

# ANNUAL REPORT 2018

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INTERNATIONAL  
PRADER-WILLI SYNDROME  
ORGANISATION

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## Mission Statement

IPWSO'S MISSION IS TO IMPROVE THE QUALITY OF LIFE FOR ALL PEOPLE AROUND THE WORLD WITH PRADER-WILLI SYNDROME AND THEIR FAMILIES.



# PRESIDENT'S REPORT

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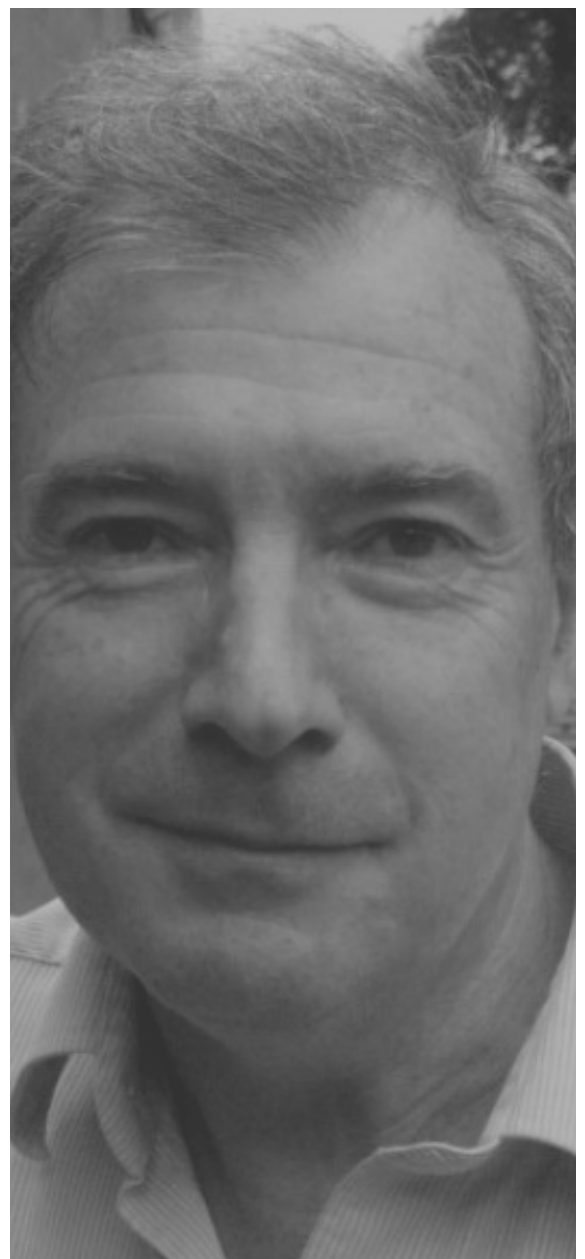
IPWSO has had a very productive year and I am happy to report, is in a good financial position to face the coming year and host our 10th International PWS Conference. Fundraising has been very positive and we have invested in a range of excellent projects.

The broad range of activities undertaken by IPWSO can be summarized under three core themes:

1. supporting people with PWS and their families across the world including funding diagnostic testing and providing information in different ways and in different languages;
2. enabling the development of local expertise by engaging with families, health professions, care providers, and policy makers in different countries through workshops, conferences and by e-mail
3. bringing together individuals and organization, particularly our member organisations, from across the world to share ideas, increase understanding at national and international levels, promote research and increase and disseminate knowledge.

## Tony Holland

PRESIDENT AND CHAIR OF THE BOARD



In this report you will read about different activities in parts of the world as diverse as South America, Europe, and the Asia-Pacific rim. The Board has also continued to focus on how we ensure that IPWSO is fit for the future with robust structures and secure administrative support. I am happy to report that figures for this financial year has been above average and IPWSO is in robust health. The American Friends of IPWSO is now formally established and under Joan Gardner's (IPWSO's Parent Delegate for the USA) and Dr Suzanne Cassidy's (past President of IPWSO) guidance they are raising funds to enable IPWSO to undertake its various tasks. I am very grateful to them, not only because of their financial support, but also because they have shown the way for other countries and we are delighted that Germany also has now established a Friends of IPWSO group. It is clear to the Board that we must diversify our fundraising – the days are gone when we can rely on one substantial grant from a single source.

Two final points, first, IPWSO is a membership organisation of National Associations. The majority of our members pay their dues both large and small, not because of what they will get out of it, but because they understand the importance of helping those who don't have access to the knowledge and expertise that many of us have. My thanks to our membership for your generosity and support and also to individuals and organisations who have contributed financially.

Secondly, don't forget our 10th international conference which is being held in partnership with the Cuban Genetics Society and will take place at the Cojimar Conference Centre, just outside of Havana. The organisation of the conference is well under way. There are and have been particular issues given that the conference is being held in Cuba, but Cuba is an amazing country and the local organisers are waiting to welcome you – do come!

Finally, my grateful thanks to the memberships of all our Boards and to our advisers and supporters who give up their time to help IPWSO.



