



# Annual report 2015

INTERNATIONAL PRADER-WILLI SYNDROME ORGANISATION



Celebrating 25 years of service around the world



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# IPWSO Boards

## IPWSO PRESIDENT

**Suzanne Cassidy, MD**  
Clinical Professor of Pediatrics  
University of California, San Francisco, USA

## VICE PRESIDENT

**Janalee Heinemann, MSW**  
PWSA (USA) Coordinator of Research & International Affairs

## TREASURER

**Don Tallon, Ireland**

## SECRETARY

**Marguerite Hughes, Ireland**

## BOARD MEMBERS

**Georgina Loughnan, Australia**  
**Jackie Waters, UK**  
**Hubert Soyer, PhD, Germany**  
**June-Anne Gold, MD, USA**  
**Mariona Nadal, Spain**

## HONORARY PRESIDENT

**Jean Phillips-Martinsson, UK**

## LEGAL ADVISER

**David Gordon, USA**

## CLINICAL & SCIENTIFIC ADVISORY BOARD

### CHAIR

**Anthony J. Holland, CBE. BSc**  
The Health Foundation Chair in Learning Disabilities,  
Cambridge Intellectual and Developmental Disabilities  
Research Group, Department of Psychiatry,  
University of Cambridge UK

### VICE CHAIR

**Susanne Blichfeldt, MD**  
Consultant (Neuropediatrics) Herlev University Hospital 2730  
Herlev Denmark

**Leopoold Curfs, PhD ,**  
Professor  
Dept. of Clinical Genetics University Maastricht/Academic  
Hospital Maastricht, The Netherlands

**Dan Driscoll, PhD, MD ,**  
Professor  
University of Florida Health Sciences Center Div. Genetics /  
Dept. of Pediatrics Gainesville, FL U.S.A.

**Moris A. Angulo, MD**  
Director of Medical Genetics Assistant Director of Pediatric  
Endocrinology  
Winthrop University Hospital Mineola, NY U.S.A.



**Janice L. Forster, MD**  
Child & Adolescent Psychiatrist Pittsburgh Partnership  
Pittsburgh, PA U.S.A.

**Urs Eiholzer, MD**  
Professor and Head PEZZ Center for Pediatric Endocrinology  
Möhrlistrasse 69 CH-8006 Zürich Switzerland

**Shuan-Pei Lin, MD**  
Professor and Director Division of Genetics and Metabolism  
Department of Pediatrics MacKay Memorial Hospital  
Taipei, Taiwan

**Ann O. Scheimann MD, MBA**  
Assistant Professor of Pediatrics Johns Hopkins School of  
Medicine Baltimore, USA

**Charlotte Hoybye, MD**  
Senior Consultant, Associate  
Professor, Department of  
Endocrinology, Metabolism and  
Diabetology, Karolinska  
University Hospital, Stockholm,  
Sweden



# IPWSO Boards, contd

## PROFESSIONAL PROVIDER CAREGIVER BOARD



### CHAIR

**Norbert Hödebeck-  
Stuntebeck PhD**

Psychologist Diakonische  
Stiftung Wittekindshof  
Germany

### CO-CHAIR

**Mary K Ziccardi**

Regional Director REM  
Ohio USA

**Hubert Soyer PhD**

Psychologist Director,  
Regens Wagner Absberg  
Germany

**Larry Genstil PhD**

Psychologist Executive  
Director Genstil Institute of  
Human Behaviour  
Israel

**John Ford PhD**

Psychologist Specialising in  
behavioural care  
New Zealand

**Fanny Cortes MD**

Clinical Geneticist Rare  
Diseases Center. Clinica Las  
Con-des Santiago, Chile

**Jackie Mallow**

Director of Admissions &  
Consultative Services at  
Prader-Willi Homes of  
Oconomowoc, USA

**Lisa Boman BA**

Director Counselor & Social  
Worker  
PWS Group Home Attendo  
Stockholm  
Sweden

## FAMCARE COMMITTEE



Georgina Loughnan, Clinician, Australia

Kate Beaver, Crisis Counsellor, USA

Damien Jones, Caregiver Manager, Australia

Rika du Plooy, parent, South Africa

Lesley Robertson, parent, Australia

Linda Thornton, parent, New Zealand

Tiina Silvast, parent, Finland

Cindy Adams-Vining, parent, New Zealand

Michelle Andrea, parent, USA

Fanny Cortes, geneticist, Chile

Sabeen Iqbal, parent, USA

## OFFICE STAFF

Giorgio Fornasier, Italy, Director of Programmes

Linda Thornton, QSM, New Zealand, Communications  
Coordinator

## FUNDRAISING COMMITTEE

Chair: Jackie Waters, UK

Hubert Soyer ,  
Germany

Marguerite Hughes, Ireland

Linda Thornton, NZ

Joan Gardner, USA

Janalee Heinemann , USA

## AUDITORS

Verena Gutmann , Austria

Gerard Meijwaad , The Netherlands





# President's Report

## Suzanne Cassidy, USA

These will be my final comments in an Annual Report during my two terms as President of IPWSO. As I mentioned in my opening comments from the first Annual Report of my presidency six years ago, I am still continually impressed with the generosity and energy of our IPWSO community, the members of the Board of Directors, Advisory Boards, Committees, staff, country delegates, and numerous others, almost all of whom are volunteers.

