

August 2010



WAVELENGTH



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Due to the fact that a great number of people with Prader-Willi Syndrome are living their daily lives in residential care, IPWSO has created a specialised Professional Provider Caregivers' Board to work with caregivers around the world. The members of this board are as follows:

Norbert Hoedebeck-Stuntebeck (chair), Germany
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From the President:

Suzanne Cassidy, MD



This is my first opportunity to communicate with those of you who belong to IPWSO as your new President, and it is my great privilege to do so. I am honored to help guide IPWSO into the next decade. Since its inception, IPWSO has been focused on bringing support, information, and some of the latest research about Prader-Willi syndrome to families, caregivers and professionals throughout the world, and we have grown enormously in numbers and influence in the 19 years since our establishment. I was delighted to be present at IPWSO's inception in 1991 in The Netherlands at the first international PWS conference and to watch IPWSO spread throughout the world. Our growth has paralleled the growth in knowledge about PWS that has, in significant part, come from the collaborations and communications established at the 3-yearly conferences by the scientists and care providers who

meet at the same time as families and caregivers have meet. This expansion of knowledge has lead to more publications and presentations at regional, national and international medical and scientific conferences, and in turn to more education and awareness about PWS for the professionals responsible for the diagnosis and physical and mental health of affected individuals. All this effort on the part of members of IPWSO has resulted in improved quality of life for affected individuals and their families.

I am a Pediatrician and Medical Geneticist from the U.S.A. I have been focused on clinical care and research on PWS since 1980, and have expended a great deal of effort in improving knowledge of this diagnosis among the medical community of the world. My aim as your President is to give IPWSO more importance and visibility in the medical world at the same time as I join the

efforts of others to help improve chapter integration and communication with the central activities, while at the same time lending my efforts to continuing the excellent efforts at country chapter support for individuals and families affected by PWS. Coordination of scientific efforts on a worldwide scale will be part of this effort.

We still have a long way to go and many countries to reach, though we now have member organizations in 83 countries!! There are many people with PWS who must be assisted, and care providers who need more knowledge about the best way to improve their health and lives. I pledge to work hard to increase the visibility of IPWSO, along with your Executive Director, Board of Directors and many other volunteers, in an effort to move closer to the goal of global assistance to people with PWS from all walks of life.



From the Editor:



All contributions to Wavelength are welcome. Publication dates: January, May, September. Views expressed in this newsletter are those of the contributors and not necessarily IPWSO.

Greetings to all,

What a wonderful month May has been with the 7th International Conference in Taipei, Taiwan, and meeting up with so many old friends and making so many new ones from all around the world. The conference had all the hallmarks of a classic IPWSO conference, but with a wonderful Taiwanese flavour. Our warm welcome and inclusion into the hearts of the Taiwan community is something none of us will forget. I know that one should speak of the highlights as being recent scientific discoveries, or new management skills, but for me, the highlight and –quite simply – the heart of the conference was to be found in the choirs, the drummers, and the stilt-walker. The choirs who sang of courage, of life, and of love; the drummers who beat out the rhythm to life, the ability to keep in time with the world and all it has to offer; and the wonderful stilt-walker who would not give up despite conditions that were not ideal, despite a tumble, he picked himself up and gave it his all. This is the spirit of IPWSO, the spirit of our families and the spirit of those we love and care for. I know many of you felt this too.

I have to mention the fantastic banquet, the welcome dinner, and the amazing food we were given! I love to try every different type of food available (well – what would you expect? I am the mother of a daughter with PWS, after all!), and especially food that I know I will never taste in my own country, so the banquet was like going on a mystery tour of culinary delights and differences. The banquet also had a little lesson on life as well – that we must embrace all cultures, all differences, and not hesitate to try something before we judge it, or else how will we know what we are talking about?

Our 7th international conference was a great success; the presentations were varied and interesting and for those attending their first IPWSO conference, I am sure you benefitted from the years and years of knowledge that have been built up since our beginnings. There are more conference reports in this Wavelength edition. Our next IPWSO conference is to be held in Cambridge, UK, in July 11-14th, 2013. This promises to be a feast – literary, geographic, historical, and culinary!

