

**Pittsburgh
Partnership**

SPECIALISTS IN PRADER-WILLI SYNDROME

www.PittsburghPartnership.com
pws@pittsburghpartnership.com
Telephone: 412 247-5822
FAX: 412 344-7717

Janice L. Forster, MD
Developmental Neuropsychiatrist

Linda M. Gourash, MD
Developmental Pediatrician

Correspondence & Consultation: 615 Washington Road, Pittsburgh, PA 15228-1909 USA

February 12, 2013

To whom it may concern:

It is our understanding that the state of Connecticut is currently negotiating with the Latham Centers to provide PWS specific housing for Connecticut residents who are currently served in Massachusetts. Also, it is our understanding that the newest arrivals to Latham Centers know that once this agreement is finalized, they will be returning to their home state. However, there are several residents who have lived in group homes in Massachusetts run by Latham Centers for most of their adult lives, and they consider Latham Centers to be their permanent home. This letter of appeal is intended to explain why a move to a new system of care would not be in the best interest for the four Connecticut residents in question. Although each of the four persons has PWS, their situations are unique. Ideally, a decision to relocate should take all of the following itemized opinions into consideration.

Consultant Opinions:

1. Individuals with PWS require a specialized placement like Latham Centers that has the **programmatic elements**, the **skilled and experienced staff**, and the **depth of experienced supervisors** to manage PWS-specific behaviors. A new home would provide the programmatic elements, but it would take some time to acquire the skill, experience and depth of supervision required to optimally manage the syndrome and each individual in their care. This learning curve of caregiving for both the syndrome and for each individual with the syndrome is universal. During the time of adjustment **which can take more than a year even in the best programs, experience tells us that we can expect an increase in disruptive, aggressive and self injurious behaviors**. These behaviors alone can delay the person's adjustment as well as the staff adjustment to the needs of the individual, while the management of acute problems crowds out long-term needs.
2. The individuals in question, as well as their parents, believe that Latham Centers provides the best opportunity for a community placement that provides exposure to peers who also have the syndrome. The Best Practice Guidelines for Standard of Care for Persons with PWS in Residential Living highlight the importance of homogeneous pairings in the care of individuals with PWS. Living with other people who have the syndrome provides an opportunity for peer

reference among those with this rare disorder and facilitates acceptance of the restrictions required for successful management and adjustment.

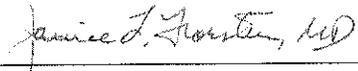
3. In a smaller PWS program, the individuals in question are at risk for psychiatric or medical hospitalization due to their aberrant behaviors. There is ample evidence to suggest that triggers for aberrant behaviors include environmental over-stimulation or under-stimulation, unfamiliarity with caregivers which leads to uncertainty, frustration and escape/avoidance of demands. Therefore, a change of living situation is likely to precipitate an adjustment disturbance with emotional and behavioral consequences. ***Situational stress is commonplace in PWS; it is the response of the experienced staff that determines for each individual whether the situation escalates to a psychiatric crisis.*** Once the person is hospitalized, further deterioration occurs due to additional environmental change. Paradoxically, psychiatric hospitalization can *add* to morbidity due to uncontrolled weight gain (as much as 20 pounds in one week), side effects of psychotropic medication (toxic reactions and over sedation), and persons with PWS are at greater risk for dying as a result of physical restraint that is inappropriately administered (failing to account for hypotonia and risk for respiratory compromise). Management of the crisis by experienced staff in the context in which it occurs is always preferred.
4. Latham Centers provides a family-like environment for persons of all ages who have PWS. It provides the necessary social stimulation that can come only from a PWS peer group. Because Latham Centers has specialized services for PWS and serves 40 adults with PWS in a community of group homes, Latham Centers has the ability to ***select for peer compatibility, developmental appropriateness and setting specificity*** to more optimally meet the needs of each person. This provides the individuals with positive role models and offers the possibility of protecting them from entrenched and maladaptive peer interactions that perpetuate behavioral reactions. This pattern is frequently seen in smaller PWS facilities.
5. Because of the rarity of the disorder, most physicians have extremely limited experience with caring for person with PWS. Parents are rightfully concerned since they have years of experience consulting with medical practitioners who do not know enough about PWS to be helpful. Latham Centers has spent years building up a network of medical and psychiatric consultants who have familiarity with PWS. This component of PWS programming is frequently overlooked or underestimated in new programs.

It is well accepted that due to the nature of the genetic deficit in PWS, the most effective tool for managing syndromal behaviors is the ***controlled environment***. Almost always, deterioration in level of function can be related to changes in environmental structure that precipitate stress. ***Stress sensitivity*** in PWS is genetically determined and related to the ***cognitive inflexibility and impulse control deficits*** that are the major underpinnings of the phenotypic behaviors associated with the syndrome. Among all individuals with neurodevelopmental disorders, tailoring the environment to the needs of the individual is one of the most important factors that determines their level of function. Compared to other individuals with neurodevelopmental disorders, people who have PWS are the ***least able to adapt to environmental changes and interpersonal challenges***. Despite this fact, it is consistently

expected that people with neurodevelopmental disabilities conform to fit in to existing opportunities in the community for living, work and leisure. In fact, the most compelling reason for individuals with PWS to be in group home residence with trained staff, who offer 24 hour-7 day per week supervision, is because the parents are not able to provide the degree of structure and predictability that these individuals require in order to be successful. Please see the attached Appendices I-V for further detail about the needs of individuals with PWS and how Latham Centers has developed the unique expertise to meet those needs.

Finally, due to the nature of the syndrome, individuals with PWS have little choice in many facets of their lives, despite the fact that they may be considered competent to consent for themselves. ***These four individuals in question and their parents choose to continue to reside at Latham Centers.*** Your consideration for the needs of each of these individuals is deeply appreciated.

Most sincerely,



Janice L. Forster, MD



Linda M. Gourash, MD

ATTACHMENTS:

- I. Description and Needs of Persons with PWS
- II. Description of Latham Centers
- III. The PWS Personality
- IV. Global Assessment of Function for PWS
- V. The Consultants

Appendix II: ABOUT LATHAM CENTERS

Latham Centers states their mission and describes program goals and attributes as follows:

Mission: *Latham Centers compassionately and creatively helps children and adults with complex special needs, including Prader-Willi Syndrome, to lead meaningful, abundant lives.*

Core Values:

- *We believe in the power of relationships.*
- *We listen.*
- *We courageously do what's right.*
- *We strive for innovation and excellence.*

Goals:

- *Remaining national and international leaders in providing treatment and services to individuals with Prader-Willi syndrome, their families and to the medical and educational providers who support them;*
- *Providing increased job/vocational opportunities to those we support;*
- *Offering an array of housing opportunities to meet the evolving needs of our individuals. (Two new adult group homes were purchased and renovated in FY12 to meet an increasing demand for our adult residential services.)*
- *Preparing students to be successful after leaving Latham Centers; and*
- *Developing new areas of expertise to complement and maintain our core services.*

Program Description:

Latham Centers, located in Massachusetts on Cape Cod, offers nationally and internationally recognized, innovative treatment and supportive services for children and adults with Prader-Willi syndrome. Latham serves approximately 45-50 children per year—40% have PWS. Latham serves approximately 45 adults per year—80% have PWS. Our primary facilities are located on a campus in Brewster, with a residential school, group homes, shared living, and supervised independent living opportunities located in Brewster and other towns on Cape Cod and in Southeastern Massachusetts.