The 2015 year has been a particularly impressive one in several ways that are outlined in some detail by others in the reports that follow.

We have made great strides in the types, breadth and amount of educational and support activities.

We have developed new materials, translated articles and brochures into more languages, reached out to more families of people with PWS and their care providers in more countries, and dealt one-on-one with more people needing help and support than ever before.

We have hosted exhibit booths at medical conferences, provided diagnostic testing for PWS where it was not available, and connected professionals caring for people with PWS with experts.

We have provided professional delegates with publications in medical journals selected by our Clinical and Scientific Advisory Board in



order to improve knowledge of developments in caring for their patients with PWS.

We have developed and made available to families and providers alike videos of presentations by experts on various important topics concerning the care of people with PWS (thanks to a grant from PWSA Ireland).

We have reached out to countries that are beginning the process of developing an Association with information and expertise. And much, much more.

Many of these activities cost money, something that IPWSO has had little of in recent years. IPWSO is in part supported by membership dues paid by the country Associations (33 Associations pay IPWSO dues and are thus voting members of IPWSO). For a few years, IPWSO also received sponsorship funding from the first pharmaceutical company that produced growth hormone for PWS,

# President's Report contd

Pfizer, but a change in the way that pharmaceutical companies can support outside organizations resulted in a few years without IPWSO receiving that funding. Those were slim years for IPWSO. But 2015 was different.

In 2015, Pfizer once again provided us some sponsorship funding. Also in 2015, our Fundraising Committee became very active and several successful fundraising campaigns were launched. In addition, thanks to the efforts of a member of our Fundraising Committee, Joan Gardner, IPWSO obtained a grant from a foundation in the USA (the K.B. Andersen Foundation) that provided equal ("matching") money for each donation from a new donor and any increased donation from a former donor, a total of \$15,000 US. And through the generosity of people and organizations around the world, we were able to earn all \$15,000!

Due to these factors, in addition to assuring support for our ongoing activities, we were able to obtain professional translation of several of our materials into languages for which we previously did not have any

materials. And we added a new program to provide travel grants for people to attend IPWSO's 2016 International Conference.

We also plan for grants to help support country-wide conferences where language is a barrier to benefitting from the international conference.

I cannot emphasize strongly enough the importance of donating to IPWSO to help it continue its educational and support activities. Even a very small donation can make a big difference! Please consider showing your appreciation and support of IPWSO by donating. Giorgio Fornasier explains how in his report.

Since these are my last comments in an Annual Report as President, I want to give particular thanks to a few of the people who have worked so hard to achieve IPWSO's mission during the past 6 years and been a real pleasure to work with. First, I need to thank Janalee Heinemann, a remarkable and multi-talented woman who has been my advisor at the same time that she has provided so much to IPWSO as its Vice-President. I cannot imagine someone who has worked harder for the benefit of people and families affected by PWS. Her support and friendship have been invaluable.

Linda Thornton, IPWSO Communications Coordinator, has been so important in spreading information about events, distributing educational materials and reports, and supporting the Board and, most importantly, people who reach out to IPWSO for assistance. She has brought our website, social media, and newsletters to a higher



Suzanne Cassidy with Janalee Heinemann and others at the IPWSO conference in Cambridge

# President's Report contd

level than ever before.

Giorgio Fornasier, our Director of Program Development, remains the person who can communicate in almost every language, who knows best the needs in countries around the world, and who keeps track of IPWSO's finances. Through donations following his singing concerts, Giorgio is the most generous of all donors around the world, and his financial support has kept IPWSO going under many difficult circumstances.

Marguerite Hughes, IPWSO Board member and Secretary, has provided assistance well beyond what a Board Secretary typically does. Her intelligence, organizational ability, and insight have been indispensable.

Jackie Waters, IPWSO Board Member, has served as head of the Fundraising Committee and as liaison from the Board to the Canadian organizers of the 2016 IPWSO Conference, working hard and effectively in both positions.

And there are many others who deserve, and will receive individually, my gratitude for their devotion and efforts. Much as I would like to thank them here, space does not allow me to.

This coming year is the 25<sup>th</sup> anniversary of the establishment of IPWSO, and the Board feels very proud of its accomplishments thus far. We are also enthusiastically anticipating many further great advances in achieving our mission to improve the quality of life for people with PWS all over the world. There are many countries that still know little about PWS and offer little support to affected individuals and their families. But each year IPWSO has contributed to progress and enlightenment. We want to continue to further this progress in every possible way.

I sincerely hope to see many of our delegates and others in Toronto at the 9<sup>th</sup> IPWSO International Conference, July 20-24, 2016. It has been an enormous honor and pleasure to have served as your President for the past 6 years. Thank you for this great opportunity.

## FUNDRAISING REPORT

**Our Fundraising Committee worked hard over this year to raise a total of just over \$30,000.**

This was helped by a fantastic grant from the Kathrine B Anderson Trust of the St Paul Foundation in USA who promised a dollar-for-dollar match up to \$15,000. Thanks to some very generous donors and members of our own Board, we achieved this through a campaign set up to represent the 103 member countries of IPWSO, "**103 Hours for 103 Countries**". This was monitored via social media and we successfully raised \$15,000 over a period of 103 hours.