10<sup>th</sup> International Prader-Willi Syndrome  
Organisation Conference  
13 – 17 November 2019, Havana, Cuba



# TREASURER'S REPORT

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I would like to start the treasurer's report of 2018 with a big thank you to Marguerite Hughes and Giorgio Fornasier who have been instrumental in collecting all relevant data and accurately providing me with all necessary information to calculate the balances and cash flows in our IPWSO accounts. I am very happy to report that the year 2018 has been very successful.

## Developments in 2018

In the year ending in December 2018 we raised a total of \$13,819.94 in our US account and 44,479.22€ in our Euro account. As can be seen in the balance pie charts, the largest part of our income - a total of \$12,012.00 on our US account and 38,738.92€ on the Euro account - was made up of donations. The second largest portion - \$1,168.05 and 5,740.00€ - was generated through membership fees.

Combining income with the expenditures of \$30,480.43 and 26,028.52€ the total comes to a surplus of 18,450.70€ in the Euro account and a negative of \$16,660.48 in



## Hubert Soyer

TREASURER

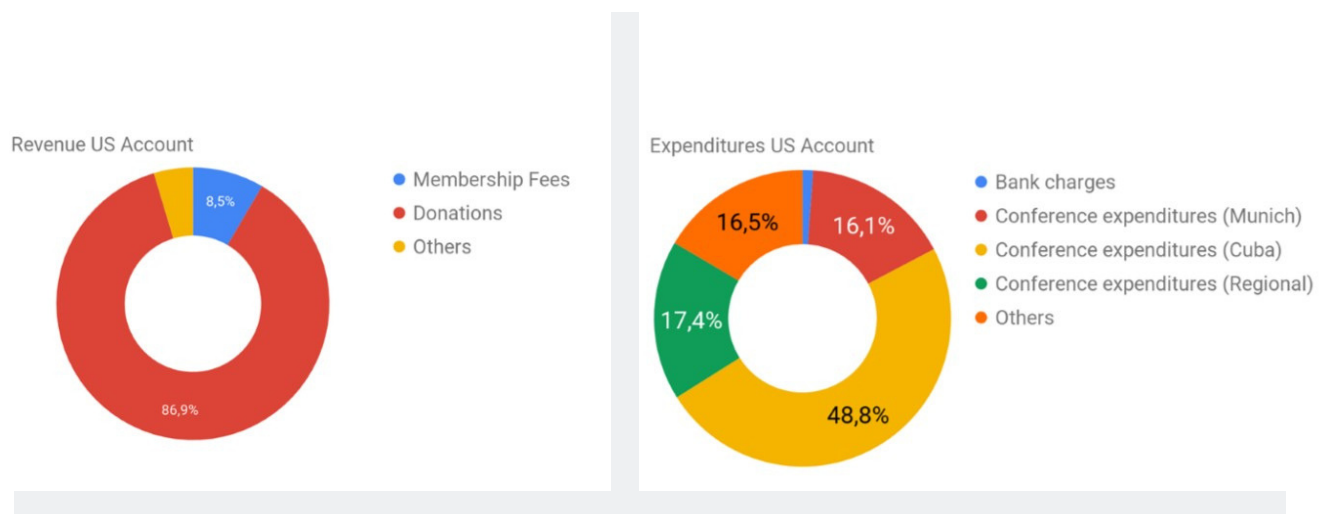
the US account in the year of 2018. Accounting for last year's balance, the US account holds a healthy \$94,178.65 and the Euro account a total of 60,242.80€.

Considering the above figures I am very happy to report that all balances look healthy and we are in a great position to start another successful year 2019. To allow for more long term planning, I encourage further consolidation of our funding.

All of this would not have been possible without the continuous support and help of our members and donors. I would like to address a special thank you to:

PWSA Austria, PWSA Belgium, PWSA Chile, PWSA Denmark, PWSA Germany, PWSA New Zealand, PWSA Norway, AFDVS Mugnai (Italy), PWSA Switzerland, PWSA USA, Sasse Elektronik GmbH (Germany), Millendo Therapeutics, Soleno Therapeutics, Levo Therapeutics, and Friends of IPWSO, USA.

A special thank you goes to Tony Holland and Marguerite Hughes for their constant commitment, their leadership, for helping wherever help is needed and for their endless energy. The tireless efforts of all our contributing members, volunteers and donors have led to stable balances that will help us make the 2019 IPWSO Conference in Cuba a success and I am looking forward to seeing you all there.



## AUDITORS' NOTE

Gerard Meijwaard (The Netherlands)  
Jim Gardner (USA)

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 2018.

# FUNDRAISING

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Fundraising in 2018 focused not alone on meeting IPWSO's budget targets, but also on securing sufficient funding to enable the hosting of the 10th International PWS Conference in Cuba in 2019.

Once again IPWSO's membership remained central to its fundraising efforts. IPWSO is grateful to the 29 member associations that paid subscriptions during 2018 and particularly grateful to the associations in Austria, Belgium, Chile, Germany, New Zealand, Norway, Switzerland, and the USA, which also provided additional donations. While all of IPWSO donations are greatly appreciated, I would like to single out IPWSO's small but dedicated group of individual monthly donors for special praise. Regular monthly giving remains an ideal source of income for IPWSO as it provides a fixed and sustainable income source and IPWSO continues to encourage others to support its work in this way.

