

IMPACT OF THE SYNDROME

STATISTICS:

It has been estimated that one in 12,000 – 15,000 people have Prader-Willi syndrome. Based on 2004 population statistics, this equates to about 19,000 – 24,000 individuals in the US with the syndrome. PWSA (USA) knows of 5,057 persons with PWS in the US (as of the end of 2004), leaving a possible 15,000 or more of those dealing with PWS without diagnosis and/or support services.

IMPACT ON THE CHILD/ADOLESCENT:

The problems for the person with the syndrome are significant and tragic. A child born with the syndrome has to deal with a constant and insatiable drive to eat--a drive not satiated with food. The child will continue to eat and seek food regardless of the amount consumed. Unless some type of significant intervention is provided, the child will become extremely obese. Extreme obesity leads to life threatening health problems and social ridicule. And the child is born with hypotonia (weak muscle tone), which limits the ability to participate in normal physical activities. Equally significant are the emotional issues that arise, particularly as the child ages into adolescence. Such problems as emotional outburst and temper tantrums increasingly alienate the child from family and peers. In addition, the child with Prader-Willi usually has speech and communication difficulties that tend to increase their frustration and withdrawal. As a result of these factors, the child with Prader-Willi often becomes obese, frustrated, estranged from family, ridiculed by peers, and unable to participate in normal school activities. Essentially, they become socially isolated. The impact on the victim is severe and, eventually, potentially life threatening.

The following are a sample of expressive quotes from those with the syndrome:

- **An eight year old boy explained,** *"My hand reaches into the refrigerator and I try to stop it but I can't"*
- **A teenage boy wrote,** *"I have a lot of questions to ask Jesus. Did God make desserts? Is it a sin to pick? (at sores or nails) Sometimes I pray to Jesus to not gain weight. I want to be happy. I don't want to do wrong things. I'm thinking if I don't have a girlfriend or not get married, I pray I have one in heaven."*

- One young man wrote. "We all have Prader-Willi syndrome. We feel that we aren't human. Humans have freedom. We aren't human because we can't have children. If we aren't human – we are Aliens. If we aren't human, where oh where is our UFO? We want to go home."
- **A mother of a 16-year-old son said her son cried to her and said,** *"Mom, why do I have to live this way?"*

IMPACT ON THE SIBLINGS – For siblings, reaching into the refrigerator for Kool-Aid or having an after school snack is a luxury reserved for neighbor kids. Most have to learn to sneak snacks, guard food, and tell on their brother or sister if they get into food. As they get older, siblings are often too embarrassed to bring friends home due to the locks on the cupboards and refrigerator, and their PWS sibling's unpredictable outbursts. Because many with the syndrome are living longer and parents are aging, adult siblings may be left with the dilemma of how to get care for their siblings.

1) PWS: Facts Regarding Prevalence Rates, Whitman 1997. PWS in a Swedish rural county: epidemiological aspects. *Developmental Medicine and Child Neurology*, Akefeldt, Gilbert & Larsson (1991); A PWS in Norway: An epidemiologic & sociomedical study. In S. Cassidy, PWS & Other Chromosome 15q Deletion Disorders, Springer-Verlag. (1991)

The following are a few examples from our call logs of sibling issues:

- Daughter with PWS is 13 years old. Mom is concerned about her 16-year-old son. Wants him to be able to go to the national conference to meet other siblings. Mom states, *"He has to watch after her after school. He is more like a jailer than a brother."*
- Sister with PWS is 325 pounds and 5'1". She is wheelchair bound and at risk of losing placement. Parents are deceased, and her sister doesn't know what to do with her. She and her husband tried to keep her for two years, but couldn't cope with the problems.

IMPACT ON THE PARENTS – If the parents of an infant or toddler are "lucky" enough to get an early diagnosis, they not only have the grief issues and survival worries of many parents who have to adjust to having a multiply handicapped child -- but also have to deal with the horror stories of the older child with the syndrome. The stories on the Internet – such as the one of the 360-

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pound girl who eats out of garbage cans – are forever etched in their minds. They look at their beautiful, passive, tiny baby and initially are usually overwhelmed with grief and concern. Ironically, during the first year parents have to struggle to get their child to eat because of the weak muscle tone. They may have to deal with decisions on gavage feeding versus NG tubes, or G tubes. Often babies are on oxygen. The toddler needs PT, OT, and speech therapy.

For the parents of the older child, survival often becomes the name of the game. Kitchens are locked tight and guarded with vigilance. Mealtimes are a constant struggle where desserts are unheard of and the child with PWS intently watches everything his/ her parents and sibling eats, and ends the meal with the inevitable, "It's not fair...!" and the beginning of a tantrum. 1 As children are surviving longer, the problems are getting more complex.