This June was World Cup month in South Africa, which brought great excitement to that country, but let us not forget that 2010 also marks the 20th birthday of South Africa's PWS Association, and we offer our warm congratulations to everyone there! Many of us will remember the founder of their Association, Birgit Schroder-Hadsbjerg, who, along with a small group of concerned and dedicated parents, established the association on 12 March 1990. Birgit was chairperson for 6 years and was also a founding member of IPWSO. She was greatly involved with all things PW until her sudden and untimely death in 1998. Her son, Jassim, was 7 years old when he was diagnosed with PWS at a time when very little was known about the syndrome in South Africa. It was through Birgit's dedication and determination that her son's needs were met and awareness of the syndrome was raised. Jassim now lives in Denmark, together with his girlfriend of many years, Heidi, benefitting from a residential programme which supports him in managing his food and finances.

Of the first five families who were founding members of PWSA (SA) all are still on the current membership list and Rika du Plooy (IPWSO parent



delegate) has taken on the role of Chairperson and editor of their newsletter, People With Strength. She is ably assisted by Wilna Basson, secretary, and Dr Engela Honey, their medical adviser, will be well-known to many of our PW professionals. The membership of the SA Association as grown since those days and awareness is starting to develop throughout the country. A new website [HYPERLINK "http://www.praderwilli.org.za"](http://www.praderwilli.org.za) www.praderwilli.org.za brings information and networks to a wide interest group and SA is now looking positively at further growth, particularly in the residential area.

I am delighted to have been asked to come and speak to groups in Durban, Pretoria, Cape Town, and Bloemfontein, and present my workshops on "Understanding the Person, Understanding the Syndrome" in August this year and, hopefully, together we will raise awareness even more.

Linda Thornton
National Director, PWSA (NZ)
Secretary, IPWSO



The lives of our children will be different

By DR LOISEL ULLOA BELLO, CUBA



Dr. Loisel with his family.

Sometimes, life can change in a heartbeat, without a warning: On July 9 2009 the lives of my family and my daughter Gabriela just did that: the uncertainties that for so long followed us started to vanish when our friend Janet contacted me with details of an organisation called IPWSO. Gabriela was then 5 years old and despite having a clinical suspicion since she was 9 months, we were unable to make a diagnosis because we did not have the equipment and means for the required test in my country.

The first thing to happen was that we received the means and instructions to get a blood sample and send it to the BIRD laboratories. Finally on August 25 we got a diagnosis to Gabriela's disability. Now we knew what it was, but the information I could find was very limited and, especially as the initial contact with the IPWSO was always through friends because I didn't have an internet account or a fax machine (even the results of Gabriela's test were sent to a friend by fax!) it was not possible to get much more information until September 2009 when I started to communicate directly with IPWSO and Giorgio, using the email account I got at the hospital where I work. From this moment I tried to keep updated on the disability management and the preparation of the family for living with a child like Gabriela. More IPWSO documentation was received in December with a friend of the family who carried it from Sweden (previously sent documentation by regular mail never arrived!). In the received package we got a lot of information in Spanish from the PWSA USA as well as a DVD for family guidance. When we started using the 'tools' provided by all this information

the change in Gabriela's behavior was amazing and life for our family became so much easier. This is something we will be eternally grateful for.

But the story does not end here, because around this time a tortuous path to attend the seventh international conference on PWS in Taiwan May 21-23 began. Something that for most of the participants took no more than a few weeks, took for me 7 months! In October 2009 I received all documentation IPWSO needed to be submitted to the Ministry of Public Health (MINSAP) to which I belong. All the paperwork started then and finally on March 24, 2010, after 4 long months of waiting, I got the permission to leave the country, something difficult to achieve for a medical doctor. I believe then that the more difficult part was over but...getting the visa to Taiwan was not precisely a walk in the park! There is no Taiwanese embassy or consulate in Cuba, so the visa application had to be done in the Dominican Republic. This requires travel there and, of course, a new visa application, this time to Dominican Republic which would take a few weeks that I did not have. Thanks to the unconditional support of the IPWSO and especially Dr. Shuan Pei Lin and Ms. Frances who lobbied with the government in Taiwan to allow me to get the visa in Guatemala instead, I finally got the clearance to apply for the visa in Guatemala. I want to mention the efforts of two families, the family of Julia Bonnelly in Dominican Republic and Mayra Urizar in Guatemala, both IPWSO delegates in their countries who gave unconditional support during the visa process in their countries.

Finally after a long 42 hours flight journey in which I had to go through Costa Rica, Guatemala, Mexico, Netherlands, Thailand I arrived to Taiwan, with a warm welcome from all members of the IPWSO board and all of the participants of the conference, I understood then that I was among one big family regardless political views, race or social class: the great global PWS family.

