



IPWSO
International
Prader-Willi Syndrome
Organisation

IPWSO Health ECHO abstract

October 12, 2021: Caring and Living with Prader-Willi syndrome in Italy

Please note this document is abridged from audio transcription of the Zoom session. Some errors resulting from the transcription process may be present.

IPWSO hosts: Georgina Loughnan (GL) Tony Holland (TH)

Presenters: Maria Guilia Marini (MGM), Risto Lapatto (RL)

Video link

Maria Guilia Marini, [Caring and Living with Prader-Willi syndrome in Italy](#)

PDF available [here](#).

[Q&A Following Maria's Presentation](#)

TH: Maria, I want to go back, right to the beginning of your talk, which you refer to the idea of disease illness and sickness. I don't know whether this is so in Italy, but in the UK, we would think of a disease as something being curable, or at least treatable and I wonder whether that is the right language for us to use when we talk about a condition that you're born with and you're going to have for the rest of your life. I know you were using those terms because that's what's used in this academia, but I wonder whether we shouldn't be talking about, maybe "condition", possibly "disorder" although that's problematic. Do you think there is something wrong with the concept of using words such as disease, referring to something like Prader Willi syndrome?

MGM: I would say disease is like not to be able to do something or not to be at ease to do something. So, probably we have put the medical brand on it. Well, this is the etymon. I don't know if you know Oliver Sacks. Sacks' focused his efforts not on the things that you're not able to do, but on the things that you can do, and how to develop the other abilities. I think that this is possibly the way to come out from a very simple reductionist model.

Because this is saying that there is something which is deviating from normality. We have to align what we consider normal; I hate the word normal, I can tell you. I wrote a chapter in my book Narrative Medicine stigmatizing the word normal.

But let's consider the disease so we may consider that maybe we should invent a new word, which is the contrary of disease, that may be "at ease". You give a good point, because otherwise we're always focusing on the biomedical.

TH: It's interesting because some of the organizations in in in this Prader-Willi world, they have as their campaign logo, "finding a cure" and I always have trouble with that. I think we should find solutions to some of the challenges experienced by people with Prader Willi syndrome. I'm not certain we should use the phrase "finding a cure" and that's why, as you know better than anyone, language matters a lot.

GL: It's just a little bit like the term disabled. I know some people from India who never use the word disabled it's always differently abled, which is such a nicer term.

MGM: Answering to Georgina, well I love the concept of multiple abilities. So maybe we should focus on this. And I just want to give the voice to our people living with Prader Willi; just a few percent of them wanted to have a cure. They want to have marriage, children, and all this, they were like in a dream scape. They are happy as they are, if they are acknowledged as they are.

Last week I was listening to a narrative of a child who was already born with the rare disease. So, this rare disease would have been bringing her to Nyctalopia or not to see things during night to see blurred color, blurred vision. Well, the prognosis of this disease is that maybe one day she should become blind. But what was funny in her narrative she was saying, "I was feeling, absolutely normal with my color with my blurred vision so it was the others who said that I was not normal, so the others made me feel not normal."

So, no I don't like the wording "finding a cure" because we cannot say. But this is just epidemic.

I have a question for you - Do you think that maybe it would be wise to understand better the, the leading and caring of the people with Prader Willi, and the relatives in such a multiple voice way also in other countries?

GL: I agree. I think so definitely

TH: I think it's a really interesting idea how much culture makes a difference here. I have to say my suspicion is that it may not make as much difference as we think it might. I don't know why I think that but I think it's a very interesting question.

MGM: I know that there are also some healthcare providers here, what do they think about this kind of study? Because we have a huge debate between what is evidence based, what it is not evidence based, this is done according the guidelines of the World Health Organization, we didn't want to stress it, to point it out, according to the guidelines of narrative research from 2016 by the WHO they are the same level of an evidence based medicine study, and the quality is given by listening to the different points of view and interpreting the text. So really being like the translator of this text.

I don't know if you think that they might be useful or, this is just that you know something ranking lower in comparison to other research. I think that it has to be integrated definitely, so they are the same level, and they intertwine.

GL: Thank you Marie, you've given us many things to consider, and especially from a care givers perspective, I think they're at the heart of having more time to do all the narratives, which I think is fabulous.

Case presentation (abridged)

GL: Risto Lapatto is a paediatric endocrinologist from Helsinki University Hospital, Finland. He will introduce himself a bit more during his presentation and also give information about the Finnish healthcare system.

Case presented: PWS in Finland – A case presentation illustrating what works and what does not

RL: I'm a paediatric endocrinologist and have been seeing Prader-Willi children and also adults for a couple of decades and so I'm very pleased to be able to join you and have enjoyed these talks very much so thank you very much for organizing them and hopefully one day we will meet face to face.

