



Annual Report 2016

INTERNATIONAL PRADER-WILLI SYNDROME ORGANISATION



IPWSO

Twenty-six years of service around the world



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Cover photo: Mahdi from Morocco

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IPWSO Board 2016 Back row: James O'Brien, Verena Gutman, Hubert Soyer, Mariona Nadal, Georgina Loughnan

Front row: June-Ann Gold, Tony Holland, Marilyn Dumont-Driscoll, Marguerite Hughes, Amalia Barlart

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Health Foundation Chair in Learning Disabilities

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IPWSO Boards, contd

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Linda Thornton, New Zealand, Communications
Coordinator
Tiina Silvast, Finland, Design Consultant

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Peter Davies (Australia)

Ellie Silbertstein (Argentina)

Jackie Waters (UK)

Constanze Lämmer (Austria)

James Loker MD (USA)

President's Report

Tony Holland

This is my first annual report in my capacity as President of IPWSO. I took over officially from Suzanne Cassidy at the IPWSO General Assembly held at the 9th IPWSO International Conference in Toronto in July 2016 - the 25th year since IPWSO was founded. At the same time a new Board was elected with a mandate to expand and develop the work that IPWSO undertakes. It is a pleasure to take on this role and for us as a new Board to pick up the challenges and to continue the work started by others who have contributed to IPWSO over the last 25 years.

The IPWSO international conference held every three years is a major highlight of the IPWSO calendar bringing together people with PWS and their families, those working in the health and social care sectors of many different countries, and researchers and clinicians. The different components of the Conference seek to expand our understanding of the needs of people with PWS and to inform and support individuals, families, and organisations from across the world. These conferences are always in partnership with a local organisation, in this case FPWR Canada. We are very grateful for all the work they and others did. It is impossible to do full justice to the conference with its different strands and themes but, as a clinician and researcher, I did get a very strong sense that we are moving into a new era where novel and potentially important treatments are being developed and are being tested – the use of oxytocin by nasal spray and the trials of medications to suppress appetite being examples. However, central to the good support of people with PWS remains an accurate and early diagnosis, well-informed families, access to affordable and informed healthcare, and the availability of guidance and long-term support over the lifespan. This is far from available across the world even in wealthy countries. There remains much for IPWSO to do in partnership with existing and new National Associations.

In the year that the International Conference is held it tends to dominate the agenda but the work of IPWSO goes on as is illustrated well in the other reports. The Clinical and Scientific Board (CSAB)



now led by Dan Driscoll; Famcare led by Georgina Loughnan; and The Professional Providers Caregivers Board (PPCB) led by Norbert Hödebeck-Stuntebeck have all been active. Linda Thornton, IPWSO Communications co-coordinator, and Giorgio Fornasier Director of Programmes, are central to the work of IPWSO and act as the first point of contact for families often at times of crisis. They are the 'face' of IPWSO and their enthusiasm and experience is invaluable to us. IPWSO's reach across the world continues to expand with new contacts over the year from Singapore, Morocco, the Gaza Strip in Palestine, China, and India. Material has been translated into 15 different languages and lists of clinical and scientific papers published throughout the year, prepared by Joyce Whittington, continue to be circulated regularly through IPWSO contacts and via National Associations. Our CSAB provide thoughtful and helpful answers to the very diverse range of questions we receive. Throughout the various activities of 2016 the aim of IPWSO continued to be both supporting people with PWS and their families, and raising awareness about PWS with clinicians and national agencies.

The work of IPWSO would not be possible without your financial support. We are very grateful to our Member Associations for the fees they pay and often for additional and generous donations they make to IPWSO. We also receive individual donations from Trustees and from friends and colleagues. With the

President's Report contd

help of Joan Gardner all donations in 2016 were matched by an equivalent amount by the KB Andersen Trust up to a value of US \$15,000. Thank you to the KB Anderson Trust and to everyone for this support.

The new Board looks forward to the challenges ahead and on behalf of the new Board I would like to thank Susie Cassidy as Past President and the previous Board for all the work they have done. Giorgio informed us at the Toronto conference that he will be retiring at the end of 2016, he will continue to be very involved in the arrangements for the IPWSO Cuba conference in 2019 and to fund raise for that conference by continuing to give concerts and through his contacts. Giorgio has been an inspiration to us by his example and

through his singing. I look forward to reporting in 2017 on the work we have done in the coming year.

Tony Holland



PWS Walk in Rio de Janeiro

Fundraising Report

Marguerite Hughes, Vice-President and Chair of IPWSO Fundraising Committee

Securing funds for IPWSO's work has been a challenge throughout its 25 year history and remained so during 2016.

