



# Annual Report 2017

INTERNATIONAL PRADER-WILLI SYNDROME ORGANISATION



*BIRD laboratory, Vicenza, Italy, where all our PWS genetic testing is done. Board member Verena Gutmann talks with research scientist*

IPWSO

Twenty-eight years of service around the world



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# IPWSO Boards

## IPWSO PRESIDENT

Anthony J. Holland, MD, CBE (United Kingdom),  
Health Foundation Chair in Learning Disabilities

## VICE PRESIDENT

Marguerite Hughes, Ireland

## TREASURER

Hubert Soyer, Germany

## SECRETARY

Marilyn Dumont-Driscoll, USA

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June-Anne Gold, MD, USA

Mariona Nadal, Spain

Amalia Balart, Chile

James O'Brien, Australia

Verena Gutmann, Austria

## HONORARY PRESIDENT

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## LEGAL ADVISER

David Gordon, USA



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University of Florida  
Health Sciences Center  
Div. Genetics / Dept. of Pediatrics  
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Consultant (Neuropediatrics) Herlev University Hospital 2730  
Herlev Denmark

#### **Leopold Curfs, PhD**

Professor  
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

#### **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

#### **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**

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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, IUSA

#### **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



## PROFESSIONAL PROVIDER CAREGIVER BOARD

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Stuntebeck PhD**

Psychologist Diakonische  
Stiftung Wittekindshof  
Germany

### CO-CHAIR

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Regional Director REM  
Ohio USA

**Hubert Soyer PhD**

Psychologist Director,  
Regens Wagner Absberg  
Germany

## AUDITORS

Jim Gardner, USA

Gerard Meijwaad, The Netherlands

**Larry Genstil PhD**

Psychologist Executive  
Director Genstil Institute of  
Human Behaviour  
Israel

**John Ford PhD**

Psychologist Specialising in  
behavioural care  
New Zealand

**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



## 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  
Michelle Andrea, parent, USA  
Fanny Cortes, geneticist, Chile  
Sabeen Iqbal, parent, USA

## OFFICE STAFF

Linda Thornton, New Zealand, Communications  
Coordinator

Agnes Hocht, UK, Administrator

## FUNDRAISING COMMITTEE

Chair: Marguerite Hughes, Ireland  
Hubert Soyer, Germany  
Linda Thornton, NZ  
Joan Gardner, USA  
James O'Brien, Australia

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## IPWSO ADVISERS

Joan Gardner (USA)

Engela Honey MD (South Africa)

Tomoko Iwasaki (Japan)

Janalee Heinemann MSW (USA)

Suzanne Cassidy MD (USA)

Barbara J Goff (USA)

Kate Woodcock (UK)

Tony Goldstone MD (UK)

Dan Beaver MD (USA)

Joyce Whittington (UK)

Clare Wall (New Zealand)

Peter Davies (Australia)

James Loker MD (USA)

Ellie Silbertstein (Argentina)

Jackie Waters (UK)

Constanze Lämmer (Austria)

Nick Finer (UK)

Colin Pearson (UK)



# President's Report

## Tony Holland, UK

### President's Report 2017

The work of IPWSO takes many forms, varying over time depending on the requests we receive and the opportunities that come our way. Although at the beginning of 2017 we made the decision to be conservative in our commitments because of initial uncertainties about funding, we have made certain that we have maintained the core of IPWSO's work of providing free genetic testing, advice to families and clinicians, and information in different languages. We have worked hard to diversify our fund-raising and to further develop the governance of IPWSO so that we can expand our endeavor in the knowledge that we are financially secure and have a governance structure fit for the future. This is even more important now given the concerns that have been expressed in the press about international charities and how they operate overseas. We are, of course, very much smaller than these organisations and we do not directly employ people. However, we must be certain that we spend our money wisely in a manner that is open to scrutiny and at all times we act responsibly. As part of our journey towards charitable status I have been looking at past Annual reports and governance documents. All the work done by previous Boards is impressive, and we have sound policies and procedures, and our work is open to scrutiny. This is how it should be.