In September IPWSO launched its appeal for corporate sponsorship for the 10th International Conference.



**Marguerite Hughes**

VICE PRESIDENT AND CHAIR, FUNDRAISING COMMITTEE

By the end of 2018 over \$70,000 had been committed by a total of 6 sponsors. Two of these sponsors, Millendo Therapeutics (Platinum Sponsor) and Levo Therapeutics (Gold Sponsor), made their conference grants to IPWSO during the 2018 calendar year and funding from the remaining sponsors is due to be transferred to IPWSO in 2019.

For a second consecutive year Soleno Therapeutics was a major donor to IPWSO with a \$15,000 grant made in December 2018 to cover the costs of operating IPWSO's free diagnostic testing program and an awareness booth at the European Society of Paediatric Endocrinology (ESPE) Conference in 2019.

In 2018 IPWSO received its first ever grant from Friends of IPWSO in the USA. This grant enabled IPWSO to host an information booth at the International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASSIDD) conference. IPWSO expects to submit additional funding proposals to Friends of IPWSO for consideration in 2019. 2018 also marked the establishment of Freunde und Förderer der IPWSO e.v., which aims to encourage support for IPWSO in Germany. All of us in IPWSO are very aware of just how much work is involved in establishing new entities such as Friends of IPWSO and Freunde und Förderer der IPWSO and greatly appreciative of the efforts of everyone involved. Thanks in particular to Joan Gardner, Suzanne Cassidy, and Hubert Soyer for leading these initiatives.

As part of a review of fundraising it seems appropriate to acknowledge the outstanding commitment of the many people from around the world who are affiliated with IPWSO. Not only do IPWSO's board members, advisors, CSAB, PPCB and Famcare members freely contribute their time to IPWSO, but in many cases they also pay the costs associated with their IPWSO work. These in-kind donations have included the cost of flights, accommodation and travel expenses. Without the generosity and commitment of IPWSO's many volunteers it could not have succeeded in helping so many people with PWS and their families during 2018.

Finally, I would personally like to thank IPWSO's President, Tony Holland, for his unwavering support of the fund-raising committee and all its efforts during 2018. Thank you also to Amalia Balart, Joan Gardner, June-Anne Gold, James O'Brien, Linda Thornton and Hubert Soyer.

## How you can help us

- **Set up a monthly donation at: [www.ipwso.org](http://www.ipwso.org)**
- **Join our mailing list and keep up-to-date with our work**
- **Follow us on Facebook and Twitter**
- **Volunteer!**

**Go to our website for further information [www.ipwso.org](http://www.ipwso.org)**

# CLINICAL & SCIENTIFIC

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This year, the CSAB has continued to prepare pdf files listing scientific papers published over the previous three month period in peer-reviewed journals. These lists have been circulated widely at the international level. The range of research being published is extensive and includes basic science and a broad range of clinical topics from genetics, to endocrinology, and to behaviour and mental health. There are papers reporting on clinical trials of specific interventions and the application of some of the advanced neuro-imaging techniques. A special note of thanks should go out to Joyce Whittington for her help in preparing this material. These are all available on our website.

The CSAB developed a standardized form this year for consultation queries and during this year members of the CSAB have responded to requests from Bulgaria, UK, Canada, New Zealand.