Throughout the event I was adding new knowledge on PWS, daily enriching my knowledge on the management of the syndrome, new treatments, and family support. It was an unforgettable experience



This little princess will now have a better life, thanks to the dedication of her parents and the long journey made by her father to learn from international experts, what it means to have PWS.

in which I had a unique opportunity to meet the most experienced professional on PWS. Thanks to my participation in this event, all the knowledge I gathered there and the support of the IPWSO, the Cuban SPW family, as we decided to call it, changed to the better and we will start nationwide work to help all of our members. In September we will have our first national conference, with the main objective of sharing all the knowledge received during this event here in Taiwan

and a future vision of helping all the PWS families in the Caribbean area. Been part of the seventh international conference on PWS in Taiwan was a dream come true for me, my family and all the other Cuban PWS families. Thanks to the support of the IPWSO and Giorgio especially (his significance for the Latin-American PSW families can not be put in words), the lives of our children will be different.

I would not leave without showing a picture of my girl ...

Dr. Loisel Bello from Cuba with Janalee Heinemann, Vice President of IPWSO, USA, Dr. Shuan Pei Lin the host of the conference from Taiwan (right) and Dr. Leopold Curfs the Chair of IPWSO Scientific Advisory Board from Holland (left) at the 7th PWS Conference in Taiwan.





Maria Paula



We send our deepest condolences to our parent delegates and their family in Colombia, Jorge Restrepo, Nora, Daniel and Ricardo, for the loss of their beautiful 20 year old daughter, Maria Paula. Her family writes:

"Maria Paula has left us alone on April 1 this year, and we deeply lament her passing. The cause of her death was an insufficiency to the kidneys, a disorder she developed when she was two years old following a bout of severe pneumonia. Maria Paula was our angel to whom we gave all our love. She was a happy girl and her tenderness, tenacity, happiness and strength were always present in her life. During her illness, she taught us how to be happy while suffering. She became a close friend of all the doctors and nurses who followed her with as much love as they could. Maria Paula had a wonderful ability with jig-saw puzzles which we had always encouraged, and this is what she did until the end. During the last ten days of her stay in hospital, she completed her last five puzzles.

Maria Paula attended high school until the day before she was hospitalized, and she was going to graduate in November this year. We were all hoping for the

realization of her dream to become a teacher of small children. She was already preparing her own educational material to make her dream come true. She will realise her dream, because we are going to donate all the works she prepared to a local kindergarten. Maria Paula has left us with a great calmness and serenity in the knowledge that we did our very best to make her life a happy one. We can only say today that we are proud we had her for our daughter.

We wish to thank God, because He gave us this Angel and if it would be possible to come to this world and be born again, we would ask God for another daughter like our adorable "gordita". We wish to send a big hug to all families and children with Prader-Willi in the world and be sure that Maria Paula from Heaven will be with you all, continuing the mission she started during her life on earth.

To Giorgio and all of you, our gratefulness for the support you offered, and for the pleasure of meeting some of you during the Conference in Argentina in 2005. We feel your presence by our side now. We send some of our recent pictures of Maria Paula, together with us and during Christmas time together with a chef – you can imagine how happy she was!

From our country, Colombia, we are available to share our experience and our life with Maria Paula which allowed us to be a happy family and to make her happy too.

A big hug to everybody.

Jorge Restrepo, Nora, Daniel and Ricardo"



What a special time in Taiwan!

What do I remember? Beautiful flower arrangements with the scent of perfume lilies which welcomed us every day at a well organised conference! The assistance and friendliness of the supervisors, the spirit of caring and sharing among the attendees, excellent presentations by the presenters and the milestone which was reached by the committee of the Caregivers Conferences, in announcing the completion of their hard work. Thank you to each and everybody who worked so hard and my appreciation to Li-Ping Tsai, Shaun-Pei Lin and Helen HC Chin and other hosts and task forces. May we again have the privilege to meet in 2013 in the United Kingdom.

Rika du Plooy (South Africa)



Every international conference has its own special flavor. In Taiwan, the hallways were lined with dozens of meter high pink blossom flower arrangements sent by sponsors and friends making a stunning visual memory. Charming volunteers were stationed at every corner, even in hotel lobbies, to guide us to meetings. Added to this was an amazing chorus of children who have rare disease. We will remember the Taiwanese people as very special!

