People With Strength

Newsletter for parents by parents

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Prader-Willi Syndrome Association of South Africa Non-profit Organisation No. 035-837-NPO, PBO Exemption No. 930 016 853, PO Box 2399 Brooklyn Square 0075, www.praderwilli.org.za

We are fortunate to have Janice Forster in Cape Town

We trust that the knowledge you gained from the lectures as well as the information at your disposal in this special edition of *People With Strength* will help you to understand this complex genetic condition. Let us always remember: behind the syndrome, behind the mass of information, is a sensitive human being, a person in his or her own right with an individual personality and a complex emotional life, who deserves to be accepted for what he or she is.

CONTENTS

WHAT IS PRADER-WILLI SYNDROME?

- Genetic disorder
- Floppy baby (low muscle tone)
- Feeding difficulties
- Cognitive impairment
- Increased appetite
- Obesity
- Food related behaviour problems
- Obsessive-compulsive features

Chairperson: Rika du Plooy rikadup@mweb.co.za 012 344 0241
Medical Advisor: Dr Engela Honey engela.honey@up.ac.za 012 319 2269

AAN DIE LESERS

Baie welkom aan almal by hierdie spesiale lesings, waar ons die voorreg het om te deel in die ervaring en kennis van dr. Janice Forster. Dr. Forster is 'n bekende in die PWS-kringe en van haar artikels is al voorheen in *People With Strength,* die nuusbrief van PWSV (SA), geplaas. Ons dank aan haar vir die tyd in die Kaap wat sy afstaan om inligting met medici, ouers en ander te deel.

In hierdie spesiale uitgawe van die nuusbrief is 'n klompie opvoedkundige materiaal saamgevat. Die Prader-Willi-sindroom is so kompleks dat dit moeilik was om te besluit watter inligting om te plaas. Die meerderheid artikels is prakties van aard en vyf is uit die pen van Famcare. Hierdie internasionale komitee het ten doel om aan ouers van volwassenes met PWS, wat in die huis woon, ondersteuning te bied. Ouers met jonger kinders kan ook waardevolle wenke hier kry. Meer oor hierdie komitee elders in hierdie uitgawe.

Ouers en betrokkenes moet hulself met kennis oor PWS bemagtig. Antwoorde is nie altyd beskibaar nie, maar soek die eienskappe van jou kind in die inligting en probeer om hom of haar werklik te verstaan. Begrip dra by tot meer effektiewe bestuur van die probleem, maar dit bly 'n voortdurende groeiproses en 'n soektog na inligting.

Ouers word aangeraai om inligting oor PWS met ander te deel. Moenie huiwer om hulp te vra nie. Begin vroegtydig en bou aan 'n oop en eerlike verhouding oor die sindroom, wat jou kind raak, met almal om jou. Ek vertrou dat daar in hierdie artikels vir elkeen iets waardevols sal wees. Hierdie uitgawe is ook elektronies beskikbaar. Kontak gerus die voorsitter: rikadup@mweb.co.za

PWSV (SA) is trots om deel te wees van die *International Prader-Willi Syndrome Organisation* (IPWSO). Hierdie organisasie is 'n niewinsgewende ondersteuningsgroep en is in 1991 gestig. IPWSO ondersteun alle PWS-organisasies wêreldwyd. PWSV (SA) is een van 102 lande wat lid is van IPWSO. Elke drie jaar bied IPWSO 'n internasionale konferensie aan. Die *9th International PWS Conference* word in 2016 in Toronto, Kanada aangebied. Vir meer inligting oor IPWSO: www.ipwso.org

Dr Suzanne Cassidy, die president van IPWSO, word deur die *American Society of Human Genetics* (ASHG) op 'n spesiale wyse vereer. Lees meer oor hierdie bekwame vrou in hierdie uitgawe.

Dit is een van die doelstellings van PWSV (SA) om aan ouers en ander belanghebbendes ondersteuning te bied. Sluit gerus by PWSV (SA) aan – ouers, familie, vriende, professionele persone en versorgers. Almal is baie welkom. Meer inligting oor lidmaatskap op die laaste bladsy van hierdie nuusbrief.

Vriendelike groete
Rika du Plooy
Voorsitter

TO OUR READERS

Welcome to all who decided to attend the lectures of Dr Janice Forster. She is well known in the world of PWS and some of her articles were previously published in *People With*

Strength the newsletter of PWSA (SA). We thank Dr Forster for putting aside a day in the lovely Cape to share her knowledge and experience with health care professionals, parents and others who are involved with and interested in PWS.

In this special edition of the newsletter you can read some educational material. The Prader-Willi syndrome is so complex that it was difficult to choose from a broad variety of articles. Most of the articles in this edition are of a practical nature. Five are from the pen of Famcare. This international committee has been formed for the purpose of supporting families who have their adult child with PWS living in the home with them. However, parents of younger children should also take note of the valuable hints given by this committee. It is imperative that the correct behaviour must be learned from a very young age. More information about this committee is given elsewhere.

We implore parents to gain as much knowledge as possible on PWS. Answers aren't always available, but try to trace the characteristics of your child in the information provided. You may be able, eventually, to understand him or her better or even fully. Understanding the problem contributes to a more effective way of management. But keep searching for knowledge and answers.

Parents are encouraged to share information on PWS and you are welcome to hand this special edition to everybody concerned. Share information with others and do not hesitate to ask for help. Start now by building open and honest relationships about the Prader-Willi syndrome with everybody around you. I trust that every reader will find something useful in all these articles. This special edition is also available electronically. Please contact the chairperson: rikadup@mweb.co.za.

PWSA (SA) is proud to be part of the *International Prader-Willi Syndrome Organisation* (IPWSO). This international organisation is a non-profit support group for all countries and was founded in 1991. IPWSO supports all PWS associations worldwide. IPWSO has 102 member countries of which South Africa is one. Every three year IPWSO holds an International PWS Conference, hosted by different nations. The 9th International PWS Conference is to be held in Toronto, Canada and will be hosted by the Federation for PW Research and the Canadian PWS Association.

Dr Suzanne Cassidy, the president of IPWSO was honoured by the American Society of Human Genetics (ASHG) for excellence. Learn more about this exceptional lady in this edition. More information on IPWSO: www.ipwso.org

PWSA (SA) aims to support all those who are involved with individuals with PWS. Parents, family, friends, health care professionals, carers are invited to join the PWSA (SA). Please find information on membership on the last page of this newsletter.

Sincere regards,
Rika du Plooy
Chairperson

The Australian and New Zealand Prader-Willi Syndrome Associations invite you to Melbourne in April 2015, to join them at the 3rd Asia Pacific Prader-Willi Syndrome Conference. Email: conference@pws.org.au.

Dr Janice Forster delivered two lectures at the Red Cross WM Children's Hospital, Cape Town on 3 October 2014.

WHO IS JANICE FORSTER?

Dr Forster is a Child and Adolescent Psychiatrist in private practice, specializing in Developmental Neuropsychiatry. She has had over 30 years of clinical experience evaluating and treating individuals with intellectual disabilities. Together with Linda Gourash, MD, she is co-founder of the *Pittsburgh Partnership, specialists in PWS,* providing consultation and education to families and organizations who care for people with PWS. Dr Forster has assessed and managed the severe manifestations of PWS at all ages and across all levels of care. Currently, she serves as a consultant to the Prader-Willi Syndrome Association of the USA and the International Prader-Willi Organisation (IPWSO). Also, she is a member of the Scientific Board and the Professional Provider Care Giver Board of IPWSO.

AMERICAN SOCIETY OF HUMAN GENETICS TO HONOUR SUZANNE CASSIDY

Edited by Sara Dwyer, Editor, The Gathered View, Volume 39, Number 5 September-October 2014

PWSA (SA) congratulates Dr Suzanne Cassidy with this well-deserved award. Dr Cassidy is currently President of the International Prader-Willi Syndrome Organisation (IPWSO). She also has served for many years on the PWSA (USA) Scientific Advisory Board.

The American Society of Human Genetics (ASHG) has named Suzanne B. Cassidy, M.D., Clinical Professor of Pediatrics in the Division of Medical Genetics at the University of California, San Francisco, as the 2014 recipient of the annual Award for Excellence in Human Genetics Education.

The ASHG award recognizes an individual for contributions of exceptional quality and importance to human genetics education internationally. Awardees have had long-standing involvement in genetics education, producing diverse contributions of substantive influence on individuals and/ or organizations. Dr. Cassidy will receive her award during ASHG's 64th Annual Meeting in San Diego.

Dr. Cassidy is well-known for her clinical and research leadership in Prader-Willi syndrome. She has also played key roles in the medical genetics education of medical students, residents, and genetics trainees as well as of patients and their families. She has developed a variety of education materials, including three editions of the textbook Management of Genetic Syndromes and clinical genetics training programs across the country.

"Dr. Cassidy has worked tirelessly to improve genetics education and support patients in a variety of roles – as a teacher, mentor, physician, author, and advocate. This award celebrates her contributions to science, medicine, and the patient experience," said Joseph McInerney, M.A., M.S. and Executive Vice President of ASHG.

Throughout her career, Dr. Cassidy has received numerous honours for her research and teaching, including election to several advisory boards, founding editorship for clinical genetics in the journal Genetics in Medicine, and visiting professorships at institutions in the

United States and abroad. In addition, she was a member of the founding Residency Review Committee for Medical Genetics when it was first recognized as a medical specialty, and belonged to the American Board of Medical Genetics and Genomics, which accredits genetics training programs and certifies medical geneticists.
Since its foundation, Dr. Cassidy is past chair and continues to serve as a member of the PWSA (USA) Scientific Advisory Board. She also gives tirelessly as president of the International Prader-Willi Syndrome Organisation.
Congratulations on this well-deserved award from the American Society of Human Genetics. This is a very prestigious honour few people ever achieve.

