

USA
**PRADER-WILLI
SYNDROME
ASSOCIATION** | *Still hungry
for a cure.*

Prader-Willi Syndrome Association (USA)

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Prader-Willi Syndrome

Increased Awareness of PWS Means...

- More Diagnoses and Earlier Identification which leads to...
- Early Intervention Services and Appropriate Treatment and Management Strategies which leads to...
- Improved Quality of Life for Individuals Born with the Syndrome and their Families



William, 2 yrs



Aspyn, 6 mo

Prader-Willi Syndrome

Documented cases of PWS
go back to the 17th Century



Prader-Willi Syndrome

- In 1956, PWS was identified as a constellation of symptoms by Swiss physicians A. Prader, H. Willi and A. Labhart
- PWS is a lifelong, life-threatening, non-inherited genetic disorder that results from a defect on Chromosome 15
- PWS occurs in 1 in 12,000-15,000 births, or approximately 25,000 people in U.S. Of these, 75-80% are either undiagnosed or unknown to PWSA(USA).
- In NV, there are only 39 known cases of PWS; 13 of which are in the greater Las Vegas area. There are an additional 70-75% either undiagnosed or unknown to PWSA(USA).
- PWS equally affects all races and both sexes.

Prader-Willi Syndrome

- Two major types of PWS – Deletion (70%); Uniparental Disomy (UPD) (25%); Other (5%)
- PWS is now easily diagnosed with a blood test
- PWS is one of the 10 most common conditions seen in genetics clinics, and is the **most common genetic cause of obesity**
- Without intervention, PWS leads to obesity-related medical problems and eventual premature death

Clinical Features

PWS can be thought of as a Two-stage Disorder

Stage 1 – Infancy Stage

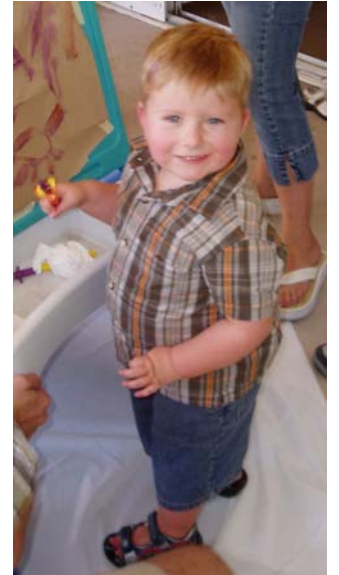
- Low birth weight and subsequent failure to thrive
- Severe muscle weakness (hypotonia), excessive sleepiness
- Suck/swallow problems, reflux, respiratory problems
- Subtle dysmorphic facial features such as “triangle” shaped mouth, narrow forehead, almond-shaped eyes
- Underdeveloped sexual organs (small labia majora in girls, small penis & undescended testes in boys)
- Delayed motor/physical milestones



Clinical Features

Stage 2 – Hunger Stage

- Between ages 2-6, hyperphagia symptoms begin with a preoccupation with food and/or compulsion to eat
- Voracious appetite begins as brain does not receive/process signals of feeling “full”
- Slow metabolic rate causes rapid weight gain
- Low energy level makes it difficult to exercise
- Emotional lability and behavioral manifestations



PWS is a spectrum disorder. All symptoms vary in degree from person to person.

Physiological Characteristics

- Hypotonia - Weak Muscle Tone
- Abnormal Growth (short stature, small hands & feet)
- Problems with Strength, Balance, Coordination, Motor Planning
- Hyperphagia - Dysfunctional Appetite Regulating System
- Respiratory Issues
- Gastroenterological Issues—Gastroparesis, Slow Bowel Motility, Inability to Vomit
- Hyper- & Hypothermia - Irregularities in Body Temperature Regulating Systems
- Incomplete Sexual Development
- Hypopigmentation
- High Pain Threshold, Bruise Easily
- Disordered Sleep
- Cognitive Limitations
- Speech and Language Issues (Dyspraxia & Apraxia)
- Dental Problems (decreased saliva production can cause severe problems)
- Skin Scratching and Picking
- Temperament and Behavior Issues with Older Children Through Adulthood
- Social / Psychological / Psychiatric Problems
- Other common characteristics may include: scoliosis, eye abnormalities (strabismus), medication sensitivity, orthopedic problems