Next year (2016) we may have the same opportunity of matching funds from the KBA Trust, but this time the funds must come from new donors, or be from further 'new' money from previous donors. This will be a challenge for IPWSO, but we are ready to face it.

Thanks to our committee: Suzanne Cassidy, Joan Gardner, Marguerite Hughes, Jackie Waters, Linda Thornton and a very special thanks to Giorgio Fornasier for his ongoing generosity and support through his concerts.



# Vice President's Report

## Janalee Heinemann, USA

IPWSO's medical support in 2015 increased in several areas. The PWS Medical Alert booklet is now in 17 languages, and the Prader-Willi syndrome Android app provides comprehensive medical information wherever you are in both English and Spanish. Other medical support provided by IPWSO include:

- The number of articles on the website translated in various languages has increased.
- The IPWSO blog, Facebook, and medical articles are sent to the professional delegates in over 100 countries
- Individual email requests for help from countries all over the world are responded to daily.
- More medical articles in layperson's language for families have been made available.
- Educational literature is distributed to hundreds of physicians and researchers at medical conferences,
- Support of educational conferences in less developed countries.
- Free diagnostic testing at the BIRD laboratory in Italy

### Creating Awareness, Education and Support

#### ESPE 2015 in Barcelona Spain:

Thanks to a generous sponsorship we received from Pfizer Europe of an unrestricted educational grant, IPWSO had an awareness and



educational booth at ESPE (European Society for Paediatric Endocrinology) in October 2015. It was encouraging to see the increase in interest in PWS by endocrinologists who attended from around the world. All 460 flash drives with medical and other PWS educational materials in multiple languages were distributed to endocrinologists from 41 countries.

As we have seen in the past, physicians from many countries were also very grateful to learn about our free diagnosis for countries that do not have the option of DNA methylation testing. IPWSO provides this service in collaboration with the Baschirotto Institute for Rare Disorders (BIRD). Some new countries that were especially appreciative of learning about this option were Vietnam, Bahrain, and South Korea. Countries that utilized the free testing services in 2015 include Lithuania, Pakistan, Algeria, Peru, India, Nicaragua, and Ungheria/Hungary.

Also at ESPE, Dr Maité Tauber, one of our PWS experts from France gave two presentations on



# Vice President's Report contd

the syndrome, and there were 17 posters presented on PWS. There was also a lot of interest shown about PWS from several pharmaceutical companies who are doing clinical trials, or are interested in doing clinical trials.

I am a professional, but also the parent of an adult with PWS, so my commitment is understandable. What is most impressive to me, is the many medical professionals who are so committed to PWS that do not have a family member with the syndrome. We have been blessed with their love for our children and dedication.

The professional that stands out the most to me regarding her dedication to PWS, and especially to IPWSO, is our IPWSO president, Dr Suzanne Cassidy, who will be completing her term limit in

July 2016. I have worked hand-in-hand with Dr Cassidy over the last 6 years for IPWSO and, probably more than anyone else, can attest to her incredible dedication to this international cause. She has worked tirelessly on almost a daily basis as our volunteer president. As most people reading this will know, Dr Cassidy was highly acclaimed for her work with Prader-Willi syndrome long before she took over the role as IPWSO president. She has been involved with PWS since the early 1980s. Dr Cassidy was also the organizer of the NATO Advanced Research Workshop on Prader-Willi Syndrome, The Netherlands, May 1991. This was the first meeting of the International Prader-Willi Syndrome Organisation (IPWSO). In fact, beyond PWS, she is recognized as one of the top geneticists in the USA, and won the Recipient of Excellence in Human Genetics Education Award from the American Society of Human Genetics, October, 2014. It has been my honor to work with such an amazing person on such a worthwhile cause.

Another special recognition goes to Dr Moris Angulo from New York, who is on the IPWSO Clinical and Scientific Advisory Board. Dr Angulo is dedicated to helping educate families and professionals dealing with PWS in Spanish-speaking countries – with no expectation of being reimbursed for his time or travel expenses. He has never forgotten what it was like to be a poor little boy on the streets of El Salvador. His love for our families shines through like a beacon of light to our families in less developed countries.

As my term on IPWSO's board is also finishing this year, may I take this opportunity of thanking all the wonderful people I have worked with over the past 6 years.



Dr Moris Angulo and child

# Treasurer's Report

## Don Tallon, Ireland

I am pleased to present this report in respect of the year ending December 31<sup>st</sup> 2015. I wish to thank my fellow board members and particularly Giorgio Fornasier and Marguerite Hughes for the timely and accurate information that they provide me with in relation to details of income and expenditure together with clear minutes of our board meetings.

