

May 2005

# HEALTH ALERT

## PRADER-WILLI SYNDROME

### OVERVIEW

### PRADER-WILLI SYNDROME

The Ontario Prader-Willi Syndrome Association serves the needs of individuals with PWS, their families and caregivers. We are a non-profit association established in 1982. The individual that you are assessing has Prader-Willi Syndrome and we have provided you with materials that we hope will be helpful in your decision-making process. If we can answer any other questions, please don't hesitate to contact our office.

Prader-Willi Syndrome is a genetic disorder with an incidence of 1:12,000 births. It presents with many medical and behavioural complications that are not well understood because of the rarity of the condition. The most life-threatening of these are related to obesity because of hyperphagia-an overwhelming sense of hunger and lack of satiation that is present at all times.

When combined with a diminished I.Q., learning difficulties and other behavioural/psychiatric conditions which are also characteristic of those with PWS, it is unrealistic to expect self-management by the individual with PWS. Their impulse is to seek food even though they are often quite eloquent in being able to express how sickly they feel because of their increasing obesity and how anxious they are to take charge of the situation. Repeated experiences show that this is not realistic and that all individuals with PWS require support and supervision in order to control their weight.

Ideally, providing a situation in which the environment is structured to be "food friendly" through a controlled diet as well as structured exercise program is the most effective tool to prevent these deaths from happening. However, providing this structure can be challenging and at some point in

their lives many individuals will experience severe medical problems related to obesity which can be life-threatening. At present, there are no long term medical options for treatment in Ontario. Currently, individuals with PWS who are deemed to be in crisis are sent to a specialized PWS program in Pittsburgh. However, access to this program can be quite daunting and depends on having a viable living situation to return to which can also be very limited.

### EMERGENCY MEDICAL CONDITIONS

### CARDIO-PULMONARY DETERIORATION

"The insidious nature of cardiopulmonary deterioration in persons with PWS whose weight has reached the obese range is evident by the fact that cor pulmonale and hypoxia are frequently undiagnosed. Cardiorespiratory decompensation often occurs seemingly without warning in patients who appeared

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to be tolerating their obesity without imminent risk.<sup>1</sup> It is the current opinion of the Ontario Prader-Willi Syndrome Association that all obese individuals presenting for medical treatment, especially those presenting for emergency care be examined for cor pulmonale and hypoxia prior to release in order to prevent sudden death. Diagnosis can be made with a chest x-ray or echocardiogram which would show an enlargement of the central pulmonary artery. A sleep study should also be performed to rule out obstructive sleep apnea which can be a cause of sudden death in this population.

## **ACUTE GASTROPARESIS**

Persons with PWS are subject to weakness in the stomach muscles because of low muscle tone. There can be a delay in emptying or slowing of stomach contraction and as stomach contents build up abdominal distension occurs and can stretch to the point that blood supply is cut off causing necrosis of tissue. This condition often pres-

ents in PWS patients with vague central abdominal discomfort, pain, heartburn or vomiting as well as bloating of the abdomen. It may be a direct result of binge eating and the individual does not have to be obese to be affected. Also, individuals with PWS show a high pain threshold so that level of complaint may not be indicative of the seriousness of the condition.

Possible treatment options include a gastric or stomach emptying test, an x-ray or CT scan of the abdomen to look for abdominal distention & endoscopy. Emergency surgery may be required to rule out necrosis and quick action necessary to prevent death.

## **INFORMATION REGARDING HOSPITAL ADMISSION**

It is imperative that any medical admission take into account the food foraging behaviours of the individual with PWS and provide appropriate supervision otherwise further weight gain will occur. OPWSA does take the position that that families

make every effort to take steps so that they are able to provide appropriate controls for an individual in crisis including access to a decision making process through a power of attorney and/or the substitute decision-making acts for both property and health. As well, families may need to seek guardianship over health care and financial control and may require your cooperation in this important process. We hope that you will be able to assist them so that the individual with PWS can have the best quality of life possible.



## **The Ontario Prader-Willi- Syndrome Association**

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