



IPWSO
International
Prader-Willi Syndrome
Organisation

IPWSO Caregivers' ECHO abstract summary Mar 23rd, 2022: Managing routine medical checks and other regular requirements

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

IPWSO host: Patrice Carroll (PC)

Presenter: Susanne Blichfeldt (SB)

Video link

["Managing routine medical checks and other regular requirements in PWS"](#) Susanne Blichfeldt M.D, Denmark

PDF of the PowerPoint is available [here](#).

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

PC: Thank you, Susanne we're lucky to have you. I'm going to start, I have a question for you, Susanne. We're seeing more and more people with PWS age, and we have a number of individuals who are over the age of 50. Is there anything new that we know about the aging process in people with PWS from a medical standpoint? Are we seeing trends, is there anything we should be paying closer attention to? We really haven't seen people with PWS in their 60s and 70s, until now. So, what should we do differently?

SB: Well, it's an often-asked question, and we do not know so much, we do not have many surveys about the oldest. But I think that medical checks, as I wrote is important to have. Also to measure for example the thyroid hormones, because we know that also in the general population the thyroid hormones can go down. Another thing, and this is my own impression, if the sex steroids - for example in men, if they have very low testosterone, I have the impression (but I have not documented for this) that they look much older.

But then again, it's also often asked about the mental state and dementia. I think you should be very careful about that. In Down syndrome, we know that Alzheimer's disease is common, but in PWS we do not have documentation for that and I have had more cases where the staff suspected it could be dementia and when it was

investigated, it was not dementia, but it was the depression. And I think you should be very much aware of if depression can rise with age. Because when you have dementia, you lose the ability to do things that you could do before. But if you have depression, you can still do it, but you don't do it, because you are depressed. So, it's very important to distinguish. But I just think we have to follow them carefully and of course, perhaps they are aging sooner than others but we do not know.

My son is 42, he is not overweight, when I walk with him in the forest he is walking faster than I am. But I don't see any aging process coming so soon in him, but I have seen it in others. And I do not know if there are any chromosomal differences. There's so much to look for, and we do not know, but I just think, careful medical examination and be aware if they are depressed. Another thing here about depression and inactivity, it is the age when the parents die. And I think many with PWS are very much connected to their parents. And if they lose both parents the sorrow can come delayed. We have seen that also with other people with mental problems, and perhaps after some months, they all of a sudden change their mental state or something. So be aware of this, what has happened around them, and they can become more passive. We are facing a new situation with all those becoming old.

Q: When you had your stomach slide up, we're talking about the stomach. Is there any reason why, if you were worried about the child or the adult having consumed a large amount of food, why you wouldn't use medication to provoke vomiting?

SB: Because it doesn't work. We have seen cases where two even three, four, or five doses and they do not vomit. And I think the reason is that the muscle tone, you know, there are smaller muscles around the stomach, and the stomach wall, and the muscles are not strong enough, these muscles, to react to this medication. So, what you see when you give this medication that usually works in other children, but those with PWS they do not vomit when you give it to them. So you should not stand and wait too long time before you empty the stomach with a tube.

And another thing is that you give sometimes when they have eaten something dangerous, then you sometimes give active coal in really acute situations. But I think you should be very much aware of this distended stomach because the thing is that they do not complain, they just say, "Well, I do not want my lunch today." We had one case in Denmark, a man, he didn't want his lunch, and the caregiver insisted, she called an ambulance, and he walked into the ambulance, and he came to the hospital and she still she had a paper (for the doctors) about PWS, and insisted to have a scan, and he had the hole in his intestines and he was operated on, and he's still doing well, I'll see him next weekend, and he is more than 50 now. So, and it was also published on the IPWSO website. So, you have to insist that this threshold is very high, and they do not complain. But not wishing to eat, we have seen situations where they resist eating because of some psychological situation, but not if they just say, "Oh, I don't think I

want to eat." We had another example, a young man, he tried to explain to the caregiver that he did not want to eat because he was at the dentist yesterday. He looked for a reason that he did not want to eat but actually, he died the day after.

Comment: My son has also, when you were talking about constipation, he suffers quite badly with constipation or faecal compaction. And the only signs I ever have is that he will leak. And then I just know that he is having an episode, otherwise I'd have no idea. No pain, nothing else.

SB: Also, those who came to the Danish hospital with distended stomach, we saw in many cases that sometimes the reason was constipation. But to my experience (but again it's not documented, or scientifically evaluated), but I see the problem is much bigger in the adults than childhood. And then again it can give rise to and rectal picking.

Q: About sleep apnea. You were talking about masking. How accepted have CPAP masks been accepted by people with PWS in your experience?