Latham Centers originated in 1970 to provide residential care and treatment for girls and young women, ages 8-22 years of age, who had come from environments of abuse and neglect that resulted in severe behavioral and emotional problems. Later, a home for boys was added to the program. Now, individuals continue to be referred to Latham Centers from failed placements in foster care or other residential treatment facilities, including psychiatric hospitals. Some of these individuals have been diagnosed with bi-polar disorder, Asperger's Syndrome, Autism, or Reactive Attachment Disorder. Latham Centers provides a beautiful physical setting and an embracing environment where the people in our care can learn life skills

and coping mechanisms for the challenges that have brought them to us and feel safe enough to have the opportunity to recover from their past traumas.

Within this comfortable setting, we have brought together a highly skilled professional team of experts, including physicians, social workers, special education teachers, physical therapists, occupational therapists, speech therapists, clinical psychologists, and residential and vocational counselors. This energetic and devoted team works together with each Latham resident and their family members to develop the right approach and best services for that person. With this individualized approach, each person's progress is evaluated continually, and our programs and services are adjusted to meet their changing needs.

Historically, the level of expertise in managing the most severe psychiatric and behavioral difficulties served the program well when services for PWS were added under the Latham Centers umbrella. The Gilbough Center was established in 1981 for the treatment of individuals diagnosed with Prader-Willi syndrome (PWS)—a rare genetic, life threatening, obesity disorder of the 15th chromosome with no known cure. PWS is a complex syndrome that manifests itself with severe idiosyncratic symptoms including insatiable eating behavior (hyperphagia), mild to moderate mental retardation, emotional and behavioral problems, and excessive and repetitive behaviors (self-injurious skin picking). It also has unique physiological difficulties including hypotonia, decreased motivation to exercise, decreased caloric needs, abnormal sleep, diminished pain response and abnormal temperature regulation that require specialized environmental management. The syndrome occurs once in about every 12,000 – 15,000 births, and is found in all races and ethnic groups. The Gilbough Program is internationally-recognized; it provides services to adults with Prader-Willi Syndrome (PWS).

Examples of community based group homes:

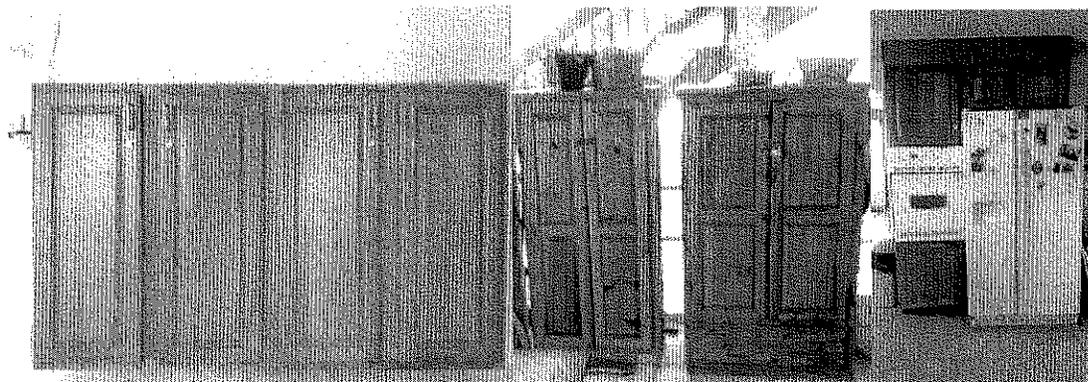




Gilbough maintains eight community residences, group homes and other shared or independent living environments across Cape Cod in localities from Sandwich to Brewster—all providing 24-hour supervision and expert care for our residents.

Food access is locked and secure in each of these houses. This is a life-saving aspect of residential living for PWS whether the person lives at home with their family or in group home residence in the community. The requirement for locked food access is mandated in the International Best Practice Guidelines, and group homes around the world have locked refrigerators, freezers, pantries or locked kitchens. Today, locked food access is the reason that people with PWS can have the potential to live full lives, because in the past, weight related mortality and morbidity was high; nearly half of individuals with PWS died during adolescence.

Locked food access:



Today, Latham is a leading treatment center in this field with an international reputation. It is the only specialized resource for PWS on the East Coast. In April of 2011, the agency earned accreditation by the prestigious international **Council on Accreditation**. Latham Centers is licensed by the **Massachusetts Department of Developmental Services**. Latham School for children is accredited by the **Department of Elementary and Secondary Education and the**

Department of Early Education and Care. The agency also works closely with the **Department of Children and Families** and the **Department of Developmental Disabilities**. Based on a reputation for excellence and innovative treatments, Latham Centers partners with other states beyond Massachusetts to provide life-changing services to students and individuals. Latham Centers, Inc. is the largest 501 (c)(3) non-profit on Lower Cape Cod. It is an equal opportunity provider and employer for more Private fundraising accounts for less than 3% of Latham's operating revenue.

In 2011 Latham Centers took on the green initiative of establishing a confidential, electronic system of record keeping. Therapeutic consultants, educational staff, nursing, supervisors and direct care staff can access an online system to chart on each person's progress multiple times per day. This confidential information is accessible by supervisors and administrators as well as parents and guardians. For example, residents are weighed daily, and weight charts can be isolated from the data base to provide information on weight gain that occurs during home visits (a common problem among family's who have children of any age in residential living). Also, parents who are anxious about their child's adjustment to the program can log on to THERAP to monitor their weight, sleep and behavior. As such, Latham Centers has the highest degree of transparency between staff and parent/guardians compared to any other program serving PWS in the country.

Since 1970, Latham Centers has been working together—with our students, residents and their families, and outstanding professionals—to bring hope and change to young people and adults who face ongoing life challenges. At Latham, they receive respect and discover new potential to become active members of their communities.

About Gilbough Program:

The Gilbough Program, founded in 1981, is one of the leading residential treatment centers in the country for Prader-Willi Syndrome (PWS). We have made it a part of our commitment to those with PWS to seek out nationally recognized consultants with an expertise in PWS to improve our programming and care. Latham Centers consults with the world renowned Pittsburgh Partnership on the most difficult and challenging PWS cases. The Partnership consultants are Dr. Janice L. Forster, Developmental Neuropsychiatrist, and Dr. Linda M. Gourash, Developmental and Behavioral Pediatrician (see Appendix III). As a team, Latham is able to engage these two highly trained and specialized doctors on all aspects of the syndrome—from program initiatives, program review and clinical and medical consultations—for the children and adults we serve.

