

December 2010



# WAVELENGTH



*Piece of artwork of the art exhibition by Taiwanese children, Prader-Willi Conference, Taiwan 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



**W**arm greetings to you all, throughout the world! It has been just a few months since I took over the President's role from Shuan-Pei Lin, and these have been extremely busy and productive months. The new Board of Directors has been looking at our priorities and working on a fundraising program which we hope will broaden our financial base and allow us to increase our support to member organizations and projects. Giorgio, as our Chief Executive, has been hard at work making contact with organizations and individuals around the world and providing education and support by phone, email, and in person. I have been particularly impressed at the incredible spread of knowledge and interest that he and Janalee (our new Vice President) accomplished at the ESPE (European Society for Paediatric Endocrinology) conference recently in Czechoslovakia, and I hope that many of you will have read of their experiences in our new IPWSO Blog, organized by Linda Thornton. Such meetings always create a great deal of interest in the syndrome and in the work that IPWSO does, and spread information on how to achieve the best

outcome for people with PWS.

Because of our presence at ESPE and other conferences, Giorgio informs me that we have welcomed delegates from seven new countries to our fold since the last issue of Wavelength: Bangladesh, Iran, Kenya, Macedonia, Russia, Ukraine, and the Palestine Territory. This is a fantastic rate of growth, and brings the total number of IPWSO member countries to 90! We get stronger all the time and, thanks to the Internet, awareness of our pivotal role related to PWS is spreading rapidly.

One of the many wonderful outgrowths of the IPWSO conference in Taipei this past May has been the newly-formed Provider and Caregiver Advisory Board. This highly energetic group completed the production of a widely-distributed CD, "Best Practice Guidelines for Residential Care", the culmination of 3 years work, led by Norbert Hödebeck-Stuntebeck. It was born from the vision of Pam Eisen some years ago. This is a vital piece of work and an excellent resource for care-providers all around the world. As these are general "guidelines" they can be adapted for each country and every culture.

Also resulting from our Conference in Taipei, and due to the wonderful generosity of our Taiwan hosts, IPWSO has been able to distribute DVDs of the entire conference to delegates at no cost. These DVDs provide an excellent resource for those who were not able to attend and those who want to have it for reference. Many people have found it very useful, including me, and the Board is hoping that donations from people who receive it will help reimburse the cost of replication and distribution.

We are drawing to the close of another busy year, and our thanks must go to our major sponsor, Pfizer, to our hard-working Board of Directors, to our CE, Giorgio, and to all our hardworking contributors for supporting the work of IPWSO throughout the world. It has been a pleasure getting to know my board and some of the very active members these past few months and I am looking forward with great interest to 2011. In the meantime, I wish a all of you a very happy holiday season, and for us all I wish a very happy, healthy and successful New Year.



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.

## From the Editor:



Recently, a young woman with PWS who was attending local Tai Chi classes with her caregiver, was approached by one of the older women there and asked if she would like to address their group about her disability, “because”, the woman said, “it would stop the others talking and making wrong assumptions.”

This is what the young woman stood up and said:

“My name is ..... and I have Prader-Willi Syndrome. I read about someone in a book called Kenneth, and I discovered he had the same lot of behavioural problems I went through. When I lose the plot really badly I feel like no one else understands what I am going through.

I am now going to tell you how it feels for me to have Prader Willi. Well it is a food disorder. This is how it affects me. If you imagine your stomach is a grocery bag and that it never fills up like your stomach. My stomach has a hole in the bottom of the bag, which I am calling my stomach and it never fills up unless it is excessively over fed. It is not the Mother’s fault as it has nothing to do with their pregnancy. As I say the mother can’t prevent their child from having

pws. I have body parts missing from the brain and from my tummy and from my womb. This doesn’t let my body function normally. Prader Willi people have the tendency to eat all the time. They don’t do it intentionally, to be greedy or rude they just can’t help themselves. So if you see people or know people with Prader Willi overeating just remind them politely with a gentle reminder or go and tell their caregiver as they should have 24/7 support. If anyone has access to the internet go and look up Prader Willi Syndrome. You will come across a very good web site which my Mum has set up.