The following are a few samples of the many, many calls that come into the PWSA (USA) office daily.

- Son is 300 pounds and 5' tall. The calf of his leg was measured at one time at 27" around and is now much bigger. They had to slit all of his pants up the side to get them on. The edema is so bad that the fat hangs over his ankles and he can't put on shoes. He wants to go to church and sing in the choir, but has nothing to wear. Parents are elderly and struggling to care for him. They are resolved that he will die within a year. Dad said, *"We try to live a good life, but I have to ask God, Why? Why?"*
- Son 33 yr., 300 lbs. and 5 foot tall, died in January. He choked trying to swallow a potato whole (trying to sneak it and afraid he would be caught). It had to be surgically removed, and he was on a respirator for 4 days before he died. Dad states, "He consumed our lives. We spent our lives trying to keep him from food." Dad said that they begged for a group home and were told they couldn't afford it or that he wouldn't fit in. *"We struggled with PWS all our lives. Now we have to live with guilt for the rest of our lives."*
- Daughter, in 20's. Mom called to thank me for my help in getting placement for her daughter, and how much my support meant to her. She said, if it weren't

for... 's placement, *"I'm sure there would be a murder and suicide by now."*

- Boy, 10 yrs., 240 lbs. School counselor called. Mom charged with child abuse (because of his weight). He steals and hoards food. Mom won't talk to anyone about the problems.
- Daughter, 24 yrs., weighs 180 but had been 250 lbs. Daughter was not diagnosed until she was 18 yrs. old. Daughter states a worker in the group home molested her. Was discharged and is back at home. Husband just died. Mom is 64 yrs. and alone with the problem. Wanted to know how to do locks. Had a locksmith put a lock on the freezer, but he drilled into the Freon line and ruined the freezer. Mom called back in the afternoon. Her daughter had just scared off the second man who was going to attempt to put locks on the freezer and refrigerator.
- Daughter, 29 yrs., 500 lbs. Has never been diagnosed. She has all the classic symptoms. They had even tried the stomach stapling and it didn't help (typical with the syndrome). Mom saw the Leeza Show and was crying with relief and gratitude that there was a name for her daughter's problem.
- Son, 16 yrs., 320 lbs. Just officially diagnosed with PWS. Hides food everywhere, e.g., under his pillow. Becoming aggressive towards mother. Took out a knife and tried to hurt himself. Embarrassed about his small penis and his obesity. Others stare and point at him. He has no social life. He cried and said, *"Momma, why am I like this? I don't want to be this way."*
- Daughter, 12 yrs., 135 lbs. Daughter was put out in the hall in second grade due to a tantrum. She was abducted and raped -- was found in the woods by the police. Mom was told she couldn't sue the school. She had been expelled in the first grade. Mom was just reported to the child protection services because she asked the teacher to not give her daughter milk and cookies, but to give her a diet coke as a substitute for a snack (typical and appropriate request). Marriage under stress.
- Son, 25 yrs., 295 lbs., had been as high as 436 lbs. He stole a \$.90 package of donuts, was arrested, and is going to court.

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- Daughter, 15 yrs. Four years ago, her daughter was burned over 25% of her body because at school they taught her if she was on fire, she should, "stop, drop, and roll". She set herself on fire because she wanted to practice it. When they were struggling to get an IV in her, everyone including the doctor was in tears. Daughter just said, "*When is lunch?*" They are still regrafting and having significant problems due to the picking.
- Daughter, 42 yrs., 165 lbs. and 5 foot tall. Behavior and food seeking problems. Mom states she is always looking for food and doesn't sleep much. Dad died last year. Mom is 60 years old and has her own health problems - I asked her how she was holding up, she said, "*Actually, I haven't held up. I am seeing a psychiatrist. I am so alone.*"
- Son, 16 yrs. Mom called from a pay phone. She had son admitted through the ER because of his volatile behavior. When she tries to stop him from eating (his weight is life threatening), he becomes aggressive with her -- swearing, and throwing things at her. The hospital said if she refuses to take him home, they would charge her with child abandonment.
- Daughter, 17 yrs. Mom was charged with abuse after her daughter called 911 on her for locking her in the basement to keep her from going out of her second floor window again in the middle of the night. The last time, she got out at 4:30 am and stole food. Her father is deceased. Mom struggling alone with the problems.
- Daughter, teen. Mother wrote to tell about how grateful they were to get a conference grant. They had never stayed in a hotel before. She said, "It is something we will remember and appreciate for years to come." She told of an incident that day in high school when her daughter with PWS misbehaved and got very loud. Afterwards, when they discussed it, her daughter said, "*What would you do if everyone has doughnuts and then they give you an ancient orange and expect you to be grateful?*"
- Son, 11 yrs. Mom had just read the Indianapolis Star and realized that her son "*fit the description of PWS to a tee*". She was crying with relief that there was an answer for why her son was the way he was. She said, "*I have gone through hell for 11 years.*" That

morning, she found him sitting in the tub with 5 pounds of ice cream all eaten. He will sneak boxes of cake mix, mix with water, and eat. She tried to lock food and he busted all the locks off. He picks at sores so much the school will call her and tell her she has to make him quit. He has all the classic characteristics of PWS.

