A MEDICAL REFERENCE GUIDE

For Parents and Caregivers

What Is Prader-Willi Syndrome?

A disorder of chromosome 15

Occurrence in the Population: 1:12,000 to 1:15,000 (both sexes, all races)

Major Characteristics: low muscle tone, poor growth in early infancy, small external sexual organs, excessive eating developing in early childhood which could lead to obesity if not externally controlled, learning difficulties and difficult behaviors

Major Medical Concern:
Extreme obesity and consequences relate to over-eating.

Cause and Diagnosis of PWS

Prader-Willi syndrome (PWS) is caused by a loss of genes on chromosome 15 normally contributed by the father (paternal). This can occur in three ways: (1) approximately 70% of PWS cases have a deletion of part of the long arm (15q11-q13 region) of chromosome 15, thus missing these genes on the paternal side of chromosome 15; (2) approximately 25% have maternal uniparental disomy (UPD), meaning two chromosome 15s from the mother (maternal) and no paternal chromosome 15; and (3) 2-5% have an error in the "imprinting" process that makes these genes from the paternal chromosome 15 to be present but nonfunctional.

PWS is not inherited, but occurs by chance, and there is no known cause. The reoccurrence of PWS in the same family happens only in rare cases of the "imprinting" error referred to above. All children in whom PWS is suspected should have genetic testing, which is very accurate using current techniques. If diagnosis is confirmed, it is recommended that the family receive genetic counseling.

Weight and Behavior

Weight

Individuals with PWS have excessive appetite due to failure of the brain to tell them when they are full. Overeating and obsession with food usually begin before age 6. It is important to understand that this urge to eat is an actual physical disorder, overwhelming, difficult to control and requiring almost constant supervision.

Individuals with PWS require fewer calories due to their low muscle tone, low metabolism and inactivity. To create the proper calorie diet, seek out a registered nutritionist / dietitian who is familiar with the syndrome or willing to learn. Regular weigh-ins and a periodic diet review are needed. The best meal and snack plan is one that the family and caregiver are able to apply routinely and consistently. Weight control depends on restricting food from the individual with PWS and may require locking the kitchen and food storage areas. Daily exercise (at least 30 minutes a day) is a must for weight control and health. To date, no medication or surgeries have been found that would eliminate the need for strict dieting and supervision around food. Growth hormone treatment, because it increases muscle mass, may allow for a higher daily calorie intake and a better quality of life.

Behavior

Infants and young children with PWS are usually happy and loving, showing few behavioral problems. Most older children and adults, however, do have difficulties with behavior control, often happening when going from one activity to the next or with unanticipated changes. These behavioral problems usually begin at about the same time as the obsession with food, though not all behavioral problems are food related. Behavioral difficulties usually peak in adolescence or early adulthood. Daily routines and structure, fi rm rules and limits, "time out," and positive rewards work best for behavior management.

Developmental Concerns

Physical Development

Average developmental milestones are typically delayed one or two years. Although the low muscle tone improves, problems with strength, coordination and balance may continue. Physical therapy and

occupational therapy help to promote muscle development and improve coordination. Foot and/or ankle supports may be needed, Growth hormone treatment, which increases muscle mass, may improve muscle development. Exercise and sports activities should be encouraged.

Speech Development

Low muscle tone may create feeding problems and delayed speech. Speech therapy should be started in infancy to help with feeding issues and speech development. Sign language and picture communication boards help to reduce frustration and aid communication. Products to increase saliva may help with pronunciation problems. Speech development is usually delayed, and articulation problems may persist throughout life. However, speech may become an area of strength for some individuals with PWS, particularly those with UPD (both of the chromosome 15 pair from the mother).

Understanding

Individuals with PWS typically have learning problems. Areas of concern may include short attention span, inability to understand simple directions and difficulties with abstract thinking. Common strengths include long-term memory, reading ability and understanding language. Early infant stimulation is recommended; special education services and support should be started in infancy and continued through adulthood. By federal law, physical, occupational and speech therapies should be available and covered by your local school system.