What then have we done over the past year? Early in 2017 we achieved what the previous Board had set in motion, establishing a group of



IPWSO advisors. Some of the Advisors are parents of children with PWS and others have professional or business backgrounds, bringing very different skills. We wish to expand this group further and, in particular, to identify people from across the world who may be able to help. During the year we have asked for advice from several advisors and we are very grateful for their support. During 2017 Joan Gardner established Friends of IPWSO and in 2018 they will be in a position to raise funds to support specific IPWSO projects. Marguerite Hughes, June -Anne Gold, Marilyn Dumont-Driscoll, and myself from the Board, and Dan Driscoll, Chair of CSAB, together with the past President, Suzie Cassidy, were at the USA Association meeting in Orlando to meet with the newly established Board of the Friends of IPWSO. Thank you to all who have made this possible.

The ignorance about PWS was vividly highlighted early in 2017 when it became apparent that the Chinese photographer, Li Song, was announced

as one of the winners of the Sony World Photography competition for his photographs of an 11 year-old boy, Li Hang, with PWS. The photographs vividly illustrated Li Hang's struggles with food and with making friends. They also showed him receiving an alternative treatment, 'flame therapy'. When the exhibition was open in London the organisers agreed that information would be made available about IPWSO. Also, at around that time Susie Cassidy had been invited to attend a conference in China and she was able to meet with some families of children with PWS.

Members of the Board have been involved in various activities on behalf of IPWSO over the year. We provided financial support to a meeting held in Morocco for families from across North Africa. The meeting was arranged with support from the French PWS Association, and Mariona Nadal attended on our behalf. The Professional Providers and

Caregivers Board (PPCB) of IPWSO together with Amalia Balart, organised a conference in Chile attended by over 200 families and professionals. Georgina Loughnan and James O'Brien represented IPWSO at the Asia Pacific Meeting of the International Association for the Scientific Study of Intellectual and Developmental Disabilities (IASSIDD) in Bangkok. From the IPWSO stall they handed out over 70 memory-sticks with information about PWS and spoke to well over 100 delegates. They went on to meet with families of children with PWS in Bangkok and in Singapore.

With Giorgio Fornasier's retirement from IPWSO, Verena Gutman has taken over as our link with the Bashchiroto Institute for Rare Disorders (BIRD) and she met with the Bashchirottos and with the staff who work there. Over this past year they have received 24 samples for analysis from places as diverse as Georgia, Peru and Hungary. I am grateful to Verena for taking on this liaison role and to the staff at BIRD for the work that they continue to do on IPWSO's behalf. A reliable diagnosis of PWS is so important as information and understanding can then follow, and this can make a huge differences.

To develop our links with the wider 'rare diseases' community, Mariona Nadal attended a Rare Diseases International conference. We recognise it is important for IPWSO to engage with such international umbrella organisations if we wish to inform policy development at an international level.

We therefore propose to develop these links



*PWS conference in Morocco*

further in 2018.

Marguerite Hughes has chaired our fundraising sub-committee and her report summarises the work this committee has undertaken. We started 2017 concerned about funding, but progress has been made in diversifying our fund-raising activities. My special thanks to our member organisations for their on-going support and for the generous additional donations that some national associations have given us. Also, my thanks to the Organisations, Trusts, and individuals who have donated money during 2017. It goes without saying that without this support none of what we do would be possible. Thank you also to Marguerite and the members of the sub-committee for their hard work and persistence.

Linda Thornton is the first point of contact for families and others who contact IPWSO wanting to know more about PWS. This advisory and support role, particularly to families in parts of the world where there is no Association and there is very limited knowledge about PWS, remains the core of what we do. As we expand our booklets and leaflets and translate them into many different languages, so then we can better help families and professionals. Contact from individuals is not limited to coming from countries who do not have their own National Association and where there is an established National Association, often the best thing we do is to put people in contact with them. During 2017 Linda has had enquiries from Algeria, Argentina, Austria, Bulgaria, Canada, Colombia, Denmark, Dubai, Egypt, France, Georgia, Hong Kong, Hungary, India, Italy,

Malta, Morocco, New Zealand, Peru, Puerto Rico, Poland, Qatar, Russia, South Africa, UAE, Ukraine, and Vietnam. Some of the enquiries go to members of the Clinical and Scientific Advisory Board chaired by Dan Driscoll. The Professional Providers and Caregivers Board and Famcare continue to provide guidance and to prepare material. Thank you to the members of the CSAB, PPCB, and Famcare Boards for all that they do.