DIFFERENCES BETWEEN THE GENETIC SUBTYPES OF PRADER-WILLI SYNDROME

By Dr Suzanne Cassidy

Permission is granted by PWSA (USA) and *The Gathered View* to publish this article.
***The Gathered View* (ISSN 1077-9965), September-October 2012**

WHY DO DIFFERENT GENETIC CHANGES CAUSE PWS?

PWS is always due to the same thing: deficiency in the expression of certain genes on chromosome 15. Specifically, the deficient expression is from genes that are normally expressed from the member of the chromosome 15 pair that was inherited from the father, because the copy of these genes on the chromosome 15 inherited from the mother is normally not expressed ("imprinted"). This lack of expression on the maternally-inherited copy is the result of a chemical reaction called DNA methylation. So, in the general population, the genes relevant to PWS are only expressed (like reading a blueprint) from the chromosome 15 inherited from the father, and in PWS, that copy is missing (when there is deletion or uniparental disomy); or else the chromosome 15 inherited from the father is erroneously switched off ("imprinted") like the genes on the chromosome 15 inherited from the mother (an imprinting defect).
A deletion is a missing piece of a chromosome. In PWS, the missing piece is at the top of the long arm (called q) of chromosome 15, in a region called 15q11.2-q13. There are three common points along the chromosome in which the break occurs, two at one end with four genes between them and one at the other end. The type of deletion missing the larger piece of genetic material is sometimes called a Type 1 deletion, and the smaller deletion is sometimes called Type 2. A few deletions are larger or smaller than the common deletions, but most genetic testing for PWS does not identify those differences.

Maternal uniparental disomy (also called UPD) is a situation in which both of the members of the chromosome 15 were inherited from the mother (and the genes relevant to PWS are therefore not expressed from either copy). The chromosome 15 that should have been inherited from the father—the one with the expressed genes for PWS—is missing. This situation is somewhat more common among children with PWS born to women over age 35 years.

An imprinting defect is a condition in which there is one chromosome 15 from the mother and one from the father, but the one from the father behaves as though it were inherited from a mother, at least as far as the genes for PWS are concerned. Those genes relevant to PWS are switched off in both the chromosome 15 inherited from the mother (as is normal) and the one inherited from the father (which is not usual). Most cases of imprinting defect

are random and of unknown cause, but a small percent are due to a small deletion in the i1mprinting center which is responsible for switching genes nearby off and on (applying or erasing DNA methylation). In some families that have a child with PWS due to an imprinting center deletion, there is a significant risk to have another affected child. Therefore, in families planning future children, further genetic testing and genetic counselling are important.

A few people who have PWS are found to have a **"translocation**", which is a rearrangement of two chromosomes such that part of one is stuck onto part of another. Most of the time that this happens in someone with PWS, the translocation involves one chromosome 15 and there is a deletion on the rearranged chromosome. In that case, the individual in effect is like people with PWS due to a deletion. Occasionally, a translocation can cause uniparental disomy 15, and the individual in effect is like people with PWS due to UPD. Sometimes with a translocation, there is an effect from another missing piece of chromosome in addition to it causing PWS.

WHAT ARE THE DIFFERENCES IN THE EFFECTS OF THE DIFFERENT GENETIC CAUSES OF PWS?

Researchers, by studying many people with each genetic type of PWS (primarily the most common deletion and uniparental disomy types) have identified a few differences between the groups. Most of these differences are in the frequency or severity of findings in the two groups. People with an imprinting defect appear to be most similar to those with UPD.

Before mentioning these differences, it is very important to explain that these are GROUP differences, not individual differences. *There is no feature that is exclusively found in one of the three genetic categories.* And it is also important to recognize that even within a genetic type there is a lot of variation among people affected with PWS, so there is a lot of overlap in the genetic groups. For example, even though the people with UPD as a group have a slightly higher mean intelligence quotient (IQ) than people with deletion as a group (8 points, with an IQ of 100 being the average in the general population), there is a very wide range of IQ within each genetic group (about 40-90). So in an individual, knowing the genetic type is not very helpful in knowing the future abilities or problems of the individual.

In general, and as a group, people with a deletion are more likely to have fair colouring (hair, eyes, skin), whereas those with UPD more closely resemble their parents. People with a deletion are more likely than those with UPD to have the characteristic facial appearance of PWS. And they are more likely to be skilled with jigsaw puzzles.

In general, and as a group, people with UPD are more likely to be born late. As noted above, they have a slightly higher average IQ than the group with deletion. In addition, they have somewhat milder behaviour problems (temper tantrums, stubbornness, repetition and controlling behaviour). However, it appears that people with UPD as a group have a significantly higher incidence of autistic characteristics and of psychiatric disorders, including psychosis, than those with deletion.

In addition to these differences, there are some studies suggesting somewhat worse behavioural problems in those with a Type 1 (slightly larger) deletion versus those with a Type 2 deletion. There are also some studies that have not shown these differences.

It is important once again to stress that knowing the genetic type does not predict the manifestations of PWS in an individual. It is primarily important for genetic counselling purposes, as far as we know today.

WHY ARE THERE CLINICAL DIFFERENCES BETWEEN THE DIFFERENT GENETIC TYPES?

The genetic or biological basis for these differences has not been determined. In people with a deletion, a number of genes are missing, and some of those genes are normally expressed from both members of a chromosome pair. In people with UPD or an imprinting defect, the genes are not actually missing, but are just not being expressed (turned into messenger molecules, regulators or proteins). This presence of some genes that, as far as we know, are not the ones causing PWS but are also in the chromosome segment included in the deletion, may have an impact on the clinical manifestations in an individual. There is still much to learn about the genetics of PWS!

THE DIAGNOSIS OF PRADER-WILLI SYNDROME IN SOUTH AFRICA

Dr Engela Honey is at the Department of Genetics at the University of Pretoria and she is also the medical advisor of PWSA (SA)

The suspected diagnosis of PWS is usually made by a paediatrician or other health care professional based on suggestive clinical symptoms such as poor feeding and hypotonia after birth and uncontrollable eating behaviour and weight gain later in life. (Consensus diagnostic criteria for PWS, Holm et al 1993) This suspicion must be confirmed with genetic testing and most of the diagnostic tests are available in South Africa.

The preferred test is the “methylation analysis” which detects all the major genetic subtypes of PWS (deletion, uniparental disomy and an imprinting mutation) in more than 99% of cases. This test is unfortunately not able to distinguish between the different subtypes and for that further expensive testing is necessary. This is therefore not routinely performed for establishing the diagnosis. A “FISH” (fluorescent in-situ hybridization) test will pick up a deletion but will miss the other subtypes. In very rare cases the syndrome can be due to a large deletion which will be detectable by standard chromosome studies and in those cases it is recommended that chromosomes should be done on the parents.

Abroad a new test called a micro-array is commonly used in place the FISH test but this test is unfortunately not available in South Africa. A test called a MLPA (“multiplex ligation probe analysis”) which test for microdeletions and duplications (small pieces of chromosome material missing or extra) has recently become available in South Africa and can also be used replacing the FISH test.

It is however important that all genetic testing is accompanied with genetic counselling done by a qualified professional who can explain the implications of the diagnosis, the different genetic tests and their limitations as well as the risk of recurrence in future pregnancies.

Testing in South Africa is centralized to a few laboratories but all the major laboratories will offer testing but will outsource the test to the relevant laboratories. The cost for testing may vary between laboratories and will be available on request.

PEOPLE WITH STRENGTH

All the newsletters since 2002 are loaded on the website of the PWSA (SA).
Please view on www.praderwilli.org.za

GASTROPARESIS: THE NEWEST THREAT

by Lisa Graziano, M.A., PWCF Executive Director
Janalee Heinemann, M.S., PWSA (USA) Director of Medical Affairs
Ann Scheimann, M.D., M.B.A., Gastroenterologist and PWS Specialist

An urgent request from the PWSA (USA): *Please share this important information with families that may not be PWSA (USA) members or do not go to the PWS internet sites.*
This article will be posted in the medical section at www.pwsausa.org

If you're a member of the Prader-Willi California Foundation and the national Prader-Willi Syndrome Association (USA), then you have read about or discussed at a meeting of some sort this thing called gastroparesis. There have been articles about the fact that it exists, alerts about it (Medical Alert: Gastrointestinal Issues in Individuals with PWS), and now a peek at the likely incidence rate. What we haven't yet received enough information about is how do we know if our child/adult has it and what can we do about it. This is the focus of this article.

Please know that this will likely be a difficult read for those of us who care for and love someone with PWS. Knowledge is power, however, and so if the information contained within this article helps inform care providers enough that they can keep someone with PWS safer, then the read is worth it.

In 1999 The Gathered View included an article about the discovery by PWS specialist Rob Wharton, M.D., of what he termed Acute Idiopathic Gastric Dilation. What Dr. Wharton saw in his patient was that for some unknown (idiopathic) reason the stomach (gastric) was quickly (acute) pushed out (distended), causing the stomach tissues to die. If not immediately treated with surgery, this condition may lead to death.