SB: Personally, I know only a few cases of adults wearing masks, but they accept it very well. It happened to be rather high functioning people. But more than 10 years ago there was a Scandinavian study about growth hormone deficiency in adults, and they all had a sleep test done. and among 40 people was found three cases who needed a mask, who had a central apnea. And they all said after they had the mask that the daily function became much better. And there was a mother in Norway, who lived with her adult daughter, and she said, "I just got a new person in the house.", she just became much more active because she had much better sleep.

But you should be aware if the sleep apnea is a so called obstructive, I have not written so much about it, but you have to go to the doctor who check the mouth and throat, in case it is because the passage of the air from the mouth into the lungs is blocked by the tonsils, for example, and then you have to remove the tonsils. Also you do such a sleep study in all children before you start growth hormone because the growth hormone can also cause swelling of the tonsils. So, you have to make the distinction between the obstructive, and the central apnea.

When I think about it, I know a case but I don't do not know him very well, and he has many psychological problems, but the mask was accepted perfectly. And they get much more energy, if they have enough air during sleep.

PC: Several years ago, we had a handful of older women who had a sudden onset of hypothermia. Do we see that in other people? Is it an age thing, is it a gender thing, or is it just a random?

SB: Well, I have not experienced cases in Denmark. But I know in the Clinical and Scientific Advisory Board, it has been mentioned many times. And I think you have to

be aware that, you can have this hypothermia together with depression. But it has something to do with the temperature regulation, and again we're back to the autonomic nervous system, and there's so many things that we do not know here, but we have to be aware of it. In these cases, I would also ask if they have the hypothermia because of hypothyroidism, but I think in the States you measure the thyroid hormones. Also, I do not know if it's about the estrogen, do they need estrogen, usually in an older woman, you do not produce so much estrogen, it's a normal thing in your body. So, you just have to check everything. It is a PWS thing, I have not mentioned it, I should have mentioned it, it can happen.

Comment: I would just mention that as they get older, I think we have to remember to look for the common other health issues that are unrelated to PWS that that we would see in the general population like cancer, and yes, probably dementia as well, but we have a patient who developed Multiple Sclerosis, and it took a long while for them to decide what that was, but now she's on medication and doing very well.

We've had a couple of people die from cancer. One man was losing weight very well because he had lots of exercise and a good meal plan, but then he started to lose a little bit more weight and everyone thought, "Isn't this wonderful?", and then we thought, "No there's something going on." So just the simple things that we would notice in other people, don't forget to look for those in people with PWS as well.

SB: Yes, thank you so much, of course it's so important to think about how with age, we can all have many difficult situations and more and more diseases can arise, and of course you should not look for only PWS things.

I have another question for you. As you know, we are a small country and we have only around 140 person all in all, with PWS in Denmark and half of them are adults. But I made a very brief survey last year, where I asked about physical activity in the primary homes, and it came up that there lived, much more women with PWS than men in our country. It was a double then amount of women than men, and we could not explain it. And you know it's a small number still, but do you have any experience around that you have more adult women than adult men with PWS?

PC: It's about even. Years ago, we would have more males in our children's program only because they're placed more frequently, because some of their behaviours look a little scarier than the young girls, so we had more boys. But I don't know if we have more males in our adult program.

Comment: I can answer that Patrice, we have more older females, who are 40 to 67. And then the males are, we have 2 older males in that age group and the rest of the males are around 30 to 45.

SB: We looked at recent death. And there was no difference between men and women. The eldest that we have in Denmark, she's 71 now. She's doing fine.

PC: And actually, you're right, because, I know I don't think anyone from Oconomowoc (PWS home in Wisconsin, USA) is on here, but they have far more women than men in their adult program.

SB: And then I then again I ask, are they treated for their hypergonadism? I think it's so important that the testosterone level is high enough, not for sexual activity but for body function and this I have to mention again, that these hormones are given not for being sexually active and I do not think it influences that. In some cases, perhaps, but generally, it's for the body function, and it's not for only looking like a male but it's for the muscles and bones, and the inner organs, that testosterone for men is so important. If the men do not have enough testosterone than they can have anemia, too few blood cells and so on.

PC: Sometimes, people are hesitant because they think that it will increase aggression in males. Have you seen that happen? I haven't either.

SB: No, no. And we have discussed that in IPWSO and I remember 20 years back when we in Scandinavia talked about it and other countries they were more, much more hesitant. And therefore also, you know, if you give some testosterone and then you have a behavioral problem, then you say, "Oh it was because of this testosterone.", and you take it away. But it was perhaps because there was a change in the group home, the staff was new, or something else.