In 2016 \$7,538 was received in subscriptions from 34 member countries (IPWSO's 70 associate members are not required to pay subscriptions). We are very grateful to all the members that pay subscriptions (and even more grateful to those that also give donations) as these enable us to provide support to parts of the world where there is very little help available for people with PWS and their families.

Our fundraising started well during 2016 with a much-appreciated \$20,000 donation from Pfizer International. The KB Andersen Foundation also provided good news by offering a \$15,000 matching grant opportunity for the second consecutive year. This meant that up to \$15,000 of donations from donors who had not donated to IPWSO in the previous year (or donations in excess of what donors had given the previous year) would be matched by the KB Andersen Foundation. We worked hard to secure this \$15,000 in "new money" and finally met the target and secured the full matching grant in December.

However, there was bad news too as Pfizer Europe, which has long been an important IPWSO donor, did not provide any funds in 2016. This meant that for the first time since 2004 we were unable to host an information booth at the European Society for Paediatric Endocrinology conference.

IPWSO's budget for 2016 shows that we achieved our fundraising target of \$55,000, although this money came from a very small number of sources. In addition to Pfizer International and the KB Andersen Foundation, most of IPWSO's fundraising income came from fundraising concerts held by Giorgio Fornasier and the generosity of IPWSO's current and past board members.



The IPWSO board recognises the need to diversify and find more sustainable sources of funding so in 2016 took the decision to register IPWSO as a charity in England and Wales with a view to becoming eligible to apply for funding from new sources.

The IPWSO board also recognises the need to expand its individual donor base. While it was heartening to hear at our 2016 conference how much support there is for IPWSO to continue to provide help around the world, IPWSO's challenge is to convince some of these supporters to provide financial help to make its work sustainable.

No reflection on IPWSO fundraising would be complete without recognising the huge contribution made by Giorgio Fornasier who, in addition to all his other work for IPWSO, has for years been IPWSO's greatest and most consistent fundraiser. Giorgio stepped down from his role as Director of Program Development at the end of 2016, but has generously agreed to continue to manage IPWSO's accounts in a voluntary capacity until the process of registering IPWSO as a charity is complete. Giorgio will also help IPWSO organise its 2019 conference, which will take place in Cuba from 27 November to 1 December.

Marguerite Hughes

Treasurer's Report

Hubert Soyer, Germany

As of the IPWSO Conference in Toronto in July 2016 I have had the honour to serve as the new treasurer of IPWSO. I would like to take this opportunity to thank Don Tallon for handing over a well managed balance to me, for handling all balances for the first 6 months of 2016 and for helping me compile this report. Further, I would like to thank Don and Marguerite for helping me catch up with my new role, Jim Gardner and Gerard Meijwaard for double checking all accounts and transfers and Giorgio Fornasier for providing me with timely and accurate information about income and spending.

I am very happy to present this report for the year ending in December 2016.

Developments in 2016

We've raised a total of \$ 56,986.32 in our US account and 28,250.75 € in our Euro account. Combined with an expenditure of \$ 50,121.95 in the US account and



34,217.11€ in the Euro account I am happy to report a surplus of \$ 6864.37 in the US account and a slight minus of 5966.36 € on the Euro account. We had 19,529.65 € and \$ 54,126.66 in donations and 5827.81 € in membership fees. I would particularly like to thank our major sponsors, Pfizer, PWSA USA, the PWSA Switzerland, Giorgio Fornasier and numerous other benefactors. A big thank you also goes out to all the members who contributed through their constant support and membership fees.



IPWSO Conference, Toronto:

Tony Holland, Dan Driscoll, Marilyn Dumont-Driscoll, Moris Angulo

Audited Financial Statement

BALANCE (2016) - Euro Bank Account	
Revenue	
Donations (Including IDONATE)	19.529,65 €
Membership fees	5.827,81 €
Others	2.893,29 €
Total revenue	28.250,75 €
Expenditures	
Bank charges	-450,50 €
Conference expenditures	-11.105,39 €
Staff salaries	-14.216,58 €
Others Outgoing	-4.844,64 €
BIRD Tests	-3.600,00 €
Total expenditures	-34.217,11 €
Saldo (2016)	-5.966,36 €
New Saldo (1.1.2017) (Saldo + last year balance)	43.677,33 €

BALANCE (2016) USD BANK ACCOUNT	
Revenue	
Sum of membership fees	\$0,00
Sum over donations	\$54.126,66
Others	\$2.859,66
Total revenue	\$56.986,32
Expenditures	
Bank charges	-\$385,32
Conference Expenditures	-\$37.409,63
Staff salaries	-\$12.327,00
Total expenditures	-\$50.121,95
Saldo 2016	\$6.864,37
New Saldo (0/01/2017) (Saldo + last year balance)	\$75.275,37

AUDITORS' NOTE

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 2016.