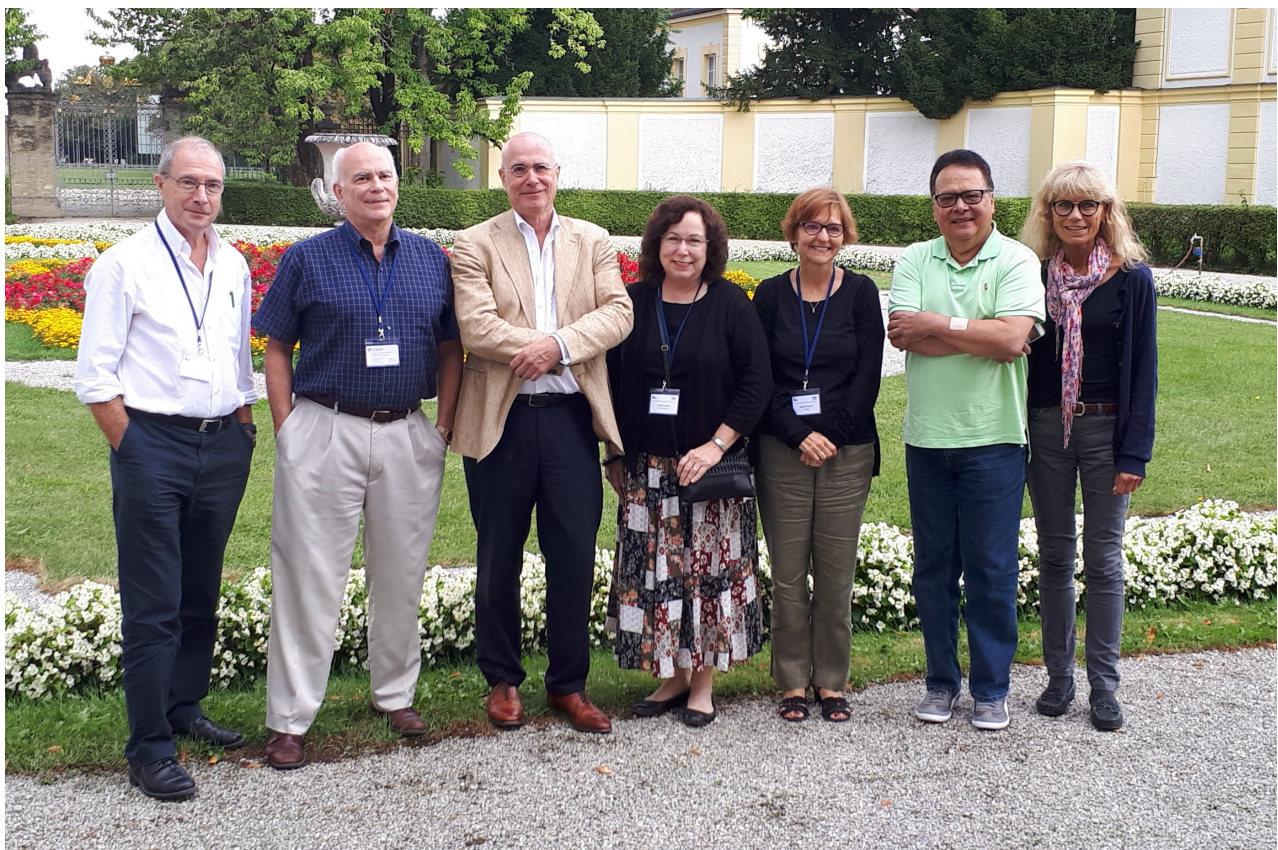
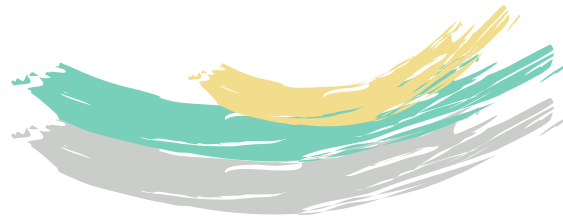
**Dan Driscoll**

CSAB CHAIR



Thank you to the many members of the CSAB who have responded to these requests. IPWSO office keeps a record of these enquiries.

The CSAB met in Munich, Germany in September 2018 to plan the upcoming IPWSO meeting in Cuba in 2019 as well as work on Medical Overviews and Checklists for infants, children and adolescents with PWS similar to what is now available for adults with PWS for families and health care professionals on the IPWSO website. These guidelines are expected to be completed by Spring 2019 and will become available on the IPWSO website.



**Clinical & Scientific Advisory Board present at the  
Caregivers' Conference, Munich**

Left to right: Tony Holland (UK), Dan Driscoll (USA), Leopold Curfs (The Netherlands),  
Janice Forster (USA), Ann Scheiman (USA), Moris Angulo (USA), Charlotte Höybye (Sweden)

Absent: Susanne Blichfeldt (Denmark), Maithe Tauber (France), Urs Eiholzer (Switzerland), Shuan-Pei Lin  
(Taiwan), Harold van Bosse (USA), Susie Cassidy (USA)



# PROFESSIONAL PROVIDERS CAREGIVERS BOARD REPORT

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I thank all members of PPCB (and most of them have been on the board since 2010) very much for all their engagement and time they spend for PPCB and for people with PWS in all parts of the world.

To realize goals and motivate our group it is important to find new people who will support our work in this field. So we are very thankful that Damien Jones from Australia has now joined the PPCB. With his knowledge over many years in different fields of PWS and his enthusiastic personality he has enriched our group from the beginning. Thank you very much Damien!

An important step in 2018 was developing the caregivers delegate group. Michael Pethe, our PPCB member from Switzerland, has been supporting this group of nominated caregivers from the national associations which is now more than 10 people from different countries. One of the results of their face-to-face meeting in Munich in the summer of 2018 was the idea of a summer camp for people with PWS from all over the world in Denmark in 2020.



## Norbert Hödebeck-Stuntebeck

CHAIR, PROFESSIONAL PROVIDER CAREGIVER BOARD

As the planning for this develops, we will keep you in touch through our newsletters. Thank you very much Michael for that!

Since the PPCB started in 2010, we have always had a highlight of the year. In 2018 it was our Caregivers conference in Munich, at Castle Fürstenried. We are grateful to Hubert Soyer for his tireless work and ideas to make this event so successful. We had 120 people from 26 countries from all over the world in Munich. It was also the first time that the whole IPWSO board and the whole Clinical Scientific Advisory Board (CSAB) were involved in the conference. Consequently the conference was of a very high quality which was seen in the discussions in the workshops and in the lectures to the audience.

Because of this success, we hope to bring these groups together so that together we can develop methods and solutions in assisting people with PWS in the future.

To all - and it was a large number – who were involved in arranging this event, we say a very big thank you very much. As I have said in my annual reports over the years: "after the conference is before the conference." So we will be concentrating on preparing an attractive program for the caregivers conference as a part of the IPWSO conference in Havana, Cuba in the end of 2019. We hope to see many of you there to discuss future development of the support for people with PWS over all aged and over the whole world.