Latham Centers uses the Children's Institute (Pittsburgh, PA) diet plan (The Red Yellow Green Diet), and it has been individualized by a dietary consultant from Tufts University in Boston. Together with our consultants, the residents and their families, we have created a successful diet plan that works. Many in our PWS community have lost substantial weight and

maintain healthy weights. They've also found improved health through exercise, with fewer illnesses and weight related problems, and higher self-esteem.

Exercise is a priority for individuals with PWS in order to maintain a healthy weight, to combat excessive daytime sleepiness, to promote breathing, to enhance insulin metabolism, to consolidate sleep, and to relieve stress. Community integration with Special Olympics is enjoyed by everyone. Individual exercise goals are met on a daily basis through walking, swimming or biking in the community, weather permitting. Exercise equipment is located in the homes for days with inclement weather.

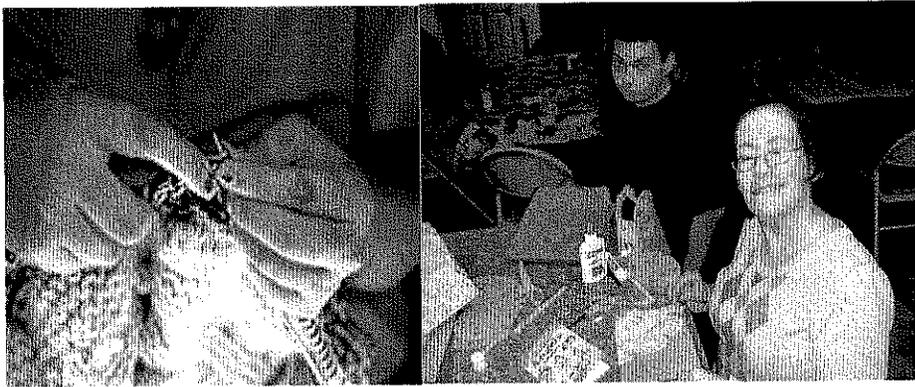


Medals at Special Olympics

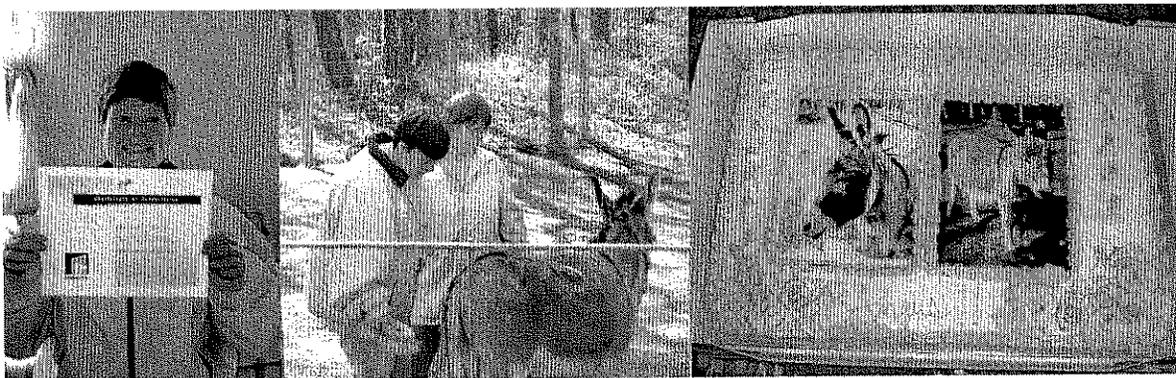


Making Exercise Fun!

A recent initiative for individuals with PWS is to provide sensory motor enrichment to combat the compulsive skin picking associated with the syndrome that can become chronic and life threatening. Although 1:1 supervision and intensive behavior programs might be required to manage skin picking, the staff at Latham Centers/Gilbough have found that vocational and leisure skills built into the daily plan can prevent or diminish the sensory hunger so characteristic of the syndrome.



Sensory motor activities and donkey therapy



The anisotherapy program provides sensory motor stimulation through grooming and caretaking activities with donkeys. Residents also exercise the donkeys providing an opportunity for movement and sensory integration for the person with PWS.

Gilbough offers our residents the same level of care and support that is a hallmark of all Latham's programs. Residents enjoy living in attractive, comfortable homes situated in the local community. Each home is a reflection of the individuals who reside there. Residences may consist of a group home with other housemates, or in a home they share with a caregiver. Staff support is provided 24 hours a day and we pay careful attention to monitoring and securing food, grocery shopping and preparing meals. It is a constant struggle to maintain autonomy among individuals with PWS while the life threatening nature of the disorder requires a high degree of staff vigilance and intervention. It is the large number of individuals in the PWS community at Latham Centers that makes this possible. Although they may live in group homes, Latham residents with PWS come together for social and vocational activities. Knowing a group of referent peers with the same syndrome is essential to the adolescent and adult task of forming and maintaining ones sense of self and accepting the nature and limitations of the disability. When everyone with PWS in the community receives the same level of structure and environmental security, regardless of where on the Cape the group home resides, then

management of syndromal behaviors is optimized. Our consultants (Pittsburgh Partnership, Forster and Gourash) have taught us that a *no doubt, no hope and no disappointment* paradigm works very successfully for individuals with PWS, not only for food related behaviors, but for the majority of non-food behaviors that are associated with the syndrome. ***When people with PWS have FOOD SECURITY, they can use their brains for other things.*** As such, a goal for each individual is *maximum function with support*.

Our professional team brings together academics, vocational training, medical and social services, the arts, sports, technology, and more. Our residents compete in Special Olympics, engage in meaningful work and vocational activities, and learn to explore the vast and interesting community in which they live.

The Top Ten Reasons Latham excels in PWS residential placement:

- 10. Cape Cod is one of the most beautiful places in the world.** Our main campus, where our children's program is housed, is in the historic town of Brewster while our adult houses are situated in various towns and villages of the Cape. Each town has its own character, appeal and unrivaled natural scenery.
- 9. We have been working with PWS for a long time.** Experience is worth a lot and we have a lot of it. Since 1980 we have been working with individuals with PWS and have been honing our skills since then. Each day we learn more and each day our services improve.
- 8. There is strength in numbers.** For many of our students and individuals, coming to Latham is like discovering a home they never knew they had. For the first time many of them now have peers that are also diagnosed with PWS. Having friends and relationships with people who really understand you builds bonds of belonging, as well as a sense of security.
- 7. When the food is secure, you can concentrate on the rest of your life.** All Latham buildings and residences are food secure. Obviously, this includes all cooking facilities, but it also extends to offices, events and staff belongings. By doing our part to make "food" a non-issue, our students and individuals find it easier to focus on other aspects of their lives. Unlike other environments our individuals have no hope in securing additional food sources here.
- 6. Delicious, nutritious meals.** Our PWS menu provides high quality nutrition that is delicious and varied on a menu that is structured and predictable. Our students and individuals know well beforehand what the meal is and how much of it they are going to get. This consistency is crucial to what we do.