Gerard Meijwaard (The Netherlands)

Jim Gardner (USA) April 2017

Clinical & Scientific Advisory Board



Dan Driscoll, MD, USA

Clinical & Scientific Advisory Board for the IPWSO Annual Report



After the 2016 IPWSO Conference and change of Board members, the following people now make up our Clinical & Scientific Advisory board.

Membership of CSAB

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. 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 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: twins, gastroparesis, epilepsy, rectal prolapse, gastric bypass surgery, nutrition, physical exercise, “acquired” or PWS-like diagnosis, therapies, treatments and general information.

The request come from a range of countries across the world, for example, Hungary, Japan, New Zealand, Ireland, Australia, UK, USA.

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.

Other matters

Preliminary discussions are underway with our counterparts in Cuba in preparation for the Scientific Conference at the IPWSO meeting in Havana, Cuba in 2019 (November 27 to December 1) 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

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 20th to the 24th 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 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.

Professional Providers Caregivers' Board



In the PPCB Board, we have continued to communicate regularly via internet or telephone meeting throughout the year 2016 and used the conference in Toronto for our yearly face to face meeting with the board members.

We welcomed our new board member, Micha Pethe, who has worked as a team-leader in a group home in Switzerland for many years and has organized, along with the Switzerland PWS association, a variety of PWS events. Before he joined the board, he was the Caregiver Delegate for Switzerland. We are very happy to have him join our board and share his new ideas and motivation with us.

When we have a look into the future, we see several tasks and events on the horizon. There is the participation at the South American PWS conference in September 2017 in Santiago de Chile, where some

of our board members will support the conference with lectures and workshops and share their knowledge and area of expertise with this part of the world. We hope that it will be the beginning of a long and successful process in these countries and we will strive to support our South American colleagues as much as possible.

Lastly, the PPCB began to discuss the first ideas and planning for the IPWSO conference in Cuba, which I am sure will be a significant and impressive event!

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

Norbert Hödebeck - Stuntebeck PhD

Chairman PPCB



Famcare



Georgina Loughnan

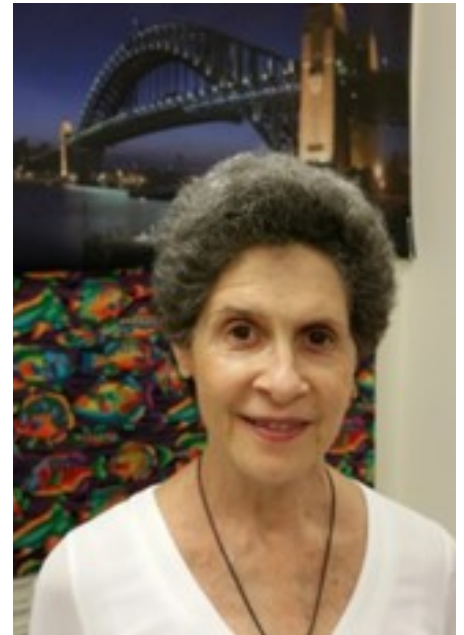
During the past year Famcare has added three new articles to our collection. “Transitional Years” addresses vulnerable times in the lives of adults with PWS. This is a huge topic and so we have attempted to address significant periods of change. By highlighting the importance of ‘attention to detail’ throughout the processes of transition, stress and related anxiety often experienced by the person with PWS, and their families, can be prepared for and hopefully, reduced.

Our article “Duty of Care” aims to empower families to seek out the best possible options for their person with PWS, with the knowledge that each person has the right to live and work in an environment that is appropriate to their needs.

“Personal Care” addresses the sensitive topic of daily hygiene and grooming. These seemingly simple tasks can be the cause behind much tardiness and related angst that is endured daily in the family home of someone with PWS, by all members of the family.

This brings the number of Famcare articles to twelve. We are continuing to work on having all articles translated into as many different languages as possible, in addition to English. At present we can provide several of our articles in Arabic, Dutch, German, Hindi, Italian, Russian and Spanish. We welcome any assistance with translations that you may be able to offer. If there is a language you feel we should be providing, please let us know!

Famcare is currently preparing a short information card on the importance of food limitation, for families to carry with them at all times. This card asks other people to assist you in limiting the access to extra food or drinks for your person with PWS, by providing a



“snapshot” of what PWS is all about.

