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

<table>
<thead>
<tr>
<th>Contents</th>
<th>Page</th>
</tr>
</thead>
<tbody>
<tr>
<td>Characteristics and need for multidisciplinary</td>
<td>2</td>
</tr>
<tr>
<td>approach ............................................</td>
<td></td>
</tr>
<tr>
<td>Birth ................................................................</td>
<td>3</td>
</tr>
<tr>
<td>0 - 12 months ..........................................</td>
<td>4</td>
</tr>
<tr>
<td>1 - 5 years ............................................</td>
<td>5</td>
</tr>
<tr>
<td>5 - 11 years ...........................................</td>
<td>6</td>
</tr>
<tr>
<td>11 - 17 years ..........................................</td>
<td>7</td>
</tr>
<tr>
<td>17 years plus - adult services ........................</td>
<td>8</td>
</tr>
</tbody>
</table>
Information for health professionals:
Prader-Willi Syndrome is a rare genetic condition which has multiple medical, developmental, learning, behavioural and emotional implications. This myriad of issues overlap and so children require a multi-disciplinary approach. The following pages summarise the key issues for routine consideration by health professionals. These are in age bands.

PWS characteristics which have ongoing health implications may include:
- Hypotonia
- Feeding difficulties in infancy often requiring tube feeding
- Lifelong hyperphagia emerges and can cause extreme weight gain. Dietary restrictions are compounded by reduced calorie requirements
- Multiple endocrinopathies including Growth Hormone insufficiency and hypogonadism commonly
- Longstanding reduced exercise endurance
- Scoliosis
- Squint
- Dysfunctions in temperature regulation, pain threshold, vomiting (may be absent); central and obstructive apnoea
- Secondary implications for incidental illness and health care: interpretation of symptoms; in general anaesthesia; surgery
- Delay and disorder in motor, social, emotional and language development
- Mild, moderate or sometimes severe cognitive disabilities and specific learning disabilities e.g. slow processing of information; disordered executive functions; difficulties switching between activities;
- Complex behaviours and mental health risks: e.g. severe emotional lability and outbursts; skin picking; major anxiety; adult psychoses

Medical alert: A medical problem in a person with PWS may present in an atypical way and could be overlooked if carers and professionals are not aware of this possibility e.g. pain in accidents or illness; temperature accuracy in illness; lack of vomiting in serious abdominal conditions; management of general anaesthesia and surgery. Leaflets for parents to give to professionals are available on this website

Multi-Disciplinary advice and review throughout childhood: Care should be co-ordinated by a paediatrician with an interest in endocrinology with access to a tertiary Paediatric Endocrinologist or a tertiary specialist PWS clinic and include regular reviews by a Paediatric Dietician and members of a Child Development Team plus access to behavioural and emotional advice, local and specialist mental health services. Genetic information and advice early. Links between health professionals and schools.

Tertiary and specialist PWS clinics include Birmingham, Brighton, Chelsea and Westminster, Glasgow, Staffordshire & more are developing
### Age: Birth - early weeks

- Information to parents about PWS. [www.pwsa.co.uk/new-diagnosis](http://www.pwsa.co.uk/new-diagnosis), [www.pwsa.co.uk/family-life](http://www.pwsa.co.uk/family-life) and [https://www.pwsa.co.uk/Healthandsocialcare](https://www.pwsa.co.uk/Healthandsocialcare)
- Feeding: anticipation of difficulties related to hypotonia & sucking etc. May need NGT. Will benefit from early feeding expertise and Dietician involvement
- Refer to Genetics Consultant
- Refer to paediatric physiotherapy re hypotonia and motor development
- Arrange follow up by local Paediatrician with Endocrine expertise and a Specialist PWS clinic if available
- Inform Child Development Team
## PWS Multidisciplinary health care age: 0 – 12 months