Again with Giorgio's retirement we have been more dependent on Linda and my thanks to her for all her efforts. Towards the end of 2017 the Board agreed to fund Agnes Hoctor for six hours a week to provide additional support. Agnes works with me in the Department of Psychiatry at the University of Cambridge. One of her main tasks will be to provide administrative support for the next international conference. During 2017 we started the preparation for the 10<sup>th</sup> IPWSO conference which will be held in Havana, Cuba from the 13<sup>th</sup> to 17<sup>th</sup> November 2019. Groups have been established between IPWSO representatives and the local Cuban organisers to oversee the different parts of the conference. We will keep you informed about developments and key dates through the website and newsletters.

We ended 2017 financially sound. Thank you to the Board and to the many others who have been supportive over the year. In 2018, there will be the next in what has been a series of conferences over several years aimed at those working in support services for people with PWS. These meetings are



organized by the Professional Providers and Caregivers Board (PPCB). This year's meeting, will be held in partnership with Wittekindshof and Regens Wagner, two organisations in Germany who provide support to people with PWS, will be from the 28<sup>th</sup> to 30<sup>th</sup> August 2018 at the Fürstenried Palace in Munich. As with previous meetings the hope is that representatives from provider organisations from many different countries will be able to attend to share ideas.

Our priority for 2018 is to complete registration as a charity. This requires establishing a new legal entity and then transferring the assets to this new

organisation, which will be official a Charity. We will be consulting further with our members as this process progresses. By doing this we will increase the options for fundraising. IPWSO as always continues to provide advice and support around the world and by the end of 2018 everything should be in place for an outstanding Cuba conference the following year – we look forward to seeing you there!

*Tony Holland*



**IPWSO Board**

Back: James O'Brien (Australia) Verena Gutmann (Austria) Hubert Soyer (Germany) Mariona Nadal (Spain), Georgina Loughnan (Australia)  
Front: June-Anne Gold (USA) Tony Holland (President, UK) Marilyn Dumont-Driscoll (USA) Marguerite Hughes (Ireland) Amalia Balart (Chile)



# Fundraising Report

## Marguerite Hughes, Ireland

### Vice-President and Chair of IPWSO Fundraising Committee

IPWSO raised over \$60,000 during 2017 and achieved a budget surplus for the year. Its cautious approach to expenditure, in particular its decision not to immediately fill the position vacated by Giorgio Fornasier in 2016, was largely responsible for the surplus. A new staff member will commence employment with IPWSO in January 2018.

While the overall sum raised was small, it is encouraging that past sources of funding remained consistent and income was also received from several new sources.

As in previous years, IPWSO's membership subscriptions and donations from member associations remained essential to its financial stability. Over \$7,000 was received in subscriptions from 29 member countries (the majority of the 104 countries affiliated with IPWSO are associate members and are not required to pay subscriptions). Almost \$15,000 was received in additional donations from the PWS associations in Australia, Austria, Belgium, Canada (FPRW Canada), Chile, Ireland, Italy, Norway, South Africa, Switzerland and the USA. IPWSO is very grateful for the ongoing support it receives from these associations.

Pfizer has been IPWSO's most consistent corporate donor and in 2017 contributed a very generous \$20,000. This represented the 8th consecutive year in which IPWSO received funding from Pfizer.

IPWSO was also delighted to receive a \$10,000 contribution from Soleno Therapeutics, which is



currently commencing Phase III clinical trials into DCCR, an oral tablet for the treatment of PWS.

Individual donations and fundraising efforts deserve mention too. Giorgio Fornasier, although retired since 2016, continued to hold fundraising concerts for IPWSO during early 2017. IPWSO's current and past board members once again responded generously to IPWSO's fundraising appeals. So too did many new donors who initiated fundraising events and made personal donations. Particular thanks are due to one anonymous donor who donated \$5,000 in matching funding to encourage donations from others.

In late 2017 IPWSO initiated an appeal for regular givers who would be willing to donate a small amount to IPWSO each month. To date people from 3 continents have responded and set up monthly donations ranging in amount from \$10 to \$100. Along with support from IPWSO's member associations, these individual monthly donations represent the most sustainable form of income available to IPWSO currently. IPWSO is very grateful to those who have already signed up to its

monthly giving appeal and will continue to solicit this type of support in 2018.

IPWSO has observed with great interest the development of Friends of IPWSO in the US and Tony Holland and I were delighted to meet with the board of Friends of IPWSO in Orlando in November. We take great encouragement from the commitment and professionalism of the Friends of IPWSO board, particularly its Chair and founder, Joan Gardner. Going forward IPWSO hopes to submit targeted grant requests to Friends of IPWSO.