Cognitive Characteristics

Most individuals with PWS have decreased intellectual functioning. Average IQ typically 55-70. Distribution generally 5% IQ 85+; 25% Borderline MR; 35% Mild MR; 25% Moderate MR; 25% Moderate MR; 5% Severe MR.

Decreased abilities in:

- Picture recognition
- Mathematics
- Short-term memory
- Daily living skills despite IQ



Areas of Strength:

- Friendliness, affectionate, desire to please, desire to nurture
- Long-term memory
- Recognize and evaluate shapes and figures
- Integrate stimuli in spatial relationship
- Puzzle solving

Treatment & Management Strategies

There is no known cure for PWS. But there are various medications, treatments and therapies that can help manage, reduce or even eliminate some of the symptoms.

Treatment consists of a multidisciplinary treatment approach. A typical family will see a multitude of professionals including:

- Geneticist
- Endocrinologist
- Pulmonologist
- Feeding Specialist
- Ophthalmologist
- Dentist
- RC Case Workers
- Behaviorist, Psychologist, Psychiatrist
- Neurologist
- Urologist (boys)
- Gastroenterologist
- Nutritionist
- Orthopedist
- Attorney
- PT, OT, ST, Social Skills Therapists
- Residential Staff



Treatment & Management Strategies

- Early Diagnosis – Genetic Testing
- Growth Hormone Therapy - FDA approved for use in children with PWS. Ongoing studies support FDA approval for use in infants and adults, with many of the following improvements seen:
 - Increased height and growth rate
 - Increased hand & foot size to normal proportions; more “normalized” facial characteristics
 - Decrease in body fat and body mass index (BMI)
 - Increase in muscle development
 - Improved respiratory function
 - Improved physical performance
 - Increase in resting energy expenditure
 - Improved cholesterol levels
 - Increase in bone mineral density
 - Improved cognitive functioning
 - Increased self-esteem

Treatment & Management Strategies

- Ongoing family education on the syndrome, along with treatment and management strategies via national PWSA (USA) and PWS NV S.H.A.R.E.
- Physical, Occupational, Sensory Integration Therapy, Oral Motor/Speech & Language Therapy, Social Skills Therapy
- Marriage and Family Therapy to help family members manage high and chronic levels of stress, chronic grief, sibling issues, extended family support (or lack thereof)
- Oral Hygiene products designed to relieve dry mouth symptoms
- Weight Management Strategies, including restricted calorie diet, 24/7 environmental controls
- Exercise regimen

Treatment & Management Strategies

- Behavior Management vs. Behavior Modification Strategies
 - Collaborative Problem Solving Approach as outlined in The Explosive Child, Ross Greene, Ph.D.
 - “No Hope for Food” principles
 - Positive, supportive, routinized, calm environment
 - Praise! Praise! Praise!
- Special Education
- Estate Planning, Special Needs Trust & Will, Conservatorship
- Sex Hormone Replacement Therapy in Adolescence
- Meaningful Work and Hobbies
- Residential Living Arrangements
 - Family, Group Home, Supported Living

Outlook today is more hopeful than ever before

- Normal life expectancy with weight management and 24/7 environmental control of food
- Media attention increases the public and medical community's awareness of the syndrome
- National obesity epidemic has influenced research to better understand the body's appetite regulating systems. We hope this will lead to the development of medications that will treat PWS-specific hyperphagia, as well as benefit general public.

- Early Diagnosis
- Appropriate Early Intervention Therapies & Management Strategies

Improves the quality of life for our children and their families



William, 3 yrs



*Aspyn,
14 mo*



*Aspyn,
17 mo*