Over the following years, particularly with closer examination by PWS/GI specialist Ann Scheimann, M.D., it has become clearer that a great number of other people with PWS have a stomach that empties too slowly. In fact, Dr. Scheimann now believes **it is highly probable that a significant number of people with PWS have some degree of a slow emptying stomach.** The medical name of this disorder is gastroparesis: the muscles in the wall of the stomach work poorly and prevent the stomach from emptying properly. As a result, food stays in the stomach longer than it should. Over time, the volume of accumulated food in the stomach can cause the stomach to become full. Like a balloon that has too much air, the stomach of someone with PWS that contains too much food can respond in one of two ways: it will rupture or the food will push so hard against the stomach lining that it "compresses and weakens" the cells in the stomach. Both of these conditions cause massive internal infection and can quickly lead to death. *(Please note that there has typically been a prior eating binge with most incidents of GI necrosis and death.)*

Other important factors to consider are that some medications such as narcotic pain relievers and anticholinergic medications (group of bronchodilators) can also cause the stomach to empty too slowly (as well as cause dry mouth symptoms). Abnormally high blood glucose (sugar) levels or undetected hypothyroidism can also slow stomach emptying; therefore, it is important to control blood glucose levels and screen periodically for hypothyroidism.

The symptoms of a slow emptying stomach are primarily nausea, vomiting, abdominal fullness after eating, and/or pain. But for persons with PWS who often have a blunted pain threshold and an absent vomit reflex, symptoms of gastro-paresis or Acute Idiopathic Gastric Dilation can be extremely difficult to detect.

At the same time the stomach empties too slowly, the bowel intestinal tract seems to empty too slowly. This means that digested food that the body turns into waste product and must eliminate from the body as faeces/stool is not entirely eliminated, leaving too much stool in the intestinal tract.

Many parents and care providers believe that because their child or adult has a bowel movement every day, this means they don't have a slow emptying bowel. This is not necessarily true. Even with a regular daily bowel movement, the intestinal tract may not empty appropriately. As the colon becomes more backed up with retained stool, the ability to evacuate stool is less effective. Over a long period of time, continuous, constant hard pushing has resulted in some people with PWS experiencing rectal prolapse. (The feeling of constant fullness and pressure on the anus or itching of the skin from irritation from bile acids present in the stool can contribute to reasons that some people with PWS insert their fingers into their anus or pick at it.)

As the colon becomes more impacted with retained stool, emptying of the stomach commonly slows down. This means that the risks of gastric rupture or dilation are dangerously elevated.

How to Detect Gastroparesis and Slow Emptying Bowel

How do we know if the individual with PWS we're caring for has gastroparesis or a slow emptying bowel? What are the signs? What are the symptoms? What do we look for? The answers are, unfortunately, that there probably aren't many easily recognizable signs or symptoms.

Because the abdominal core muscles are generally weaker in persons with PWS, the stomach can often appear to be more rounded. If food is not emptied quickly enough, the stomach can look rounded (distended) and feel "too firm" to the touch. On the other hand, for those who are taking growth hormone medication and are therefore leaner, the stomach can already feel "firm" to the touch.

The definitive test to identify delayed stomach emptying is the Gastric Emptying Study which measures the amount of time it takes for food to empty from the stomach and enter the small intestine. The test is done in the nuclear medicine section of a hospital. The patient fasts overnight and eats a breakfast that contains a tiny amount of radioactive material. The patient then lies flat and still on an exam table under a large "arm" that measures the amount of food particles that evaporate from the stomach over a period of time; generally four hours is the appropriate amount of time for the emptying study following a mixed meal of liquid and solids. There are no side effects from a gastric emptying study; the radioactive material is not absorbed into the body and is eliminated in the stool. The test can be difficult for kids under the age of 10 to complete because it is critical that the person lie perfectly still throughout the duration of the test. It is important to make certain that other factors such as constipation and/or thyroid disease are well controlled prior to completion of the test.

Treatment Strategies

1. As with all treatment of PWS symptoms, the first approach is to always provide Food Security: a) a healthy, low-calorie, low carbohydrate diet; b) meals and snacks served at structured times/sequences throughout the day; and c) all access to food restricted.
2. Request from PWSA (USA) information about GI issues in persons with PWS.
3. If there are GI concerns present, consider consultation with either a paediatric or adult gastroenterologist, dependent upon age. Provide the physician with your GI issues documents.
4. Discuss the pros and cons of a Gastric Emptying Study.
5. Discuss the use of medications such as metoclopramide (Reglan) and erythromycin to improve stomach emptying.
6. Discuss an assessment for stool build-up (e.g., palpitation, x-ray). The Bristol Stool Chart can be used to screen/track progress with management of constipation.
7. Normal stools should be Bristol Class 4 (See Table on website) http://en.wikipedia.org/wiki/Bristol_Stool_Scale
8. Discuss the use of over-the-counter medications such as Miralax to improve stool elimination and over-the-counter probiotics to help regulate the balance of helpful organisms (micro-flora) in the intestines.
9. If there are challenging issues for your primary GI specialist physician, suggest the GI specialist contact a PWS GI specialist by contacting PWSA (USA).

We continue to learn more about the gastrointestinal and bowel emptying issues of PWS. We have a lot of questions and some theories, but no definitive answers. As we do, we will inform you. Janalee Heinemann.

INFORMATION FOR SCHOOL STAFF: SUPPORTING THE STUDENT WHO HAS PRADER-WILLI SYNDROME

Compiled by Barb Dorn, Crisis Counsellor PWSA (USA) www.pwsausa.org
All students with PWS are individuals. Each has varying strengths and needs.
This chart does not reflect the behavioural needs of all children and young adults.

Common Behaviours often seen in Students with PWS	Possible Management Strategies
Rigid Thought Process It is common for people with PWS to receive and store information in a very orderly manner. There is a strong need for routine, sameness, and consistency in the learning environment.	• Foreshadow changes and allow for discussion. Do this in a safe area where they can share feelings. (The student needs to time to adapt to this change) • If there is a change - use visuals; put things in writing – lists, schedules • If able, communicate changes in personnel ahead of time – but not too far ahead. • Don't make promises you can't keep • Break down procedures into concise, orderly steps. • To resolve "stubborn issues" try using "compromise". Both the student and the

	educator have to come up with a totally new solution. Not only is this a successful problem-solving strategy – it can also be a form of diversion. • Provide praise when being flexible
Perseverative or Obsessive Thinking This is the tendency to get "caught" on one issue or thought to the point where it overshadows the main theme of the learning or social event. This behaviour can contribute to difficulty in transitioning from one topic/activity to another. Students often have an great need to complete tasks. It can lead to loss of emotional control.	• Use reflection – have student restate what you said • Put in writing; use visuals. Carry a small notebook if needed. • Less is best – give less amount of work at one time rather than more. Add to the work as time allows. • Avoid power struggles and ultimatums • Ignore (if possible) • Don't give more information than is necessary especially too far in advance. • Use "strategic timing" – time the activity that the student has difficulty ending right before snack or lunch. • Set limits. "I'll tell you 2 more time, then we move on to next topic. This is #1."
Tenuous Emotional Control Any combination of life stressors can lead to emotional "discontrol". The result may be exhibited as challenging behaviours such as tantrums – yelling, swearing, aggression, destruction, self-injury. During these episodes, reasoning is lost. Recovery of control takes time and is often followed by sadness, remorse, and guilt. Because of a problem in sequence processing, students are not always able to turn what not to do into what to do.	• Be aware of "hallway over stimulation" – especially before the school day begins. Have student enter the building at a less popular entrance. If possible, have arrival time be 5-10 minutes after school starts. Dismiss early. • Start the day off on the right foot by allowing time to go over the schedule for the day and work through any changes there may be. Putting the new schedule in writing often helps to decrease anxiety. • At the start of the day – set daily goals with the student. Limit to no more than 3. • Communicate behaviours you wish to see. Make it a cooperative task that provides concrete behaviour expectations. Put goals in writing. Avoid the word "DON'T"… focus on the word "WILL". (Ex. "Please talk in a quiet voice …instead of "Don't yell". When I feel frustrated, I **will** tell Mr. Smith or another adult.") • Provide positive attention and praise when student is maintaining control, especially in difficult situations. Celebrate success! • Encourage communication and acknowledging feelings. Words are important – LISTEN carefully! • Include the student in behaviour plans. Having

	their input elicits cooperation and a sense of support. • Be a role model. "I always say "darn" when I am angry. Let's try that for you …darn, darn, darn". Practice when the student is **not** agitated or angry. • Depending on the student and the situation – use humour. It is often effective. • Anticipate build up of frustrations and help him/her to remove self to "safe area" • Create a key word or phrase that will alert the student that it is time to go. • Practice using these words/phrases when the student is calm. • Develop a plan and **teach the student what to do** if he/she feels angry or frustrated. Many students substitute a means of releasing this pent up anger – long walks/exercise, ripping paper, tearing rags, popping packaging bubbles… • **Don't try reasoning during times when out of control. Limit discussion.** • Have a plan in place if student becomes more violent. Safety for all is a priority. • Consistency in approach is imperative • Provide positive closure. Don't hold a grudge. • If using consequences – they should be immediate and help the student learn from the outburst – saying "I'm sorry", sending a note to say they are sorry ...
Food Craving and Diet Restrictions For people with PWS, the message of fullness never reaches the brain – they are always hungry. In addition to this craving for food, food is metabolized at a rate that causes extraordinary weight gain. Food must be monitored and the individual supervised in all environments.	• Make sure lunch is placed with a bus driver and /or an assistant on the ride to school. • Educate and inform all people working with this student – including bus drivers, custodians, secretaries and volunteers. • If the student states he/she has not had breakfast – call parents or caregiver before giving more food. (Often times they say this to get more food.) • Supervise in lunchroom and in all food related areas – including vending machine areas. In some cases, student may need to eat in classroom (with peer/friend) • Many require supervision in hallways or near unlocked lockers at all times. • Avoid allowing the student to have money. Lock up all sources of money – including purses.