Most memorable of all, however, was the genuine caring and interaction of all attendees from those with brand new diagnoses to scientists to veterans like us. It is this warmth which makes hours of sessions, even about the difficult aspects of PWS, energizing instead of discouraging. I stand in awe of those, who came as one person from a country, determined to return home build a PWS infrastructure in the health and education systems. Armed with the information of the Taiwan conference and the support of the IPWSO members whom they met, there is no doubt that they will be successful.

P.S. It is also amazing that Dorica Dan will be opening a new center for PWS, residential, therapies and vocational a few hundred yards from the city hall in Romania. Truly, IPWSO's empowerment of individuals is extraordinary!

Thanks,
Joan Gardner
(U.S.A.)





The Right to Make Your Own Decisions

By Professor Tony Holland, President PWSA (UK)

This article is based on a paper written in response to the death of an adult with PWS who had become very severely obese. This case of Chris had earlier received national publicity as he had briefly been detained in hospital under the English Mental Health Act. The national newspaper concerned had campaign against this, with both Chris's and his mother's support, arguing he should be free to live where he chooses and not have to diet.

The situation in the UK and in democratic countries in general is that adults (those 18 years and over) have a right to make those decisions that affect their lives. In the case of health treatment it is individual consent that renders an investigation or a health procedure, lawful. For a person to give valid consent he/she must be appropriately informed, have the capacity (ability) to consent and be able to communicate his/her decision and be free from coercion. To treat an adult who has the capacity to consent without his/her consent would be unlawful and probably an assault. In different countries the policies and practices and legislation of that country will address issues of decision-making in children or the particular circumstances when the above may not apply and, for example whether adults may be admitted to hospital to have treatment without their consent, for example, for assessment and/or treatment for mental illness.

For adults with PWS there is potentially a real difficulty in this transition from being a child and under the responsibility of parents, to being an adult having the opportunity to make their own choices. Ideally, as someone with PWS leaves school, a care strategy is devised whereby with their agreement they move into a food supervised setting that can ensure considerable independence but can manage the food environment - in this respect prevention of obesity is by

far the best option. However, if the person concerned does not wish for this and/or such options are not available the type of problem that arose with Chris, occurs. Under the legal framework of England and Wales at the time of Chris's death (prior to the passing of the Mental Capacity Act 2005), there were only three possible approaches. These were summarised in our paper in the Journal of Medical Ethics: Holland, A.J. and Wong, J. (1999) Genetically determined obesity in Prader-Willi Syndrome: The Ethics and Legality of Treatment. Journal of Medical Ethics, 25: 230-236.

With the Adults with Incapacity (Scotland) Act 2000 and the Mental Capacity Act (England and Wales) 2005 the situation within these three countries have changed, although essentially there



are still just three options. These are: 1) to accept that dieting is a matter of individual choice for people with PWS, as it is for the rest of the population, and therefore one can inform, encourage and cajole, but not force dieting; 2) to determine whether the person has the capacity to make such a decision and if it is determined he/she does not then there is the responsibility to act in his/her best interests - if necessary (in England and Wales) referral to the Court of Protection under the Mental Capacity Act may be required for their ruling on the issues; and 3) to use the powers of the Mental Health Act (or equivalent in other countries) to lawfully detain the person with PWS in hospital. Whether these options are appropriate and

possible in any given country will depend upon the laws of that country. For some countries Guardianship legislation may, for example, be available and appropriate, at least in some circumstances.

The balance in this situation is always this tension between respect for autonomy and need for care. The solution is ideally through co-operation and consent but if that is not possible there are only limited options as described above. Key to the argument about capacity to make decisions is whether PWS is seen to be different from general obesity. In the above paper we argued that people with PWS may be different, whilst they may have the capacity to consent to many things in their lives, however, when it comes to food the insatiable hunger so distorts their thinking that it renders than incapacitated with respect to decisions about dieting. Under those circumstances, it can be argued, that there is a duty to take over control of dieting in their best interests. Thus, one could defend limiting access to food on the grounds that it was in his/her best interest. However, as Chris's case illustrated the framework provided by the law is only a small part of the issue - of perhaps greater importance is having the 'PWS informed' support and services available in the first place.

What are the lessons from this situation of Chris? First, support is complicated and there may be a very difficult tension to resolve - autonomy versus care. Secondly, if appropriate local social care services had been available it would have maximised the chance that such situations would not have arisen as Chris and others with PWS may have been more willing to live in an appropriate environment. Thirdly, difficult decisions may have to be made and if necessary legal advice sort to resolve such problems. Knowledge of PWS is key to this.