In the name of Mary K. Ziccardi, Fanny Cortes, Jackie Malow, Lisa Boman, Hubert Soyer, Larry Genstil, Michael Pethe, Damien Jones, John Ford, let's go on!



IPWSO Boards and participant care-providers at the conference in Munich, August 2018

# FAMCARE REPORT

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The Famcare articles are now included in material that is distributed at IPWSO awareness events, such as attendance at the International Association for the Study of Intellectual and Developing Disabilities Conferences. They can also be made into booklets for distribution at PWS conference or meetings. All our work can be seen on our website: [www.ipwso.org/famcare](http://www.ipwso.org/famcare)

We have had a productive year and published four new articles, and updated one of our earlier articles:

"When Rituals become Problems"

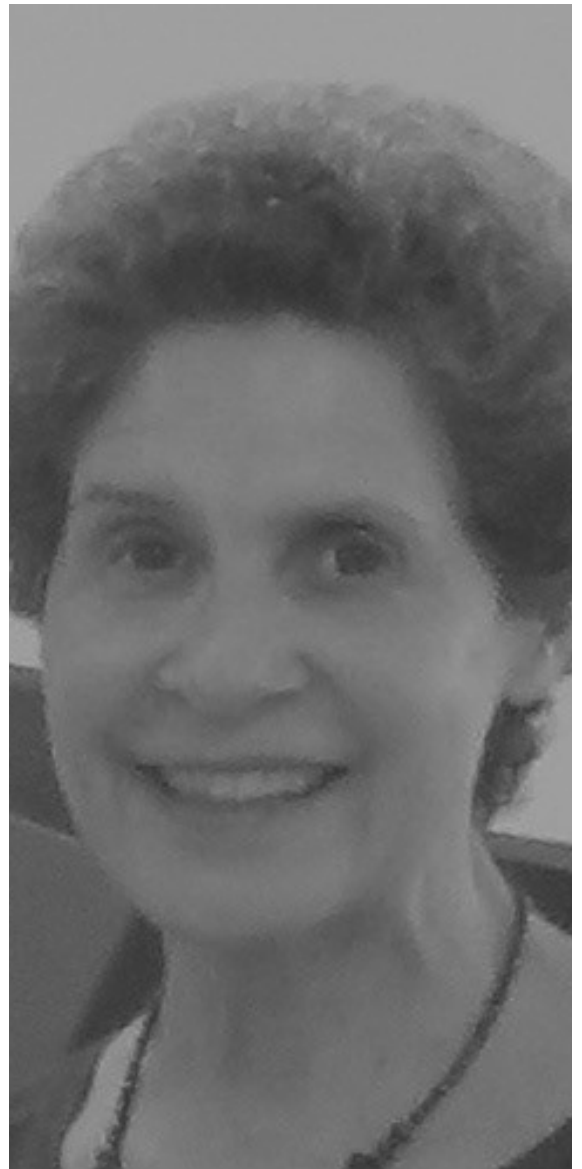
"Transitional Years"

"Life Plan"

"Law Enforcement"

"PWS Awareness for Professionals" (updated).

We continue to increase the number of Famcare articles that are translated in languages other than English. We now have Arabic, Dutch, German, Italian, Hindi, Mandarin, Russian and Spanish translations of several Famcare articles with more to be translated each year.



**Georgina Loughnan**

CHAIR OF FAMCARE



These articles are now included in material distributed at IPWSO awareness events, such as attendance at the International Association for the Study of Intellectual and Developing Disabilities Conferences. James O'Brien, an IPWSO Board member and President of the Australian PWS Association, and I attended the November 2017 Bangkok Asia Pacific IASSIDD Congress and had the pleasure of meeting with Thai families and professionals involved in PWS.

In the latter part of the year, Tony Holland and I attended a PWS conference in China. This was the first meeting that anyone from IPWSO had attended and therefore quite significant.

The Famcare committee continues to include parents and professionals from around the world. All written material is reviewed by an international professional advisory committee as well as the IPWSO Board of Directors. The articles may be printed for distribution at conferences or regional meetings, and present well in a booklet format. Famcare is currently working with Parents Programme subcommittee for the family programme of the next IPWSO Conference, in November this year in Cuba. *"It is a privilege to be involved in this work for what is expected to be a wonderful conference"*, Georgina said.



Famcare is always open to new suggestions for topics. Our committee comprises parents of adults with PWS and professionals. Georgina states, *"Parents speak openly from experience and our work is based on this parent-to-parent wisdom."*

Dr Dr Duangrurdee Wattanasirichaigoon.  
Georgina Loughnan and James O'Brien with  
families in Bangkok

# IPWSO PROJECTS

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## Long-distance Education

James O'Brien, IPWSO's Board member from Australia, was granted a Churchill Scholarship to study long-distance education systems and how they can be put to good use in countries such as his own, and how IPWSO might benefit from similar software in the support it offers around the world. He met individuals and attended meetings not just related to PWS, and visited organisations with staff who have the skills to inform the way that IPWSO could work and communicate in the future. He held workshops on PWS in India that brought families and professionals together.