Since college years my weight has always been an ongoing issue and during my older years there was just too much temptation around shops like buying lollies chips, icecreams, all the yummy stuff that tastes great but it’s not healthy for me or anyone else.

My life time goal would be to look after my parents when they old and when they can no longer look after themselves, carry on at trying to lose weight and to give up smoking and to be there for my sisters children and to be a Aunty and to keep up the good work by controlling my anger and to present no more assaults and not getting into trouble with the police.”

This was written by my daughter. She is 26 years old and has been through more in her short life than most people who live til their 90s. Having PWS is such a tough thing to deal with and most of us, as parents and caregivers, just deal with each situation as it presents itself, but actually being inside the skin of someone with the syndrome is almost too hard to imagine. It made me smile to think our daughter might want to care for us (read: cook for us) in our old age, but made me sad to think one of her goals in life was not to get into any more trouble with the police. I don’t know anyone else with that ‘goal’.

So as this year draws to a close, I think of my many friends with PWS and their parents and caregivers, and I hope that the season of Christmas, however it is celebrated, is a good one for you all. I hope those of you who are snowed in, will be kept safe and warm, and those of us whose celebrations will be in the warmth of summer, also keep safe.

With every good wish that 2011 brings peace , happiness and joy,

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

# STRENGTH IN NUMBERS

– the Romanian PWS Association does it their way

The Second Eastern European Conference on PWS held in Zalau on 29-30 October 2010. Chaired by Doric Dan, and supported by APWR, IPWSO, EURORDIS, Health Ministry, Innovation Norway, partners and local

to bring awareness of specialised needs to the Ministry of Health. Practical, sensible, and able to get results, this model is hugely successful in countries where knowledge of disabilities is less recognised.

disciplines of: weight management, physical therapies, speech therapy, behaviour therapy, and educational interventions, and with a large network of professional help (international and national), the Romanian PWS Association



authorities, this conference drew international speakers from Denmark, UK, USA, Italy, Norway and France.

This conference had a slightly different slant from most of our international PWS Conferences in that the part played by EURODIS, which is the umbrella for all Rare Disorders, was to look at the model of the Romanian PWS Association as a model for all new associations.

The Romanian PWS Association was established in May, 2003, and the Information Centre for Rare Genetic Diseases was opened in Romania on 16 October, 2005, as the first resource centre of its kind in this country. Because of the isolation of the very many smaller groups of genetic disabilities, this umbrella organisation was imperative

Regular meetings and conferences make certain that voices of the smaller groups, such as the Prader-Willi organisation in Romania, are heard, and needs can be acted upon – the bigger the group, the louder the voice. Starting out with only 4 members, this organisation has grown hugely over the last 7 years; joining with other international organisations for Rare Disorders, has given this young group the strength it has needed. Still in its infancy, it seeks to provide information, resources, education, and support to every rare disorder looking for such help.

As the PWS Association was the first group organised under this umbrella, it is being looked to as a model for future associations. Based on the five main

has certainly broken the groundwork for other groups wishing to form their own association.

Dorica Dan believes that this strong growth can be attributed to the combined knowledge gained under the larger international umbrella of EURODIS. Their own national organisation of RONARD has now been formed (Romanian National Alliance for Rare Diseases) with 32 founding members and networks with many other agencies.

For small countries where PWS is less recognised, Dorica believes this is the best possible way to go – associate and align your group with other rare disorders, and together become a strong, tight-knit group with more power to get things done.

# Emanuela's Story

Emanuela was born on the 11th November, 1998 in Skopje, Macedonia. Growing up in a small country where PWS is only a theoretical case of study, not a lot of doctors were familiar with the actual statistics about this syndrome. As a child she was given the wrong diagnosis and was treated for Cerebral Paralysis until the age of 10, when her pediatrician recognized the syndrome and the test came positive. This is one of many cases where doctors in Macedonia, because of their lack of knowledge about this condition in general, misdiagnose a child with PWS. As she was growing up, her parents had hard times dealing with the behavioral problems and especially the food management. They had no one to ask help from. Nobody could understand their everyday problems or give them answers.

All the hard work that Emanuela, her family and the doctors did to help increase the quality of her life didn't seem to give results. In the most hopeless times, out of nowhere, a voice of support and a chance for better life knocked on their door. This is when Emanuela's pediatrician got an e-mail from the IPWSO, that Mr. Norbert Hödebeck-Stuntebeck from Germany was willing to help her and make her life better.

