development team</li> <li>• <b>Leaflets for parents to give to professionals</b></li> <li>• Refer to community paediatrician</li> <li>• Liaise dietician, physiotherapist, health visitor, GP, other professionals</li> <li>• <b>Information about PWS for families</b></li> </ul>	<ul style="list-style-type: none"> <li>• Assess growth</li> <li>• Support catch up growth with caution to avoid overfeeding Support timely weaning</li> <li>• Ensure standard DOH vitamin supplementation</li> <li>• Consider important specific nutrients for weaning - iron</li> <li>• Education for families on nutritional needs and fundamental principles of diet in PWS</li> <li>• <b>More dietary information</b></li> </ul>	<ul style="list-style-type: none"> <li>• <b>Physiotherapist:</b> Hypotonia &amp; motor delay</li> <li>• <b>Consultant community paediatrician:</b></li> <li>• Developmental oversight</li> <li>• Aware of feeding needs and support</li> <li>• Refer to and liaise: physiotherapist, dietician, endocrine paediatrician, S &amp; L therapist as needed etc</li> <li>• <b>Information for health professionals</b></li> <li>• <b>Information about PWS</b></li> <li>• <b>More physiotherapy information</b></li> </ul>	

**Age: 1 - 5 years**

<b>Hospital Paediatrician</b>	<b>Paediatric Dietician</b>	<b>Child Development Team</b>	<b>Other Services</b>
<ul style="list-style-type: none"> <li>• Regular reviews by a paediatrician with endocrine expertise and a tertiary endocrine service for children with PWS</li> <li>• <b>Growth and Growth Hormone therapy</b></li> <li>• Undescended testes: referral</li> <li>• Assess for respiratory symptoms and sleep apnoea</li> <li>• Other medical: scoliosis; squint; vision</li> <li>• Medical investigations: endocrine, nutrition</li> <li>• <b>More dietary information</b></li> <li>• <b>Information about PWS</b> features incl: feeding, may not vomit, high pain threshold, temperature regulation, skin picking etc</li> <li>• Liaison with professionals: dietician, physiotherapist, community paediatrician, child development team, school SENCO etc</li> <li>• <b>Dental review</b></li> <li>• Provide detailed advice for EHCP</li> <li>• <b>EHCP medical input</b></li> <li>• Support DLA application</li> <li>• <b>Information about PWS for families</b></li> <li>• <b>Leaflets for parents to give to professionals</b></li> </ul>	<ul style="list-style-type: none"> <li>• Assess growth</li> <li>• Provide fundamental dietary intake information</li> <li>• Annual 24hr recall or weighed food diary assessments for nutritional adequacy</li> <li>• Review and discuss fundamental principles of diet in PWS</li> <li>• Annual biochemical assessment of nutrients to include : iron, zinc, Vit D, folate, B12</li> <li>• <b>More dietary information</b></li> </ul>	<ul style="list-style-type: none"> <li>• <b>Physiotherapist (PT) :</b> motor delay &amp; hypotonia, footwear , any need for orthotics</li> <li>• <b>More physiotherapy information</b></li> <li>• <b>Community paediatrician</b> Developmental oversight &amp; related referrals: <b>speech and language therapy</b> (SALT), OT, PT</li> <li>• Information and advice to parents and preschool: incl. PWS development, learning, behaviours, food supervision - <b>Information for Nurseries</b></li> <li>• EHCP medical advice</li> <li>• <b>EHCP medical input</b></li> <li>• <b>DLA information</b></li> <li>• School transition: meeting; health, advice</li> <li>• Links with hospital paediatrician , dietician and other professionals</li> </ul>	<ul style="list-style-type: none"> <li>• <b>Behavioural support and advice services</b></li> <li>• Special Educational needs</li> <li>• <b>EHCP medical input</b></li> <li>• IEP</li> </ul>