Slide: Early diagnosis and start of treatment

- Healthy parents, uncomplicated pregnancy, delivery normal, 1st child,
 - The girl was floppy, and PWS was diagnosed (UPD) at 2 months of age
 - Paed endo team took over the coordination of care

 - GH started early, therapies (physio, psych, speech, occupational)
 - Evaluation by other specialists (ENT, lung, neuro, etc)
 - Mother very supportive
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RL: I am going to present a case that will show some of the problems we have here and some things that work well. So this girl was, was the first, very eagerly wanted, child of a family, and turned out to be very floppy at the beginning and the investigations are quite extensive and fast these days because of some neurological diseases that require treatment very early if you want to be successful so the floppy babies are really investigated rapidly and this was 20 years ago but even she got her diagnosis in two months so I think nowadays we do much better and probably I would say that 95% of PWS cases are diagnosed before first month of age. And we tend to do the genetic diagnosis at the very beginning so that then we can explain the parents what is going on.

Then the care is typically taken to the endocrine team, because of the growth hormone but then the problem that I'm seeing these days is that the younger generation they are more and more focused on one particular area of medicine and people with PWS

really need lots of different things and I think the sub specialists care is not always the best and will speak later of my work with the PWS Association of Finland.

Often they turn to me and ask me for a second opinion or complain that whoever is taking care of the child or the adult is too focused on, for example endocrinology rather than the whole person, so this is a little bit of generation gap, I'm seeing these days in Finland and the younger doctors they are more focused so there's something that needs to be done in this respect, and then the growth hormone is nowadays started very early and we do have the therapies available as needed.

We have to remember that people with PWS, at least here, are very different. So, some are very capable and basically attend normal nursery schools, etc, with very little extra help, whereas others are almost out of bed patients and luckily very few these days so this has changed but still the, the variation among people with PWS is quite enormous.

We do the ear, nose and throat evaluation, and lung function. The sleep studies here are organized by the lung physicians so we do the sleep study. Of course, the neurology etc. So this all gets done and then the follow up is as required.

And of course the families are very important and in this case the mother, who actually is a midwife, she's been very supportive to her daughter over these years but, one unfortunate thing that I have recognized here is that if a child has a chronic condition, it can be cancer or asthma or diabetes or PWS or whatever, the rate of divorce is very high and also in this case the parents split up and I'm sure there's some psychology that the stress gets too much, but it's really sad. There are things that the doctor wonders like, can I do something? Of course, there are many things that the doctor has very little to offer, but just can listen and be supportive.

I think that we probably would need almost a new profession of people who would be the care giver, like also somebody who works with the patient all the years and supports and coordinates things etc and doesn't have to be a doctor necessarily.

Slide: Finnish healthcare system and PWS

- Experience variable
- Special services for mentally handicapped
- No centralisation
- Prescribing restrictions
- Paediatric vs. adult subspecialties
- Integration of other services: education, social etc.

RL: The Finnish health care system, I won't talk about it much because it's changing all the time. With PWS sometimes there is an anti-centralization philosophy here so it's not possible to say that PWS is treated in this place and that's it so basically, it's treated everywhere. And this leads to the problem that all rare diseases, and even not so rare diseases, the experience of doctors and nurses gets very variable around the country.

We do have special services for mentally handicapped but certainly not all PWS people go to that service, at least not in the beginning, and that also has some problems that it is not evenly distributed with resources around the country.

The prescribing is restricted so that you need to be a paediatric endocrinologist, to be able to prescribe growth hormone so is why these people are treated by the endocrinologist, but the endocrinologist could of course be the consultant and someone else be the prime.

Then we have a big split between paediatric and adult subspecialties. In paediatrics we all work together so whether you're a paediatric endocrinologist or haematologist or whatever, we are all in the same paediatric hospital working together, and it's very easy to consult, or refer a patient to a colleague. Whereas the adult subspecialties have basically their own companies and if the adult the renal patient needs an opinion of, for example heart surgery, then they need to refer the patient to a totally different unit and this patient has to travel etc. So that gets problematic with people who have a multitude of problems so if you just got one single problem then it's easy on the adult side, whereas the people who have more than one problem they have difficulties because the care is split into so many different subspecialties.

Then, we try to integrate them, the medical service with other services like education and social work. This sometimes is successful and sometimes it's not. I think the more you do, the better it gets but you need to know the other players and what they can do and what's possible but if you set up meetings and you talk to these people then the patient usually benefits so I think that is something that we have to pay attention to and that's the direction we try to move with our health care, that we are more in collaboration with these other services.