"The aim of the project was to research methodology for delivering an affordable, replicable and sustainable awareness training program that can be delivered into cities, regional centres, rural and remote communities across Australia. This should also benefit training for PWS. The vast distances that separates the individuals, coupled with scarce resources due to our small membership, poses huge challenges in relation to delivering training and parent support services."

James' report will be available in 2019

**JAMES O'BRIEN**  
BOARD MEMBER



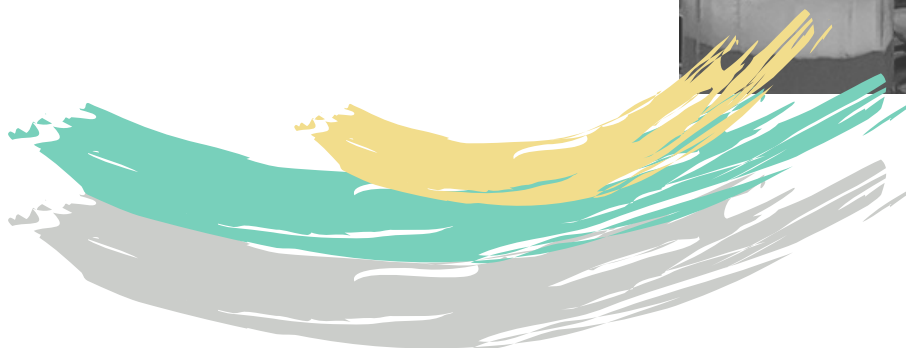
# Vietnam

An email from a paediatrician working in a centre for disability in Hanoi reached IPWSO's office asking for information and help with Prader-Willi syndrome. After much correspondence between the Board and Do Thuy Lan - Psychiatrist and founder and director of Morning Star Centre. IPWSO decided to grant an educational scholarship for Do Thuy Lan to attend the Caregivers' Conference in Munich, in August 2018 where she would have the opportunity to listen to, and learn the best practices for caregiving and understanding the challenges that make up Prader-Willi syndrome.

As a result of this and Lan's further request for support, IPWSO applied for funding from Friends of IPWSO (USA) to provide a workshop in Hanoi for medical professionals, parents, teachers, and caregivers to further improve the knowledge of PWS throughout that country.

Leading this educational workshop is Georgina Loughnan, Board Member, and it will take place in October 2019.

With the ongoing support of Friends of IPWSO, the board is confident that supporting these kinds of workshops will bring much-needed knowledge to countries where this is currently limited.



## Dr Lan du Thuy

PSYCHIATRIST, VIETNAM



# Baschirotto Institute for Rare Disorders

IPWSO's board member from Austria, Verena Gutmann is the contact person for the Baschirotto Institute for Rare Disorders (BIRD) which is the diagnostic laboratory IPWSO uses for its free diagnostic service. The laboratory is in Vicenza, Italy and this service is a cornerstone of IPWSO's work. IPWSO subsidises all requests for free diagnoses to patients who live in countries where this is otherwise unavailable.

This year a total of 41 diagnoses were made from samples sent from around the world including countries such as Bulgaria, Nicaragua, Peru, Venezuela, Hungary, Georgia, India, Lithuania, Pakistan, Armenia, Iraq and Italy. Although it is never good news to learn of your child's disability, this early diagnosis provides families with knowledge and support that they would not otherwise have ever gained.

The agreement with BIRD was originally established through the friendship of the Baschirotto family and Giorgio Fornasier (a past President of IPWSO) with the result that this service has benefited hundreds of families around the world, and will continue to do so.



## Verena Gutmann

BOARD MEMBER, AUSTRIA

# IPWSO ABROAD

## IASSIDD conference

IPWSO President, Tony Holland attended the European meeting of the International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASSIDD) in Athens in August where IPWSO was hosting a stall funded by the Friends of IPWSO (USA).

*"This meeting brought together many different disciplines working in the field of intellectual disabilities and there was a considerable interest in the work we do and in our material."*

In his research hat, Tony also attended the Foundation for Prader-Willi Research (FPWR) annual conference held in Las Vegas where a colleague was presenting some of their research findings. *"What is striking is how there are now many trials taking place that offer hope that there will be treatments to reduce the hyperphagia as well as interesting work on the use of oxytocin. It is important that IPWSO has close links with FPWR and like-minded agencies across the world."*

Tony also attended as an invited speaker, a conference in Rio de Janeiro, Brazil. *"It was very impressive to see how the National PWS Association of Brazil has brought families and professionals together, particularly in Sao Paulo and Rio de Janeiro, and started specialist clinics to provide support and raise awareness. One major challenge in a country such as Brazil is simply the size of the country – how to reach families living outside the main cities,"* he said.