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 conference or regional meetings.

A future Famcare project is session time within the next IPWSO Conference in Cuba in 2019!

We continue to welcome feedback and suggestions for future articles and services that Famcare could provide. Please communicate with us through the IPWSO website.

Georgina Loughnan

Programme Director's Report

Giorgio Fornasier, Italy

In 2016 we had to face the very high costs for the organization and participation to the 9th International PWS Conference in Toronto (Canada) in July.

IPWSO received during this year a generous grant from Pfizer, private donations from Austria, Belgium, Switzerland, Ireland, Australia, Sweden, Germany and especially from USA. Moreover I personally raised and donated IPWSO 6,985 Euro through my concerts in Italy.

We assist anybody who contacts us, whether they are members and pay a membership fee or not. When particular or complicated cases are brought to our attention, our Clinical and Scientific Advisory Board with leading international specialists on PWS assist families and professionals at no cost, even if the person contacting us is from the poorest country in the world.

From 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.

In 2016 we covered the costs of methylation tests made at the Laboratory at BIRD in Italy for the following countries:

Algeria: 1 test
Armenia: 1 test
Azerbaijan: 1 test
Colombia: 2 tests
Georgia: 2 tests
Jordan: 3 tests
India: 8 tests
Nicaragua: 4 tests
Pakistan: 4 tests
Peru: 5 test
Serbia: 1 test
Hungary: 4 tests

As said above, the organization of the International PWS Conference in Toronto was very costly to IPWSO and I noticed that the majority of our members could



Giorgio and Maurizia Fornasier

not attend because they couldn't afford to. We are happy that General Assembly voted for Cuba to host the 10th International PWS Conference in November 2019, where costs will be more reasonable and where we expect more families to join in, especially from Latin America.

We do need more help and financial support from our member countries and individuals. Any amount donated to IPWSO is welcome and can be transferred easily by using a wire transfer to our bank, sending a cheque to my address, using PayPal or Western Union and also the new I donate system you can access by visiting our website.

CIAO!

I decided to fully retire at the end of this year after 22 years I served IPWSO as Board Member, President, Executive Director, Director of Program Development and Accountant. I wish to thank everybody in the world that made my life richer and richer.

Good luck!

Giorgio Fornasier

Communication Coordinator's Report

Linda Thornton, New Zealand

One of the most exciting things to happen in 2016 was IPWSO's 7th International PWS Conference hosted by FPWR in Toronto.

With careful management of our income we were able to offer scholarships to cover air fares, registration and accommodation to the conference. We had over 18 applications and selected seven. Those selected were:

Gabriella Lemmers Vieira from Brazil, mother to a 4 yr old son, and one of the founding members of the PWS Association in Brazil. After the conference and with the information and support gained, Gabriella was able to announce that Brazil had now become a fully subscribed member to IPWSO. Congratulations Brazil!

Gloria Queipo MD PhD

Gloria is a Human Geneticist, molecular biology and is from Mexico. She has worked in the field of PWS for 10 years and is the Professional Delegate for Mexico. Her work covers most of the families in Mexico.

Marta Lopez USA/Spain

Marta is a MA student of Developmental Psychology, studying in the USA, and is the Caregiver Delegate for PWSA Spain. She is part of the multidisciplinary team supporting families and has worked over the past 5 years to help with leisure time activities and physical activity at PWS camps.

Asmaa Chakkour, Morocco

Asmaa is sister to her brother with PWS and is instigational in establishing the PWS Association in Morocco. After the conference in Toronto, she returned to her home and set about organising the first PWS conference in alliance with PWS France. She has become a lynch-pin in raising awareness in her country.

Fernando Briones, Spain

Father to an adult son with PWS, Fernando established the Federation for PWS in Spain, supporting families around the country and, importantly, in Spanish-



speaking countries around the world.

Simone Ito, Brazil

Paediatric endocrinologist at the Sao Paulo University, Simone was instigational in establishing the first out-patient clinic for patients with PWS in Brazil

Neil Gumley, Australia

A Direct Care Disability Manager working for the Department of Human Services in Melbourne Victoria. Neil is involved with the implementation and direct management of the first PWS specific residential home in Victoria.

We have welcomed Brazil as a fully subscribed member country, we have also welcomed Morocco as a newly formed Associate Member and we also welcome back Singapore under a revived leadership. Of course our aim is to have all our Associate Member become fully subscribed members and hopefully we can support many more new Associations in the coming year.

Requests for information keep me busy and my thanks go to all our IPWSO Boards and IPWSO Advisers who help find answers to those many questions!

Linda Thornton

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.