5. Our staff are leaders in the field. As said earlier, experience can be priceless. Latham staff are highly trained; specifically in working with individuals with PWS. Each staff member attends a week long orientation, monthly trainings, weekly staff meetings, exposure to outside experts on PWS and the support of other highly, trained and experienced colleagues. Our staff are the ones who make the program exceptional.

4. Lots and lots of vibrant opportunities. Both our adults and children's programs offer numerous experiences, outings, activities and events specifically tailored for individuals with PWS. The lives of our students and adults are full of engagement, learning and enjoyment. Whether it is learning through vocational opportunities, enjoying a horseback riding class, creating a work of art or attending a dance with your friends-each day has a wonderful opportunity in it.

3. The Latham program treats the whole person. This is what sets Latham apart. Prader-Willi Syndrome is expressed differently in individuals and needs individual treatment. It is not just a "food disorder" or an issue of creating a solid behavior plan to address issues. It is far more than that and the treatment needs to be fully integrated and knowledgeable of the syndrome. Our entire program has been created specifically to support the students and individuals in our care.

2. The program is a lot of fun. Our students and individuals create wonderful lives while they are with us. As staff members, it is a rare privilege to witness how a new member of our Latham community can blossom in this environment.

1. Latham gets it. As a parent, teacher, referring agency or individual with PWS, if you have talked to the outside world about PWS, you understand what this statement means. Here at Latham we have been in this special field for thirty years and understand the many special challenges of the syndrome.

Latham Centers is an international leader in the treatment of PWS with more than 30 years of experience. Latham Centers is one of only a handful of providers in the world with expertise specific to PWS. Experienced staff participate in ongoing training by professional experts to maintain the knowledge and skills required to manage every aspect of the syndrome. Food is controlled and diet is strictly regulated. Vocational tasks and leisure activities are specifically designed to address the unique sensory motor needs of this population. Latham Centers provides nursing services and case management. Community physicians and therapists provide medical and psychological services to the residents with PWS. Leadership staff present

regularly at PWS conferences regionally, nationally and internationally. Parents involved in national and international PWS associations have asked Latham

Appendix I: General Information about Prader-Willi Syndrome:

The International Best Practice Guidelines for Standard of Care for Persons with PWS in Residential Living states that:

All guidelines should take into account the assessment of the individual to assure health and safety and to promote quality of life.

The individual's opinion [or that of the guardian] should be taken into account when implementing these guidelines whenever possible.

*The preferred form of living arrangement is in a PWS-specific environment. This allows for **consistency** in treatment and a sense of fairness to the individual. All attempts to create a family like environment including their own space regardless of group size is important for the person with PWS to have **a sense of belonging** as well as a place to disengage from group living whenever necessary. It is vital for the individual to have a choice of vocational opportunities. Enjoyment and fulfillment of one's own day is crucial for anyone to feel productive and an important member of their community.*

*Optimal success for the person with PWS will be based on the type of supports that are in place. Those supports require an understanding of the unique needs associated with PWS. All areas of a person with PWS' day should have proper supervision to assist with those unique needs. There needs to be a **24 hour type of supervision** in place in either the form of direct caregiver support, alarms or security systems that alert caregiver or monitoring agency that the individual has either left the designated area(s) or that they are in need of assistance. A combination of the above may also be used.*

At times additional support may be needed due behavioral issues or need for assistance from another caregiver. Each supportive environment, both home and vocational, should have a system in place that allows for quick response from another caregiver to assist in the need at hand.

***Routine and consistency** is essential to the successful living for a person with PWS. **Inclusion** of their ideas and preferences should be part of the planning of the structure of their regular routine. Meals should be managed and structured in a formal way to ensure that everyone knows the plan of how meals are to be planned, monitored, and served. It is necessary for every person with PWS to have a **diet plan** that can be followed by all. **Exercise** should also be a part of their daily routine including some levels of movement or fitness every day with the amount of time to be determined on individual basis. **Healthy weight management** is also an important part of monitoring the progress of supporting a*

person with PWS. Weights should be taken at a minimum of once per week based on history.

Structure of the home for a person with PWS should have some form of **house rules for living with others**. This allows for everyone to understand and remain consistent with those guidelines, expectations, and boundaries that are needed when in a group living environment. Another area is the **complete security of food, money, and medication**, which should be locked and managed by caregivers.

To assist in overall self-esteem it is important for everyone **to feel a part of his or her community**. Inclusion of those activities within the community should be individually assessed to determine environmental supports that may be needed. Understanding the person's own level of independence will assist in maintaining required needs for safety. Living in a community should include good neighbor relationships, it is important to create systems explaining about PWS.

It is essential to have **individual behavior management plans** ranging from positive motivation to crisis management. This once again allows for consistency and proper understanding of that specific individual's needs when that person may be in a crisis or to simply avoid or redirect the situation.

Training of caregivers is key to the success of the individual with PWS. Caregivers need a good understanding of the complexity of the needs of the syndrome as well as the compassion that is needed when supporting a person with PWS.

Maintaining healthy relationships once the person with PWS is outside their family's home is important for the person with PWS. Contact and involvement with family and friends may need additional support and guidance from caregivers. That support may consist of teaching relationship building and in some incidences may require additional counseling. Human sexuality may also require support and education.

See **Standards of Care and Best Practice Guidelines** for residential care of persons with PWS which was published by the International Prader-Willi Syndrome Organization in 2012. (<http://www.ipwso.org/publications/standards-of-care-best-practice-guidelines/>)

Neurobehavioral:

Prader-Willi syndrome is a complex and heterogeneous neurodevelopmental behavior disorder known to be caused by missing or unexpressed genes on the 15th chromosome. Subtle but critical

feedback circuits that enable typical individuals to modify physiological and behavioral responses to environmental challenges are faulty in PWS. The behavioral phenotype of PWS is one of the most striking and complex of all known genetic disorders. Persons with PWS do not learn from negative experiences in such a way as to prevent behavioral problems in the future. This is a complex dynamic that varies from person to persons but an overestimation of the potential for persons with PWS to learn from experience and to change their behavior is one of the most common sources of conflict with the caretaking environment.