Slide: Finnish Healthcare system and PWS

- Public (Tax, compulsory insurance)
- Health centre (GP)
- District hospital (secondary, 25)
- University hospital (tertiary, 5)
- Private (self, voluntary insurance)
- Mostly just outpatient clinics
- Area where you live is important
- Some freedom of choice

GL: We also have this strange division between private and public sectors. So public, using taxpayers money, is divided so health centers where a GP type of service is given and then we have district hospitals and University Hospitals and all children with PWS end up in either district hospitals or University Hospitals because the University Hospitals have their own district so that that means that are secondary services for that particular area. All these will have a pediatric endocrinologist taking care of the patient. But when the person becomes an adult, and luckily it's not very strictly defined, when somebody becomes an adult, so we can treat them when they are

young adults, up to 25 or so, in our hospital. But at some point we need to move them out. Unfortunately, there's no adult service available in these hospital settings so they end up in health center, unless they have other medical conditions that would qualify for hospital service. So most of the adults with PWS are looked after by GPs and then we try to help them. But it would be very nice to have an adult service for them.

The private side can compensate this, to some extent because some doctors even though they work in public, they also work private. Then, if the patient is willing to pay, they can then see whoever doctor they want. But the private is of course very limited and it's basically just appointment and that's it. So the other services that we need are not that easily available in the private sector.

Slide: The girl grows

- General health remains good, weight ok
- Manages at school with extra help, completes secondary school (9th grade)
- .
- Spontaneous puberty to Tanner stage 3-4
- Independency is encouraged and she trains to travel alone, etc.

RL: So in this case, the girl grew and her health was always very good. We don't have a weight problem in children, and not very much in adults either so I think the understanding and the control of food and certainly some treatment, maybe the growth hormone does play a role. I've been very pleased to see that the growth and the weight gain in the patients that we serve here is very good.

She did manage to complete Secondary School, which is nine years in Finland, the first or second grades together, with extra help. This is nice and several PWS people have done this and are going to do this so they're doing very well in school, with the help. But the problem is that school gets competitive after that and there are some disappointments when they, they get some dreams or the parents may have some dreams of what the child can do and then once the school gets too difficult than whatever extra help you give it's not enough.

So what are they doing after the ninth grade is sometimes a problem and this needs to be thought of ahead of time so that they can prepare themselves. Families need preparation. There are educational services available and that is again very much dependent on where you live.

She also entered puberty spontaneously which is a nice thing and then it got stuck which is another disappointment that everything's looking good and then suddenly stops. This tends to happen quite often, of course the boys do less well in this sense than girls. We encourage all young patients to become independent, teach them how to treat themselves and what they need, and this girl also became sort of independent and learned to use the bus herself. But this then led to a situation which could have been catastrophic because she is so nice and kind that somebody just talked to her and

then she was discovered two days later in another city so somebody needs to protect these vulnerable young teenagers who look like adults but are not.

We are not sure, we believe that there was no sort of sex or misuse or rape, luckily but this is very difficult how much freedom we can allow, versus to control what's safe and what's good. So this is one issue we have because our law is very strict so you cannot restrict people so they are allowed to go and do whatever they want basically and even young people who are less than 18. So 18 is no magic number in law here. If the child is considered capable of making decisions they can make decisions earlier. As we're now seeing with the vaccinations to Corona so everyone 12 and up can decide themselves whether they take it or not.

So this is an issue with PWS because they are very kind and nice people and if someone talks with them they are, they can do risky things that they shouldn't do.

Slide: The adult

- She is healthy
- Lives supported but alone

- Studies in a special occupational school, what next?

RL: Now, years have moved on, she is now about 20 years old and is very healthy and lives in a good place because the PWS Association, in cooperation with a building company bought land first and then they constructed the building. But we have these special homes for PWS people and they are, of course, fantastic, if you can get a place in them. That's limited and I think there are three or four in the whole country. Some PWS people certainly live in other places, but these are good because they get peer support and there is enough support but everyone's got their own little apartment and can do alone, what they want to do, and they control the food, they eat together and so on. So these have been successful but it was all thanks to the association who took the initiative and started this risky project but I think even money wise, you don't make a win but you don't lose a lot of money so this is good and I hope that in the future we will have more of homes like this.

She also studies in a special occupational school, and these studies will be completed next year. What she will do then is unknown, so whether anybody will employ her or whether she will get what's called self-safe employment from the city - they employed people with handicaps and the people get the feeling that they do something meaningful and they also get a small salary so that remains to be seen.