Although IPWSO succeeded in achieving a budget

surplus, over two thirds of its income was only received in December, which meant no cessation in pressure for the fundraising committee. I would like to sincerely thank the members of IPWSO's fundraising committee for their dedication and hard work. Thanks to Hubert Soyer, James O'Brien, Linda Thornton, Joan Gardner, Amalia Balart and June-Anne Gold.

Finally, I would like to sincerely thank all of IPWSO's donors and supporters whose contributions continue to make its work possible.

### ***IASSIDD—International Association for the Scientific Study of Intellectual and Developmental Disabilities***



James O'Brien and Georgina Loughnan represented IPWSO at the 4<sup>th</sup> Asia Pacific Congress of IASSID held in Bangkok, Thailand in November 2017. "This was a great opportunity to meet with like-minded people, those dedicated to the improvement of the lives of people with disabilities. Although many people who approached our information booth had heard of PWS, they were keen to increase their knowledge and eagerly accepted the information FLASHDRIVES that had been prepared for distribution. The IPWSO brochures were widely distributed and contact details were

exchanged by those who were keen to keep in touch. The hospitality and warmth of the conference organisers was fabulous and complimented by the graciousness of the Thai people we met.

"The absence of a presentation, either poster or oral, on PWS, highlighted the importance of future submissions to this conference as it seemed a most appropriate forum for presentations on PWS.

"During our stay in Bangkok and Singapore, we had the pleasure of meeting with the IPWSO professional and parent delegates and their families. Professor Duangrurdee Wattanasirichaigoon is a geneticist supporting the families of children with PWS, in much more than just genetic counselling! She meets regularly with them and lobbies for their needs. She was also a part of the organising committee for the IASSID congress. Sontaya, Preeyathida, Pink and their families were a delight to meet. They are working hard to maintain communication between and support for people with PWS in Thailand. They noted the need for more communication with parent of adults with PWS, as well as the ongoing need for more government support. The subsidisation of growth hormone is still greatly desired."

# Treasurer's Report

## Hubert Soyer, Germany

As last year, I am proud to present the new treasurer's report for the year 2017. A lot happened in past year and I would like to express my gratitude to Marguerite Hughes and Giorgio Fornasier for helping me aggregate all the relevant data and providing me with the necessary information on balances and cash flows in the IPWSO accounts.

### Developments in 2017

In the year ending in December 2017 we raised a total of \$41,382.88 in our US account and 20,798.65€ in our Euro account. Furthermore, we received 2,130.00€ in donations to our Petty Cash account which was closed at the beginning of October 2017 and merged with our Euro account.

Comparing these incomes with the expenditures of \$5,819.12 on the US account, 22,032.00€ on the Euro account and 2,781.88€ on the Petty Cash account we can present a surplus of \$35,563.76 and a minus of 1.885,23€. A great part of our income was generated through donations of \$39,880.00 and 11,271.25€ while membership fees amounted to \$1,069.71 and 6,195.25€.

I would especially like to thank Pfizer, Soleno, PWSA Australia, PWSA Austria, PWSA Belgium, FPWR Canada, PWSA Chile, PWSA Ireland, PWSA Italy, PWSA Norway, PWSA South Africa, PWSA Switzerland, PWSA USA, the Presentation College, Elena Da Lan and all the other donors for their generous contributions as well as Giorgio for donating the proceeds of numerous concerts. Furthermore, I am very grateful to all the members for contributing through their constant hard work and membership fees.

Given the numbers above, I am pleased to report



that we have completed a successful year 2017 with all balances being healthy.

At this point I would like to direct a special thank you to Tony Holland and Marguerite Hughes, who have been tirelessly advancing the process to receive charitable status for IPWSO in the UK - to create a stepping stone to long term funding opportunities.

For the coming year I hope that we will be able to maintain the level of donations for preparing the IPWSO Conference 2019 in Cuba, to get the charitable status for IPWSO and to find many other opportunities to efficiently use these funds to support people with PWS and their families as well as researchers and caregivers.