- Son, 8 yrs. Mom crying quietly on the phone, and said, "*I need help.*" He gets up in the night and forages for food. His tantrums are accelerating and he is showing more signs of obsessive-compulsive behavior. His teacher called last night and said she didn't think she could teach him anymore. Mom said, "*He has to be watched every minute.*" They have a working farm and she has a full time job to make ends meet, but doesn't know how she can continue to work. Today, he locked some of their chickens in an airtight container, and they died. She said when she found them, "*I just sat down and sobbed.*"

MEDICAL IMPACT OF THE SYNDROME

"*Life-long and life threatening*" are the words that first smack parents in the face when they get a diagnosis of Prader-Willi syndrome. As an infant, the medical problems are dramatically different than in the second phase. Due to the weak muscle tone, feeding the infant with PWS becomes an all-consuming job. Parents learn about gavage feeding, G tubes, NG tubes, etc. Respiratory problems are also an issue of concern at all ages, plus working to get the many therapies, such as growth hormone, that may make a difference in the quality of life.

The older child with the syndrome faces all of the life-threatening issues related to morbid obesity, plus problems unique to PWS. Due to the poor gag reflex, many have choked to death. The poor thermostat and high pain threshold can cause death due to a serious infection.

If a person with PWS greatly distends his or her stomach, it can stretch to the point that it cuts off its blood supply, thus causing necrosis. This can be a very life threatening condition if not quickly diagnosed and treated.

The following are examples of calls that come into the office regarding medical impact of PWS:

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- Son 4 yrs. weighs 95 lbs. He is in ICU on *"death's doorstep."* The obesity has caused upper airway restriction and an enlarged heart.
- Mother found her 3-yr-old daughter dead. The autopsy revealed she had PWS. Death was due to weight related obstructive sleep apnea and a fatty liver. Mother grieving and angry the doctors didn't diagnose her daughter earlier.
- Son 15 yrs. has significant sleep apnea when he gets overweight. Has seizures so severe his heart has stopped. He has kept sores (from picking) open on his body for 6-7 years.
- Group home lost two members with PWS within a couple of months. One young man got up in the middle of the night and ate so much he died.
- Twenty-year-old young man died suddenly at camp. The camp staff was not trained adequately and didn't respond to his complaints of pain and vomiting. When a counselor found him unconscious on the floor of the bathroom and ran to get the nurse, she said, *"Just tell him breakfast is ready and he will get up."* By the time the ambulance was called, he was dead.

IS THERE HOPE?

For the parents of the young child, it is a new era. With an early diagnosis and a lot of support, life holds promise of a brighter future. This is best described in the words of one young mother:

"Lifelong and life-threatening.' I still remember the terror that I felt when I read those two descriptive phrases about the diagnosis that we had just received for our precious baby daughter, Erin. Prader-Willi syndrome. Whoever heard of Prader-Willi

syndrome? This has to be a mistake. Sure she's weak. Sure she sleeps a lot. No, she doesn't smile much; but then she doesn't cry much either. But how does all of that add up to 'lifelong and life threatening'? I can accept 'developmental delays' and I can live with them. Delays have to do with time, and we have plenty of that. So where do they get off telling me about this Prader-Willi thing? ***"The fear, anger, and disbelief of receiving a diagnosis of Prader-Willi syndrome are still as vivid in my mind as they were the day the doctor called us with the test results.***

So much has changed since then, but the memories of that day will be with me forever. Going through the materials that the doctor gave me was as difficult as anything I have ever done. The picture that was painted by that information portrayed Erin's future as being as black and dismal as could possibly be. If she were destined to have all of the problems on those lists, what point could there possibly be to her life?

Since that time, I have spent countless hours researching the issues involved in PWS and talking with other parents — especially the parents of young children. Due to early diagnosis and aggressive early intervention, I now know that I can expect a drastically different life for Erin than the one I pictured when I received the original information on PWS. Research is currently going on that could make major inroads into the medical and behavioral challenges that come with PWS. It doesn't cancel the diagnosis or the issues she may face but knowing that certain things will probably be real challenges for her, I can set up a plan to meet each of these issues head on. ***This allows me to be proactive instead of reactive and greatly increases the chances that Erin's life will be full...both of health and happiness. She deserves no less!***