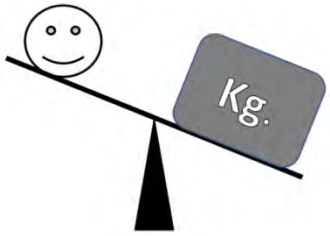
	Money buys food! • Address any stealing or trading of food in private. • Follow guidelines for treats or eating of extra food. Communication with home is very important. • Follow calorie-controlled diet. If a special calorie diet is needed and served by the school, a prescription must be obtained from a health care provider and should be a part of the student's educational plan. • Don't delay snack or lunch; if this is necessary discuss ahead • Limit availability and visibility of food. Be aware of candy dishes. • Avoid using food as a reward or incentive. • Be aware of smells – there is nothing like the smell of popcorn to make a student with PWS agitated. • When going on a field trip or other outing, discuss all food-related issues ahead of time. Will you bring snack along or will it be purchased? If purchased – what will it be? Will the outing interfere with the time of a meal or snack? • Obtain weekly weight by school nurse if indicated. • Daily exercise should be a part of student's schedule. **If a student with PWS is caught with food in his/her possession that is stolen – DO NOT ATTEMPT TO PHYSICALLY TAKE IT AWAY. Try to compromise, trade or other forms of negotiation. Do not threaten. Evaluate what happened. Institute measures to prevent reoccurrence.** • If it is discovered that student has had a binge episode and eaten a large quantity of food – contact parent immediately. This could result in a health emergency. • Encourage to eat slowly – student may choke from eating too fast.
Poor Stamina People with PWS tire more easily and may fall asleep during the day. Morning is typically their optimal learning time, when energy level is highest	• Get person up and moving. Send on errand. Take a walk. • Schedule high energy, mobilizing activity after lunch • Offer items /activities which stimulate large muscles and deep breathing - balloon blowing, party blowers

	• Provide scheduled rest time or a quieter activity if needed
Scratching and Skin Picking These two behaviours are often seen in individuals with PWS and may be worse during times of stress. Combined with a higher pain threshold, these behaviours can result in tissue damage if not controlled.	• Use diversion - provide activities to keep hands busy (colouring, computer time, play dough, hand-held games, magazines, book…) • Keep nails short. Apply lotion liberally – it keeps skin slippery and soft making it more difficult to pick. Applying lotion can also be an effective diversion. • In extreme cases, provide constant supervision – even in the bathroom. Limit time in the bathroom. • Cover area with band aide or similar covering • Don't just tell him/her to stop picking – it won't work. • Apply mosquito repellent before any walks or outside activity.
Difficulty with Peer Interactions While children want and need other children and value friends, they often lack age-appropriate social skills. They often face challenges in issues of fairness and comparing themselves to others, often resulting in frustration and anger.	• Many do better in small groups. Benefit from verbal cues and guidance. • Pre-plan outings. Keep time short • "Supported recess or social outings" – planned activities with a friend • Include child in planning activities that are of interest to him/her (board games, puzzles, computer games…) • Provide social skill classes that emphasize sharing, taking turns… • Role play and practice appropriate social situations. • Clearly state and write do's and don'ts for social interactions w/ friends of opposite sex

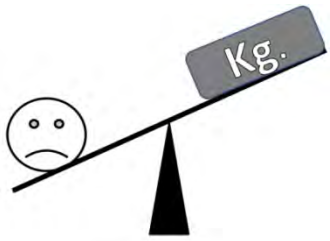
Students with Prader-Willi syndrome are very caring, sensitive and conscientious. They want very much to be successful, have friends and be a part of their school community. Although they face some unique challenges, with proper support and understanding … they are playing, learning, working and living successfully in our communities.

PRADER-WILLI SYNDROME - FOOD SECURITY - BASIC CONCEPTS

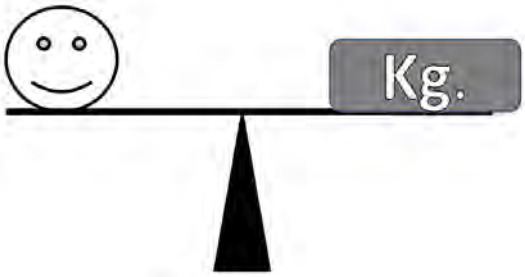
By: Linda M. Gourash, MD & Janice L. Forster, MD of the Pittsburgh Partnership Specialists in Prader-Willi Syndrome From: www.pittsburghpartnership.com



Families and professionals often mistakenly believe that the patient cannot be happy unless he has as much food as he demands. Because efforts to limit food, if attempted without establishing food security cause increased stress and behaviour problems.

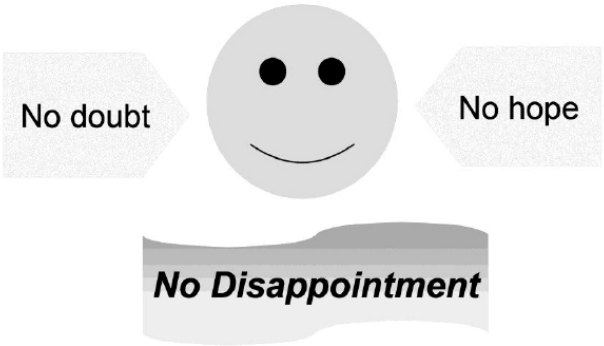


When *Food Security* is fully implemented, weight and behaviour are both managed successfully and simultaneously.



REMEMBER: "FOOD IS STRESS"

Food Security includes:



NO DOUBT

The person with PWS is able to relax and think less about food when he knows the plan for his food each day. This is achieved by a predictable routine for the day in which meals are scheduled reliably among his other activities. Focus on the sequence of events and not the time of each meal. Advanced planning assures the individual of what activities will precede the meal and which will follow. Advanced menu planning provides him with expectations which will be reliably fulfilled.

NO HOPE (NO CHANCE)

As children get older, opportunities for food acquisition increase and they require more measures to assure that they are not hopefully scouting for food all day. Chances to obtain food are stressful and therefore, as much as possible, should be eliminated. The measures taken will depend on the individual's history and capability of food acquisition.

Successful Behaviour Management of PWS means that uncertainty about food must be eliminated as much as possible. Advance planning of meals, a schedule of all the day's events with the place of meals clearly identified, reminders of these plans and a behaviour program which requires completion of one task before the next activity (including meals) is begun, all contribute to successful behaviour management.

FOOD SECURITY CHECKLIST FOR THE FAMILY

NO DOUBT

- ☐ My child has a menu posted. He/she always knows what he/she is eating for the next meal.
- ☐ My child takes his/her lunch to school/work.
- ☐ My child is rarely disappointed about food. He always gets exactly what he is expecting. ☺
- ☐ My child sometimes corrects others about his/her diet. ☺
- ☐ My child knows when he/she is going to get a treat well in advance. There are no surprises.
- ☐ My child never receives unplanned treats
- ☐ My child rarely asks about what he/she will be eating. He/she already knows. ☺
- ☐ My child knows when his/her meals are scheduled during the day. ☺
- ☐ I never threaten my child that a meal will be delayed or changed in any way.
- ☐ My child has scheduled zero calorie treats built into his /her daily schedule.
- ☐ My child knows that if his usual menu is disrupted for any reason he can always count on the same "alternate".

NO HOPE

- ☐ My child does not have free access to calorie free foods or beverages other than water.
- ☐ During meal preparation another member of the family is assigned responsibility for watching my child with PWS.
- ☐ My child rarely argues/tantrums about food. ☺
- ☐ When we go to a buffet at a restaurant or party my child knows that I will be preparing his/her plate.
- ☐ My child has someone assigned to be with him/her during lunch at
- ☐ [As far as I know] My child has not successfully stolen extra food in the last 2 weeks. ☺
- ☐ We have a plan for every special occasion and my child knows what the plan will be well in advance.
- ☐ Even though my child knows and expects his/her diet, I know that he cannot be trusted to maintain it him/herself. ☺
- ☐ My child never prepares his/her own plate.
- ☐ When we "dine out" or "order out", we get the menu in advance so that my child

school/work.

- ☐ My child does not keep his/her own money.
- ☐ At this moment there is no unlocked food anywhere in my home.
- ☐ knows exactly what he/she may order.
- ☐ My relatives/neighbours never offer my child food. I have successfully explained

☺ = signs of success. If you have all of these signs of success your food security is complete!

FOOD SECURITY CHECKLIST FOR THE SCHOOL/WORKPLACE

NO DOUBT

- ☐ This student/worker has a menu posted. He/she always knows what he/she is eating for the next meal.

OR

- ☐ This student/worker brings his/her lunch to school/work.
- ☐ During any food preparation another member of the team is assigned responsibility for watching this student/worker with PWS.
- ☐ This student/worker is rarely disappointed about food; he/she always gets exactly what he/she is expecting. ☺
- ☐ This student/worker sometimes corrects others about his/her diet. ☺
- ☐ This student/worker knows when he/she is going to get a treat well in advance. There are no surprises.
- ☐ This student/worker knows that if his usual menu is disrupted for any reason he/she can always count on the same “alternate”.
- ☐ This student/worker rarely asks about what he/she will be eating. He/she already knows. ☺
- ☐ This student/worker knows his/her schedule every day.
- ☐ This student/worker knows when his/her meals are scheduled during the day.