What would you do?

By Jackie Waters, PWSA (UK)

I have a grown-up daughter, 32, who, even though she has PWS, was never too much trouble as a child, and her weight was well-managed. In fact, so well-managed by both her and me that I thought that we could perhaps relax the rules a bit as she came into adulthood.

Gradually, she took over cooking meals for herself, shopping and going out alone - though she does still live at home. Contrary to most stories I have heard about people with PWS, the result of relaxing rules was not an immediate weight gain, but a long, slow, almost unnoticeable increase in weekly terms. But now, after some 12 years of "independence", she has put on around 70 lbs (32 kgs) which is affecting her health and well-being - she has diabetes and oedema.

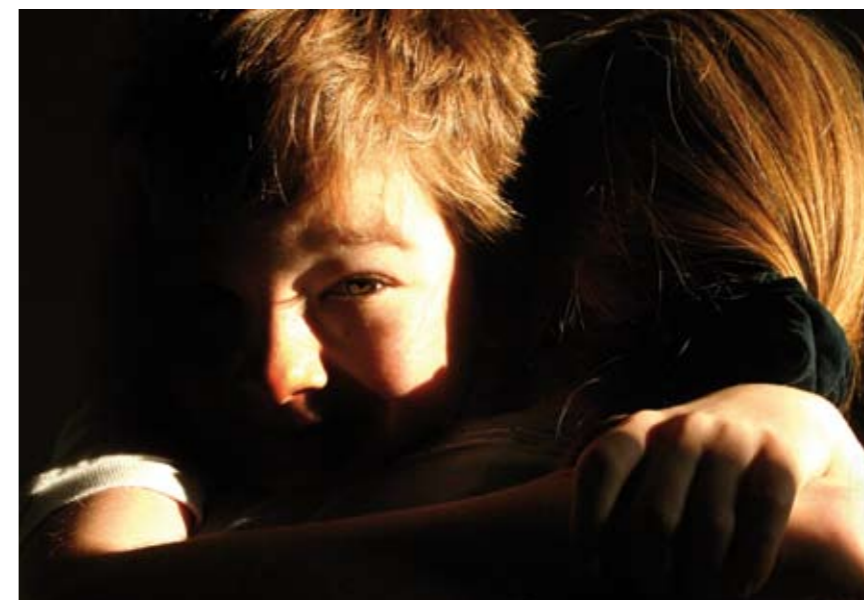
Her general stance now is, "You can't tell me what to do" and "You don't know what it's like to be me." The independent attitude extends beyond food - her room is a complete mess, but she refuses help to tidy it up - most days she loses something in the mess, and a cry of "Muuuuuum!" goes up, followed by lots of yelling and crying. She needs me then to help her find it! Similarly, when out and about, something may go wrong, and she phones me on her mobile for me to sort it out for her. It is like living with a perpetual 13 year old.

She has refused so far all efforts to place her in a specialist PWS home - of which we have several in the UK. She has had a couple of respite stays in one and was not impressed. She says she doesn't want to live with other people

with PWS. In fact she shared a room at residential college with another girl with PWS for one year and they had quite a few "bust-ups" but have remained firm friends, still seeing one another a couple of times a year.

Did I do wrong by letting her have this independence? If anyone should have known better it is me - as I am the author of several books and leaflets on the best way of managing PWS, and they all advocate environmental controls and restricted access to food!

I think she would say I was right. She has an interesting life, including boyfriend and part-time job, and she feels in control of what she does. I, on the other hand, worry about her increasing health problems, and have to deal with the day to day problems of someone who is constantly in the kitchen cooking for herself, refusing to eat with me and my partner - our mealtimes are a set of complicated logistics in a very small kitchen of two people trying to cook entirely separate meals. And there are many other "issues" which require daily management/intervention from me - but only when she wants me to!



But in another sense, I have had less personal stress by not trying to impose a dietary regime on an unwilling and fairly intelligent person - and the inevitable battles that would have produced.

With PWS, there are no easy answers, and how we manage it depends on several factors:

The way in which PWS manifests itself - we know it has many variables and degrees of severity

The personalities of both the parent(s) and the child - and how they relate to each other

The culture in which the family lives - where there are no residential homes, that is not even an option, and the onus lies completely on the family to deal with the situation as best they can.