PWS is a rare genetic disorder associated with morbid obesity and a pattern of cognitive and behavioral difficulties unique to the syndrome. The majority of individuals with the syndrome have the paternal deletion subtype (70 %) on chromosome 15. The cognitive function of individuals with PWS is rarely commensurate with their intellectual potential because of impoverished judgment due to impaired perspective taking (inability to perceive another person's point of view), deficient working memory (inability to hold two or more ideas in mind at the same time for comparison judgment), and self-centeredness/egocentrism that interferes with empathy. Thought perseveration, cognitive inflexibility, and a lack of behavioral inhibition occur frequently, and these interfere further with adaptive problem solving. The behavioral phenotype consists of oppositional behavior (arguments, tantrums, targeted retaliation, manipulation) in response to failed anticipation or disappointment. There is evidence to suggest that individuals with PWS have aberrant drive mechanisms beyond those associated with food. In some individuals, excessive and repetitive behaviors can present more of a challenge for management than food related behaviors. Unlike true OCD, people with PWS are not disturbed by their excessive and repetitive behaviors, and they do not experience disgust or shame. Emotional dysregulation can result in self-injurious behavior or rage attacks; occasionally there are psychotic underpinnings to these high intensity behaviors. Although many adults with PWS can benefit from psychotropic medication combined with psychological therapies and/or behavioral management, *the syndrome-specific intervention is a controlled environment*. Effective environmental management includes a trained team, 24-hour supervision, calorie control, mandatory exercise, FOOD SECURITY¹, a daily plan with consistency and predictability, low expressed emotion, non-contingent reinforcement and appropriate behavioral interventions. Environmental management is essential to act as a buffer for the stress sensitivity of the PWS person and their physiological inability to adapt to change. Having too much freedom with too many choices results in overwhelming anxiety that further interferes with already poor problem solving abilities. Maximizing structure, consistency and predictability can minimize the false

¹ FOOD SECURITY pertains to the psychological state where there is no doubt what food and the amount of food that the person will have and no chance that the person can access additional or different food. When these conditions are established, there is no disappointment about food, and temper tantrums about food are avoided.

expectations that lead to disappointment, emotional dysregulation, and behavioral problems. The goal for adaptive function in PWS is *maximum function with support*, not independence through self-regulation.

Weight Regulation:

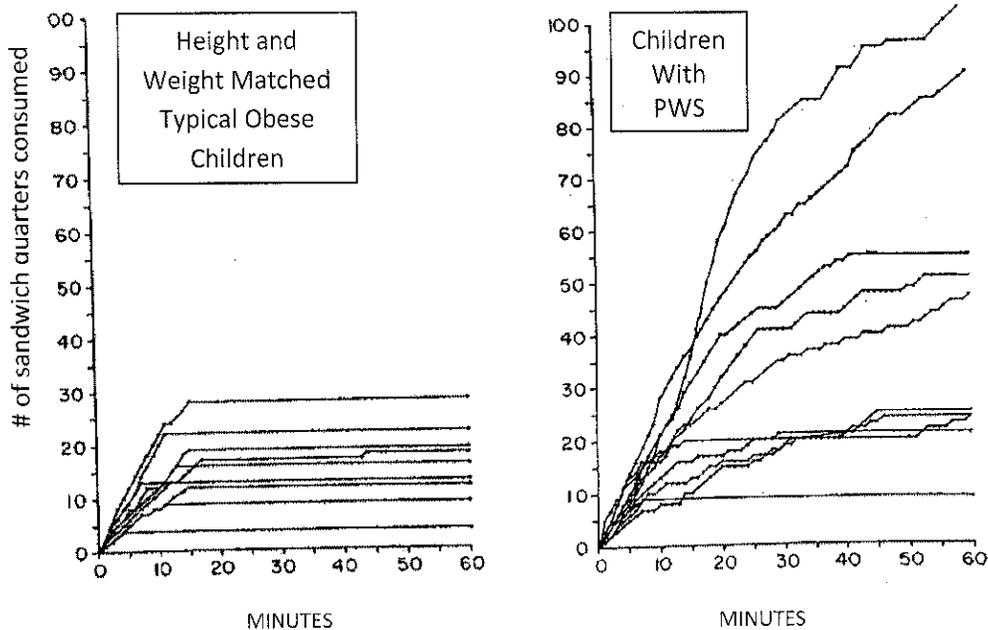
The most striking feature of PWS is a severe abnormality in the regulation of body weight. After an initial period of failure to thrive, poor weight gain and disinterest in eating that lasts during the first 1-2 years of life, children with the disorder begin to show a perceptible weight gain despite normal calorie intake and slow linear growth. If unchecked, they will consume far more calories than they burn; some have become dangerously obese by the age of 3-4 years. Prior to the availability of early diagnosis and effective interventions (mainly education and support of the families) half of persons with PWS died by age 15 years of age of obesity related complications, primarily from respiratory failure (in effect, suffocation). Obesity is prevented *only* by constant vigilance and locking of food in the home. Obesity complications are still the most common cause of death even in the USA. Among persons with PWS, acute gastric dilatation and necrosis occur both spontaneously and following ingestion of large amounts of food. There is at least one death per year due to gastric rupture caused by excessive intake. Persons with PWS have low calorie needs; they require 50-80% of the calorie needs of a typical person the same size. Most adults with the syndrome require only 1000-1200 kilocalories per day. Management of the syndrome is widely recognized to require that external controls be placed on persons with PWS so that they cannot acutely or chronically ingest unhealthy amounts of food. In addition to an unhealthy weight gain, food is problematic for persons with PWS in other ways. There is a close association between incomplete control of access to food and disruptive/aggressive behavior. When food is completely controlled the patient is able to relax and decrease his or her focus on and anxiety about food. When food is incompletely controlled behavioral incidents increase greatly in frequency because the perceived possibility of being able to obtain food creates anxiety and irritability. False expectations or food seeking endeavors that are ultimately thwarted can result in explosive anger with aggression.

Persons with PWS need *Food Security*. Food Security is a concept referring to the *psychological* security of the individual with respect to food. It encompasses far more than locks and planned menus; it requires the skillful handling of all food-related potentialities, even conversations about food. That is, all *potential* or *imagined* food access must be managed as competently and completely as real access to food. Weight control alone does not guarantee psychological food security. When food is potentially accessible, persons with PWS typically become anxious and obsessive about obtaining it, and this anxiety leads to volatile emotions and unpredictable

behavior. Since food is ubiquitous in our society and because persons with PWS are frequently willing to go to great lengths to obtain food (shoplifting, burglary, lying/ manipulation, searching through and eating garbage), all aspects of their lives must be managed in a safe environment with a trained team of individuals who understand the syndrome based upon education and experience. Experience is crucial because there is a tendency for people not to believe the syndromic behaviors until they see and experience them first hand, not once but many times. When persons with PWS enter a hospital ward, respite care situation, or detention facility, where management principles pertaining to PWS are rarely understood, the person's weight gain is alarming. We have experienced the difficulty in getting people, even medical professionals, to understand the low calorie diets and environmental controls needed for patients with PWS. Multiple failed attempts at placement and repeated crises are a common history for adults with PWS.

