Prader-Willi California Foundation
Supporting People with Prader-Willi Syndrome

Information for Regional Centers: How Prader-Willi Syndrome Impacts Your Consumer

Prader-Willi syndrome (pronounced PRAH-der-WILL-ee) is a rare, complex neurodevelopmental disorder caused by the lack of function of specific genes on chromosome 15, now referred to as the "PWS region." The condition occurs randomly and affects 1 in 20,000 people. There is no known cause, and there is no cure.

PWS is most known for causing a constant, insatiable drive to eat or obtain food called **hyperphagia**, and substantial **maladaptive behaviors** including self-injurious behaviors, impulsivity, perseveration, obsessive-compulsive symptoms, extreme rigidity, oppositional defiant disorder, property destruction, and verbal and/or physical aggression. PWS affects nearly *every* major system in the body and impairs growth, muscle tone, bone development, respiratory function, cognitive development, body temperature, pain threshold, sleep cycles, metabolism, and sexual development.

The severity of symptoms varies from person to person, and can even vary within the individual throughout their lifetime, but because PWS is a genetic disorder and every cell in the body is missing critical genetic material, no one escapes PWS's life-limiting and life-threatening symptoms. Individuals with PWS require specialized care throughout their lifetime from a multi-disciplinary team of specialists. **California's Regional Center system plays a critical role in helping individuals diagnosed with PWS and their families receive the supports and services they need.**

Early Start Services

By definition, every child age birth to three years who is diagnosed with PWS is eligible to receive Early Start services.

PWS's severe hypotonia (low muscle tone) begins in utero therefore babies are typically born via C-section, often as an emergency delivery. Varying degrees of intervention are typically necessary to combat the neonate's disinterest in feeding and ensure the infant's survival. The hospital's NICU team will likely first employ specialized feeding nipples, then move to nasal-gastric tube feeding, and as a last resort, may need to insert a G-tube to deliver nutrients directly into the baby's stomach. Babies should be released from the hospital as soon as proper feeding strategies are in place. Once home, this is where the Regional Center's support begins!

Essential Intervention and Therapies for the Infant and Child with PWS Age Birth to 3 Years

There are a number of essential medical and therapeutics interventions a child with PWS needs as soon as a diagnosis is made. **Regional Centers can assist the family by helping them identify and secure what are often referred to as the Core Therapies** for the child with PWS. These Core Therapies include:

- **Recombinant human growth hormone** (rhGH) is a synthetic, manufactured protein identical to natural growth hormone, used to treat pediatric and adult growth deficiencies by improving linear growth, hand and foot size, normalize facial features and body composition, improve muscle tone, bone mineral density, respiratory function, cognitive development, and more.
- **Oral Motor Therapy** to improve infant feeding by improving strength, coordination, and muscle control in the lips, tongue, jaw, and cheeks. As the child ages, oral motor therapy should continue in the form of Speech and Language Therapy to combat dyspraxia, a common diagnosis in PWS, and improve chewing, swallowing, and speaking (articulation).
- **Occupational Therapy** (OT) to help improve fine motor coordination and sensory-integration processing.
- **Physical Therapy** (PT) to help improve larger muscle development and movement, strength, balance, coordination, posture, endurance, and sensory-integration processing
- **Other medical and therapeutic needs** of infants and children with PWS include periodic assessment for and treatment of orthopedic abnormalities, eye abnormalities, oral and dental abnormalities (including lack of normal saliva production).

Transitioning from Early Start Services to Lanterman Act Eligibility at age 3 Years

Caring for the young child with PWS requires extraordinary sustained energy and attention. By the time the child turns 3 years old, families are often exhausted and overwhelmed. But for most, the challenges of raising their loved one with PWS are only just beginning. **Regional Centers can help families by making the process of evaluation and transition from Early Start to full Lanterman Act services as smooth as possible.**

Families recognize that the Regional Center is required to make an assessment of their child to make a determination as to whether California's eligibility criteria are met. Sometimes the determination will be easy to make. Some individuals with PWS will be **dually diagnosed with Autism Spectrum Disorder** and will meet criteria for this eligibility category. Some individuals with PWS will be **dually diagnosed with Epilepsy or Seizure Disorder** and will meet criteria for this eligibility category. The majority of individuals with PWS will test with Mild to Moderate Intellectual Disability (IQ 36 to 69), with 3% testing as Severe or Profound (20-35), and **will meet criteria for Intellectual Disability**

About 3% of individuals with PWS will test with an IQ above 69 points. Research now reveals that "children who began [growth hormone] treatment before 12 months of age had higher Nonverbal and Composite IQs than children who began treatment between 1 and 5 years of age." For those individuals whose IQ tests above 69 points, it will be important for Regional Centers to assess for eligibility under the "**Fifth Category.**"

PWS causes substantial disability and significant functional limitations in major life activities as specified in the Fifth Category, particularly and *without exception* in the areas of:

- **Self–care**
- **Mobility**
- **Self–direction**
- **Capacity for independent living**
- **Economic self–sufficiency**

In the areas of "**Learning**" and/or "**Receptive and Expressive Language Skills**" a few individuals with PWS may not exhibit significant enough functional limitations. For example, while most individuals with PWS have extensive Expressive Language skills deficits, their receptive language skills may not be sufficiently impaired to meet criteria in this area which requires substantial deficits in *both* skillsets.

Without exception and despite intellectual functioning, individuals with PWS require treatment from a multidisciplinary, interdisciplinary team of specialists including an endocrinologist, geneticist, neurologist, gastroenterologist, ophthalmologist, pulmonologist, orthopedist, urologist, speech and language pathologist, physical therapist, occupational therapist, behavior therapist, social skills therapist, psychiatrist, special education teacher, resource specialist, job coach, and/or other specialists.

PWCF wants Regional Centers to know that regardless of IQ score, **Prader-Willi syndrome meets the criteria for a "substantial handicap"** as defined in California Code of Regulations Title 17, Section 54001 as "a condition which results in a major impairment of cognitive and/or social functioning. Moreover, a substantial handicap represents a condition of sufficient impairment to require interdisciplinary planning and coordination of special or generic services to assist the individual in achieving maximum potential." **We know the earlier and greater support we provide to persons with PWS, the fewer symptoms they may exhibit!**

Please refer the family or care provider of someone with PWS to the Prader-Willi California Foundation and the national Prader-Willi Syndrome Association | USA (www.pwsausa.org) so that we may provide the ongoing education and support they will need throughout their loved one's lifetime. If you suspect your consumer may have PWS please refer them to PWCF so that we may help them obtain accurate genetic testing.

Please contact PWCF with questions about PWS and when we may be of support or assistance to you, your team, or the individuals and families you support. **800-400-9994 | info@pwcf.org | www.PWCF.org.**