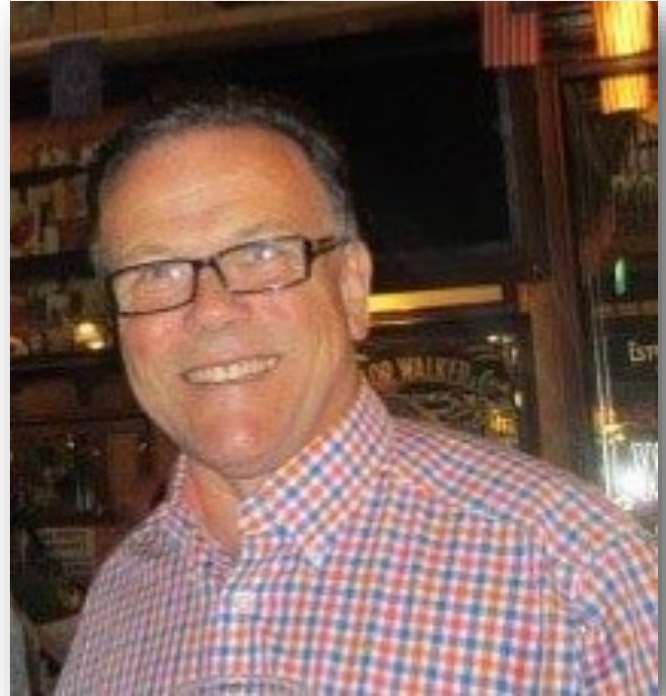
### Developments during 2015

Our combined fund raising efforts during the period have resulted in our raising \$99,095 which is an excellent result. We are grateful to the members of our Fundraising Committee Linda Thornton, Jackie Waters, Marguerite Hughes, Joan Gardner, Susie Cassidy and Hubert Soyer for their hard work and effort to achieve such a positive result. Thanks also to our major sponsors, Global Pfizer, The KB Andersen Foundation, Joan Gardner and to those members of our board who have continued to support us with donations during 2015. Giorgio continues to support our fundraising efforts with his regular concert donations.

We enter 2016 with optimism and we are looking forward to our upcoming 9<sup>th</sup> IPWSO International Conference, "Building Global Community", in Toronto from July 20<sup>th</sup> to July 24<sup>th</sup> hosted by FPWR Canada.

### Financial Position

As treasurer, I receive regular monthly financial statements from our Director of Programmes. I note the amounts of income and expenditure



and check these against original monthly bank statements which are posted to me by the bank. I check that all the expenditure is in accordance with the guidelines laid down by the board of IPWSO.

In 2015 we were very fortunate to receive two one-time donations, one from an anonymous gift of \$ 18,500 and the other, a \$15,500 donation matching grant from the KB Anderson Foundation. Therefore our budget for 2015 shows an income for the year of \$99,075 against an expenditure of \$45,232, leaving us with \$53,843 to devote to important initiatives which we have been unable to fund in the past, including translation of several of our materials and conference grants for those requiring financial assistance to attend.

We are very grateful for those exceptional donations.

# Audited Financial Statement

## IPWSO Financial Summary at 31/12/2015.

### Audited Financial Statement

		<b><u>Expenditure Detail 2015:</u></b>
US \$ Account:	\$68,411.00	Staff Salaries: \$21,302
€ Account:	€48,894.33	Office running: \$ 1,205
Petty Cash:	€ 749.36	Phone App: \$ 6,517
Paypal:	€ 100.82	Printing \$ 1,010
Total estimated \$122,624. (Exchange Rate €=\$1.0898)		BIRD Lab \$ 1,848
		ESPE \$ 8,074
		Education Con \$ 1,100
		PPCB Fund \$ 2,986
		Bank Charges \$ 1,120
		<b>Total: \$ 45,162</b>
<b><u>Income Detail 2015:</u></b>		
Membership Fees:	\$7,145	
Pfizer ESPE Grant:	\$10,900	
Fundraising:	\$81,050	
<b>Total:</b>	<b>\$99,095</b>	

## AUDITORS' NOTES

The Auditors have reviewed the ongoing monthly financial statements and the above Treasurers' report and have concluded that these correctly represent the IPWSO financial position as of 31st December 2015.

*Verena Gutmann*  
*Gerard Meijwaard*

# Secretary's Report

## Marguerite Hughes, Ireland

For me 2015 stands out as a year in which IPWSO focused particular attention not alone on disseminating existing resources and generating new resources, but also on ensuring that as much information as possible was made accessible to as many people as possible. This is no small task given the 104 countries that make up IPWSO's membership and the many languages spoken in these countries. In furtherance of this objective IPWSO significantly increased its budget for translating materials during 2015. It also promoted and further developed Prader-Willi World, a free Android app designed to enable users to have information about PWS always at their fingertips and to enable them to connect with other families worldwide.

In addition to the core secretarial duties that the role bestows, the IPWSO Secretary has the privilege of being involved in a wide array of IPWSO's day to day activities. One activity I took



particular pleasure in involved the coordination of two PWS training films, which serve as a good example of international cooperation at work. Not alone did two professionals volunteer to present the films, but the Prader-Willi Syndrome Association Ireland and the Prader-Willi Syndrome Association (USA) collaborated with IPWSO in their production. These films are now freely available to organisations and individuals around the world.

As with every organisation, the successful operation of IPWSO requires ongoing administration by its staff and board. During 2015 the monthly board meetings, ongoing projects, subcommittee activities and regular email communications continued apace. During 2015 too IPWSO dedicated time to planning for the future and, in particular, to investigating options for pursuing formal registration as a charity or foundation.

I remain in awe of the spirit of cooperation that both drives IPWSO's work and is fostered by it. It is both a pleasure and a privilege to serve on its board.



*All videos can be seen on YouTube*



# Clinical & Scientific Advisory Board



## Anthony J. Holland

### Membership of CSAB

The membership of the Clinical and Scientific Advisory Board is set out below.