Time flew by, and it was already November, the month that she was invited to go to Germany and stay at a PWS group home for a few days. Emanuela was excited and willing to give this a chance. Having in mind her condition and all the unexpected behavior that this syndrome carries her parents had their doubts, but this was not only a huge opportunity, but also the only one.

We arrived in Gronau. Right after the first meeting with the caregivers and psychologists things changed. Her mother was surprised of the staff's knowledge about the syndrome. Their will to help her daughter was amazing to her. She finally realized that only by knowing the different psychological, emotional and organic disorders that her daughter had, ways to improve her lifestyle could be found.

Emanuela fell in love with the place. For the first time in her life she found a place, away from home where she was accepted for who she really is. Despite of the language barrier she made friends with all the people in the group home. You could tell from the look in her eyes that she was thrilled to be among people that are like her. It was way easier for her to fit in, and have a great time, while ignoring her eating habits. It wasn't hard for her to accept the house rules and the most important thing; she was happy and pleased with the healthy food she was given. The results were incredible. She could not be happier about the 3 kg she lost during this 5 day visit.

The day for our departure soon arrived and it was time for us to go back to Macedonia. As we arrived at the airport in Skopje, nothing that her father or sister had to ask was more interesting than the impressions she had from this trip. From the next day many things were changed in their home. Much of the advice her mother got about managing everyday life with

PWS were implemented immediately in their house.

Still as all parents know, at home it is not as giggly and smooth as it is with professional caregivers. They are hoping and willing for a permanent change. Emanuela wishes and waits for an opportunity for her to go back to Gronau, visit her friends at the PWS group house, and hopefully get to live with them for a while. Also helping Emanuela was a great progress for Macedonia in the field of PWS. An important benefit from this whole project, other than the good it did for Emanuela, is increasing awareness among the doctors and the whole parenting public. Maybe by letting people and doctors know about PWS, more of the statically expected 250 PWS children will be diagnosed.

I hope that one day I can see Emanuela living a happy life.

*Written by Emanuela's family and translated by Desi Kocavska, a medical student and friend of the family.*



# Guidelines on Scoliosis



BY:

AUTHOR: HAROLD J. P. VAN BOSSE, MD  
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## Monitoring and Treatment for Children with Prader-Willi Syndrome

### BACKGROUND

Children with Prader Willi Syndrome have an incidence of developing scoliosis at rates between 40-90%. Approximately 15% of children with Prader Willi Syndrome will develop severe or significant curves, requiring bracing or surgery. The earlier the curve is detected, the better the possibilities for treating the curve with casting or bracing.

There are two peak ages for scoliosis presentation in children with PWS. Under the age of 4 years, most of the curves are C-shaped, and are most likely related to the hypotonia. The second peak, centered around 10 years of age, typically is the more common idiopathic S-shaped curve. Fifteen percent of curves diagnosed before 4 years of age subsequently required surgical treatment, 41% of curves diagnosed after 4 years of age required surgical correction, as per a PWSA-USA survey of membership. Spinal deformities in children with PWS are often diagnosed late. This delay appears to be due to unique characteristics

of spinal deformities in children with PWS, rather than the presence of obesity. Fewer children with PWS now develop obesity, and often the curves are diagnosed prior to the onset of obesity. What seems to be the more important factor is that spinal deformities in children with PWS have less vertebral rotation than seen in other children with scoliosis curves of a similar size. Vertebral rotation causes the asymmetry of the chest wall seen during forward bending, usually the first sign of scoliosis. Therefore, the child with PWS may have a moderate curve radiographically, but only mild findings clinically. For that reason, there should be a much lower threshold for working up clinical findings in children with PWS, compared to otherwise unaffected children.

### CONDITIONING

Children with PWS should be encouraged to be as active as possible, particularly those activities that build core musculature, strengthen the abdominals and the back muscles. In addition to

sports and recreational activities, focused physical therapy and hippotherapy, emphasizing core muscle strengthening, may help improve a hypotonic curve in a young patient with a flexible deformity. If kyphosis is noted to develop, a physical therapy regimen should be included with specific exercises to strengthen the back extension muscles. A number of parents have also found pilates to be beneficial.