The factors we might use to measure quality of life conflict with one another in PWS: people with the syndrome cannot have a healthy life without foregoing at least some independence and personal control over their lives.

I am still looking to achieve that balance with my daughter, so that she can have the best of both worlds, as far as possible. I know that if restrictions are not placed on her soon, she will not live much beyond 40 years old, but the task remains to get her to agree to them. As I said to her social worker, "Her placement with me has broken down" (not bad after 32 years, though!). The search is now on to find a better one - if we do, I will let you know!



Rising From the Ashes

By Janalee Heinemann, IPWSO Vice-President



Janalee Heinemann with Hubert Soyer in one of the greenhouses of Regens Wagner.

I felt privileged to be one of the honored guests in July of Hubert Soyer at the Regens Wagner's 100th Anniversary Celebration in Absberg, Germany.

Regens Wagner is an outstanding, multifaceted residential setting for Prader-Willi syndrome and other disabilities. It serves and houses 250 adults with special needs in Absberg, and the company as a whole serves 6,550 throughout Germany. Thanks to Hubert, who is the director, in the fifteen years that he has been director of Regens Wagner, seven cheerful, beautiful PWS designated homes have been established on expansive and serene landscape. There are seven to eleven residents in each home. The residents all contribute to the remarkable artwork that is hanging everywhere and all have extensive opportunities to work and participate in recreational activities. Currently, Hubert is building several large

greenhouses out of town on the farm they operate, and another building where the vegetables will be cleaned, stored and sold by the residents. Attached to this building is a Bistro that will open to the community and be staffed by residents. The Regens Wagner campus is exclusive for those with special needs and yet inclusive of the larger community at the same time.

Bishop Gregor Maria Hanke (Bischof von Eichstätt) and Princess Katharina Fürstin von Wrede were at the 100th anniversary celebration along with numerous other dignitaries. There were also five of us who are representatives of IPWSO that were there to honor Hubert who has had the vision, energy, charm and devotion needed to create a beautiful world for people who have Prader-Willi syndrome. Tausend Dank! (A thousand thanks!) Hubert.

While Regens Wagner is a shining light for people with special needs, it had to rise from the ashes twice in the 100 years since its establishment. The first time was during World War II when many residents were taken away and never seen again in spite of the nuns (who are much of the heart and soul of Regens Wagner), and members of the local community hiding residents at the risk of their own lives. Then, there was a terrible fire in 1969 that destroyed most of the buildings thus it did have to literally rise from the ashes and be rebuilt.

Like a family getting a diagnosis of PWS, you have to rise from the ashes of your despair and feeling of hopelessness and create a new world for yourself and your child. It may not be the world you had planned, but it can be a world with its own sense of beauty and joy.



Sleep Problems in PWS

The first intervention is to improve sleep hygiene by introducing a daily exercise plan and shaping behavior to assure the appropriate amount of sleep and to supplement this with planned naps, if necessary.



This article is written by Janice Forster, MD (left) and Linda Gourash, MD Pittsburgh Partnership, Pittsburgh, PA (USA)

Sleep problems occur frequently among individuals with PWS. The most common problem is excessive daytime sleepiness (EDS). There are many reasons why individuals with PWS experience EDS. Sometimes EDS is related to sleep apnea that disrupts the quality and efficiency of sleep. Obstructive sleep apnea is associated with increasing body mass index (BMI). Central sleep apnea occurs independent of BMI. Many individuals with PWS experience

oxygen desaturations as a result of these apneas. It is common for our individuals with PWS to receive sleep studies to investigate the occurrence of apnea or oxygen desaturation; these sleep studies are usually performed in a sleep lab by a pulmonologist. Sleep apnea can interrupt the continuity of sleep, and these disruptions result in a decrease in both the quality and quantity of sleep. Excessive daytime sleepiness can result from this decrease in sleep efficiency. Hypoxemia (low oxygen in the blood) can occur

independent of apnea. Many of our individuals experience hypoventilation (shallow breathing). In the extreme situation, hypoventilation can cause hypoxemia. Although hypoventilation is more likely to occur as BMI increases, it occurs to some degree in everyone at rest; it is reversed with exercise and deeper breathing. Hypoventilation results in an accumulation of carbon dioxide (CO2) in the lungs. In the typical individual, rising CO2 levels (hypercapnia) automatically trigger the need to take a deep breath or sigh. Individuals with PWS are less sensitive than typical persons to accumulating levels of CO2, so hypoventilation continues. In typical individuals with obstructive sleep apnea, hypercapnia alters sleep architecture and worsens daytime sleepiness.