NO HOPE

- ☐ This student/worker is never offered food that is not planned in advance and cleared with his/her family or residence.
- ☐ I never threaten this student/worker that a meal will be delayed or changed in any way.
- ☐ This student/worker has scheduled zero calorie treats built into his /her daily schedule.
- ☐ This student/worker has no access to calorie free foods or beverages other than water.
- ☐ This student/worker rarely argues about food. ☺
- ☐ This student/worker has someone assigned to be with him during lunch at school/work.
- ☐ This student/worker has no opportunity to get food during transitions or transportation. He/she is continuously supervised or the food is stored out of reach.

- ☐ At this moment there is no unlocked food anywhere in the areas where this student/ worker is permitted.
- ☐ This student/worker does not have access to money or to vending machines.
- ☐ We have a plan for every special occasion such as birthdays or holiday celebrations and this student/worker knows what the plan will be well in advance.
- ☐ Even though this student/worker knows and expects his diet, the entire team understands that he/she cannot be trusted to maintain it him/herself. ☺
- ☐ This student/worker has not successfully stolen extra food in the last 2 weeks. ☺
- ☐ The other students/workers never offer this student/worker food. Our team has successfully explained to them why they must never do this. ☺
- ☐ Our team never uses treats as unplanned rewards

☺ = signs of success. If you have all of these signs of success your food security is complete!

HOW DOES A PERSON WITH PWS THINK?

Kate Beaver, Crisis Intervention Counsellor, PWSA (USA)

***The Gathered View* ~ Prader-Willi Syndrome Association (USA), January-February 2013**

Persons with PWS are generally concrete thinkers. Terms like, "Hop to it!" may not be understood to mean "Begin the task immediately" and may cause confusion, anxiety, and result in an unwanted behaviour.

Persons with PWS have a delay in processing the information you give them. Most children will take between 3-5 seconds to understand what you say. If too many instructions are given or the instructions are generalized, they can miss the middle part of what you said and misunderstandings occur. Instead of saying "Go get ready for bed", try breaking the process down into steps. For example, "It's time to brush your teeth", then wait 3-5 seconds before repeating the request. If after the second request they do not comply, do the task with them before asking that they do the next step.

Problem-solving skills are often impaired. This is in part due to the processing problem and not being able to put things in order of how they should be done, which also creates anxiety. Children become frustrated and anxious trying to do what you want them to without being able to know what to do first and what all it entails.

Short-term memory is often poor.

Long-term memory is usually excellent (thanks to ghrelin) so that once something is learned, it's remembered forever.

PWS is about Anxiety

Persons with PWS typically feel high levels of anxiety – all the time.

Maladaptive, unwanted behaviours are often attempts to reduce the level of anxiety the individual with PWS is feeling: skin picking (also done when feeling bored); repeated questions; excessive talking; controlling, oppositional or argumentative behaviour; sleeping. If you can reduce or eliminate the cause of the anxiety, you'll reduce or eliminate the behaviour problem! The best start is to remember they have a processing delay and they want to please you.

FAMCARE - AN INITIATIVE FROM IPWSO FOR THOSE CARING FOR AN ADULT WITH PWS AT HOME

Famcare is an international committee that has been formed for the purpose of supporting families who have their adult son/daughter/relative with PWS living in the home with them. Georgina Loughnan, an IPWSO Board member, is heading the project. She is supported by a committee made up of parents of adults with PWS and experienced professionals. The committee is also most fortunate to have a highly skilled team of advisors.

SKIN PICKING IN PEOPLE WITH PRADER-WILLI SYNDROME

By the Famcare committee, a project of IPWSO: famcare@ipwso.org

Skin picking or scratching is a common behaviour seen in people with Prader-Willi syndrome (PWS). The intensity and duration of this behaviour varies from person to person and episode to episode. It may occur in the form of scratching an insect bite until it bleeds and becomes an infected sore or it may manifest in the form of nose picking or anal picking. When this behaviour continues for a prolonged period it can cause unsightly and distressing open wounds. 75% of the people surveyed by Famcare reported skin picking in their relative with PWS. A USA study reported anal picking in 9.5 % of people who skin picked.

WHY DO PEOPLE WITH PWS SKIN PICK?

The behaviour is thought to start in response to boredom, stress and anxiety or as a form of self-stimulation. It is also known that people with PWS have a high pain threshold and do not feel pain so intensely. Once commenced it is difficult for the person with PWS to cease the behaviour without some form of external intervention, as they become “stuck” in the behaviour.

If the underlying cause of the skin picking can be identified, especially where stress or anxiety is thought to be a contributing factor, the ideal would be to investigate and discuss what is causing the stress for the person with PWS. If the stress can be removed or decreased, the skin picking should also reduce. Remember, however, that the picking may continue as it may have built up to a repetitive behaviour which will take longer to stop.

WHAT WORKS TO AVOID SKIN PICKING

There are simple and practical strategies that help to minimise the severity and frequency of skin picking, despite the fact that it may still occur spasmodically.

These include:

- routine cutting of finger nails e.g. weekly
- daily use of moisturiser for fingers, hands and arms
  - it is best if the moisturiser is applied by the person with PWS
- positive reinforcement for “healthy, good-looking skin”
- keeping hands occupied – using a “word-find” or puzzle book, computerised game use, object kneading – e.g. soft ball, worry beads, hand craft – such as knitting, crocheting, beading, unravelling woven blankets

- daily sensory stimulation of the hands and arms – e.g. massage
- maintaining a PWS appropriate and calm environment
- brushing hair daily for a number of strokes, counted – e.g. 50, encourages the growth of "beautiful, shiny hair" when the habit of pulling hair out, is a problem.

WHAT WORKS TO LIMIT SKIN PICKING WHILE IT IS OCCURRING?

Skin picking often occurs in waves. These can be hard to work through but it is possible to lengthen the time between episodes and shorten the actual periods of skin picking.

Below is a list of strategies that parents and carers have found successful to reduce this behaviour

- tell the person how wonderful their "unpicked" skin is
- tell the person you want to help their *sore* to heal
- give verbal praise for periods of time spent not picking
- offer the person a reward (not food) for time spent not picking – this can be for a few hours or lengthened to days or at the end of a week for 7 days of no picking. Drawing up a *contract* involving "periods of non picking" often works well.
- keep hands occupied at vulnerable times (see above)
  If anal picking is a problem rolling balls of toilet paper while sitting on the toilet may keep hands occupied
- try *not* to focus on or talk about the skin picking
- using distracting calming strategies and verbal re-directive strategies e.g. asking the person to tell you about something that interests them
- dress any picked sores with antiseptic cream if necessary
- cover the picked area with a gauze dressing and bandage to prevent access to the area
- maintain supervision of the person if they are only picking in secret – but nighttimes are difficult
- try using a strong solution of salt and warm water to bathe picked areas
- an Aloe Vera based cream works very well on the sores
- using Calamine lotion to cover the area and make less attention attracting
- in severe cases medication (*only* prescribed by a doctor) may be beneficial – e.g. Topiramate, topical Bactroban, ReVia (naltrexone hydrochloride)

All medications must be monitored by a doctor to prevent any reactions within the person with PWS

Skin picking does not occur all the time. Prevention is always the better option, however, techniques used to avoid skin picking need to practised regularly – as with all PWS management

COPING WITH CHANGE IN PEOPLE WITH PRADER-WILLI SYNDROME

Famcare is a project of IPWSO: famcare@ipwso.org

Most people with Prader-Willi syndrome (PWS) have difficulty coping with change. This varies from person to person and can be a real problem for those families affected. Change can be anything from a substitute teacher at school, to a different route being taken in a car trip or a new cup being used. Change can also occur suddenly and unexpectedly, as in an electrical blackout. A bad reaction to change may result in the person with PWS refusing to comply with requests, routines or plans and can quickly escalate into perseveration (repeated questions or comments), arguments and aggression. "Shutdown" is another

typical response to the anxiety associated with change. These responses can be stressful for families as they often occur at the most inconvenient times.

Why do people with PWS have difficulty coping with change?
The problem is thought to be linked with the inability to "switch" attention from one thing to another. People with PWS generally find it more difficult to switch attention. Researchers from the UK have shown that the greater difficulty a person with PWS has in switching their attention from one thing to another, the more resistance they show to change. It is also known that people with PWS tend to prefer repetitive routines and often exhibit ritualistic or inflexible behaviours. Although varied in their reactions to change, they all feel "safe" with set schedules and expectations. If a change can be predicted in advance, it is possible to use strategies to avoid much of the anxiety that may otherwise result from the change occurring.

What works to avoid anxiety related to change
These are simple and practical strategies that help to minimise the reaction to *predicted* change:

- Discuss a back-up plan in advance. For example, you could agree to buy pears if there are no apples at the fruit shop. The fact that this is discussed in advance can prepare the person for a different situation to the one they are expecting.
- Thoroughly plan all details and double-check things with the person with PWS and with others. Occasionally the person with PWS has an expectation that you are not aware of. For example, they may expect that you will be stopping for a cup of coffee on the way home, however if you are not aware of that and drive directly home, that would be perceived by them as a change. So it is always helpful to go over the details of any plans, beforehand.
- Agree on some rules for an outing. For example, a rule that "If there is any yelling for any reason we will return home." Before the outing reminds the person of the rule, using positive terms eg "your best behaviour will mean we can finish the trip and won't need to come home early". A set consequence of bad behaviour can be hard to enforce at the time, however, going home once can stop a lot of problems in the future.
- Praise the person whenever change is accepted graciously.
- Use picture-boards or written routines. Any change to routine is added to the board in advance. For example, a schedule could be written for the week. Doctor's appointments or variations in the normal week can be written on it. If the person with PWS has any problems with the plans they can tell you in advance so, hopefully, any problems can be discussed and a solution can be negotiated.