*Hubert Soyer*

# Audited Financial Statement

BALANCE (2017) - Euro Bank Account		BALANCE (2017) - USD Bank Account	
Revenue		Revenue	
Sum of membership fees	6.195,25 €	Sum of membership fees	\$1,069.71
Sum over donations (Including IDONATE)	11.271,25 €	Sum over donations	\$39,880.00
Others	3.332,15 €	Others	\$433.17
Total revenue	20.798,65 €	Total revenue	\$41,382.88
Expenditures		Expenditures	
Bank charges	-524,51 €	Bank charges	-\$240.78
Conference expenditures	-700,00 €	Conference expenditures	-\$5,000.00
Staff salaries	-14.074,00 €	Others	-\$578.34
Others	-2.363,49 €		
BIRD Tests	-4.370,00 €		
Total expenditures	-22.032,00 €	Total expenditures	-\$5,819.12
Saldo (2017)	-1.233,35 €	Saldo 2017	\$35,563.76
Saldo + last year balance	41.792,10 €	Saldo + last year balance	\$110,839.13

## 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 2017.

April 2018



# Clinical & Scientific Advisory Board



## Dan Driscoll, MD, USA

### Membership of CSAB

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

Dan Driscoll (Chair) (USA)  
Susanne Blichfeldt (Vice Chair) (Denmark)  
Moris Angulo (USA)  
Susie Cassidy (USA)  
Leopold Curfs (The Netherlands)  
Urs Eiholzer (Switzerland)  
Janice Forster (USA)  
Tony Holland (UK)  
Charlotte Höybye (Sweden)  
Shuan-Pei Lin (Taiwan)  
Ann Scheimann (USA)  
Maithé Tauber (France)  
Harold van Bosse (USA)

### Scientific papers

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 by Linda Thornton, Communications Coordinator. 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 neuroimaging techniques. A special note of thanks should go out to Joyce Whittington for her 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: Growth Hormone, Pain in PWS,

C.I.D for PWS, cervical surgery, tracheostomy, cancer and severe infection

The requests come from a range of countries across the world, for example, UK, Brazil, New Zealand, USA, Japan, Georgia.

Thank you to the many members of the CSAB who have responded to these requests. Linda keeps a record of the replies for future reference.

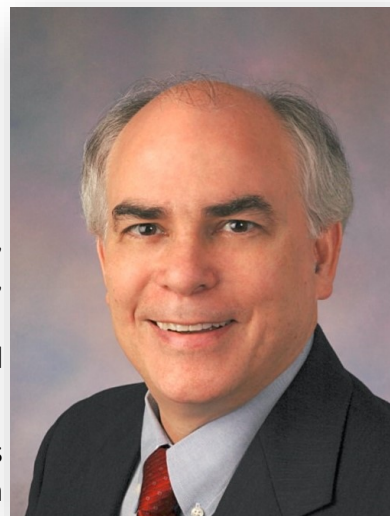
The CSAB prepared a Medical Overview and Checklist for Adults with PWS that is now available to families and health care professionals on the IPWSO website.

### Other matters

Dan Driscoll and Tony Holland travelled to Havana, Cuba to meet with members of the Cuban Human Genetics Society in preparation for the Scientific Conference at the IPWSO meeting in Havana, Cuba in 2019. We anticipate having a robust Scientific meeting in collaboration with the Cuban Human Genetics Society.

Numerous research clinical trials involving PWS are currently being conducted in different parts of the world. The CSAB is keeping abreast of these trials and will report on promising results as they become available.

The quarterly collection of papers is available on our website: <http://www.ipwso.org/news>



*Dan Driscoll*

# Professional Providers Caregivers' Board



Norbert Hödebeck-  
Stuntebeck, PhD

Chairman, Germany

2016 – When we reflect over the last year and ask ourselves “ what was the most important thing or event in our work in the PPCB in 2016?”, it is easy to answer that this was the IPWSO Conference in Toronto, which took place from the 20<sup>th</sup> to the 24<sup>th</sup> of July 2016. The Caregivers Conference was a part of the overall IPWSO conference and occurred over two full days, with more than 20 presentations, moderated discussions and expert panels by speakers from 10 countries throughout the world. In attendance were more than 80 people, representing more than 20 countries. Because we used a “call for abstracts” in advance of the conference, we were able to offer many new, interesting and important speakers for the presentations.

We began the conference with overviews of services in different countries. In these presentations, we discovered unique things in some countries but also many similar approaches, some of which developed over the years independent of each other but resulting in a similar outcome.

