Prader-Willi Syndrome

1. **What is Prader-Willi Syndrome?**
   It is a complex, rare genetic disorder that results from an abnormality on the 15th chromosome. It was first identified in 1956. Though PWS is a rare disease, it is one of the ten most common conditions seen in genetics clinics and is the most common genetic cause of obesity that has been identified. The diagnosis occurs equally in males and females and occurs across all ethnic groups.

2. **What are the effects of PWS?**
   While everyone with PWS is unique, individuals may share certain characteristics. This may include:
   - **At birth, hypotonia (low muscle tone) and failure to thrive.**
   - **As infants, delays in motor, cognitive skills and speech.**
   - **In early childhood, a compulsive eating and obsession with food may develop. This feature usually begins before age 6. The drive to eat is physiological and overwhelming and difficult to control.**
   - **Obesity is common but not universal.**
   - **Behavior issues such as repetitive behaviors, anxiety, and rigidity to schedules and routines, and temper tantrums are common.**
   - **Skills in solving jigsaw puzzles and word searches.**
   - **Especially nurturing and loving towards animals and babies.**
   - **Great perseverance in finishing tasks that can seem monotonous or routine to others.**

3. **Can PWS be treated?**

   While there is currently no cure for Prader-Willi syndrome, individuals may benefit from receiving input from geneticists, primary care physicians, endocrinologists, nutritionists, psychologists, psychiatrists, special educators, speech-language therapists, occupational and physical therapists, families, support staff and other care providers.

4. **Are there any treatments available that can help with weight management?**
   - **Early diagnosis and intervention**
   - **Growth hormone treatment**
   - **Healthy diet and daily exercise**
   - **Close supervision to prevent access to food**

5. Can Prader-Willi syndrome be inherited?

Most cases of Prader-Willi syndrome are not inherited, particularly those caused by a deletion in the paternal chromosome 15 or by maternal uniparental disomy. These genetic changes occur as random events during the formation of reproductive cells (eggs and sperm) or in early embryonic development. Affected people typically have no history of the disorder in their family.

Resources:

- www.pwsausa.org/
- http://www.pwsausa.org/chapters/new-jersey
- https://sites.google.com/site/njpwsa/home
- http://vkc.mc.vanderbilt.edu/VKC/about/
- http://www.thearc.org/
- http://www.sonj.org/
- http://www.medicinenet.com/prader-willi_syndrome/article.htm#prader-willi_syndrome_facts
- http://ghr.nlm.nih.gov/condition/prader-willi-syndrome
- https://clinicaltrials.gov/search/condition=%22Prader-Willi+syndrome%22?recruiting=false
- http://ghr.nlm.nih.gov/handbook/consult/findingprofessional

With interventions in place and careful monitoring, quality of life can be greatly improved for persons with PWS and their families