## MONITORING

Scoliosis in infants with PWS is unlikely to develop prior to the influence of gravity across the spine. Therefore, monitoring of spine should begin when the child first sits independently, usually around the first birthday. Yearly screening radiographs of the seated or standing spine should be used in addition to a clinic examination. If a deviation greater than 10° from straight is noted, radiographs should be obtained more frequently, depending on the age of the child, size of the curve, and apparent risk towards progression.

## CASTING

If scoliosis is detected in the infant, spinal casting has been shown to be effected before 3 years of age. Curves over 20° should undergo casting with the Mehta technique, with a goal of decreasing the curves to as close to 0° as possible. Even curves over 90° can obtain some correction with casting, and should be attempted in this age range. In general, a child's third birthday is often seen as the upper limit to starting casting, but in practice casting has been initiated even up to the fourth birthday. The casts are changed every 2 months in children under the 2 years of age, and every 3 months in children over 2 years of age. Casts are continued until the curve has been successfully reduced, or if correction plateaus over successive cast. The child is then braced to maintain the size of the curve.

## BRACING

Brace treatment should be considered for curves over 20° in children under 10 years of age, and for curve over 25° in children 10 years or older. The brace, a thoracic-lumbar-sacral orthosis or TLSO, should be worn 22 hours per day,

allowing an extra hour or two out-of-brace for physical activities. When the brace is prescribed, initial radiographs in brace should be obtained to verify moderate correction, as compared to the out-of-brace radiograph. Smaller spine curves in children with Prader-Willi syndrome are often flexible, although it will not be possible to obtain a 50% correction of all curves. Follow-up out-of-brace radiographs are obtained every 4 to 6 months, having the child remain out of brace over night, up until the time of the radiograph.

## SURGICAL INTERVENTION IN THE GROWING CHILD

In growing children (usually up to 10 years of age) with progressive scoliosis that cannot be maintained under the 50°, a surgical intervention is needed. Performing a definitive spinal fusion at this age may improve the child's deformity, but will restrict spine and chest growth. At maturity, the patient will be adult sized, but with a child sized chest. Therefore, implantation of an expandable device is likely required. (In practice, once I have identified a curve that will require surgery, I try to postpone intervention until the curve cannot be maintained below 50° in an in-brace radiograph.) The goal of this type of surgery is to decrease the curve's size initially, then prevent it from progressing while allowing for spinal growth. There are two kinds of expandable implants, one is the "growth rods" or "nonfusion spinal instrumentation" (NFSI), the other being the "vertical expandable prosthetic titanium rib" (VEPTR) device. The VEPTR device has proven to be problematic in children with PWS, primarily due to the low bone mineral density leading to frequent failure from rib fractures. For that reason, we recommend the use of a NFSI. For PWS, the construct that has worked well is a two segment fusion cranially, and a two segment fusion caudally, each with bilateral pedicle screws, for four pedicle screws anchor above and below. The segments are spanned with dual rods. A typical construct would be from T3 to L4, fusing T3 and T4 to act as the upper anchor, and L3 and L4 as the lower anchor (see figure). A characteristic of PWS is a cervical-thoracic junction kyphosis.

Therefore the proximal extent of the fixation should be as low as possible (T3 or T4), and a moderate amount of existing thoracic kyphosis can be accepted. Over-correction of thoracic kyphosis appears to drive the cervical-thoracic junction kyphosis towards progression.

The construct is lengthened every 6 months, to keep up with spinal growth. Near skeletal maturity (as determined by bone age), the construct will need to be converted to a definitive fusion.