Sexuality, Rights, Behavior, PWS and High Intelligence, Social Competencies , Muscle strengthening activity, Tomatis Training, Sensory Deficits and Aging were themes and topics which

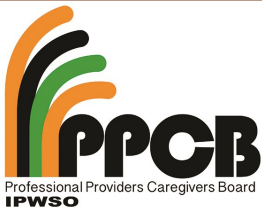


were addressed and made the conference a special event. The opportunity to participate and learn from the knowledge of other caregivers throughout the world, coming in direct contact with these expert speakers, adds to the uniqueness of the Caregiver's Conference.

The high quality of the presentations we experienced has made it our goal to collect as many of the presentations as possible to create another “Best Practice Book” so that others can share the knowledge and improve the care to many with PWS around the world.

There were many successes at this conference. However, a disappointment was that the Caregiver's Conference presenters and participants were not afforded the opportunity to have direct, formal interaction with the presenters and participants in the Scientific Conference. We are continuing to attempt to address and agree on a format in which this collaboration is possible. We believe that we work best when professional caregivers and

# Professional Providers Caregivers' Board contd



scientists collaborate, share and inspire each other to find the best ways to enhance the quality of life for people with PWS.

During the conference in Toronto, we shared another significant step towards supporting people with PWS in their environments. The second "Best Practice Book", with the focus on "Trainings", was finished and is now available to purchase. In this publication, you will find chapters about Sensory Motor Integration, Kinesthetics, Nutrition, Social Competencies, Behavior and others. Additionally, we have begun to work on a translation to German and hope to present this in summer 2017.

The year 2017 was not the "loudest" year of the Professional Provider Caregiver Board of IPWSO. This means that we were unable to organize a caregiver conference or a face to face meeting with all of our board members in 2017. This was the first time since we started with the work in

PPCB in Taiwan in 2010 that we have not met in person throughout the entire year.

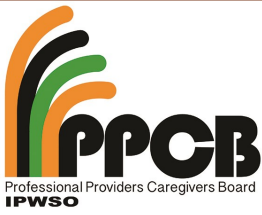
But "not loud" does not mean that we were not active!

One of the most exciting events of 2017 was to support the planning of the First South American PWS conference in Santiago de Chile, which took place on September 8 and 9, 2017. The main idea of this conference was to bring people from South American countries together with the PWS world, with the goal of developing intensive networks in these countries for the future. It was a very successful conference! Some of our PPCB members were invited to this event and each made several presentations to the audience, sharing their experiences about supporting people with PWS. The PPCB members who participated were very impressed with the intense spirit of working together and supporting each other, now and in future years.

In 2017, the PPCB began to plan another event, which will be held in 2018. We are pleased to offer the 5<sup>th</sup> Caregivers' Conference in Munich on



Meeting in Toronto



August 28-30, 2018. As in the past, this Caregivers' Conference is scheduled in between the years of the IPWSO International Conferences. The Caregivers' Conference in Munich will have the focus on the workshop method of presentation, as has been traditional for the Caregivers' Conferences in the past. This presentation style gives professional caregivers the chance to work out new strategies, methods and processes to support people with PWS in the best ways we know. To date, we are aware that more than 20 countries will be represented at this upcoming event.

Another initiative that the PPCB rekindled in 2017 was the idea of having a group of caregiver delegates from countries all over the world. Today, we have nine countries which have nominated a caregivers delegate. At the first meeting, organized at the end of 2017 as an internet call, most of the delegates were able to attend and participate. During this meeting, we discussed and decided on the rules and duties of caregiver delegates for the future ([www.ipwso.org](http://www.ipwso.org)); discussed ways to ensure that the caregiver delegates are visible on the national websites, to encourage people from their own countries to contact them and become involved, and we began to write and make available an overview about PWS-specific services in their countries and began to collect information and start to write an overview about holiday locations which

are supportive environments for people with PWS. This active network is waiting for other caregiver delegates from the countries that are not yet represented. An interested caregiver must be nominated by their national PWS association in order to be considered as a delegate. If you are interested in more information or to work with this group, please contact a member of the PPCB.

During both our PPCB internet board meetings or conversations in smaller groups of PPCB members, we discuss our prior experiences but we focus much more on the needs to develop advanced knowledge, and we have chosen to especially focus on the following content and themes in the coming years:

- \*We are aware that many positive developments in the past years have made it possible that people with PWS have the chance to live longer than at any time in the past. What are the special needs to be considered when people with PWS get older? Do they need different or separate services for living or working? Is dementia a concern, and if so, how much?