What works to limit reaction to an unpredicted, last minute change of plans?
At times, unpredicted and unplanned change does occur. If you are fortunate enough to be able to respond quickly you may be able to reduce anxiety.

Below are some strategies that parents and carers have found successful in similar situations

- Give a solution before explaining the change. For example if a favourite coffee shop is closed for renovations, start by saying, "We are going to have coffee today at another coffee shop. That'll be different, won't it! We can ask them to make it just how you like. The coffee shop we like will be open again next week but it is closed today so we can't

go there today." The last piece of information given in this example is the fact that the shop is closed and the fact that they will still get a nice coffee, comes first.

- Give a visual example of the problem or the change. For example if you were planning to move a cupboard into a room and it did not fit, showing a person with PWS the cupboard outside the door is better than trying to explain the problem in words. Simple sketches can at times be more easily understood than words.
- Be patient and calm as it may take some time for the change to be processed and then accepted.
- Try to point out the positive aspects that are involved in the change.
- Expressing shock at the change, yourself, and asking the person with PWS to help in the situation can diffuse their reaction to the change if they are given some responsibility within the event.

Managing change is an important part of managing the overall well-being of a person with PWS. Effective management of change can reduce anxiety and improve outcomes in other areas such as health and behaviour management.

Even 'good changes', such as a holiday, can be a sources of stress for the person with PWS and need to be managed as a change to routine. Perseveration, increased anxiety and worry about what will happen while they are away and so on, are often seen. Not telling the person with PWS about something 'good' coming up for them is an option, but not always a good solution.

Working out changes with the person with PWS, so that a better result might be achieved next time, can be useful. For example: " Next time, if you think your cat has been in a fight and might have an abscess, do you think it would be better to go straight to the vet instead of trying to fix it yourself?" If the answer is 'yes', then you can incorporate it into their guidelines – write it up on their pin-board, and make sure they know the new plan!

After an argument, or a 'blow-out', and when the person with PWS has become quiet and even remorseful, ask them what *you* can do to help them next time something like this upsets them. You may be surprised at their answers and often they can lead to positive results. For example: "Please leave me alone for a while," "Please listen to me," "Please don't treat me like a child," These sorts of agreements can often prevent a future argument or blow-out.

It's all about compromise

As the person with PWS grows older, their behaviours may mellow. There is an expectation from the person themselves, as well as parents, siblings and family friends that they will be treated more and more like an adult. As parents and caregivers, we know this is not always possible, but we can sometimes be a little more flexible and trusting in certain areas. It is difficult to let go, and there will be times that the trust between you will be broken, but if you compromise, safely, often you can reach an agreement. As PWS lack of judgement and poor emotional control are not visible when they are calm and happy always prediscuss consequences.

Practising increased, safe responsibility can facilitate positive behavioural change.
For example: Carrying a set amount of money for a specific, preplanned event can encourage self confidence and trust. However, success with this will take much preparation, explanation and negotiation. Having a "back-up" agreement to cover times when such a

compromise does not work successfully is essential, such as: "If you spend the money instead of using it for what we've discussed it will be best for you not to take the money next time. I will then continue to organise the payment so you are not tempted to spend it on food."
People with PWS vary in personality and cognitive ability. Some people can cope with more responsibility than others. If monetary responsibility is not necessary it is best avoided so as not to set the person up for failure.

A parent says: The main thing about compromise is that if you are going to *take something away* from a person, you must always give something back. *For example:* "Unfortunately I cannot let you keep a dog in this house because we really don't have room. But, you can keep a bird in a cage or a goldfish in a bowl, and we could always arrange for you to help walk some of the poor dogs at the SPCA who never get a chance to go out".

Appealing to their own sense of judgment ("we don't really have room") and getting them to see that fact; giving something back (goldfish, or bird, for example) and appealing to their sense of importance, of doing something for others (walking someone else's dog), seems to work.

Preparing people with PWS for change gives them the opportunity to cognitively and emotionally process what is to happen and how the change will affect them. It may take time and effort to prepare people for change but the results are usually worthwhile. However, if the person with PWS is stressed for a reason unknown to you or they are aware of anxiety you may be experiencing, difficult behaviour can occur despite your use of all of the above suggested strategies. Don't feel like they or you have failed. Keep applying positive proactive strategies. In time, improvements will be noticed.

THE BASICS OF A HEALTHY ADULT LIFESTYLE

Famcare is a project of IPWSO: famcare@ipwso.org

An early childhood diagnosis of Prader-Willi syndrome (PWS) usually provides parents with the opportunity to learn strategies to manage potentially difficult behaviour and obesity. Despite variable intellectual and physical ability, it must be remembered that a cognitive disability, hunger and reduced satiety remain with the person with PWS throughout their lives. The ability to make appropriate independent decisions regarding energy intake and choices of life rarely eventuates, even as the child with PWS becomes an adult. It will always remain the responsibility of their parent or carer to make healthy choices and maintain firm boundaries around food, money, exercise and lifestyle.

ENERGY NEEDS

Throughout this article the term "energy" refers to the calories (or kilojoules) obtained from the food and beverages consumed.
Adults with PWS still need a limited energy intake - of about 60% of the daily recommended energy intake for non-PWS adults. Remember the caloric intake guide: 7kcal per centimetre of height (18 kcal/inch) if they need to lose weight or 8-11 kcal per centimetre of height (20-28 kcal/inch) for weight maintenance. This does not change and applies to people with PWS of all ages.

HOW IS THIS ENERGY INTAKE ACHIEVED?

Restricting energy consumed requires a consistent balance of meals and snacks. This means all extra food sources need to remain inaccessible to the person with PWS. Kitchens, pantries, refrigerators, food cupboards, fruit bowls, garbage bins, shops, relatives' homes, friends' homes, neighbours' homes, work places, day programmes, food courts, shops, church functions, sporting events and independent travel all provide the opportunity for "extra" energy intake. Most people with PWS will take food or money to buy food, or eat leftover food or scraps from garbage bins, if given the opportunity. This is because they are constantly thinking about food and not satisfied by the amount they consume. This is a trait of PWS, not a personality trait of the person with PWS. A consistently secure environment coupled with psychological 'food security' is essential to achieve emotional and behavioural stability.

People with PWS certainly need choice and variety in the food they eat and what they drink but the calorie content must be determined according to their needs. The more constant the restrictions are the better the person with PWS will adapt to the restrictions. Also, it is extremely important to maintain restrictions once they are in place. This is not only to maintain a healthy weight but also to avoid over-consumption that could lead to severe gastric health problems.

CALORIE BARGAINING

This practice involves reducing the usual energy intake for a few days prior to a party or similar irregular or special event, where higher energy food will be consumed. It is a useful strategy that enables greater "socialising" while managing the extra calories to avoid weight gain. For example: reduce the quantity of meat or remove the carbohydrate from the evening meal for 2-3 days prior to an event to allow for the extra calories consumed at the party. Even if the reduction in calories does not equate exactly to what is consumed at the event – the strategy implies an energy control. Offer the person the choice to attend the event and "calorie bargain" or avoid the event. He/she may choose not to go to the event so they don't have to reduce their usual intake. Calorie bargaining gives them a choice, while retaining a constant energy input. It also helps to keep in place "mental security". It requires prior explanation and discussion with your person with PWS and does not suit everyone.

Whether you practise calorie bargaining or not, remember, it is always best if energy consumed out-of-home is discussed, planned for and chosen, before the event, wherever possible.

A parent writes: *in our home we follow the practice of "food or portion bargaining". Our daughter with PWS eats what we eat and we concentrate on low fat, low carbohydrate meals with lots of vegetables and adequate fruit. When our daughter asks for another serving of food – she is happy with my response of "no, I will keep it for you to have tomorrow." When we eat out, if possible, we discuss what food will be served and she is reminded that "I'll dish it up for you." She has a small portion of dessert, if it is served, and if she asks for more our strategy is to ask the host for a portion to take home with us for her to have the next day or later in the week, as a treat.*
She is also prepared to reduce her next meal that follows the outing meal.

The above strategies work well as a result of consistently practising them in the same way each time, so trust is established between the person with PWS and his/her family member. In many cases something that is stored for later, is not even asked for again.

Another parent writes: *when we eat out our "rule" is that she will ask for a take home box when she orders her food, and then I put half of the food into the box before she starts to eat. She knows that the other half will go home with us and she can have it the next night for dinner. For her, this is like eating out two nights in a row. We have not had any problems with this and it works beautifully for her.*

People outside the family unit also need to know about PWS so they can support and assist you in the healthy strategies you practise for your person with PWS. When people you have contact with are well-informed they can often feel less apprehensive about being with the person with PWS. The knowledge can empower them to understand the nature of PWS a little better.

EXERCISE – ENERGY OUTPUT

All people with PWS require regular effective exercise, as much as energy restriction, throughout their lives. People with PWS have more fat and less muscle. Their muscles have reduced tone and are weaker, which reduces joint stability, agility and bone strength. Exercise is the key to weight management, cardio-respiratory fitness, muscle strength and joint stability, bone health, mental health and the prevention of diabetes for all people but so important for people with PWS. Exercise also provides a distraction from focusing on food, develops emotional well-being, boosts self esteem and improves behaviour.