**Age: 5 - 11 years**

<b>Hospital Paediatrician</b>	<b>Paediatric Dietician</b>	<b>Child Development Team</b>	<b>Other Services</b>
<ul style="list-style-type: none"> <li>• Regular reviews by a local paediatrician with endocrine expertise and a tertiary endocrine service for PWS</li> <li>• <b>Growth and Growth Hormone therapy</b></li> <li>• Checks for precocious puberty</li> <li>• Medical investigations: endocrine, nutritional</li> <li>• <b>More dietary information</b></li> <li>• Assess for respiratory symptoms and sleep apnoea</li> <li>• Spine, feet, hypotonia, joint laxity, physiotherapy needs</li> <li>• Diet and feeding review with dietician</li> <li>• <b>Diet</b> and <b>Exercise</b> advice</li> <li>• Liaison with other professionals physiotherapist, dietician, community paediatrician, child development team, School SENCO (<i>special needs co-ordinator</i>)</li> <li>• EHCP reports when requested</li> <li>• <b>EHCP medical input</b></li> <li>• <b>Dental review</b></li> <li>• <b>Information about PWS for families</b></li> <li>• <b>Leaflets for parents to give to professionals</b></li> </ul>	<ul style="list-style-type: none"> <li>• Assess growth</li> <li>• Provide fundamental dietary intake information</li> <li>• Annual 24hr recall or weighed food diary assessments for nutritional adequacy</li> <li>• Review and discuss fundamental principles of diet in PWS</li> <li>• Annual biochemical assessment of nutrients to include: iron, zinc, Vit D, folate, B12</li> <li>• <b>More dietary information</b></li> </ul>	<ul style="list-style-type: none"> <li>• <b>Consultant community paediatrician:</b> Developmental reviews; referrals</li> <li>• <b>Behavioural information and sources of advice and help</b></li> <li>• Spine; vision; hearing</li> <li>• <b>EHCP for families</b></li> <li>• Liaison with: hospital /endocrine paediatrician; dietician, school , other professionals</li> <li>• Advice to transition meetings: <b>reception class and senior school</b></li> <li>• <b>Physiotherapy:</b> periodic reviews hypotonia: posture; ankles; orthotics; spine</li> <li>• <b>More physiotherapy information</b></li> <li>• <b>Exercise plans:</b> to home, school</li> <li>• <b>OT</b> - as needed</li> <li>• Consider <b>SALT</b> referral</li> <li>• <b>Skin picking information</b></li> <li>• <b>Information for schools</b></li> </ul>	<ul style="list-style-type: none"> <li>• Special Educational Needs processes:</li> <li>• <b>EHCP medical input</b></li> <li>• Educational psychology (EP) assessments: with reference to possible specific as well general learning disabilities</li> <li>• <b>Behavioural support and advice services</b> and <b>CAMHS</b></li> </ul>

**Age: 11 - 17 years**

<b>Hospital Paediatrician</b>	<b>Paediatric Dietician</b>	<b>Child Development Team</b>	<b>Other Services</b>
<ul style="list-style-type: none"> <li>• Regular reviews by a local Paediatrician with Endocrine expertise and a tertiary PWS Paediatric Endocrine service</li> <li>• <b>Growth and Growth Hormone therapy</b></li> <li>• Pubertal onset, progress and induction if needed</li> <li>• Spine re scoliosis; ankles</li> <li>• Medical investigations: endocrine, nutritional</li> <li>• <b>More dietary information</b></li> <li>• <b>Diet</b> and <b>Exercise</b> advice</li> <li>• Puberty and growing Up - Information for young person - <i>available shortly from PWSA</i></li> <li>• Young person's questions</li> <li>• Emotional / mental health issues and possible referrals</li> <li>• Information shared with school SENCO, dietician, physiotherapist, community paediatrician</li> <li>• <b>EHCP medical input</b></li> <li>• <b>Transition planning and medical transition</b></li> <li>• <b>Information about PWS for families</b></li> <li>• <b>Leaflets for parents to give to professionals</b></li> </ul>	<ul style="list-style-type: none"> <li>• Assess growth</li> <li>• Provide fundamental dietary intake information</li> <li>• Annual 24hr recall or weighed food diary assessments for nutritional adequacy</li> <li>• Review and discuss fundamental principles of diet in PWS as adult</li> <li>• Annual biochemical assessment of nutrients to include : iron, zinc, Vit D, folate, B12</li> <li>• Identification of adult dietitian for transition</li> <li>• <b>More dietary information</b></li> </ul>	<ul style="list-style-type: none"> <li>• <b>Community Paediatrician</b> <ul style="list-style-type: none"> <li>• Periodic reviews of developmental</li> <li>• social &amp; behavioural progress.</li> <li>• EHCP updates and meetings</li> <li>• School transitions : implications of PWS</li> <li>• <b>PWSA Information for Secondary Schools</b></li> <li>• Links: Hospital paediatrician and dietician; behavioural and mental health services</li> <li>• Periodic reviews: hypotonia, posture, spine and whether need for orthotics - <b>More physiotherapy information</b></li> <li>• <b>Exercise plans</b> for school and home</li> </ul> </li> <li>• Update <b>advice for EHCP</b> : all professionals</li> <li>• <b>OT, SALT</b> reviews and advice: as needed</li> <li>• Growing Up with PWS - <i>Information for young person - available shortly from PWSA</i></li> </ul>	<ul style="list-style-type: none"> <li>• Emotional, behavioural and mental health services who access PWS information or have PWS expertise: ie local / tertiary</li> <li>• <b>Behavioural support and advice services</b> and <b>CAMHS</b></li> <li>• <b>EHCP medical input</b></li> <li>• <b>Mental health</b></li> <li>• <b>Transition</b></li> </ul>

## 17 Years Plus - Adult Services

- Endocrine and weight: Adult Endocrinologist; working with a local Dietician and with tertiary PWS advice if needed
- Diet, food supervision and food environment: Dietician working in conjunction with Endocrinology, other professionals and family
- Challenging Behaviour - Challenging Behaviour and LD **NICE Guidelines**
- Mental Health Services: local and tertiary expertise - LD **NICE Guidelines**
- **The mental health of people with PWS with a specific focus on psychotic illness**
- **Mental capacity issues**
- Physiotherapy assessments and advice as needed **More physiotherapy information**
- **Educational and learning needs and EHCP to 25 years**
- **Social Care: including residential needs, respite care, financial**