Tony Holland (Chair)  
Susanne Blichfeldt (Vice Chair)  
Urs Eiholzer  
Moris Angulo  
Shuan-Pei Lin  
Leopold Curfs  
Dan Driscoll  
Janice Forster  
Ann Scheimann  
Charlotte Höybye

### Scientific papers

The CSAB has continued to prepare PDFs listing scientific papers published over the previous three month period in peer review journals. These lists have been circulated widely by Linda Thornton and also circulated at national level. The range of research being published is extensive include basic science and a broad range of clinical topics, for example, from genetics, to endocrinology, and behaviour and mental health. It is difficult to pick out and single area but there are papers reporting on trials of specific interventions and also the application of some of the advanced neuroimaging techniques. Despite a recent setback I would anticipate further papers reporting on trials focussing on the early PWS phenotype, the hyperphagia, and the social and maladaptive behaviours. Many thanks to Joyce Whittington and Suzie Tall for their help in preparing this material.



### Advice

During this past year members of the CSAB who have responded to requests for advice on the following areas:

- Mental health and behaviour including autism and pathological demand avoidance
- Uncertain diagnoses
- Genetics (including a complex example of mosaicism)
- The use of growth hormone and whether or not it is free in different countries
- Vitamin D deficiency
- Low carbohydrate diet
- Bariatric surgery
- Use of medication for temper outbursts and side-effects
- Establishment of a genetic testing service.

The request come from a range of countries across the world, for example, China and various South American, and European Countries. Thank you to the many members of the Board who have responded to these requests. Linda keeps a record of the replies for future reference.

### Other matters

A meeting of the CSAB is planned in Toronto in July 2016. At this meeting it is proposed that the Board considers the nature and extent of its remit and particularly whether there is a need for supporting research at an international level and for preparing advisory papers.

# Professional Providers Caregivers' Board



## Norbert Hödebeck-Stuntebeck

To get the email that read “Norbert, it is time for the annual report about the work and activities of PPCB in 2015” triggered different feelings and thoughts like, “oh yes, I am late with this ...” and “when I can find time to do this?” But when the first words were written, more and more of the things that we accomplished in the past year came to my mind. Following that, more feelings surfaced that reminded me that IPWSO, with the PPCB as a part of it, can be very pleased about all the activities we have supported and outcomes we have achieved. We say this with a sense of accomplishment and pride, also recognizing that there are still many big steps to take in the future.

### 2015

It is an understatement to say that the “only” one big thing the PPCB did in 2015 was the International Caregivers' Conference in November 2015 in Orlando, Florida, USA. Many countless hours of planning, skype and telephone meetings were necessary for the PPCB to structure and prepare for this conference. We benefited from our work and experiences from the conferences in all of the years since we began the PPCB in 2010 in Taiwan. The experiences and knowledge developed in past years helped us to make the conference in 2015 a most successful meeting for the nearly 60 participating professional caregivers.

Most of the attendees came from the US, and some were also from Europe, Russia, South America, New Zealand and Australia. The main part



of the one day Caregiver Conference, which was scheduled one day in advance of the national PWS conference of PWSA (USA), was organized by the PPCB Co-chair Mary K. Ziccardi. Another member of the PPCB, Jackie Mallow, served as the overall PWSA (USA) conference Co-chair. We are very thankful to both of them for their exceptional engagement for this wonderful event! The “Pam Eisen Memorial Lecture” was given this year by Janalee Heinemann and focused on medical information. This was followed by an overview of current research activities in PWS, presented by Merlin Butler, MD. Both set the stage for the high quality day that the participants were going to experience.

However, the main focus of the day was on the workshops where the participants had the chance to not only be informed but also to be involved in the themes and in the development of new ways of caring for people with PWS.

Behavior, communication, sensory integration, social competencies, Tomatis Therapy, supporting and training staff, aging and PWS

# PPCB REPORT contd

basics were the topics of these workshops. Some of this topics will be featured in a second book written by the members of the PPCB. It is our goal that this publication will be finished and available at the next IPWSO conference in July 2016 in Toronto, Canada. The book will be titled "Trainings".

Another very important realization of the PPCB members has been the acknowledgment that we need to share and exchange information between countries. Members of the PPCB may accomplish this, in part, through traveling to other countries or inviting experts to their country to learn from each other and develop and share new ideas and improved ways of caring for people with PWS. We have done this collaboratively with Macedonia, Norway,

Sweden, Australia, Austria, Switzerland, Denmark, and have plans to do it with the UK and other countries in the future.

So you must understand that completing this "annual report" on behalf of the PPCB gives me a sense of pride and accomplishment for what was completed in the past year. More importantly, it empowers the PPCB to plan ahead and continue to do what is necessary in the future to support everyone that is caring for someone with Prader-Willi syndrome. For all that the PPCB members have accomplished and for the anticipated success of their future activities, I am very thankful and looking forward to a productive future!

See you in Toronto!





# Famcare



## Georgina Loughnan

2015 has been another productive year for Famcare.

We were delighted to welcome Cindy Adams-Vining, from New Zealand to our committee! Cindy is highly experienced in PWS being the mother of 27 year old Hannah. Cindy is an active advocate and educator for PWS who has presented at several national and international conferences.

