The prader-willi syndrome

Prader-Willi Syndrome is a serious genetic disorder that begins at birth with no known cure ; causing mental retardation, short stature, low muscle tone, incomplete sexual development, and its main charecteristic, the desire to eat everything and anything in sight.

Prader-Willi syndrome was first known as Prader-Labhart-Willi Syndrome after three Swiss doctors who first described the disorder in 1956. The doctors described a small group of kids with obesity, short stature and mental deficiency , neonatal hypotonia (floppiness) and a desire to constantly eat because they are always hungry. Many other features of PWS have since been described, but extreme obesity and the health problems associated with being fat are the most prominent features. Individuals with PWS have some but not all of the same features and symptoms.

PWS is a birth defect. A defect in the hypothalamus, a region of the brain, is suspected to be the cause. The hypothalamus determines hunger and satiety. They can” t fell satiety, so they always have a urge to eat. Some PWS cases are so out of control thay will eat bottlecaps, glass, pencils, garbage, bugs, dogfood, and anything else they can stuff in their mouths.

“ The ingenuity and determination of PWS children in surreptitiously obtaining edibles is almost legendary and belies their cognitive defects. Serial weighing may be the only way to discover whether such a child is, in fact, stealing food”(Finey, 1983).

PWS occurs in about l in 10, 000 births. It occurs in both males and females equally and is found in people of all races and all nations. It is one of the ten most common conditions seen in genetics clinics.

Young people with PWS resemble each other very much. Most of the time, they look like brother and sister. Most of PWS people have almond shaped eyes, narrow foreheads, downturned mouth, thin upper lip and a small chin. Other common features are : obesity , they may be short; they have small hands and feet; have a skin picking habit, thick and sticky saliiva, incomplete sexual development, a curved spine (scoliosis), and chronic sleepiness.

PWS patients also have similar personalities: talkative, friendly, extreme attempts towards getting food, arguementivness, repetitve thoughts and behavior, stubbornness, frequent temper tantrums, and sometimes sudden acts of violence.

Most people with PWS have some degree of mental deficiency. The average IQ of people with PWS is 65 , and it ranges from 20 to 90. 41% of PWS people have IQs in the normal or borderline range. Specific academic weakness in math and writing are common, but reading and art are considered strengths. A delay in getting to early developmental milestones is common in PWS. The average IQ testing shows that people with PWS are mildly retarded, the range is from severely retarded to not retarded, with 40% having borderline retardation or just a low normal intelligence. Most affected children, besides their IQ scores, will have many, severe learning disabilities, and will show poor academic performance no matter what their IQ shows to their mental abilities.

There are many signs and symptoms of PWS that show up before birth. some are decreased fetal movement in 80-90% and having an abnormal delivery

in 20-30% due to having a really floppy baby. There are two distinct clinical stages of PWS.

Stage 1

Babys with PWS are called “ floppy babies” a lot. Thats because they have weak muscles, officially it is known as hypotonia. This hypotonia, which almost always occurs, could be mild to severe. Neonatal hypotonia makes sucking difficult, and a special feeding method called a gavage is used. A gavage the placing of a tube into the stomach through the mouth. They use it during the first days of life a lot.. Decreased caloric intake from the special feeding difficulties may lead to failure to gain weight. To keep the baby” s weight under control supervision by a professional nutritionist or a specialist who understands the syndrome might be necessary. Physical therapy is strongly recommended to improve muscle tone. When the muscle tone improves enough, an increased appetite and weight gain starts. The beginning of the second stage has begun. This hypotonia does not progress and begins to improve between 8 and 11 months of age in most cases. It improves, but it is never completly normal.

Stage 2

Stage 2 occurs between one and two years of age and is characterized by an appetite that can not be satisfied whic causes excessive weight gain. Speech problems, sleepiness, decreased pain sensitivity, skin picking habits and decreased growth are also characteristics of the second stage of PWS. The personality problems develop between ages 3 and 5 years also.

Most parents who have a kid with PWS do not have another kid affected with PWS. The cases of PWS are thought by scientist to have occured by chance

in isolated flukes of nature. But, there have been reports of families with more than one kid with PWS, but it is not common. Fewer than a dozen families with more than one affected offspring have ever been reported.

A blood sample for high resolution chromosome analysis is drawn on anyone who is though to have PWS. This will check out the chromosones. Chromosomes are packages of information found in the cells of our bodies. Each cell has a set of 46 chromosomes, which come in pairs numbered from 1 to 23. Parents contribute with one chromosome from each pair. Okay, now Prader-Willi Syndrome is caused by the absence of some genes on one of the chromosones that affect the functionimg of the hypothalamus. Many laboratories around the world are researching this. About three-fourths of people with PWS have a tiny piece missing from one member of the pair of chromosone fifteens (the one inherited by the father). The other one fourth are missing the dads contribution to this part of the chromosone by missing all of the fathers chromosone fifteen and having two copies the mother” s chromosone fifteen. The genes in this region are not functional and noone understands why.

As soon an the kid has improved muscle tone, and has increased its appetite, and is old enough to get move on the floor, than any food that can be easily gotten must be moved to a safer, out-of-reach place. To make inappropriate “ food” unavailable to the kid with PWS, parents must learn special patterns of food storage and handling Sleepiness during the day and napping a lot are some of the common features of PWS. Recently, studies have show that there is a strong link between this and sleep quality. Some of the types of sleep disorders that have been described in PWS affected people are:

disturbance to the sleep wake cycle, obstructive sleep apnea, hypoventilation syndromes and narcolepsy. Although patients with PWS fall asleep very quickly, their sleep period is significantly disrupted with frequent awakenings and abnormal patterns of rapid eye movements sleep (rems).

Obstructive sleep apnea occurs with increased upper airways resistance, either from enlarged tonsils , relaxation of the upper airway musculature, or from structural airway anomalies. Sometimes actual pauses in breathing during sleep can occur.. Narcolepsy, which involves sleep attacks and occasional loss of muscle tone, Short stature is also a common feature of almost all PWS affected people (80- 100%), but birth height is usually normal. The average adult height is 59 inches in women and 61 inches in men. Abnormal growth hormone response suggests a possible dysfunction of the hypothalamus and, growth hormone deficiency as a contributing factor in short stature. Improvement in growth rate and decreased rate of weight gain have recently been demonstrated in several growth hormone- deficient children with PWS after six months of growth hormone treatment

Other significant actions of growth hormone that have been reported is an improvement of muscle mass, muscle strength, energy expenditure, bone mineralization , sexual development , and also a decrease in fat mass , have led to further investigations in people with PWS.

Children with PWS have distinct behavioral abnormalities because of all the frustrations associated with the syndrome. These behaviors may begin as early as two years of age. They will get a variety of different eating behaviors like foraging for food, secretly eating large amounts of food, and other

attempts to continue eating. Other problems include verbally and physically aggressive behaviors such as lying, stealing, scratching and skin picking. Tantrums and unprovoked outbursts are common among children and youths with PWS.

People with mild cases of PWS can do many things their normal peers can do, such as go to school, get jobs, and sometimes even move away from home. However they need a lot of help. Kids going to school would need to be enrolled in special education programs(Otherwise they” d be eating their pencil and paper). They need to be constantly supervised.