Q: I have 2 questions. The first is that we have reported more and more choking, especially with the aging people. It's very important that caregivers must be aware of that, it is possible for many reasons. But can choking increase with the use of psychotropic treatments?

And second point, you mentioned incontinence and this is also reported more and more, and do you have any suggestion, what can we do for that?

SB: I often visit where my son lives, you know, that's personal experience, and I think many doctors should do that because I see some of them are chewing perfectly well, and some are sitting chewing with open mouth, and the saliva dropping out of the mouth while we're chewing, and then they are not chewing sufficiently and then they swallow too big pieces. That's one thing. But if you have too high doses, or perhaps you need high doses of psychotropic medication, then the muscles can be affected. Again, and your chewing is bad because of bad muscle function, and then you can worsen it with this medication. So you should be very careful. But this is only a personal recommendation - I would say, if a person has bad chewing function that you know about beforehand, and then you give medication that can further relax the muscles you can perhaps cause a bigger problem.

And about the urination. They do empty the platter, that's our experience, when they go to toilet. But before they go to toilet, the bladder can be very full. And there is a

recent publication from China, where they looked at the bladder function, and they saw that they empty the bladder, but they do it very slowly, in some cases, and also they have very high volumes of urine in their bladder before they go to toilet.

So I think, to avoid this incontinence, the first recommendation as we give for small children, is to have fixed hours for going to toilet. And again, also my personal experience with my son, I sometimes say before we go to some event or something, "Shouldn't you go to toilet?" and then he says, "Well, I do not feel that I should.", and then he goes to toilet and then when I ask him he says he urinated very much. So he himself is very much aware of it, strangely enough, that he does not feel that he needs to go to toilet, but he does because it's needed. Then of course you can have incontinence arise with overweight, and also perhaps with age, because we see old people also having incontinence because of age.

So, fixed hours for toilet. Also, if they have free access to water if they drink, a lot of water before they go to bed it can also be a reason for the problem. And then again, if you give medication to avoid urination during sleep, then be very careful that they do not drink too much before because what this medication does is that the body does not produce urine and therefore you dilute the blood and the level of the potassium becomes too low and then you can have a seizure. So be very careful with medication, and also in many cases, you have to restrict the volumes of water ingested.

Comment: I'm a psychiatrist and although I primarily am working with children right now, I'm also boarded in adult psychiatry. So I wanted to comment on the use of psychotropics. I think that, yes, you need to consider whether it's going to affect their saliva production. So many of the medications are anticholinergic and therefore you might have dry mouth associated with that. So you want to think about, watching when an individual is put on a new medicine, whether there's even less saliva than they've been making before. I agree that there may be issues with psychotropics associated with chewing and swallowing, there can be changes in mouth movements associated with psychotropic so you want to be watching for any type of unusual or different types of mouth movements, and that may inhibit or get in the way of being able to swallow well and swallow better.

And then the other question about some medications, again, can cause urinary retention, so it makes it harder to go pee. And so you might want to be thinking about that, if you're concerned about incontinence or that would cure your incontinence. I mean we sometimes treat bedwetting in children with a little bit of anti-depressant type of medicine to keep them from bedwetting. But in an older adult that may not be a good thing. So, just wanted to give a heads up on how psychotropics can have side effects and you have to balance is it helping enough versus is it hurting?

Q: I would like to ask if you have recommendation for a minimum time of physical activity every day. And it's a good idea to do it every day the same time? And what is the best motivation technique?

SB: Well, I would refer to Georgina's presentation ([The Power of Exercise for People with PWS](#)) also, but I recommend to all that they walk. The best thing is to walk half an hour after each meal. But I think it's very individual what plans you can make. One thing is that I experienced is that, and all parents say that and many caregivers say that they love to walk. And when they walk, they talk, and you are told many things. And you do not have to choose a new path every day. For those who like to walk, it's a good idea to walk the same path. Where my son lives, each morning after breakfast, they all go, what they call roundings, and then they go to the same, all of them, and they talk, and they enjoy, and it's a part of their daily life. So I think it's a good idea, in the home to have a plan that you walk every day, and walking is the easiest way to move your body, but I do not know if you have any more thing to add about this Georgina?

GL: I think as Susanne, we say that to do at least 30 minutes, at least five days a week, maybe six days a week is better. I always say have one day off, it depends on what they're doing, but walking is very good, aerobic activity is very good, but also strengthening activities are very good for their muscles. So, I actually say that exercise is not an optional extra. It's just as important as the food restriction. When doing anything we start small and gradually build up.