Famcare has now produced nine articles with the latest for 2015 being, *Strength in Boundaries* and *PWS Awareness for Professionals*. We have been most fortunate to have had most articles translated in to German, Spanish and Italian and are in the process of having them all translated into Arabic, Russian and Hindi. If you are in a position to assist with the translation of our current or future articles in a language not listed, we would be most grateful to hear from you!



Famcare articles have been written for the families who have adults with PWS living in the family home, but they provide information for caregivers outside the

family home as well as professionals who are working with adults. Much of the

information is also relevant to adolescents and younger people with PWS.

The Famcare committee is made up of an international group of parents and professionals. All written material is reviewed by an international professional advisory committee and then approved by the IPWSO Board of Directors before being accepted for publication. We ask you to share the Famcare articles with as many relevant people as possible. At a conference level they can be printed in a booklet form and distributed to delegates who may or may not have access to the website.

We welcome feedback and suggestions for future articles and services that Famcare could provide or assist with.

We continue to welcome communication via email through the IPWSO website or via our Famcare email: [info.IPWSO@gmail.com](mailto:info.IPWSO@gmail.com)

I look forward to an increasing parent-to-Famcare communication in the future.





# Programme Director's Report

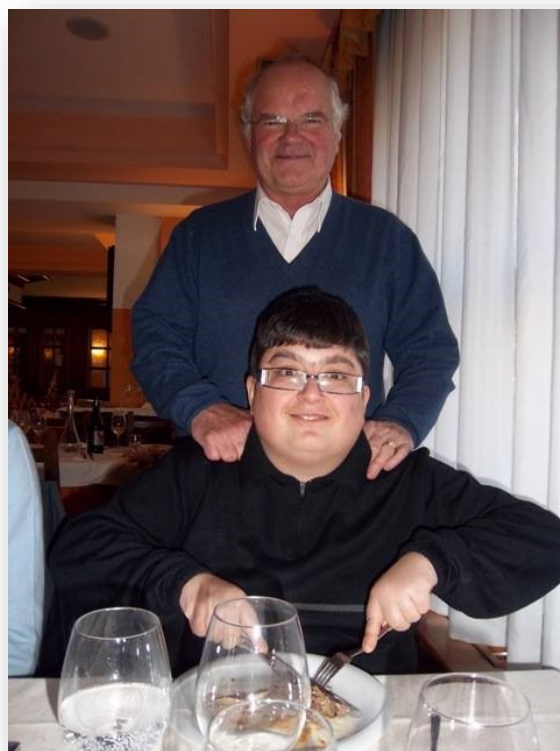
## Giorgio Fornasier, Italy

2015 has been a good year for IPWSO because we received an unexpected and generous grant from Pfizer, as well as very good private donations from Switzerland, Ireland and especially from USA. Moreover the funds I keep raising with my concerts have been reasonably good. Anyhow we must count on our own international family donations, but I regret to note that major donors are mainly among Board members and officers who promote such a fundraising activity.

As you can see on the treasurer's report we have the funds available to face the high costs we have for the organization and participation in the 9<sup>th</sup> International PWS Conference in Toronto (Canada) in July 2016. We also need to print updated educational material, and support regional events we hope take place in Northern Africa and Latin America next year.

I think it is important to remind everybody what we are doing:

1. IPWSO is represented by delegates or contact persons in 104 different countries. Only 33 are paying a membership fee every year!
2. The majority of our members do not have a National PWS Association and they get little or no support at all from their Government or National Health Institutions. IPWSO is the only reference and help families and professionals have for PWS in their own countries.
3. We assist anybody who contacts us, whether they are members or not and whether they pay a membership fee or not. When particular or complicated cases are brought to our attention, our Clinical & Scientific Advisory Board with leading international specialists on PWS assist families and professionals at no cost, even if the



Giorgio with his son, Daniele. Italy

person contacting us is from the poorest country in the world.

4. Since 2003 we have offered a free diagnostic service at the Genetic Molecular Laboratory of BIRD in Italy. As far as I know, we are the only organisation in the world offering such an important service that can save lives.

Even though we keep encouraging our members in less developed countries to organise workshops for PWS, to gather professionals and families together, creating awareness and providing key speakers from abroad, this year we had a successful event in Mexico only. We are hoping to be able to do it again in 2016 in other areas such as Africa, Asia and Latin America.

### MEETINGS AND SCIENTIFIC EVENTS

We followed with enthusiasm and interest important PWS meetings and workshops that

# Programme Director's Report contd

took place in Mexico and Cuba. We wish to thank PWSA France very much for their valuable assistance in Northern Africa which will hopefully lead to important workshops on PWS organised in Algeria and Morocco.

## **ESPE 2015**

Thanks to a generous sponsorship we received from Pfizer Europe we could participate at ESPE (European Society for Paediatric Endocrinology) that took place in Barcelonan (Spain) from 1st to 3rd October 2015. Janalee Heinemann, Mariona Nadal from Spain, and I ran the IPWSO booth. We were crazily busy with people stopping by our booth for information on PWS. This year it was much easier to distribute educational and information material because we put all files in different languages inside a flash drive memory stick bearing our logo. This was highly appreciated by the 370 doctors visiting our booth. As it always happens, we had new contacts in countries where we are not represented yet.