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<th>Hospital Paediatrician with endocrine interest &amp; access to a tertiary Endocrinologist /PWS clinic</th>
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<tr>
<td>• Growth and feeding, in liaison with dietician and other infant feeding professionals</td>
<td>• Specific caution to <strong>avoid overfeeding if tube fed</strong></td>
<td>• Physiotherapist : Hypotonia; motor delay</td>
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<td>• Sleep apnoea &amp; respiratory symptoms</td>
<td>• Provide PWS specific dietary advice <a href="https://example.com">UsefulinformationfordietitiansmanagingPWSpatients.pdf</a></td>
<td>• Consultant Community Paediatrician: Developmental oversight</td>
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<td>• Undescended testes: optimal surgery pre school</td>
<td>• Standard DOH vitamin supplementation</td>
<td>• Information for parents <a href="https://example.com">www.pwsa.co.uk/family-life</a></td>
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<td>• Squint. Start clinical examinations for scoliosis</td>
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<td>• Genetic subtype &amp; parental genetic questions</td>
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<td>• Discussion about Growth Hormone ( GH)</td>
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<td>• GH initiated by a specialist Endocrinologist - 1 year</td>
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<td>• Investigations prior to starting GH: incl level 2 sleep studies</td>
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<td>• Information: PWS implications for general health: feeding patterns early and later; may not vomit; temperature regulation; high pain threshold etc.</td>
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<td>• Parent information on PWSA web site <a href="https://example.com">www.pwsa.co.uk</a></td>
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<tr>
<td>• Leaflets about PWS for parents to give to professionals’</td>
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<td>• Multi-disciplinary needs: liaise with physiotherapist &amp; child development team, dietician, Health Visitor, GP</td>
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*Leaflets : [www.pwsa.co.uk/informationforhospitalandA&Estaff](https://example.com) & [www.pwsa.co.uk/PWStipsformedicalprofessionals](https://example.com)*
## PWS Multidisciplinary health care age: 1 – 5 years

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| • Growth  
• Diet & approaches to food in all settings - with Dietician  
• Growth Hormone: Information. Initiated by endocrine specialist from 1 year onwards  
• Review respiratory symptoms and sleep apnoea - repeat sleep studies 3/12 after starting GH / if symptoms  
• Blood tests: incl IgF1, TFT plus. Annual nutritional. Periodic Bone age  
• Testes: surgery before school age  
• Scoliosis: clinical examinations not routine X Rays  
• Impact of hypotonia on ankles.  
• Squint.  
• Parent information about PWS: vomiting, pain, temperature regulation, skin picking, behaviour  
• Leaflet for parents to give to professionals *  
• Parents: PWSA information & support **  
• Advise Dental reviews & vision tests  
• EHCP: If requested provide detailed written information about health implications of PWS in school  
• DLA: Provide written information if requested | • Regular reviews of diet & advice about all food management  
• Information on principles of diet & anticipating food management in PWS [UsefulInformationfordietitiansmanagingPWSpatients.pdf](#)  
• Consider annual food diary: 3 day or 24hr recall for nutritional adequacy  
• Annual nutritional: blood test: iron, zinc, Vit D, folate, B12 Multivitamins | • Physiotherapist: include implications of gross motor delay & disorder, ongoing hypotonia, footwear, orthotics. Scoliosis. Provide EHCP advice  
• Community paediatrician EHCP and Nursery -school transition: detailed implications of PWS (Food management; development inc social & behavioural, motor disorder; specific & general learning; health (pain threshold, temperature regulation).  
• DLA: details of impact on exercise tolerance, resistance, outbursts, safety  
• Occupational therapy: fine motor  
• Speech & Language: specific disorders common / general disorder  
• Advise parents & schools of information on PWSA UK website |

*Leaflets: [www.pwsa.co.uk/informationforhospitalandA&Estaff](#) & [www.pwsa.co.uk/PWStipsformedicalprofessionals](#)
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| • Growth monitoring  
• Growth Hormone therapy by specialist  
• Diet and all food management in liaison with dietician  
• Respiratory symptoms and sleep apnoea  
• Examine for scoliosis, not routine X rays  
• Ankles: impact of hypotonia & orthotic needs  
• Medical investigations: Endocrine: incl IGF1: Bone age Nutritional.  
• Review exercise  
• Skin picking  
• Leaflets for parents to give to health professionals  
• Information and support from PWSA UK **  
• EHCP reports when requested: on health implications of PWS: food management, exercise, pain, temperature etc. | • Review diet and management of all food  
• Information on principles of diet & food management in PWS: [Usefulinformationfordietitiansmanaging PWSpatients.pdf](#)  
• Consider annual food diary - 24hr recall or 3 day for nutritional adequacy  
• Consider Multivitamins  
• Annual blood test including: iron, zinc, Vit D, folate, B12 | • **Physiotherapy**: periodic reviews of impact of hypotonia: ankles; orthotics; spine; exercise advice & programmes for EHCP/school  
• **Consultant community paediatrician**: Contribute to transition meetings EHCP: address food; social, motor, language, behavioural, specific & general learning, temperature, pain, health  
• **SALT** specific speech & language disorders  
• **OT** fine motor  
• Advise parents & schools about information on PWSA website |