BELOW: SEVERAL STUDIES HAVE DEMONSTRATED THE ABILITY OF PERSONS WITH PWS TO CONSUME AN ENORMOUS NUMBER OF CALORIES IN A SHORT PERIOD OF TIME AND TO BE ABLE TO CONTINUE TO DO SO DAY AFTER DAY. SATIETY SIGNALING IS WEAK AND SHORT LIVED. IN THE STUDY ILLUSTRATED BELOW, CHILDREN WITH PWS WERE COMPARED WITH TYPICAL CHILDREN WHO WERE OBESE (WEIGHT AND HEIGHT MATCHED CONTROLS). OBESE CONTROLS REACHED SATIETY (VOLUNTARILY STOPPED EATING) IN 10-15 MINUTES BUT SOME CHILDREN WITH PWS WERE ABLE TO CONTINUE EATING FOR THE FULL HOUR OF THE STUDY CONSUMING AS MUCH AS 3000 CALORIES IN A SINGLE MEAL. THE AVERAGE CALORIE CONSUMPTION WAS OVER 1400 CALORIES, ABOUT 6 TIMES THE ACTUAL CALORIE NEEDS.

Zipf & Bentson *Am J Clin Nutr* 46:277-281:



All individuals with PWS have a psychiatric diagnosis: *A Personality Change Secondary to a Medical Condition, Prader-Willi syndrome.*

(see Appendix III: *PWS Personality*; Appendix IV: *Global Assessment of Functioning for PWS*). The stress sensitivity associated with the syndrome predisposes to co-morbid psychiatric diagnoses of anxiety, depression, and impulse control disorders. PWS individuals are likely to develop mood and anxiety disorders with or without psychosis that require treatment with mood stabilizing and antipsychotic medications. In addition, the phenomenon of mood and behavioral activation has been noted among many individuals with PWS in association with the use of some psychotropic medications including the class of selective serotonin reuptake inhibitors. Mood activation is associated with irritability or euphoria, increased goal directed behavior, increased risk-taking behavior with a high likelihood of negative outcome, impulsive self-injury or aggression, and destruction of property; it has also been associated with psychosis. Treatment involves the discontinuation of the activating agent with the resolution of symptoms in many cases. If mood activation persists, the use of mood stabilizers with or without antipsychotic agents is required.

Many individuals with PWS have intellectual disability. The average IQ is 60. They also have co-morbid learning disabilities. Math disability is very common, and language disorders are frequently observed. Nonverbal learning disorder and apraxia/dyspraxia spectrum occur with lesser frequency. Hypotonia is one of the diagnostic criteria for PWS. As a result of hypotonia, infants don't get the amount of sensory motor experience that they need. There is evidence to suggest that sensory processing and integration are also deficient in PWS. Stereotypic and self-stimulatory behaviors, such as body-focused repetitive behaviors, develop as a way to overcome this *sensory hunger*. Typically, these behaviors worsen with stress. Unfortunately, chronic lesions from skin picking can become infected and disfiguring.

Self-Injury:

Self-injurious behavior is extremely common in PWS but highly variable in intensity. Aberrant neural feedback causing decreased pain sensitivity and lack of disgust likely contributes to self-injurious behaviors. Skin picking is the most typical behavior, and it leads to open sores. It can increase with stress or persist in a chronic low-level pattern. Complications of skin picking include infection, bleeding and facial disfigurement. Simple low-grade environmental unpredictability well tolerated by other persons with mental handicap can be a contributing stressor to these and other problematic and dangerous behaviors.

Rectal picking/digging with or without fecal smearing or coprophagia is also common in PWS and is often intractable. It appears to be closely related to stress and, when frequent, leads to fecal incontinence, infection, hemorrhage and anemia. Stressed persons with PWS may engage in

rectal digging/picking continuously unless physically prevented from doing so. Rectal digging/picking is a complex phenomenon often misunderstood; it is incorrectly attributed to sexual stimulation or a manifestation of obsessive-compulsive disorder. Chronic rectal picking has led to unnecessary surgery such as colostomy but also to hemorrhage and need for transfusion.

Fecal smearing has been reported frequent in a number of patients with PWS. Coprophagia is less frequent but has also been reported. These behaviors generally appear when person are stressed or in a conflict situation.

During emotional outbursts, self-mutilation in the form of self-stabbing or gouging is sometimes seen resulting in deep tissue injury. At other times self-endangerment is displayed; stuffing objects into their airway or other body orifice is a recurring behavior. Removal of one's own teeth or toenails and peeling the skin from the soles of the feet are signature PWS behaviors.

Medical:

There are numerous medical problems associated with the syndrome, and their management often differs from interventions for typical persons. Parents as well as professionals who have experience working with this syndrome have many stories of how difficult it is to have the inexperienced (with PWS) medical professional believe what they are told about PWS. The following medical problems are most frequently observed:

- Hormonal insufficiency resulting in short stature, abnormal sexual development and brain development
- Orthopedic abnormalities resulting in scoliosis, kyphosis, lower extremity deformities and osteoporosis with fracture susceptibility due to osteoporosis or osteopenia.
- Sleep continuity and sleep transition abnormalities. They have breathing abnormalities, abnormal response to oxygen supplementation, abnormal respiratory response to CO₂ retention;
- They are at risk for choking *at all ages* due to oro-motor deficits in combination with impulsivity around food.
- Gastrointestinal abnormalities include abnormal intestinal motility and an abnormally high risk for gastroparesis (partial paralysis and dilation of the stomach) that sometimes leads to necrosis followed by perforation peritonitis and death.

- Glucose intolerance is common when persons with PWS are overweight; it appears to be the result of hypoinsulinemia rather than insulin resistance. This physiologic anomaly is usually mistreated and not always distinguished from type 2 diabetes which can also occur.
- Undefined abnormalities in physiologic feedback sometimes result in electrolyte abnormalities, temperature dysregulation resulting in coma with or without hypothermia.
- Atypical response to medications
- Sensitivity to supplemental oxygen which can suppress the breathing drive

Another factor complicating medical management is the universal propensity of persons with PWS to seek medical treatment excessively and inappropriately resulting in *over* diagnosis and *over* treatment of numerous minor complaints. While at the same time there is a well-documented tendency for serious conditions to be underreported resulting in a delay in diagnosis. Abnormalities of physiological feedback mechanisms may be responsible for a diminished pain response and a delayed or truncated inflammatory response. This puts patients at risk for misdiagnosis of serious injury or illness; infections and surgical intestinal problems are frequently undiagnosed until the patient is too critically ill to treat successfully. This latter problem leads to death several times each year according to the data kept by the national association.