Slide: The Finnish PWS Association

- Well established
- Council: Active parents and grandparents
- Activities: Meetings, camps
- Most families are members

- Collaboration with some housing companies
-

RL: Lastly, I'll just mention this Finnish PWS Association. I don't have a role with them but I do volunteer work with them so I attend some of their meetings and camps and they can get in touch with me and I can provide a second opinion for free and it's like goodwill work.

I'm hoping to find someone to take over when I retire that it takes some time but it's of course very nice to meet these people and know them and meet them also outside hospital and just have a chat and you know play with them that's all good. But the association is well organized and well guided, and they are active. A generational thing is that usually the active people are the ones who have children and then once the children become adults, they lose a little bit their interest. But we have had new people coming in and it's not a big organization but I think it is one of the best patient organizations in this country that I know of, and certainly also has connections with the international organization, and helps these people, a lot, who are spread out over the country which is an area a little bit bigger than the United Kingdom, but the population that is 10% of that so you can imagine that you having the two meter safety distance is easily done - we used to be 10 meters away from each other.

So, I think I would be happy to discuss this case with you and you answer any questions you might have.

[Discussion following Risto Lapatto's case presentation](#)

RL: Certainly the thing that I'm thinking is who should be the responsible person, who would be the best person to be in charge for PWS? It may not be the same person throughout the years and maybe each child needs a different person than an adult, but certainly my concern is the adults so I find it appalling that that adult services are basically non-existent. Also should we allow these people to walk free without any guardian with them or not and can we take the risk?

Comment: Thank you for your presentation, we have almost the same scenario except that our organization is not so active, for now, and we don't have the opportunity to send our adult patients to residential care units and we don't have good proceeding of their life after the age of 18. I feel the same thing with adult endocrinologists because, for example paediatric endocrinologists see the patient from the very beginning, from birth or from the third or fourth month when we establish a diagnosis and we start to to establish connection with the parents, we see them every three months. And we are very into their situation. But when they become the age of 18, when transition has to be made, the adult endocrinologists don't know what to do with this person. You don't have residential care, he's like a hopeless situation, and they just do not know what to do with them because in Bulgaria, we don't have growth hormone, for patients over 18, and this is also a big problem, and I'm trying to, to help those patients to go to their

endocrinologist just once a year. Sometimes it's impossible, especially now in the Covid time. So everywhere, it's very difficult for patients that are becoming adults.

TH: I thought that was a very nice talk because you illustrated all the issues that I think all of us have experienced, perhaps to various degrees. We have the same sort of stories in the UK. One of the things that we do have for adults is what we call community interdisciplinary or multidisciplinary community teams for adults with intellectual disabilities, and these are teams that will consist primarily of nurses, social workers psychiatrists, psychologists, speech and language therapists and, and others whose job it is to try and support people with the whole range of disciplines of intellectual disability in the community working with social care providers. The problem for adults with Prader Willi syndrome is that often they are assessed as not having an intellectual disability, and that's rather crudely down on the basis of have an IQ above 70, so some people with Prader Willi syndrome are not entitled to support from that team.

But I think if they are entitled to it, some approach like that can bring this more multidisciplinary perspective to the care and support of people with intellectual disability including people with Prader-Willi syndrome. But none of it's of any value unless you have access to good social care, either extra support in the family, or specialist support outside of the family. Because certainly in adulthood, if you haven't got the social care right then whatever you do from a health perspective, you're probably not going to help them. So I think for me, social care is critical, and then a multidisciplinary service is also a necessary part of what you offer.

GL: I probably should explain, in Sydney, in Australia we set up an adult Prader-Willi clinic 30 years ago, this year. That was in response to the fact that the children were now, adults and still attending the paediatric clinic. And it comes under the umbrella of metabolism and obesity services so it is run by endocrinologists, but we are in a public hospital. So, I'm a physiotherapist, but we have access to the sleep unit and the respiratory doctors. Although we're not all in the one department we're all in one hospital, and it's public so it's free. We've been able to use our service as a lobbying service in a way, and we've professional backing to plea for government support and also care support for people with Prader Willi syndrome.

We've tried to establish these in other places in Australia but they say it'll cost too much whereas we did not put any money into this clinic we just took hours from the different disciplines to work with patients with Prader-Willi and gradually built it up from there. So, I believe it can be done and I don't think you need a fortune, you just need people who will take the time out to start seeing people with PWS, and bring in other disciplines to help, and to learn about PWS.

Comment: From my research, we support 88 adults now with PWS across various residential services and there are other providers in the UK, so I estimate about 200 people, 200 adults are supported in some kind of residential setting with food and

money is managed. Health outcomes are difficult to quibble with. The health outcomes they tend to be very, very good. The challenge we're facing now is as a provider we've got people within the organization questioning the restrictive practices that that very model produces.