If your person with PWS is not exercising - now is the time for them to start. It is never too late! If they are only able to walk short distances then this is their starting point. Walking 10 minutes twice a day is a great start! The secret is to manage their routine to include some physical activity every day. The aim is to eventually have them doing 30-40 minutes of aerobic type exercise 5-6 days per week. Examples of exercise include: walking, swimming, cycling, dancing, rowing and similar continual exercise. When first introducing a new activity or exercise, a small reward for the completion of a planned exercise can be a good incentive. A planned meal or snack can be used as a routine reward for completed exercise. Exercising before eating or waiting for one hour after eating is recommended except when the person is known to have delayed gastric emptying. Then it's best to exercise immediately after eating to assist digestion. When introducing exercise explain that as the exercise becomes easy the intensity will gradually be increased to make them stronger. To improve their fitness, the intensity or duration of the exercise should be increased very gradually. Also remember to warm-up and cool-down. Exercising in the morning has additional benefits: increased alertness, it's done and out of the way, the person is not as tired as at the end of the day.

Incidental or purposeful activity is also important. Household chores like making their bed, hanging out the washing, collecting the mail and helping with domestic responsibilities not only keeps them active but may also improve their self esteem as you entrust them with family jobs. Adults like to be needed and given responsibilities to prove their ability. Mastery and praise can work wonders!

Where possible, it's even better if family members can join in the exercise so it is seen to be good for everyone, not just the person with PWS. Once exercise is a regular part of their day, less encouragement is required as it becomes a part of their consistent routine.

Outdoor activities are a wonderful way to combine energy expenditure and the many benefits gained from being outside. Sunshine is the essential source of Vitamin D! Walking on different terrains or surfaces also increases muscle strength. Taking in the sights and sounds of nature when in parks, forests or country areas can be educational and stimulating to the senses and mind. Walking is one of the best activities for sensory motor stimulation in adults with PWS!

A parent writes: *for my son walking outdoors is a noticeable anti-depressant and mental health aid. Getting sunlight into the eyes, moving around other people, deciding which path to take, hearing birds and seeing insects, having something different to talk about makes being active outdoors all worthwhile. Where possible, involve others in the life of your person with PWS. Positive friends, neighbours and other family members enrich their lives. A simple "hello" or smile or "how are you going?" are positive interactions for people with PWS, who generally have difficulties with friendships and community socialisation.*

A parent writes: *the only thing that really has worked for our daughter regarding exercise and sticking to it, has been a junk-mail run that is she currently doing. It takes her two hours, and she gets paid a very minimal amount (but anything's better than nothing). Being paid for a "job" has given her not only the encouragement to stick at it, but the ability to say that, yes, she has a job! Amazingly, she goes out in all weathers. Amazingly, she has stuck with it, and amazingly, she has lost 10 kg in 5 months!*

Encourage as wide a range of interests as possible!

Each person will naturally be inclined towards certain interests but "expanding their horizons," when possible and appropriate, has benefits. Encourage reading, writing, craft activities, interest in world events and different sports. Just be mindful of the availability of extra calories at any event attended and discuss the management of this beforehand. Being employed in an active way takes up another opportunity to expend energy in a way that is "part of the working day" rather than an uncomfortable chore. If they are being paid to be active this is even better!

DRESSING APPROPRIATELY

We talk about "dressing for the weather" but must remember that people with PWS do not have this ability. Parents and caregivers are their weather guides and need to encourage their people with PWS to dress appropriately for the weather – regarding clothes and shoes. People with PWS can suffer from hypothermia and hyperthermia when not dressed accordingly for the temperature.

When your person with PWS is exercising or being regularly active, be aware of any injury or pain that may cause greater problems in the future and reduce their ability to exercise. Skin picked sores on the legs or stomach may be exacerbated during exercise if they are rubbed by clothing. They may need to be covered and protected to prevent infections developing. Wearing appropriate shoes for exercise may need to be pre-negotiated. When someone with PWS has favourite shoes and is not keen to wear "joggers" these may prevent the person from exercising safely or adequately. Appropriate exercise shoes don't need to be

expensive but do need to be supportive and protective, especially if the person had diabetes. A good shoe for exercising in: 1) has a flexible sole and a good arch support 2) allows for expansion of the feet as they warm up (laces are better than velcro straps) and 3) has a good heel support

Be aware as your person with PWS settles into a regular exercise routine
- things may change:
They may have more energy! They may be happier! They may become healthier!

It's never too late to introduce exercise to your person with PWS. It may take some persistence on your part to encourage regular exercise, but if you can add it to their daily routine and make it an enjoyable experience or one that is rewarded in some way the benefits will certainly out way the initial difficulties. Exercise is not an optional extra for people with PWS. It is as important as a restricted energy intake.

A positive experience for all involved can lead to positive results for all involved.

It is exercise alone that supports the spirits and keeps the mind in vigour - *Marcus Tullius Cicero*

GOOD HEALTH CHECK LIST

Famcare is a project of IPWSO: famcare@ipwso.org

The basics of a healthy adult life for someone with Prader-Willi Syndrome (PWS) include a healthy, appropriate eating plan and regular, effective physical activity to avoid obesity, to keep the body fit and to maintain good mental health. Before any of these can be achieved the people working with the person who has PWS must have a good understanding of the complexities of the syndrome.
This "checklist" is to assist you in maintaining good health for your person with PWS. It is to be shared with other family members, professionals and caregivers who are involved with your person with PWS.

PWS is a genetic disorder which, due to a lack of expression of particular genes on the 15th chromosome. It effects several systems in the body. Below are listed some common problems seen in people with PWS, what the effect of the problem is on health and how it needs to be monitored or treated. Changes in physical health are often only detected or suspected because of changes in general behaviour so it's important to know what to be aware of and what to check!

System/organ	Effect	Monitor / Watch / Treat
Appetite regulation	Always wanting to eat / drink Abnormal interest in food Overeating	Food & beverages must be limited Overweight / obesity
Behaviour	Often perceived and misunderstood by others to be mood or mental illness. Often occurs because of stress and lack of appropriate support	Requires appropriate management strategies and rarely, medication, unless due to a diagnosed mental illness, which can occur in people with PWS.
Bladder	Poor emptying often seen Obesity may increase urinary tract infections(UTI) &/or incontinence	Timed toileting Increase awareness of full bladder feeling / empty bladder feeling Confusion may indicate UTI

Body fat - increased (with less muscle) even with healthy weight	Overweight and obesity can lead to serious complications	Energy intake must be limited Exercise must be a part of life Walking daily for 1 hour is simple & effective
Bone density (BD) (and bone strength)	Often reduced due to reduced hormone levels, decreased muscle mass and too little physical activity. Low BD increases risk of fractures.	Maintain adequate calcium & Vitamin D intake and check blood levels. Check bone mineral density every 2 years. Helped by regular weight bearing exercise.
Bowel	Constipation is common and may lead to bowel or urinary incontinence, rectal picking and slow stomach emptying	Maintain regular fluid intake. May require daily low dose laxative. Helped by exercising after eating. Prevent over-consumption of fibre.
Diabetes Mellitus (DM)	Often develops as a result of obesity but also seen in normal weight Poorly controlled DM can cause undesirable weight (muscle) loss, renal failure and loss of vision.	Maintain a healthy weight & regular exercise to prevent DM. Check fasting blood glucose levels (BGL) every 6-12 months. If known DM exists check HbA1C (average BGL of past 6-8 weeks) every 3-6 months.
Ears and hearing	Lack of concentration or response may indicate the development of hearing loss, or psychoses	Check with ageing as person with PWS may not be able to express loss of hearing. If acutely impaired check for infection first.
Eyes	Strabismus (abnormal alignment of eyes, squint); short/long sightedness	From birth or a change in vision with age Check eyes every 2 years
Feet / legs	Can develop oedema and cellulites then severe infections, when obese or inactive. Feet , hip, knee abnormalities (from birth) can worsen with weight gain and ageing	Maintain a healthy weight Maintain daily activity / exercise Check regularly for sores and infections especially if swollen or poor circulation
Lungs	Obesity, reduced breathing mechanics, scoliosis, and kyphosis can cause reduced oxygen consumption. Obesity & inactivity can lead to pneumonia, lung infections and respiratory failure. Asthma can also occur	Maintain healthy weight Maintain daily activity /exercise Medical check is required if exercise tolerance is poor or deteriorates. Blood oxygen levels may need to be checked.
Mental illness	Depression, psychosis, mood disorders	Require psychiatric assessment and may need treatment with medication. Due to brain dysfunctions in PWS dose-response and side-effect susceptibility are less predictable so lower initial doses are recommended.
Mouth	Reduced saliva causes dry, sticky mouth Reflux from stomach can harm teeth (may need antacid medication)	Poor dental hygiene – dental erosion Requires regular, effective cleaning and regular visits to a dentist.
Muscle - Reduced strength &	Weaker muscles, unstable joints Scoliosis, kyphoisis	Poor posture, slower mobilisation, reduced breathing mechanics (worse