- \* Twenty years ago, with the birth of someone diagnosed with PWS, there was very little information about it and families learned by "trial and error". Today, we are more frequently blessed with the situation that there is detailed information about PWS for the families of infants. We need to ask ourselves if the development of a child/ youth with PWS today is totally different from 20 years ago? Do they have more and improved opportunities for employment? Do they have better opportunities to engage in healthy relationships with others? Is this a new generation that faces different challenges and demands for



services?

\* When considering a new generation of people with PWS, who present so differently with chronological ages, activity levels and personalities, to name a few variables, there is much to consider. Do these differences require separate living spaces? How can and how should we handle this in the future? What are the services that are most needed and in demand?

\* All of us know that managing the challenging behaviors presented by someone with PWS is one of the most critical aspects. We need to collect and consider what programs, trainings and services are available and in which parts of the world can these services be obtained? Do the supports throughout the world communicate and work together to learn from each other? And, how should these services and supports be documented so that we can share basic information and make an evaluation of what works and what does not?

\* We are all aware that physical exercise is a basic theme in caring for people with PWS. How can we help with motivating people to participate? How can we influence communities so that exercise activities for people with PWS are available in an inclusive way in society?

\*Caregivers, and others, who know the most about the management of PWS are the best qualified to care for those who have PWS. Can we identify what programs exist throughout the

world? How can we create basic modules which are consistent and important in every country? How can we share the knowledge throughout the continents and explore the idea of “continental team”?

Over the past several years, we have found that it can be very successful to find solutions to these themes, and other important questions, by bringing those knowledgeable about the day to day care of PWS, to work together. Additionally, much can be gained by caregivers and scientists/researchers working together. If we can merge the questions of the professional caregivers and families with the knowledge and resources of the scientific organizations, we may be about to determine more concrete strategies, and ultimately, an increased chance for people with PWS to have be able to look forward to a healthier and happier life. We are looking forward to the Caregivers’ Conference in Munich 2018 and to the 2019 IPWSO Conference in Cuba, where we plan to have designated places and times spaces for overlapping dialogue and work between the professional caregivers and the scientists.

For all the things we did in the past and we will do in the future, I would like to say thank you very much! I am especially grateful to all of the members of the PPCB for their active engagement and the resources they have committed for the advancement of the ideas of IPWSO.

So, let’s go forward and follow Pam’s ideas!

In the name of the PPCB Board members

*Norbert Hödebeck- Stuntebeck*

# Famcare

## family care at home



### Georgina Loughnan, Australia

Where did 2017 go!

This was a quieter year for Famcare as health issues dictatated output.

Our new article for 2017 was “Promoting Positive Behaviour Throughout Life”. This article emphasises the fact that behavioural outbursts are a part of PWS and will occur in adults with PWS, just as they do in children. It discusses the “what” and “why” of inappropriate behaviour and gives practical tips on what to do to address it. Preparation and calmness continue to be key factors in management. This brings the number of Famcare articles to thirteen.

Despite the Famcare target audience being families who have adults with PWS living with them, the information is now widely used by caregivers and families of teenagers and even younger children.

Thanks to Linda Thornton’s diligence we are continuing to have the Famcare articles translated

into different languages. We now have some articles in Mandarin, Russian and Hindi with more to come! Please see our Famcare pages on our



website for additions to the Famcare articles.

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. Please share the Famcare articles with as many relevant people as possible. The articles may be printed for distribution at conferences or regional meetings, and present well in a booklet format.

Famcare is currently working with the subcommittee for the family programme of the next IPWSO Conference, to be held in Cuba in 2019! This is a most exciting venture to be included in what is expected to be a wonderful conference.

Please remember, Famcare is always open to suggestions of new topics and contributions to topics.

*Georgina Loughnan*

# Communication Coordinator's Report

## Linda Thornton, New Zealand

As usual the year of 2017 was full of interest with lots of requests for information from families around the world. Some of these questions brought our whole Clinical & Scientific Advisory Board (CSAB) into play. There seem to be more and more questions about Prader-Willi syndrome and how it might be 'disguised' by other issues or other syndromes, so how do we define it?

Having professional boards such as our CSAB, Famcare, and Professional Providers Board, means that anyone from anywhere can contact one of our experts with PWS skills and receive guidance. This is such an advantage and gives families confidence that their questions and concerns are being heard. Communication is one of IPWSO's major cornerstones and so often long-term correspondence makes friendships around the world.