*Leaflets: [www.pwsa.co.uk/informationforhospitalandA&Estaff](#) & [www.pwsa.co.uk/PWStipsformedicalprofessionals](#)*

**Parent information: [www.pwsa.co.uk/family-life ](#)  
**Information [www.pwsa.co.uk/family-life](#)
## PWS Multidisciplinary care age: 12 – 17 years

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| • Growth monitoring  
  • Growth hormone therapy by specialist  
  • Diet. Wider food management & anticipation of possible increasing independence in food access  
  • Puberty: onset, progress; induction by specialist if needed  
  • Examine for scoliosis & kyphosis: not routine X rays  
  • Ankles & impact of hypotonia, orthotics  
  • Medical investigations: Endocrine include IGF1, TFT, FSH, LH, Testosterone / Oestradiol; Hb A1C, LFT if high BMI. Nutritional. Bone age.  
  • Exercise & physical activity  
  • Young person’s questions (16+): plus suggest www.pwsa.co.uk/informationtofamilies/HealthyFuturesPack  
  • Behaviour and mental health & sources of advice  
  • Sexual health, pregnancy & consider contraceptive advice  
  • Plan transition to adult services : Endocrine & Dietician  
  • Leaflets for parents to give to health professionals | • Review diet and all food management  
  • Information on food management in PWS: [UsefulInformationfordietitiansmanagingPWSpatients.pdf](#)  
  • Consider food diary 24hr recall / 3 day & assess nutritional adequacy  
  • Discuss potential access to food (& money for food) and increased risk of weight gain in adolescence  
  • Liaise with school/college  
  • Consider Multivitamins  
  • Annual blood test: include iron, zinc, Vit D, folate, B12  
  • Identify adult dietitian at transition | • Physiotherapy - Periodic reviews because of on going hypotonia, low muscle strength & tendency to reduced exercise; orthotics; posture, spine  
  • Exercise programme in EHCP; ideas for home  
  • **Community Paediatrician** - EHCP updates and school meetings: social development, behavioural, mental health, food management in adolescence ( dilemmas & risks of weight gain); exercise requirements  
  • **Mental health services**  
  • PWS specific emotional and behavioural facts and phenotype  
  • Specific mental health risks and disorders  
  • Contribute to school transitions implications of PWS for schools  
  • Advise parents & schools about information on PWSA UK website |

[www.pwsa.co.uk/informationforhospitalandA&Estaff](#) & [www.pwsa.co.uk/PWStipsformedicalprofessionals](#)
17 Years Plus – Adult Services required

- **Adult Endocrinologist working with a local Dietician** and accessing Tertiary Endocrine PWS advice if needed. Sexual health and contraceptive advice
- **Dietician** advice to carers and young person on food and restrictions around all food access, supervision and a secure food environment on health grounds. Liaison with social care re **Deprivation of Liberty** (DOL) assessments.
- **Physiotherapy** reviews and advice as needed especially of orthotics & exercise programmes
- **EHCP and Educational and learning needs and resources**: Local Authority responsible for EHCP to 25 years and information about specialist colleges & supervised work settings
- **Social Care**: respite, residential care, financial, PIP, mobility.
- **Mental capacity. Deprivation of Liberty** assessments.
- **Challenging Behaviour and mental health services**: adult mental health team plus access to specialist tertiary expertise, advice and joint working. Refer to Challenging Behaviour and Learning Disability NICE Guidelines
- **PWSA UK**: [www.pwsa.co.uk/information-for-families/adultswithPWS](http://www.pwsa.co.uk/information-for-families/adultswithPWS)