## Tony Holland

IPWSO PRESIDENT

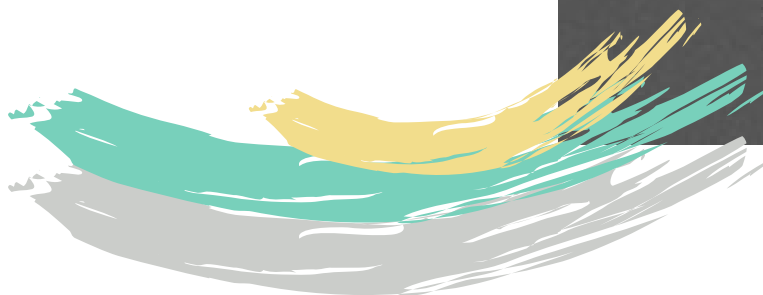
## SOUTH AMERICA

Individual IPWSO Board members have been busy. Amalia Balart together with colleagues from other South American countries has developed a PWS “expert network” and they have held several meetings with IPWSO contributing towards one such workshop in Colombia. Being able to contribute financially to countries needing IPWSO’s educational support is a mainstay of its Mission Statement and firmly fixed in IPWSO’s yearly work plans.

Amalia has been tireless in her travels around South America, teaching, educating and drawing families together to form PWS Associations. She writes, *"It would be great to be able to go ahead and translate our booklets and leaflets. Many families do not have technology (perhaps also very low income), they feel very alone."*

*"Perhaps we could help them to find other families with similar issues and help them start their own parent groups."*

Amalia felt that by being in touch with families who came to her meetings was very worthwhile and hopes that she can continue to support families in South America. *"For some, it was the first time they had met other families with a child with PWS, and it was a very eventful thing to see",* she said.



### Amalia Balart

BOARD MEMBER, CHILE

# Shanghai PWS International Forum China, 14-15 December

In December 2018, the Children's Hospital of Fudan University, National Children's Medical Center hosted the 2018 Shanghai Prader-Willi Syndrome(PWS) International Forum. Both Tony Holland and Georgina Loughnan attended. Georgina said, *"Along with our President, Tony Holland, I had the pleasure of being invited to speak as an international specialist on PWS. My presentation covered Clinical and Lifestyle Management of People with PWS."*

Other topics included in the programme were the development of early screening, a web app, mental health and behavioural disorders associated with PWS: from understanding to intervention; Novel treatment of PWS; From Research to Clinical Trials for PWS; Sleep-disordered breathing in children with PWS. The forum was attended by paediatric physicians from China and Hong Kong and included endocrinologists, geneticists and researchers. *"Our generous hosts showed us many of the sites of Shanghai and treated us to delicious, traditional Chinese cuisine."*

PWS awareness in China is growing, but treatment appears to be limited to children. Growth hormone is not funded by the government but is available if families can afford it.

*"As a result of attending this forum, I have connected with a group of endocrinologists from Hong Kong who are now initiating a local PWS database and next February, Dr Mingsheng Ma, a paediatric geneticist who attended the Shanghai Forum will spend the month with my department at Royal Prince Alfred Hospital, Sydney, to learn more about the clinical and lifestyle of those with PWS."* Georgina said.



## Georgina Loughnan

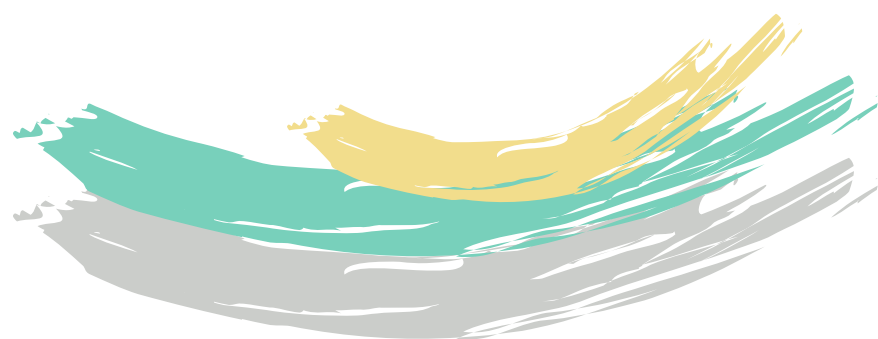
BOARD MEMBER, AUSTRALIA

# Australasian South Pacific PWS Conference 18-20 October

IPWSO funded two educational grants to the committee of the Australasian South Pacific PWS Conference which allowed Gayatri Iyer from India to attend this conference.

The conference was well-attended with good representation from Australasia and the Pacific Rim, and from IPWSO with Professor Dan Driscoll (Chair of IPWSO's Clinical & Scientific Advisory Board) attending the conference as opening speaker; Marilyn Dumont-Driscoll spoke as a paediatrician to the younger parents, Melanie Silverman on the importance of food and diet, and Assoc Prof Barbara (BJ) Goff addressing issues of schooling, education, and sexuality.

Australia and New Zealand have held conferences together for over 20 years, but only since the last 12 years have extended to include the South Pacific rim countries. It is hoped that more countries will become involved with these conferences, particularly with future involvement from Singapore, Vietnam and India.





# WHO WE ARE

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## IPWSO Board

IPWSO has a working board of up to 10 people. Board positions are held for a three-year term with a further three years if desired. Voting is held every three years at the General Assembly held in conjunction with the International PWS Conference.

Requests for nominations are posted six months in advance and a Selection Sub-committee interviews candidates and gives a slate of best nominees to the IPWSO Board. The slate is reviewed and posted on the IPWSO website. If there are no further nominations or objections, this will be the slate that IPWSO Parent and Professional Delegates will vote on at the General Assembly.