Our decision to print Medical Alerts in various languages was a great initiative everybody appreciated. Copies in pdf can be easily sent by email everywhere when needed and they can also help parents going on holidays in different countries. Next ESPE will be held in early September 2016 in Paris (France) and Pfizer Europe already assured their support to enable IPWSO to participate with our educational booth.

## **MEMBER COUNTRIES**

During 2015 we assisted individual families and professionals all over the world with prompt answers to the questions they put and submitting information they were looking for.

## **FREE DIAGNOSTIC SERVICE AT BIRD**

We covered the costs of methylation tests made at the Laboratory at BIRD in Italy for the following countries:

Algeria: 3 tests

Lithuania: 1 test

Pakistan: 1 test

Libya: 1 test

Hungary: 1 test

Peru: 2 tests

India: 1 test

Nicaragua: 1 test

We are following an interesting request that came from a laboratory in India asking for cooperation with BIRD to organize methylation tests according to latest technology by them.

## **FINANCIAL SITUATION**

We do need more help and financial support from our member countries and individuals whom we assist. Any amount donated to IPWSO is welcome and you can transfer money easily to us using a wire transfer to our accounts, sending a cheque to my address, using PayPal or Western Union and also the new Idonate system you can access by visiting our website.



Giorgio, singing to a classroom of children

# Communication Coordinator's Report

## Linda Thornton, New Zealand

When people write in the first instance to IPWSO, their letters land on my desk—or, rather, in my inbox. Thus, I read of families' despair and feelings of helplessness, or questions needing medical or scientific answers, or requests for information, and a desire to build their own knowledge. These come from literally all ends of the world. And living, as I do, in New Zealand I sometimes reach for the Atlas just to check how far these requests have come.

I work from home, so there's no sense of office pressure, and I often correspond with families for months until they feel more confident about living with PWS; many become really good e-friends. I'll probably never meet them, but we are drawn close by our common bond of PWS.

In this year, 2015, I received many questions from parents that required more of a clinical or medical answer. These I passed to the head of our Clinical and Scientific Advisory Board, Professor Tony Holland, who, in turn, sends these onto his board members. Answers come back to me promptly and these are sent back to the parents. This is a service that could well be unique among parent groups and strengthens the relationship between professional and parent. I hope that many more parents take advantage of this rich resource. It's not often you have a team of specialists at your fingertips!

I also keep the website up-to-date, collaborate with Tony Holland to send our quarterly research publications out in our Newsflash, as well as keep our Facebook page and Twitter accounts going and you will see the occasional blog from me (or Marguerite). This last year I have also edited a



new publication, "PWS and the Younger Child" which will be published in early 2016. I help run the fundraising activities and contribute to our Famcare work.

I have worked in the field of PWS for over 25 years, 12 of these having been with IPWSO. Every time there is a change of board members (which occurs every 6 years), I feel a sense of excitement at working with new people, but I strongly feel the loss of saying 'au revoir' to those I have worked with over the years. This year will see another 'changing of the guard' and I want to say how much I have enjoyed working with Susie Cassidy, Janalee Heinemann and Jackie Waters. Their steady help and advice along with our other board members, has helped steer IPWSO through some troubled global financial times and their generosity with time and support has been second-to-none. I can only stand back and admire their complete dedication to us all, wherever in the world we might be.

My job has grown over the years, but still provides me the same continuing interest and desire to know more about the syndrome that has changed the lives of thousands of parents worldwide.

# PRADER-WILLI WORLD APP

## Mariona Nadal, Spain

PWW (Prader-Willi World) is an app for Android devices (both smartphones and tablets) developed by IPWSO. Published at the end of 2014, is now a mature app with near 1,000 installations and more than 700 users, growing by around 5 new users per week.

You can find it on <https://play.google.com/store/apps/details?id=org.ipwso.pww>, or looking for “PWW” on Play Store.

On PWW you can have at hand a lot of reliable information about PWS always available in your pocket, to use at school, at the hospital, at home, whenever and wherever a crisis may occur:

**What is PWS?**,

**Medical Alert** (an important life guard)

**Famcare** (strategies that work for home care),

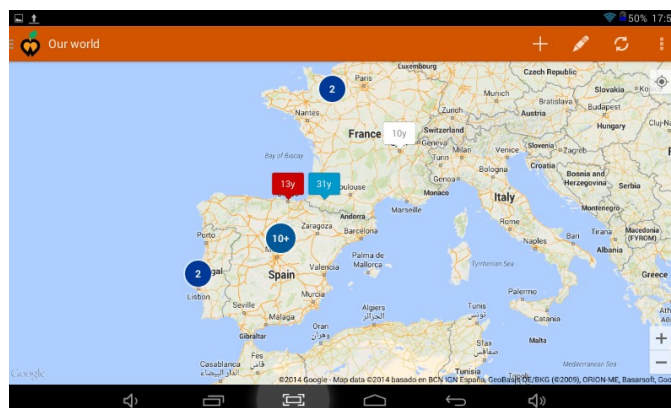
**News** (directly from IPWSO blog) or

**Useful Links** (where to find even more info).

But PWW is more than info. On PWW you can track the growth for the whole family (or just for the person with PW, up to you), on **My Growth** section. Or you can note anything important that happens on your daily life and you want to share with your therapist or doctor later:

**My Notes** is the right place to do so... you can add pictures if necessary!

Whether you have a new diagnosis near you or are a veteran on PWS, PWW helps you not feeling alone...