tone **Reduced muscle mass**	Increased sensitivity to some medications	with obesity) Requires strengthening exercises. Dose of some medications, when newly prescribed, should be lower than usual
Pain tolerance - high	Complaints of real pain are rare Real pain is often indicated by change in behaviour or activity level Check all swollen red areas of body if a fall or trauma has occurred	Undetected illness or injury from accidents All pain complaints must be investigated to exclude a cause & prompt diagnosis Insist on x-rays /other investigations after trauma or appearance of ill health or pain.
Sequential processing	Poor personal hygiene → infections	Encourage thorough washing of body May require assistance or visual cues
Sex hormones Are reduced	Lack of, or reduced sexual maturity Replacement required for bone health	Yearly sex hormone (androgen) blood test from 15 years of age Testosterone replacement (males - start with low dose)/oestrogen replacement - females), as required
Skin picking	Sores & infections	Cut fingernails every week, keep hands busy to distract from picking.
Sleep apnoea (pauses in breathing while sleeping)	Daytime sleepiness (occurs in PWS even without sleep apnoea), lack of concentration, excess irritability Can worsen with weight gain	To be assessed by sleep/respiratory specialist - may need positive airway machine or longer sleep time at night.
Stomach & Intestines Slow emptying of stomach and reduced intestines passage time often occurs in people with PWS	Distended stomach Loss of appetite – no desire to eat (often only symptom – must investigate!) Vomiting, rarely seen in PWS, often indicates serious illness	Complaints of "discomfort" from bloating and dilatation of stomach or loss of appetite *Risk of gastric necrosis* – death of stomach tissue due to reduced blood flow to stomach wall. Requires urgent medical treatment.
Temperature (body) – poor regulation and sensation	Inappropriate clothing for weather Can suffer from hypo or hyperthemia Infections, but no fever	Assist with choice of clothing for hot/cold weather If generally unwell, CHECK FOR INFECTION with or WITHOUT raised body temperature / fever
Water intoxication	Will lead to electrolyte imbalance which may cause seizures	Do not allow unlimited fluid intake
YEARLY MEDICAL CHECKS	**May need to be insisted on by family members *for* their person with PWS**	Check weight, waist measurements, blood pressure, lung function, teeth, posture. Annual blood tests are recommended from the age of 15 years. Ask for: biochemistry (including calcium, cholesterol, glucose), iron studies (including iron), endocrine (including sex hormones, thyroid, Vitamin D,)

Do not be afraid to seek a medical assessment for your person with PWS if you are concerned in any way about their health, due to changes in their manner or behaviour. Always provide information about PWS for the doctor or medical professional who is seeing your person with PWS, remembering that PWS is not well-known and includes specific traits that must be understood before treatment is prescribed. People with PWS need the ears, eyes and voice of the person with whom they live, to maintain good health and longevity!

PLEASE VISIT IPWSO's blog and become A FRIEND of IPWSO.

A variety of interesting material is available on IPWSO's blog. Papers of the International PWS Conference, held in July 2013 in Cambridge, UK, can be seen online. You are welcome to communicate directly with the Communications Director of IPWSO, Linda Thornton. She will welcome your comments. http://ipwso.blogspot.com

"MANAGING A MELTDOWN" IN PEOPLE WITH PRADER-WILLI SYNDROME

Famcare is a project of IPWSO: famcare@ipwso.org

Most people with Prader-Willi Syndrome (PWS) have meltdowns from time to time. This varies from person to person and is a serious problem for those families affected. The levels of anxiety seen in people with PWS cannot be understated. Most of the difficult, anti social behaviour demonstrated by people with PWS can be related to their inability to express and/or deal with anxiety or emotions they are feeling. They can be upset by something we would just take for granted, or it could be over something that occurred yesterday or last week, as well as something that may have just occurred but has not yet even acknowledged by others. It is often "either all or nothing". They may become highly anxious about a particular meal being served slightly differently and they may show minimal emotion when a family member dies. Preparing them for outings or events is a key strategy for addressing potential anxiety.

People with PWS think differently! They take longer to process information they are given and may become confused when given too much information at the same time. They can present as stubborn or oppositional when really they are just taking time to 1) understand the information they have received and 2) respond with an appropriate response. If they do not like what they are hearing they may not be able to express this fact, but will instead "act out" the anxiety they are experiencing.

WHAT IS A MELTDOWN?

A meltdown is when a person is unable to control themselves due to heightened emotions. The behaviour that results can be anything from refusing to communicate or move, to uncontrollable screaming and crying and in more extreme cases self-harm, violence and recklessness. Some behaviours may just be attention seeking and will cease, if ignored.

WHY DO PEOPLE WITH PWS HAVE MELTDOWNS?

People with PWS have a number of underlying issues that could cause them to have meltdowns. Firstly, most people with PWS appear to be anxious for much of the time so they may be on edge even before anything goes wrong or changes.

Their anxiety can be increased when they are around food or even when they are thinking about food or an upcoming social event. Anxiety takes many forms and can be seen in behaviours such as: avoidance, non-compliance, denial; perseveration; muddledness; frustration; argumentativeness; a drop in communication levels; compulsive behaviours.

People with PWS are ritualistic or like to do things in a particular way and therefore can become upset if things don't happen in a specific order. They have trouble regulating their emotions due to the issues they have with the area of the brain known as the hypothalamus. The hypothalamus has an effect on behaviours and is thought to impact on aggression and fear. This combination of factors is thought to increase the frequency and intensity of meltdowns seen in people with PWS.

WHAT SHOULD YOU DO WHEN A PERSON WITH PWS IS EXPERIENCING A MELTDOWN?

These are simple and practical strategies that help to minimise the impact of the meltdown:

- In the early stages of a meltdown it is important to validate the emotion that is being expressed. That is say to the person experiencing the meltdown "I can see that you are upset about what is happening. I understand that it is upsetting for you" OR "I can see that you are anxious. What is bothering you? Let's work on coping strategies".
  *Coping strategies can be slow counting or deep slow breathing.
- In the early stages you may be able to redirect the person with a firm calm voice. You may be able to work with them to solve the problem by offering two alternatives. For example if you ran out of coffee you could ask "Would you like a tea or a cold drink instead?"
- Another strategy is to guide them through the process of solving a problem. For example in the situation above you could say "Well we don't have any coffee what can we do about that?"
- Often it is impossible to stop a situation from progressing to a meltdown. If this happens remain as calm as possible during the incident. Getting upset or reacting emotionally to the situation will usually make things worse. Think before you act or speak. Speaking in a calm, assertive and confident voice can help. Avoid yelling and try to remain in control of your own emotions. Try not to show fear, anger or frustration.
- Keep yourself safe and out of harm's way. Consider the safety of others that are in the vicinity and try to discourage them from exposing themselves to harm or becoming involved. Consider the safety of the person having a meltdown. You may need to move dangerous objects away. In cases of violence or recklessness people may need to leave the area. You may need to seek help.
- Give the person time and space to calm down if possible. In a house they can be encouraged to retreat to their room until they are calm. In a workshop environment they could be encouraged to go to a safe area such as a change room or vacant office. In a public space try to encourage them to calm down in a quiet and safe area away from other people.

- If the person yells at you tell them that you can't help them unless speak in a soft calm voice. Refuse to respond or answer their question until they speak calmly. While this strategy may take time it can often help to calm the situation.
- Offer a cool drink or a wet towel. Encourage the person to wipe their face down before you discuss the situation further.
- Retreating to your own room and locking the door can be a good strategy if you are in your own house and it is safe to do so. Then you can come out and talk only once the person has spoken to you in a calm and respectful voice. This works well if they are trying to get you to agree to an unreasonable request. Be prepared to retreat back into your room if they yell or scream at you again.
- After some time the meltdown will end. It is important to be positive afterwards. Resist the temptation to immediately discuss the incident or tell them what they did wrong. You can always mention it once time has passed. For example the following day or later that night. You should try to avoid reigniting the anger or frustration. Once the incident has been discussed calmly try not to refer to it again.
- It is best to continue on with the rest of the day as far as is possible. It is important to move past the episode.

Some strategies that parents and carers have found successful in dealing with meltdowns

- As with our other children, if they threw a tantrum when they were little, we constantly used time-out in her room with the door open. We never gave in to unreasonable demands.
- We never used violence or aggressive behaviour ourselves. We built up her self-esteem from a young age.
- If possible, we would talk through the issue and try to help her to understand both sides of the issue.
- We always discuss and prepare for events or outings that could potentially cause anxiety.
- When our son becomes aggressive or violent and we can no longer reason with him, we remove ourselves from the situation and let the meltdown run its course. We remain nearby to make sure he doesn't hurt himself, but without him knowing we are there.
- Teach coping skills every day!
- The best way to learn to manage feelings of frustration is to practise often, when anxiety is minimal. Over time, these skills will be remembered during episodes of increasing anxiety. *Some effective strategies are: slow breathing, counting, using stress relief objects e.g. music, sensory balls and communication

THE PRADER-WILLI SYNDROME ASSOCIATION OF SOUTH AFRICA

The Prader-Willi Syndrome Association (SA) is a support group and was established by a small group of parents in March 1990

We invite everyone involved with individuals with Prader-Willi syndrome, including parents, family members, friends, professionals, caregivers and others interested, to become members of the association.

WOULD YOU LIKE TO JOIN THE PWSA (SA)?

Please contact:
Chairperson: chairperson@praderwilli.org.za or tel: 012 344 0241 or
Visit our website: www.praderwilli.org.za

COST OF MEMBERSHIP

Registration fee R50.00 (once-off payment)
Annual membership fee R200. R220 for members outside RSA.
You are welcome to make a direct deposit into the savings account.
Please ensure that your surname is included as reference on the deposit slip.
Please forward proof of payment to the chairperson.
chairperson@praderwilli.org.za or fax: 012 344 0241

BANK DETAILS OF SAVINGS ACCOUNT

PRADER-WILLI SYNDROME ASSOCIATION (SA)
ABSA BROOKLYN, PRETORIA
Branch number 632005
Acc. no. 11 364 1800
Reference: SURNAME, please

The views and opinions expressed in *People With Strength* are those of the authors and do not necessarily reflect the views of the management committee of the PWSA (SA).

USEFUL WEBSITES

Prader-Willi Syndrome Association UK	www.pwsa.co.uk
Prader-Willi Syndrome Association USA	www.pwsausa.org
International Prader-Willi Syndrome Organisation	www.ipwso.org
The Foundation for Prader-Willi Research (FPWR)	www.fpwr.org