So we've been working with some of our people and educating them around the risks of maybe chipping away these restrictions. But it's something of course we should be doing anyway, we should always look at the least restrictive options. But concern as we start to chip away at the model that we refer to, is it will see less better health outcomes and maybe an increase in behaviour for some people.

GL: Well thank you everyone and thank you Risto, I think that it's such a topical subject that you have brought up. And one other thing I that our Prader-Willi syndrome research foundation is trying to establish a Center of excellence for Prader-Willi syndrome in Australia, so that would have a holistic approach but it's going to take a long time.

Note: Further information on [Transition](#) is also available in our [Caregivers' ECHO resources](#).

Tony Holland Summary

I thought Maria's talk was very, very interesting and insightful because it comes at the whole topic of Prader-Willi syndrome from a very different perspective to one that, as a medic, I'm used to. She started by saying a bit about the study, and the importance of the partnerships in undertaking this to study. She described the different ideas around a Narrative Medicine approach as being either tools of understanding or tools of education and I thought that was quite an interesting perspective.

She talked about disease, the term disease being the definition, illness being the experience and sickness being the social and cultural meaning attached to that particular disease and an illness. Then she reflected on how that perspective applied to our understanding of Prader-Willi syndrome, and her study was really looking at what was described as the transaction between different actors. And so she looked at the perspectives of the people with Prader-Will syndrome, both children and adults, and she had 241 narratives and 59 follow ups. She looked at the perspectives of family and the perspectives of health professionals.

This wasn't surprising but it was nice to hear about it, that when people with Prader-Willi syndrome were asked what they wanted from life, they had a really quite romantic view and goals. She also described quite a complex relationship with food which I don't think would surprise us but interestingly, subtle difference in the way that men talked about what they eat whereas women talked more about how they how they eat. I don't know whether that would be true in the typically developing population, it might be.

But, I thought that was an interesting observation. They talked about their illness and discrimination, but also reflected on thinking about the future.

She then talked a little bit about how things have changed with Covid. When she moved on to talking about the family, I thought one of the most moving bits was the issue of communicating the diagnosis. And this was before Covid, and the diagnosis had been communicated over the telephone and in ways that really were not appropriate. Maria used the phrase cruelty of language, and I thought that was a very powerful phrase. Then she went on to discuss how in so many ways Covid has changed the circumstances for people with Prader-Willi syndrome and that of their family.

And then she went on to talk more about healthcare providers and the different perspectives that healthcare providers had that to some extent clashed with the perspectives of families, and how they saw telemedicine in actually a fairly positive light.

Then we moved on to Risto's presentation. His presentation was around the case of a young girl who's now an adult, with Prader-Willi syndrome who classically was floppy at birth and although this was 20 years ago it was diagnosed, very quickly, and how her care was primarily through endocrinology.

But then, Risto identified the classical problem that I think we all recognize, of when you need the expertise of other disciplines, how do you do that and how do you get that? He expressed concern that doctors in Finland, and the same is true here in the UK, are getting more specialized, so it becomes more and more difficult to provide a holistic approach.

He reflected on some of the issues around the Finnish healthcare system and how people's experience was variable. It wasn't centralized and the same is true in this country, that there's a divide between pediatrics and adult, and at times poor, not always, but can be poor integration with for example, education, and so on.

He then went on to describe how the person was generally doing well, was not overweight, had spontaneous puberty, but it hadn't gone to completion. I thought a really interesting point how in secondary school as the school becomes more competitive, then it becomes more of a struggle for the person with Prader-Willi syndrome. So, in a way, their skills perhaps aren't sufficient to manage the more complex environment of later school life.

He reflected on how basically she's doing well, living in support set up by I think the Finnish Prader-Willi syndrome Association, but she isn't alone. But there is fear for her future and for the opportunities, and particularly this tension between freedom and independence, and the risks that come with that and which there are no easy solution.

So I think they were the main, main points that I got from the two really interesting in talk, so thank you.

Upcoming Health ECHO sessions

Tuesday 9 th November	<i>Socialisation and Communication in People with PWS</i> Norbert Hödebeck-Stuntebeck, Psychologist, Diakonische Stiftung Wittekindshof, Germany
Tuesday 7 th December	<i>Orthopaedics in PWS</i> Prof. Dr. Ralf Stücker, Altona Children's Hospital, Hamburg, Germany

Thank you very much to everyone who attended the session and participated. We look forward to seeing you on Session 9 in November.

Ends.