SB: I know one, she has a competition about how many steps, she did in a day and she has a phone call with her mother every week and then they have a small competition about who walked the longest or the most, so it can be a stimulation to count how many steps you have done for the week.

Q: I had a question about the exercise and the aerobic piece because some individuals walk faster than others, so in terms of raising their heart rate for the weight loss benefit is there a certain, I know depending on the individual, but like percentage rate like above their resting that they should be getting for the benefit of the exercise?

GL: It's interesting because I was just reading an article today on heart rate and does their heart rate get up as high as non-affected people, and is their heart rate recovery as efficient? It's difficult. I would not say that you have to take their heart rate but certainly we look at a person and we say, are they working hard? And ideally, as I said, it all depends on the weight, and their fitness when they're starting, because everybody will start at a different level. If they are doing nothing, it might be just a matter of doing a little. And remember, it's going to be something new to them so they're not going to want to do it anyway because it's not part of their routine.

We have a nice [article on motivation](#) that I could probably send you of ways of getting them to start, but just start small and then gradually build up and tell them that you're

going to be increasing it because you're challenging them and they need to improve their fitness and obviously the exercises are for a lot more things than just weight loss it's for muscles, it's for to keep them occupied, it's to give them a focus, other than food.

SB: And, again, to motivate, perhaps, with a step counter you can make competition with yourself - today I walked so long and tomorrow I want to walk so long.

Patrice Carroll Summary

PC: We have been talking about the management of routine medical checks and other requirements. We talked about PWS compared to other syndromes. Typically, mothers have normal pregnancies - the syndrome is caused by lack of genetic signals. The brain difference in satiety and temperature regulation, how balance is poor, body perception is poor, connection between brain cells differ. We spoke about the autonomic nervous system, the stomach, intestines, the level of stress, sleep, and the sensation of temperature.

We talked about medical problems, and the recent Dutch study, that there are more physical issues. Body composition - muscles and bones are slender and calorie needs are lower than normal. The hypothalamic dysfunction - the body does not produce sex hormones normally, and hypothyroidism.

We spoke about the growth hormone, and the importance for muscle development. It does not change appetite. And then of course the debate on whether or not to continue with growth hormone into adulthood.

We spoke about diabetes, type two diabetes treatment is weight loss. Sleep can be abnormal, sleep apnea is common. We spoke about temperature abnormalities and cases of severe infection without fever, and how they'll they need guidance to pick the appropriate clothing.

The need for calcium and vitamin D lifelong, and the importance for calcification of bones. Be aware of vision, hearing, skin, etc. many won't feel or report changes in their body function. Many have a squint, and vision can change in young adulthood and certainly we need to check that over the age of 40. Check hearing when you feel it's necessary and if there's any changes, and over the age of 50,

The mouth, the tests for swallowing - they produce little saliva, they often have reflux and won't report it. Chewing and poor motor function. Swallowing can be incomplete. The muscle function is poor. The need for every six month check from the dentist, especially the harm of acid on their teeth, and choking is a well-known cause of death in people with PWS

Acid reflux to the stomach, the stomach is easily distended. Vomiting is difficult, if not impossible. They'll rarely complain of descended stomach. Slow passage - 1.5 to 2 times slower than the average person for the intestines. Rectal picking can start with constipation.

We spoke about the urinary system, and that they often don't feel that they have a full bladder.

In bones, osteoporosis is common, always ask for an extra X ray if you feel that fracture is a possibility. Feet and legs, swollen legs. Diuretics will not help, weight loss is the cure for that.

I think the quote of the day for me is when speaking about hygiene and learning, "What is seen is remembered and what is told is forgotten. I really, really liked that and I'm going to steal it from you.

We spoke about aging and PWS and looked at thyroid, hormones, how men who have low testosterone look much older. I didn't know that, that's actually pretty interesting. We don't know if they are more likely to have dementia. It could also be depression, so look at that.

And then had really great questions regarding vomiting, sleep apnea, and then circled back to choking. We spoke about psychotropics and the changes in saliva production and mouth movement and to be cautious of that around chewing and swallowing and choking.

And I think that pretty much sums up what we spoke about. It was really wonderful session.

Remaining Caregivers' ECHO Sessions

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| Weds 20 Apr | Education of PWS, behaviour appreciation, and positive support strategies Patrice Carroll, Director of PWS Services, and Brittni Kliment, Director of Program Marketing and Admissions, Latham Centres, USA |
| Weds 18 May | Looking at different diagnostics Norbert Hödebeck-Stuntebeck, Psychologist, Germany |

Thank you very much to everyone who attended the session and participated. We look forward to seeing you on Session 11 in April.