Extraordinary Stress Sensitivity:

Even when food issues are properly managed, persons with PWS are extremely sensitive to perturbations in their psychological environment. Changes of routine, real disappointments or false expectations precipitate emotional outbursts that may escalate to destructive and aggressive behavior, self-injury or self-endangerment. Many of these behaviors are preventable in the correct environment with skillful management of the typical PWS personality traits: perseveration, anxiety, emotional dyscontrol, etc. Families and caregivers learn to manage these traits by creating a secure, low stress (for the child) environment. Frequently the entry into school, camp, residential care and/or hospital settings, including psychiatric or medical hospitalization, precipitates or exacerbates rather than resolves crises. ***Change of any kind, no matter how minor, can precipitate a crisis.*** Persons unfamiliar with the syndrome are unable to make the counter-intuitive judgments needed to respond effectively to the person's needs. Commonly used behavioral management techniques that are effective for other persons with developmental disabilities will often escalate rather than calm persons with PWS. When persons

with PWS display volatile behavior, they are typically over-sedated with medications when managed by persons unfamiliar with the syndrome. Death has resulted from respiratory compromise during over sedation or physical restraint or a combination of these interventions. In short, when professionals and caretakers fail to understand the syndrome and respond inappropriately, life-threatening consequences can occur.

Need for Experienced Caretakers and Specialized Care:

The outrageous behavior, seen out of the context of the Prader-Willi Syndrome, that is, outside of the context of having seen the behavior many times in many persons with the disorder, is likely to provoke irritation and anger resulting in a punitive attitude or harsh vocal tone. In an environment of less experienced caretakers, this leads to caretaker responses that perpetuate the stress and maladaptive behavior rather than reduce it. This creates a vicious cycle of additional stress that perpetuates maladaptive behavior.

Caretakers are stressed when dealing with a person who is verbally abusive, assaultive, self-injurious and manipulative. It is critical for them to feel supported by persons who have experienced what they are experiencing. They need supervisors who are able to help them to interpret the behaviors properly and to offer practical advice on how to respond. A supervisor who has not had this practical, personal experience is at a disadvantage and may *appear* to be non-supportive. Then, the staff loses confidence in their supervisors and vice versa. The result is a systemic breakdown with a loss of communication and anger; this precipitates staff turnover, which ultimately increases environmental stress. *The manifestations of PWS behavior have put many systems of care into crisis.*

Mismanagement of food issues as well as nonfood issues is common among treatment teams who have limited PWS experience. Many of the errors made by professionals and/or caretakers are as simple as saying the wrong thing at the wrong time or phrasing the response in the wrong way. Even physicians unfamiliar with the behavioral phenotype of the syndrome should never see a patient without the assistance of an experienced PWS team member. Privacy rights sometimes interfere with this precaution, but failure to observe this rule can result in over-management, serious noncompliance with medical recommendations, or sudden emotional outbursts and *dangerous* behavior.

Attempts to combine the residential care of persons with PWS with persons with those who have other developmental disabilities, even in the hands of experienced providers, have been largely unsuccessful. An environmental setting dedicated to serving individuals with PWS has been the

most consistently effective model for providing residential care and vocational programming, especially for the more challenging cases.

Layering of experience in the hierarchy of supervision:

A system of care for PWS that has experienced supervisors and administrators is able to respond to the needs of the PWS residents and direct care staff with maximum success. Studies have shown that caring for individuals with PWS creates the highest level of stress for caregivers as well as administrators compared to those who work and manage systems of care dedicated to individuals with other or mixed developmental disabilities. Ongoing supervision and assistance in crises by experienced staff is one way to guarantee effective responding as well as minimization of staff burn out. A new house in a new community will take time to develop caregiver expertise (in a trial and error fashion) and a relationship with the community at large, unless the staff and supervisors have experience working with individuals with PWS.

Appendix III: PWS Personality

Abstract presented at PWSA-USA Scientific Conference 2006

The PWS PERSONALITY: What is "within normal limits" for PWS?

Janice L. Forster, MD and Linda M. Gourash, MD

The Pittsburgh Partnership Pittsburgh, Pennsylvania

Among individuals with PWS, some temperamental and behavioral characteristics are so commonly present that they are considered to be part of the **behavioral phenotype**. These features are the "background noise" when considering psychiatric diagnoses in patients with PWS; changes in the severity of these symptoms are clues to changes in mental status. *All of the characteristics of this phenotype can become exaggerated with stress.*

The behavioral phenotype of PWS defines the PWS PERSONALITY; it corresponds to the DSM IV Axis I Diagnosis of *Personality Change Due to a Medical Condition (310.1)*. There are five domains of psychiatric/behavioral symptoms in the PWS PERSONALITY: food related behaviors, oppositional defiant behaviors, cognitive rigidity/inflexibility, anxiety/insecurity, and skin picking.

PWS PERSONALITY: FIVE DOMAINS of PSYCHIATRIC/BEHAVIORAL SYMPTOMS in PWS

I. Food related behaviors (in order of ascending severity):

- overeating of typical food
- eating atypical food (frozen, raw, spoiled food or pet food)
- sneaking food in the home
- night time foraging in the home
- arguing or manipulating to get food
- tantrumming to get food
- opportunistic food theft (shoplifting from a store or stealing food from school or work)
- planned food foraging expeditions in the neighborhood or community
- nonconfrontational, invasive food access (breaking locks on cabinets, refrigerator or freezer)
- confrontational food access (using verbal or physical threats or actual aggression to access food)

IV. Cognitive rigidity/inflexibility:

- perseveration, "sticky thinking"
- inability to tolerate uncertainty
- difficulty with transitions or changes
- selective interests (jig saw puzzles, word searches)
- impaired judgment
- single mindedness: difficulty taking multiple view points
- egocentrism

II. Oppositional defiant behaviors:

- noncompliance
- argumentativeness
- tantrums/shut downs
- manipulation
- lying/confabulation

III. Anxiety/insecurity:

- stress sensitivity
- somatic complaints
- stimulus seeking
- constant need for reassurance
- collecting and hoarding
- affective lability

V. Skin picking:

- occurs commonly, but not universally
- as a habit behavior, opportunistic typography:
 - arms, face and scalp
 - nasal septum
 - pulling out toe nails
 - peeling skin from the soles of the feet
 - trichotillomania
- as an intense, severe, reactive behavior in the presence of ongoing stress:
 - gouging, self mutilation
 - targets rectum and/or vagina