Sleep studies also measure sleep architecture: the phases of sleep. Typical individuals experience many cycles of NREM (non-rapid eye movement sleep) and REM (rapid eye movement) sleep through the night. Each sleep cycle lasts 90-120 minutes. At the onset of sleep, NREM sleep (restorative) dominates the cycle; as the night progresses, REM sleep (dream sleep) dominates the cycle. The coordination of these sleep cycles is controlled in the hypothalamus. If a typical individual sleeps only 4 hours but usually requires 8 hours, they satisfy their requirement for NREM sleep, but they need to make up for the loss of REM sleep. The sleepiness that they experience the next day is associated with the intrusion of REM sleep into their wakefulness that is taken away by a planned nap or possibly experienced as an involuntary “cat nap.”

Due to hypothalamic dysfunction, individuals with PWS have intrinsic abnormalities of sleep cycles. They have more sleep cycles per night and more REM periods. But there are a lot of arousals during REM that fragment and decrease the overall amount of REM sleep, increasing their “sleep debt” on a daily basis. Further, their sleep efficiency may be compromised by the occurrence of apneas. Because excessive daytime sleepiness has been correlated with the intrusion of REM sleep into wakefulness, individuals with PWS may appear to have narcolepsy-like symptoms.

Narcolepsy is a sleep disorder originally described as sleep attacks, an irresistible urge to sleep. However the majority of individuals with the condition display EDS. Narcolepsy is a disorder affecting the cycles of sleep resulting in sleep fragmentation, inefficient sleep, and the intrusion of REM sleep into wakefulness. In fact, the associated symptoms of narcolepsy are related to this REM phenomenon: cataplexy (the sudden loss of muscle control in response to strong emotions); hypnagogic hallucinations (lucid, vivid dreams at sleep onset); hypnopompic hallucinations (lucid, vivid dreams just before awakening); and sleep paralysis (inability to move voluntary muscles during the transition between wakefulness and sleep). REM sleep is associated

with a loss of voluntary motor control that prevents the acting out of dreams. Sleep paralysis and cataplexy are the manifestation of this loss of voluntary motor control intruding into wakefulness. Narcolepsy occurs with or without cataplexy; and occasionally, cataplexy occurs without narcolepsy. For example, because antidepressants suppress REM sleep, their discontinuation has been noted to cause REM rebound and the precipitation of attacks of cataplexy. Narcolepsy is diagnosed by clinical findings and by the results of another kind of sleep test: Multiple Sleep Latency Test (MSLT). In contrast to the nighttime sleep studies that most of our individuals with PWS receive, the MSLT is performed 4-5 times through the day in the sleep lab at 2-hour intervals, measuring the onset of sleep and REM periods during naps. In narcolepsy the time that it takes to fall asleep is very short, and the time that it takes to experience a REM period is similarly shortened. A hereditary form of narcolepsy has been described, but narcolepsy also occurs as a result of central nervous system dysfunction when the function of the hypothalamus is impaired. Some individuals with PWS have also been diagnosed with narcolepsy. Because most individuals with PWS who have EDS receive nighttime sleep studies to look for sleep apneas, the findings associated with the daytime MSLT have not been systematically explored. Of all of the narcolepsy symptoms, cataplexy is the most difficult to treat and may require consultation with a neuropsychiatric sleep specialist. The incidence of narcolepsy-like symptoms in PWS appears to be higher than what is seen in the typical population (1:2000), but there have been no formal studies.

The treatment of excessive daytime sleepiness and narcolepsy are similar. The first intervention is to improve sleep hygiene by introducing a daily exercise plan and shaping behavior to assure the appropriate amount of sleep and to supplement this with planned naps, if necessary. The second intervention is to treat the source of sleep disruption; CPAP (continuous positive airway pressure) or BIPAP (bi-level positive airway pressure) are needed for obstructive sleep apnea or intermittent hypoxemia. Then, the daytime use of stimulant medication (either



CPAP, continuous positive airway pressure

methylphenidate or dextroamphetamine derivatives) and/or modafinil (Provigil) is recommended. Both stimulants and modafinil may be administered multiple times per day if needed, but not too close to bedtime as they might interfere with

sleep onset. Modafinil is approved for treatment of narcolepsy and excessive daytime sleepiness, but a copy of the sleep study may be required to authorize its use for EDS. Many individuals who have EDS for any reason have difficulty with

attention span and memory. Therefore, improving sleep efficiency and using stimulant medication to treat residual symptoms will improve overall level of function.



