**CASE DEFINITION FOR PRADER-WILLI SYNDROME**

Report any child up to and including 18 years of age with newly diagnosed Prader-Willi syndrome confirmed clinically and/or genetically (methylation and/or FISH [fluorescent in situ hybridization] test).

A diagnosis of PWS should be strongly suspected in children less than three years of age with a score of five points (four from major criteria) or in patients over three years of age with eight points (five from major criteria).

**PWS clinical score:**
- < 3 years: 5 points (4 from major criteria)
- > 3 years: 8 points (5 from major criteria)

A clinical diagnosis of PWS relies on a score derived from the following major and minor criteria:

<table>
<thead>
<tr>
<th>Major criteria (1 point each)</th>
<th>Minor criteria (½ point each)</th>
<th>Supportive criteria (no points)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Infantile central hypotonia</td>
<td>Decreased fetal movement and infantile lethargy</td>
<td>High pain threshold</td>
</tr>
<tr>
<td>Infantile feeding problems/failure to thrive</td>
<td>Typical behavioral problems (temper tantrums, stubbornness, stealing/begging for food, anxiety regarding food, food seeking, perseveration about food)</td>
<td>Infrequent vomiting</td>
</tr>
<tr>
<td>Rapid weight gain between 1 and 6 years</td>
<td>Sleep disturbances / sleep apnea</td>
<td>Temperature control problems</td>
</tr>
<tr>
<td>Characteristic facial features</td>
<td>Short stature for the family by 15 years of age</td>
<td>Scoliosis/kyphosis</td>
</tr>
<tr>
<td>Hypogonadism: genital hypoplasia, abnormal sexual development</td>
<td>Hypopigmentation for family</td>
<td>Osteoporosis</td>
</tr>
<tr>
<td>Developmental delay</td>
<td>Small hands and feet for height age</td>
<td>Unusual skill with jigsaw puzzles</td>
</tr>
<tr>
<td>Hyperphagia/obsession with food</td>
<td>Narrow hands with straight ulnar border</td>
<td>Normal neuromuscular studies</td>
</tr>
<tr>
<td>Cytogenetic/molecular abnormality of Prader-Willi chromosome region</td>
<td>Esotropia, myopia</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Thick, viscous saliva</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Speech articulation defects</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Skin picking</td>
<td></td>
</tr>
</tbody>
</table>

**SECTION 1 – DEMOGRAPHIC INFORMATION**

1.1 Date of birth: ____ /_____ /_______  1.2 Sex:   Male ___  Female ___ 

1.3 Date of child’s diagnosis of Prader-Willi syndrome:  ____ /_____ /_______

**SECTION 2 – DIAGNOSTIC INVESTIGATION RESULTS**

2.1 Karyotype: Normal ___ Abnormal ___ Unknown ___ 

If abnormal, please specify: 

<table>
<thead>
<tr>
<th>n-methylation test:</th>
<th>FISH (fluorescent in situ hybridization) test:</th>
</tr>
</thead>
<tbody>
<tr>
<td>Yes ___ Date ____ /_____ /_______</td>
<td>Yes ___ Date ____ /_____ /_______</td>
</tr>
</tbody>
</table>

No ___ Unknown ___  No ___ Unknown ___
SECTION 3 – MEDICAL HISTORY

3.1 Ethnicity (mother): Asian ___ Afro-Canadian ___ Caucasian ___ Other _____________________________
3.2 Ethnicity (father): Asian ___ Afro-Canadian ___ Caucasian ___ Other _____________________________
3.3 Anthropometric data:

<table>
<thead>
<tr>
<th></th>
<th>Height (cm)</th>
<th>Weight (kg)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Father</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Mother</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Patient (actual)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

SECTION 4 – HISTORICAL AND PHYSICAL FINDINGS (major criteria – 1 point each)

4.1 Infantile central hypotonia: Yes ___ No ___ N/A ___
4.2 Infantile feeding problem / failure to thrive: Yes ___ No ___ N/A ___
4.3 Rapid weight gain between 1 and 6 years: Yes ___ No ___ N/A ___
4.4 Characteristic facial features: Yes ___ No ___ N/A ___
4.5 Hypogonadism – genital hypoplasia, abnormal sexual development: Yes ___ No ___ N/A ___
4.6 Developmental delay: Yes ___ No ___ N/A ___
4.7 Hyperphagia/obsession with food: Yes ___ No ___ N/A ___
4.8 Cytogenetic/molecular abnormality of Prader-Willi chromosome region: Yes ___ No ___ N/A ___

SECTION 5 – HISTORICAL AND PHYSICAL FINDINGS (minor criteria – ½ point each)

5.1 Decreased fetal movement and infantile lethargy: Yes ___ No ___ N/A ___
5.2 Typical behaviour problems (temper tantrums, stubbornness, stealing/begging for food, anxiety regarding food, food seeking, perseverance about food): Yes ___ No ___ N/A ___
5.3 Sleep disturbances/sleep apnea: Yes ___ No ___ N/A ___
5.4 Short stature for family by 15 years of age: Yes ___ No ___ N/A ___
5.5 Hypopigmentation for family: Yes ___ No ___ N/A ___
5.6 Small hands and feet for height age: Yes ___ No ___ N/A ___
5.7 Narrow hands with straight ulnar border: Yes ___ No ___ N/A ___
5.8 Esotropia, myopia: Yes ___ No ___ N/A ___
5.9 Thick, viscous saliva: Yes ___ No ___ N/A ___
5.10 Speech articulation defect: Yes ___ No ___ N/A ___
5.11 Skin picking: Yes ___ No ___ N/A ___

SECTION 6 – HISTORICAL AND PHYSICAL FINDINGS (supportive criteria – no points)

6.1 High pain threshold: Yes ___ No ___ N/A ___
6.2 Infrequent vomiting: Yes ___ No ___ N/A ___
6.3 Temperature control problem: Yes ___ No ___ N/A ___
6.4 Scoliosis/kyphosis: Yes ___ No ___ N/A ___
6.5 Osteoporosis: Yes ___ No ___ N/A ___
6.6 Unusual skill with jigsaw puzzles: Yes ___ No ___ N/A ___
6.7 Normal neuromuscular studies: Yes ___ No ___ N/A ___
SECTION 7 – MANAGEMENT

7.1 Is the child currently being followed by one or more subspecialists? Yes ___ No ___ Unknown ___

If yes, please indicate by which of the following subspecialists:

- Occupational therapist: Yes ___ No ___
- Physiotherapist: Yes ___ No ___
- Speech therapist: Yes ___ No ___
- Special educator: Yes ___ No ___
- Nutritionist: Yes ___ No ___
- Gastroenterologist: Yes ___ No ___
- Psychologist: Yes ___ No ___
- Psychiatrist: Yes ___ No ___
- Ophthalmologist: Yes ___ No ___
- Endocrinologist: Yes ___ No ___
- Other (please specify): ____________________________________________________________________

SECTION 8 – MEDICATIONS (including growth hormone)

8.1 Is the child presently on any medication? Yes ___ No ___ Unknown ___

If yes, please specify: ____________________________________________________________________

SECTION 9 – REPORTING PHYSICIAN

First name ___________________________ Surname ___________________________

Address ________________________________

City ___________________________ Province/Territory ___________________________ Postal code ___________________________

Telephone number ___________________________ Fax number ___________________________

E-mail ___________________________ Date completed ___________________________

Thank you for completing this form.

(PWS 2003-01)