Our Facebook group is constantly active and links into our website as well as our popular blogs. This year we had contributions from many of our



member countries, introducing us to their lives and children with PWS. The blog mixes parent stories with more serious items that challenge us all when raising a child or caring for an adult with



PWS.

Life is always changing, new advances in science and research always happening, but one thing stays constant and that is the love we have for our children and the desire to do the best we can. Sometimes things don't work out, but solutions come from surprising sources. Take, for example, our friends in South Africa one of the very first countries to stand up and be counted when IPWSO was born. For years their PW Association functioned as best as it could in the face of adversity until they recognised there was another way of reaching out. It's a great solution for any country finding it difficult to manage an Association on their own. Their story is on the next page. It's another way of standing tall!

You can contact us with your concerns, or just to say 'hi', we're always here!

*Linda Thornton*



# From

## South Africa

*South Africa was one of IPWSO's formative member countries. When several of us gathered together in Nordwijkerhout (The Netherlands) and decided after our first international conference in 1991 that we must continue as a global organisation, South Africa stepped up. Birgit Schroeder became one of the first IPWSO Board members.*

*It was difficult to get PWS recognised in South Africa and over the years the PWSA (SA) decided that it would be a wise idea to merge with a larger organisation. So this is what they did in 2017. This is their story:*

### **Merge with Rare Disease SA**

There was a certain amount of admin involved in dissolving the old Non Profit Organisation, transferring funds to Rare Disease SA to “hold” for us and establishing a new way of functioning under the umbrella of Rare Disease SA.

### **Awareness campaign**

The campaign was run through March, April and into May 2017. It was organised through Rare Disease SA using their media contacts and

although we only paid for one media release the article prepared by Janet Legemaate was used countless times through many different forms of media; magazines, Sunday papers, radio and online magazines. It was hard for her as one of the magazines, the mostly widely read magazine in the country (a bit like UK's Hello Magazine) misrepresented what she said. However overall it was a very successful campaign

We added to this by running a campaign through our Facebook page for May PWS Awareness month as well.

### **Communication**

We wanted to try and offer a platform for parents to better communicate over the small day-to-day events. The Facebook page wasn't really working due to it being an open page and as such parents didn't really post comments on articles or upload their own posts. Also many people in South Africa don't use Facebook very often. It is difficult for us to get-together as South Africa is big and families are very widely



There is no  
cure for PWS  
- but it can  
be managed

**PWS Awareness –  
May 2017**





distributed. It is interesting that so many of our PWS families live in rural areas compared to the normal population. We tried to organise some get-togethers in Cape Town as there are quite a few families here but haven't had much luck getting people to come to a social event.

We decided to offer a "WhatsApp" group. We thought if we had many interested parties we'd split it up into three regional groups. However to start with we only had 20 people so we had one group. It has grown quite a lot now and we have about 50 people contributing. Parents are loving it as they can communicate so easily across the miles and it is as if they have found their "tribe".

Facebook is working well as a method for keeping families updated on the latest news about PWS from around the world and of local events and happenings relevant to us. Our following is continually growing and from Facebook they join our "Whatsapp" chat group.

### PWS Home

One of the mom's with an older son has taken on the challenge of starting a home for adults PWS in SA. Once again it is tricky due to the large distances between people but she has found a

few parents who are willing to go in together and start a home in Rural Western Cape. She has made great progress but it should still take a while to get up and running

2018:

In 2018 we would like to have a follow on media campaign and advocate for our medical aids to pay for GHT with the help of Rare Disease SA. We would also like to improve our member database as we have quite a large following on Facebook but a small list. We would also like to establish a comprehensive list of resources for all the different regions in South Africa.

In terms of getting people together the only time since I've been involved that we have had a good turnout of families to an event is when we have had a guest speaker from overseas who is an expert on a particular area of PWS. So I just wanted to mention that if you ever hear of anyone in the field coming out to South Africa on holiday or for any other reason, please put them in touch with me. If they would be willing to do a talk it would be fantastic. Just taking a chance here ☺

If there is anything else being done in other smaller (population-size) countries like South Africa that has worked really well, please let me know we are open to learning and getting great ideas from elsewhere.

It is wonderful to be part of IPWSO and all the great work that you do.

*Karin Clarke*



# 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 (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.