Studies into Oxytocin

Prof Stewart Einfeld (Brain & Mind Research Institute (BMRI), University of Sydney), Dr Adam Guastella (BMRI, University of Sydney), A/Prof Arabella Smith (Children’s Hospital at Westmead) and Prof Iain McGregor (University of Sydney) are investigating the treatment effects of a hormone, oxytocin, on physical, behavioural and cognitive aspects of Prader Willi Syndrome.

They hope to learn if an intranasal administration of oxytocin improves over-eating, eating non-food substances, weight, rages/tantrums, skin picking, daytime sleepiness, obsessionality, and emotional empathy. It is an 18 week trial (8 weeks of oxytocin nasal spray and 8 weeks of a placebo nasal spray, with a 2 week washout period in the middle).

The trial is open to people with PWS aged 12 years to 30 years, and participants will be required to visit the BMRI on three occasions.

For further information please email: pws.research@sydney.edu.au



Book Review:

Reviewed by Linda Thornton

A Handbook for Parents & Carers of Adults with Prader-Willi Syndrome – 2nd edition

Edited by Jackie Waters and published by the UK PWS Association, “ this handbook is primarily for parents of adults with PWS, but it will be of use to anyone who lives with, or cares for, a person with PWS on a regular basis, or who wishes to learn more about the syndrome.

Throughout the book, emphasis is placed on the fact that individuals with PWS share common traits, but they can also be very different from one another.

Care has been taken to ensure that the needs of people with PWS within their adult status are addressed, but this can be difficult to reconcile with their daily lives because so many people with the syndrome require a high level of support and even control to ensure that they lead a healthy and fulfilling existence.”

Once again, the PWSA UK has produced an excellent handbook covering the issues that affect us as caregivers and parents. The book covers the characteristics of PWS, health, sexual development and ageing issues, dietary management, psychological issues and behaviour management, ethical and legal issues, social life and relationship, education, training and work, as well as residential care and supported living.

There is a section on benefits, allowances and services which is specifically for the British system, but this also gives good, solid information and offers sensible information on questions to ask, what to do with Wills and Trusts,

etc that will relate to any country.

The booklet presents a very positive style of dealing with even the most demanding of issues and the framework of support given to parents with lists of questions to ask if you are not satisfied with the way things are going for your son/daughter in residential care, at school, or in the community.

The booklet is available from the PWSA (UK) for 8 pounds sterling plus postage, and can be ordered from the HYPERLINK ”http://www.pwsa.co.uk” www.pwsa.co.uk website.

Exercise and Physical Activity for Children with Prader-Willi Syndrome

Written by Kirsty Reid and Peter SW Davies of the Children’s Nutrition Research Centre at the University of Queensland, this extremely timely booklet is an excellent guide to the critical importance of exercise for those with PWS. It describes why children with PW are likely to become obese, and throws the old chestnut of a “lower metabolism rate” out of the window:

Many believe that children with PWS have a lower basal metabolic rate (amount of calories burned at rest) than other children. This belief is unfounded. Studies have shown that once a child’s height, weight, body composition and age are accounted for, the basal metabolic rate of a child with PWS is similar to that found in other children.”

It accepts, however, that children with PWS have more fat mass, and less lean mass, thus making exercise more difficult.

Preventing obesity, modifying eating behaviours, knowing how long it takes to burn off the food that we eat (for example, did you know that a simple banana muffin will take 111 minutes of cycling to burn off that amount of calories?) There are helpful hints on how to set out (and stick to) an exercise plan, how to introduce exercise to your child, the type of exercise that might be best for your child, and a whole range of resources of where to go to join activities or clubs.

The book is well laid-out, easy to read, lots of pictures and insets of helpful hints. There is also a chapter on Growth Hormone treatment which reports on a study done that shows an increase in spontaneous activity when a child is on this treatment.

This book should be on everyone’s bookshelves, without a doubt. It is currently at the printers and will soon be available from the PWS Associations in Australia and New Zealand, or from the IPWSO website. Limited copies will be available at the Taiwan conference.

Thanks should be given to the University of Queensland, the Children’s Nutrition Research Centre, the Royal Children’s Hospital Foundation, and to Pfizer for their generous contributions.