## DEFINITIVE SPINAL FUSION

In the older child, a definite fusion should be performed for curves exceeding 50° out of brace. A curve of this size has a 95% chance of progressing, even after skeletal maturity. Whereas in idiopathic scoliosis the lower age threshold for a definitive procedure is 10-12 years of age, children with PWS often have a later growth spurt, with delayed maturation, possibly related to the use of supplemental growth hormone. For that reason, it is advantageous to wait until 12 years of age for girls and 14 years for boys prior to fusion. Bone mineral density in children with PWS is frequently low, so multi-segmental pedicle screw constructs are recommended, maximizing the number of fixation points. The scoliosis is usually a kypho-scoliosis rather than the lordo-scoliosis seen in idiopathic scoliosis, but care should be taken not to over correct the kyphotic deformity, as this may secondarily exacerbate the cervical-thoracic junctional kyphosis. Also, as with the expandable implants, consideration should be given to keeping the upper level of the fusion no higher than T3 or T4, if possible, to prevent proximal junctional kyphosis from developing.

## SURGICAL AND

## ANAESTHETIC

## CONSIDERATIONS IN PWS

In children requiring anesthesia, for spinal casting, hardware implantation, or even planned interval lengthens of a NFSI, the special characteristics of children with PWS should be well understood by the entire treatment team.

### Respiratory

The children have a number of

respiratory issues, related to their hypotonia, as well as their obstructive and/or central sleep apnea. They are at high risk for postoperative pneumonia. The literature reflects a high rate of complications with anterior spinal procedures, and it is strongly recommended to avoid entering the chest. Preoperatively, a pulmonary function test is required, as is a sleep study in many cases. In cases of obstructive apnea, a tonsillectomy may be required preoperatively.

Postoperatively, extubation may need to be delayed for a few hours, or even over night, until a patient has sufficiently awoken for a strong respiratory effort. Thereafter, the patient may require CPAP or BiPAP when sleeping, possibly for a few nights to a few weeks. Their hypotonia causes them to have a weak cough, and chest physical therapy should be aggressive.

### **Bone Mineral Density**

Children with PWS have decreased bone mineral, which may lead to loss of hardware fixation and/or pseudoarthrosis. Pre-operatively, it is important to have vitamin D and calcium levels optimized. Children with PWS who have been actively managed by their endocrinologist will likely have been on long term growth hormone and possibly sex steroid replacement. This treatment can optimize bone strength by puberty, but only if vitamin D and calcium levels have been sufficient. It is a good idea for patients to supplement their vitamin D and calcium intake, especially if surgery is being considered. Although radiographs

notoriously underestimate bone mineral loss, a sure sign of an abnormality is if the vertebral density is nearly that of the soft tissue shadows, seen best at areas of overlapping bowel gas. Another warning sign is a diet low in dairy products (milk, yogurt, cheese). A DEXA scan may be helpful if 1) there is concern that the bone mineral density is critically low, and 2) there is enough time prior to surgery for the treatments to have an effect. In those critical cases, intravenous pamidronate and vitamin D may make a meaningful difference.

Operative planning should maximize the number of spinal fixation points for performing the instrumentation for fusion. A multisegmental pedicle screw construct, with fixation at nearly every level has worked well.

### **Pain Tolerance**

Children with PWS have an increased pain tolerance, which may be helpful when attempting to mobilize them after the surgery. It could also be a reason why they awake slower from anesthesia.

### **Food Seeking and Gastrointestinal Issues**

The family and hospital staff needs to be vigilant for food seeking behavior, and its possible life threatening consequences. Preoperatively, the NPO interval may be violated, making induction of anesthesia dangerous. Postoperatively, the children should not be fed until they have normally active bowel sounds and flatus. Usually, they are hungry much earlier than other post-operative patients, but their ileus resolves a day or so later than expected.

Also, the patient's preoperative calorie restrictions should be well known by the hospital's nutrition staff, and observed postoperatively. All treating staff should be aware that children with PWS do not vomit, nor lose appetite. Should either of those occur, they require an emergent nasogastric tube insertion, followed by an abdominal radiograph to evaluate for gastric dilatation, and possibly a CT scan to rule out free air. Gastroparesis leading to gastric necrosis is unfortunately a common cause of death in children with PWS.

### **Skin Picking**

A constant threat to the surgical incision is the habit of patients with PWS to skin pick, in essence scratching their wounds until they break down, leading to a dehiscence and a deep infection. This is probably the most common post surgical PWS complication seen. The skin picking may take the form of rubbing their back against a wall or furniture. In many cases, we have used a light post-operative brace to protect the skin for the first several weeks.

If the above points are carefully observed, surgery can be performed safely and uneventfully in children with PWS.

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