## Other Boards

The Clinical & Scientific Advisory Board and Professional Provider Caregiver Board have a longer-serving membership and prospective new members are run past the IPWSO Board before being selected.

All Boards meet regularly via Skype meetings and reports are shared.



## IPWSO Boards



# IPWSO BOARD



Back Row: James O'Brien (Australia), Verena Gutmann (Austria) Hubert Soyer (Treasurer, Germany), Mariona Nadal (Spain), Georgina Loughnan (Australia)

Front Row:

June-Anne Gold (USA), Tony Holland (Chair, UK), Marilyn Dumont-Driscoll (Secretary, USA), Marguerite Hughes (Ireland), Amalia Balart (Chile)

## Clinical & Scientific Advisory Board

### CHAIR

Dan Driscoll, PhD, MD

University of Florida

Health Sciences Center

Div. Genetics / Dept. of Pediatrics

Gainesville, FL USA

### VICE CHAIR

Susanne Blichfeldt, MD

Consultant (Neuropediatrics) Herlev University Hospital 2730 Herlev Denmark

Tony Holland

Emeritus Professor

Department of Psychiatry, University of Cambridge, UK

Professor Leopold Curfs, PhD

Dept. of Clinical Genetics University Maastricht/Academic Hospital Maastricht, The Netherlands

Moris A. Angulo, MD

Director of Medical Genetics Assistant Director of Pediatric Endocrinology

Winthrop University Hospital Mineola, NY USA

## CSAB Board contd.

Janice L. Forster, MD

Child & Adolescent Psychiatrist Pittsburgh Partnership Pittsburgh, PA, USA

Urs Eiholzer, MD

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

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

Harold van Bosse, MD

Orthopaedic Surgeon

Shriner Hospital, Boston, USA

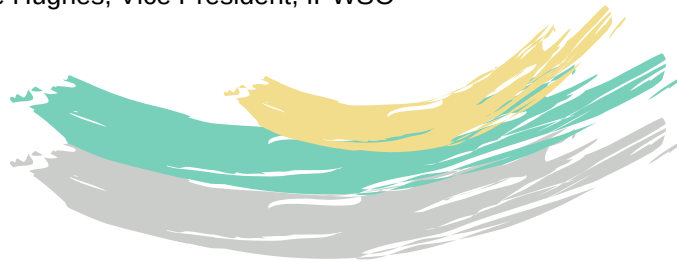
Maithé Tauber, MD

Professor of Pediatrics at the University of Toulouse and Chief of Endocrinology and Medical Genetics at the Children Hospital of Toulouse, France

Suzanne Cassidy, MD

Clinical Professor of Pediatrics and Medical Genetics at University of California , San Francisco, USA

Ex Officio: Marguerite Hughes, Vice President, IPWSO



## 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

# PPC Board contd

Fanny Cortés MD

Clinical Geneticist Rare Diseases Center. Clinica Las Condes Santiago, Chile

Jackie Mallow

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

Lisa Boman BA

Director Counselor & Social Worker

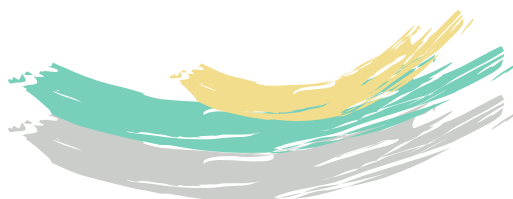
PWS Group Home Attendo, Stockholm, Sweden

Damien Jones

Caregiver Manager, Australia

Michael Pethe

Careprovider, Switzerland



## Famcare Committee

Chair: Georgina Loughnan

Physiotherapist, 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

Michelle Andrea

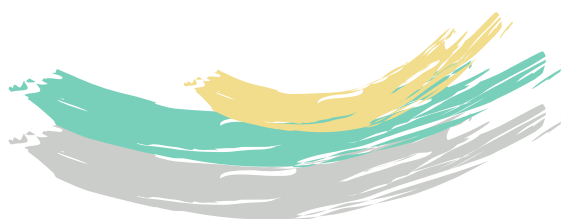
Parent, USA

Fanny Cortes

Geneticist, Chile

Sabeen Iqbal

Parent, USA



## IPWSO Office

Linda Thornton

Communications Coordinator

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information@ipwso.org

Agnes Hctor

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Website: [www.ipwso.org](http://www.ipwso.org)

Conference website: [www.ipwsoconference.org](http://www.ipwsoconference.org)

### IPWSO AUDITORS

James Gardner (USA)

Gerard Meijwaad , The Netherlands

### LEGAL ADVISERS

David Gordon (USA)

Colin Pearson (UK)

# Fundraising Committee

Chair: Marguerite Hughes, Ireland  
Hubert Soyer, Germany  
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# 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 down-turned 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 (near-sightedness) 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.

Treatment is primarily symptomatic and most importantly consists of:

- Early diagnosis and intervention
- Lifetime weight control through diet, environmental controls (lack of access to food), and exercise
- Growth hormone replacement therapy, when available
- Behavior management, with consistent limit-setting
- Special education and sheltered or supported employment

For a full description of Prader-Willi syndrome, please refer to our [website](#)

# International Prader-Willi Syndrome Organisation



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