On **Our World** you can find other families near you, and you can ask them to be in touch with you. How many friendships have grown around PWS!

Another place to find support is on **Contact** where you will have the contact data from IPWSO and IPWSO member entities and delegates on your country.

Helping PWW is helping other families with children with PWS. So your help is not only wanted, but needed.

It's really important to fill the map with a lot more of families, so any new diagnosed family may find someone willing to talk with them in their area. Even if you don't need to be contacted, please, offer yourself so other can contact you if "they need" to, so, add your marker on **Our world**.

Finally, if you like PWW and/or if you like IPWSO work, you can visit the **Magic Area** and make your contribution to help Prader-Willi World and IPWSO to be bigger and stronger and then help better to improve quality of life for people with PWS and their families.



# PRADER-WILLI SYNDROME

## CLINICAL FEATURES

The pattern of characteristics associated with PWS is consistent among those affected by it, although there is significant variability in severity and not all affected individuals have every characteristic. As a simplification, the disorder is marked by distinctly different presentation in infancy versus later in life.

**Early phase:** Initially, the neonate presents with slightly low or normal birth weight and poor suck with subsequent failure to gain weight and grow well, very limp muscles, and delayed developmental milestones. Genitals are often small in both sexes, and one or both testes are undescended in males. Because of the weak, limp muscles and poor suck, infants are almost always unable to nurse and often require special feeding techniques such as special bottles or tube feeding (gavage). The milestones of lifting the head, sitting up, crawling, walking, and speech all tend to be delayed. On average, independent sitting is achieved at around 12-13 months, walking at 24-30 months, and tricycle riding at 4 years. The first word may appear around 21 months and sentences around 3-1/2 years.

**Second phase:** There follows a period of normal eating behavior and improvement in muscle tone and activity. However, this in turn is followed by a phase that is usually marked by an increased appetite and often a compulsion to eat. This often occurs in toddlers or young children. Life becomes dominated by a voracious appetite and drive for food. Individuals with Prader-Willi syndrome apparently do not feel full (lack a sense of satiety). They can eat a tremendous amount of food without becoming nauseated or having indigestion --and many have never vomited. This striving for food combined with a lower than normal calorie requirement and, frequently decreased activity related to the hypotonia will result in rapid weight gain if not controlled externally, leading to obesity and eventually the consequences of obesity: respiratory difficulties, heart disease, diabetes, and other problems.

## CHARACTERISTICS

**Physical characteristics:** There is often a characteristic facial appearance with a narrow forehead, almond-shaped eyes, and a thin upper lip with downturned mouth. Scoliosis (S-shaped curvature of the spine) may develop. Many individuals have fairer coloring of hair, eyes and skin than other family members.

**Growth deficiency:** People with PWS typically have short stature for the family and small hands and feet. Early growth may be below normal, and there is failure to have a pubertal growth spurt. Recent studies have documented insufficiency in growth hormone, and treatment can correct this problem.

**Incomplete Sexual Development:** The small genitalia that characterize affected individuals are more easily identified in males, but affects both sexes. Males often have undescended testicles and a small, poorly delineated scrotum; females have small labia minora and clitoris. Pubertal development is typically delayed and incomplete, and fertility is extremely rare and has occurred only in females.

**Intellectual disability:** Learning and cognitive development are delayed. Although there is wide variability in abilities, the average IQ is in the 60's. Most affected persons function in the mild intellectual disability range independent of tested IQ, and most are learning disabled.

**Speech and language difficulties:** Cognitive capacity and probably also the extent of hypotonia affect this ability. Speech therapy may be helpful.

**Balance and Coordination:** Fine motor skills usually are well developed, while gross motor skills remain limited. High pain threshold and irregularities in body temperature control. Most people with PWS are unaware of injury and infection because of blunted sensory mechanisms. Unexplained high or low temperatures may occur, and there is often insensitivity to environmental temperature.

**Skin Scratching and Picking:** Many individuals with PWS pick and scratch at sores and insect bites which, if not controlled, may become chronic sores and result in infection.

**Behavior and Temperament:** Affected children tend to be loving, happy and compliant in early years, and then subtle changes typically lead to mood swings and behavior difficulties over time. This often includes temper outbursts, stubbornness, rigidity, argumentativeness, and repetitive thoughts and behaviors. Social skills are often impaired. True psychosis occurs in a minority of affected older teenagers and young adults, but it usually responds well to treatment. Depression may result in later years when self-image issues emerge, particularly over the conflict between the drive for independence and the need for management.

**Other Findings:** People with PWS may experience excessive daytime sleepiness and sleep apnea, particularly if obese. Eye abnormalities such as strabismus (cross-eyed), myopia (nearsightedness) or amblyopia (lazy eye) are common.

## TREATMENT

There is currently no cure for Prader-Willi syndrome, nor is there a drug that can be prescribed to alleviate all of the symptoms. As of this writing, there is no known substance that has been demonstrated to lessen the drive to eat, although much research is being conducted to find such a substance. Good and consistent management of entire lifestyle is required for life.