FACT SHEET

Prader-Willi Syndrome

What is Prader-Willi Syndrome?

Prader-Willi Syndrome (PWS) is a genetic disorder that occurs in approximately 1 out of every 15,000 births. PWS affects males and females with equal frequency and affects all races and ethnicities. The syndrome is recognized as the most common genetic cause of obesity.

The symptoms of PWS are caused by dysfunction of a portion of the brain called the hypothalamus. The hypothalamus is a small endocrine organ at the base of the brain that plays a crucial role in many bodily functions such as hunger and satiety, temperature regulation, vomiting, fluid balance, puberty, and fertility.

How is it manifested?

Infants:
- hypotonia (weak muscle tone)
- difficulty with feeding because of poor sucking ability
- delayed motor & language development
- respiratory difficulties

Children and Adults:
- short stature, small hands and feet (without growth hormone treatment)
- intellectual impairment
- learning deficits such as poor short term memory, difficulty with auditory discrimination
- increased risk of obesity because of a persistent sense of hunger and lack of satiation (hyperphagia), low metabolic rate (60%) and high fat-to-muscle ratio
- serious health problems if weight is not controlled (diabetes, cardiac & respiratory complications)
- behavioural problems

Who is affected?

The current incidence of Prader-Willi Syndrome is 1 in 15,000 live births.

How is it diagnosed or detected?

A suspected diagnosis of PWS is usually made by a physician based on clinical symptoms. The diagnosis is then confirmed by a blood test. Families who are seeking a diagnosis or who have concerns about the risks should work with a genetics specialist who is knowledgeable about PWS and the latest in testing. The geneticist will arrange to have blood samples sent to an appropriate laboratory for testing.

Additional Resources:

ONTARIO PRADER-WILLI SYNDROME ASSOCIATION - http://members.allstream.net/~opwsa
The Ontario Prader-Willi Syndrome Association (OPWSA) is a non-profit charity. Their mission is to enhance the quality of life for individuals with Prader-Willi Syndrome. This website contains a source of information for those interested in and affected by Prader-Willi Syndrome.

CANADIAN PRADER-WILLI SYNDROME ORGANIZATION (CPWSO) – www.pwsacanada.com
CPWSO is a national, charitable association, dedicated to serving individuals affected by Prader-Willi Syndrome (PWS), their families and interested professionals. Some of the organization objectives include promoting broader geographic interest in PWS, seek increased diagnosis in infancy, to liaise with the International Prader-Willi Syndrome Organization and with national affiliates around the world who share our goals.

MY CHILD HAS BEEN DIAGNOSED WITH PRADER-WILLI SYNDROME - WHAT NOW?
This is a leaflet put out by the Prader-Willi Syndrome Association of USA, for parents with a new diagnosis.
www.pwsausa.org/Brochure/nowwhat.htm