Growth

Failure to thrive in infancy may require tube feeding. In order to ensure proper nutrition, infants may require special bottles or nipples (e.g., the Habermann feeder). Infants should be closely watched to make sure they get enough to eat for proper weight gain. Growth hormone levels in individuals with PWS are typically low, causing shortness, lack of puberty and increased body fat, even in those with normal weight, The need for growth hormone therapy should be considered in all individuals with PWS.

Sexual Development

Sex hormone levels (testosterone and estrogen) are typically low. Undescended testicles in boys may require HCG, testosterone, or if needed, surgery. Early pubic hair is common, but puberty is usually late and incomplete.

Other Common Concerns

Having crossed-eyes is common and requires early intervention and possibly surgery.

Curvature of the spine (scoliosis) can occur unusually early, and may be difficult to detect without x-rays. The curvature may speed up with growth hormone therapy.

Weak bones (osteoporosis) may occur much earlier than usual and may cause fractures. Provide adequate amounts of calcium, vitamin D and weight-bearing exercises. Bone density tests are recommended.

Non-insulin dependent diabetes can occur. This is usually caused by excessive obesity and will improve with weight loss. Routine screening is recommended.

Other obesity-related problems include decreased breathing, high blood pressure, right-sided heart failure, bed sores and other skin problems.

Sleep disturbances include decreased breathing with lower blood-oxygen levels during sleep, and/or daytime sleepiness. Airway obstruction may occur with or without obesity.

Skin picking is a common characteristic. It usually occurs in response to an existing sore or itch on the face, arms, legs or rectum.

Dental problems may include soft tooth enamel, thick sticky saliva, poor teeth brushing and teeth grinding.

Quality of Life Issues

General health is usually good, and life expectancy may be normal if weight is controlled. The constant need for food restriction and behavior management may be stressful for family members.

Adolescents and adults with PWS can function well in group and supported living programs if the necessary

diet control and structured environment are provided. Employment in sheltered workshops and other highly structured and supervised settings is successful for many. Residential and vocational providers must be fully informed regarding management of PWS.

Medical Alerts for Treatment of Individuals with Prader-Willi Syndrome

Negative Reactions to Medications

Individuals with PWS may have unusual reactions to standard dosages of medications. Use extreme caution in giving medications that may cause sleepiness because longer and more severe responses may occur. Water intoxication (too much body water) has occurred with the use of certain medications, as well as from drinking too much fluid.

High Pain Tolerance

Lack of typical pain signals is common and may cover up the presence of infection or injury. Someone with PWS may not complain of pain until infection is severe, and they may have a difficult time telling you where the pain is. Report any slight changes in condition or behavior to a medical professional for investigation into the cause.

Breathing Problems

Individuals with PWS may be at increased risk for possible breathing problems, such as low muscle tone, weak chest muscles and airway obstruction while sleeping. Anyone with significant snoring, regardless of age, should have a medical evaluation to look for obstructive sleep apnea.

Lack of Vomiting

Vomiting rarely occurs. Medications used to induce vomiting may not work, and repeated doses may cause poisoning. Due to the excessive eating and the possible eating of uncooked, spoiled or otherwise unhealthy food items, lack of vomiting is of particular concern. The presence of vomiting in someone with PWS may signify a life-threatening illness; therefore, if vomiting does occur, a medical professional should be contacted.

Severe Stomach Illness

Abdominal bloating, pain and vomiting may be signs of life-threatening stomach problems which are more common in individuals with PWS than in the general population. Rather than localized pain, there may be a general feeling of unwellness. If an individual with PWS has these symptoms, contact a medical professional immediately.

Body Temperature

Unexplained high or low body temperatures are common. High body temperature may occur during minor illness and in surgical procedures requiring anesthesia. Fever may be absent even with severe infection. Blood work, including a CBC (complete blood count), may be helpful in determining the severity of the illness.

Sores and Bruises

Because of the common habit of skin picking, open sores may be present and prone to skin infection. Individuals with PWS also tend to bruise easily.

Excessive Appetite

Excessive appetite and overeating may lead to life-threatening weight gain, which can be very rapid and occur even on a low calorie diet. Individuals with PWS must be supervised at all times and in all settings where food is available. Those who have normal weight have achieved this because family and/or caregivers enforce strict control of their diet.