Appendix IV: Global Assessment of Functioning (GAF) Scale for PWS

Code

- 100 Superior functioning in a wide range of activities; life's problems never get out of hand;
I sought out by others because of positive qualities; understands diagnosis of PWS, but
91 does not require any adaptations; healthy weight; no psychiatric symptoms.
- 90 Absent or minimal symptoms; good functioning in all areas; interested and involved in
I a wide range of activities; socially effective; satisfied with life; no more than everyday
81 problems or concerns; aware of PWS issues, integrates diet/exercise into daily program.
- 80 If symptoms are present, they are transient and expectable reactions to psychosocial
I stress; no more than slight impairment in social/occupational/school function; aware of
71 PWS issues and compliant with diet/exercise program; asks/accepts help when needed.
- 70 Some mild symptoms (e.g., sad mood, sleep/wake problems, mild food behaviors) OR
I some difficulty in social, occupational or school function (e.g., difficulty with transitions;
61 noncompliance, theft within the household), but has meaningful interests and activities.
- 60 Moderate symptoms (e.g., labile affect, thought perseveration/repeatedly asks) OR
I moderate difficulty in social, occupational or school function (e.g., argumentative, temper
51 tantrums, shutdowns, bully or victim, lies/barters for food, collects/ hoards, mild skin picks).
- 50 Serious symptoms (e.g., suicidal ideas, cognitive rigidity, tells stories, steals food outside the
I home) OR serious impairment in social/occupational/school function (e.g., moderate and
41 persistent skin picks, inserts objects into body orifices, tantrums with aggression/destruction).
- 40 Some impairment in reality testing or communication (e.g., tells whoppers, grandiose/fixated
I ideas, dials 911, planned food acquisition) OR major impairment with school, family, work
31 (e.g., injures small animals, refuses to attend program skin, picks require medical attention).
- 30 Behavior is influenced by delusions or hallucinations or serious impairment in judgment or
I communication (e.g., erotomanic attachments, engages in rectal picking, runs away,
21 disrobes in public, stays in bed all day, suspended from school/work/community program).
- 20 Some danger to self or others (e.g., attacks self/others with sharp objects, swallows inedible
I items, confrontational food theft, homicidal/suicidal threats, gouges self) OR poor hygiene
11 (e.g., smears feces, soils/urinates inappropriately) OR refuses meals/preferred activities.
- 10 Persistent danger of hurting self or others (e.g., persistent rectal picking or attempts, property
I destruction, arrest, suicidal behavior/self injury, homicidal behavior, fire setting) OR inability
1 to care for self (e.g., stays in bed; urinates/soils bed) OR confusion; requires constant care.

Appendix V: The Consultants

The consultants have provided patient-centered consultation to PWHO as well as to other facilities in the USA, Europe, Canada, New Zealand and Israel and have toured other PWS-dedicated facilities in the USA and Europe. (France, Norway, Finland, Germany) and are therefore familiar with the challenges associated with providing quality care to persons with PWS and with the limited options available in both the USA and throughout the world. They are both invited speakers to the International Caregivers conference for PWS held biannually in Germany. Dr. Forster edited the *Standards of Care and Best Practice Guidelines* for residential care of persons with PWS which was published by the International Prader-Willi Syndrome Organization (<http://www.ipwso.org/publications/standards-of-care-best-practice-guidelines/>). Both physicians are contributing authors for this publication.

Janice L. Forster, MD:

Affiliations: Pittsburgh Partnership, Specialists in Prader-Willi Syndrome; Prader-Willi Syndrome Association (PWSA) USA Clinical Advisory Board (www.pwsausa.org); International Prader-Willi Syndrome Organization (IPWSO) Scientific Advisory Board (www.IPWSO.org); Founding Board member: Professional Provider Caregiver Board (2010-2017), Contact email address: janiceforstermd@aol.com; pws@pittsburghpartnership.com.

Dr. Forster is an ABPN board certified General, Child and Adolescent Psychiatrist in private practice in Pittsburgh, PA who specializes in Developmental Neuropsychiatry. She has had 30 years of clinical experience in the evaluation and treatment of individuals with developmental disabilities. During the ten years that she served as a psychiatric consultant to an inpatient rehabilitation program, she evaluated over 250 individuals with PWS and has managed the most severe manifestations of the disorder across all levels of care. She serves as a consultant for the Prader-Willi Syndrome Association of the USA and serves on the scientific board of International Prader-Willi Organization. Dr. Forster is co-founder of Pittsburgh Partnership, Specialists in PWS. She has presented by invitation nationally and internationally on the behavioral phenotype of PWS and psychiatric assessment and management of children and adults with the syndrome. Dr. Forster is featured in a DVD produced by the PWSA and IPWSO; her presentations have been recorded for more widespread distribution. Dr. Forster edited the *Best Practice Guidelines for Standards of Care* persons with PWS in residential care which was published by the International Prader-Willi Syndrome Organization (<http://www.ipwso.org/publications/standards-of-care-best-practice-guidelines/>)

Dr. Forster is a graduate of the University of Pittsburgh School of Medicine (1977); she completed her combined Pediatric Internship, Child and Adolescent Psychiatry and General Psychiatry residency through the Western Psychiatric Institute and Clinic of the University of Pittsburgh Medical Center (UPMC) in 1982. She is a diplomat and committee member of the American Board

Appendices: Pittsburgh Partnership Advocacy Letter

of Psychiatry and Neurology. She serves on the Steering Committee for Certification in her specialty of Child and Adolescent Psychiatry.

Linda Gourash, MD:

Affiliations: Pittsburgh Partnership, Specialists in Prader-Willi Syndrome; Prader-Willi Syndrome Association (PWSA) USA Board of Directors (2006-2009); PWSA-USA Clinical Advisory Board; Clinical Advisor and Speaker, International Prader-Willi Syndrome Organization (IPWSO) (www.IPWSO.org); Society for Developmental and Behavioral Pediatrics; Contact email address: pws@pittsburghpartnership.com

Dr. Gourash is a board certified in Pediatrics and in Developmental and Behavioral Pediatrics. She has worked with behavioral disorders and medical problems in the developmentally handicapped for over 30 years beginning as full time faculty in the departments of Pediatrics and Psychiatry of the University of Pittsburgh School of Medicine. She began her focus on Prader-Willi Syndrome upon becoming the program medical director of the Prader-Willi Syndrome and Behavioral Disorders Unit of The Children's Institute in 1999 (This took place after a 6 month mentoring relationship with her predecessor). Dr. Gourash led a team of clinicians managing hundreds of children and adults with PWS in medical crisis from some combination of extreme obesity, diabetes, obesity hypoventilation, right heart failure and respiratory failure, severe behavioral dyscontrol, psychosis and other mental disorders

Dr. Gourash is co-founder of the Pittsburgh Partnership (2004). Together with Dr. Janice Forster she teaches and writes about the practical management of persons with PWS and provides consultation for physicians, agencies and schools in managing complex cases of PWS. Their "Psychiatrists' Primer for PWS" and many other monographs are widely used by clinicians in the USA and overseas and is available as a pdf file from the PWSA-USA website. Their DVD, "**Food, Behavior and Beyond**" is used in the USA, Europe and South America for training caretakers and is very popular with parents. Much of their work has been translated by volunteers in other countries for use by parents and professionals.

Dr. Gourash is a graduate of Georgetown University School of Medicine and did her post-doctoral residency and fellowship training at the Children's Hospital of Pittsburgh (University of Pittsburgh School of Medicine).

Disclosures:

Prader-Willi syndrome (PWS) is served by a limited number of professionals and facilities throughout the USA and the world. Being a small, nationwide community of several thousand families and professionals, familiarity is commonplace. Dr Forster has been a consultant to LathamCenters since 2007, and Dr Gourash has been a consultant since 2012. Both of the

Appendices: Pittsburgh Partnership Advocacy Letter

consultants have served on committees, workgroups, advisory boards and as conference faculty with senior members of the Latham Centers management team.

Full CVs available by request.