



Self-Management – Turner Syndrome Transcript

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Turner Syndrome is where there's a loss of all or part of the X-chromosome and all or some of the body's cells, this leads to various things happening like short stature, normally infertility, cardiac problems, kidney problems, and also osteoporosis can be an issue. This is because the ovaries haven't developed and obviously hormone deficiency such as you would get in post-menopausal women.

There are various challenges, the main one is to do with height not just the actual practical problems like finding clothes and getting things off the top shelf but also other issues like the lack of spatial awareness. I found learning to drive very difficult. I often bump into things because I can't judge how far away they are.

I tend not to cross roads unless there's a proper crossing. Driving took a long time and it was just a case of keep practicing. Other issues that I think that have been difficult and practice does help but I tend to make sure that I've got a nice map with me if I go anywhere that I don't know, and plan beforehand and make sure that I've got the person who I'm meeting's phone number, so if I have any problems I could ring. But its practice and planning, and just making sure that you know what you are doing beforehand.

Also reading people's facial expressions is difficult so I don't always realise when somebody's in a bad mood or like other Turner's women I can maybe be a bit tactless and not realise it. I think its hard work for other people because they don't realise the issue and you don't realise that they're annoyed but they think that it's quite obvious that they're annoyed but it's not always to me or I sometimes don't get subtleties in what they're saying or you maybe take things too literally which is difficult.

I mean I understand sarcasm and I understand irony but it's reacting to it which is sometimes difficult. How I cope with that is just trying not to react to it, to just be polite and not get upset and if I see somebody else getting really cross, just backing off.

I think the social issues, if you're brought up with Turners are different because you know that those are issues and people can teach you coping strategies and they know how to treat you. They know that you maybe find certain issues difficult and therefore they make allowances for that though like maybe phrase something differently. It's not so much that you're treated differently but there's more understanding now of the challenges that are faced.

Healthcare has changed a lot since I was originally diagnosed. Issues are taken seriously. There's more awareness of what those issues are and there's much more consultation with the patients about what they think is the way forward. Beforehand it used to be that the doctor would tell you how they wanted to proceed whereas now there's much more 'Well, I think that this is the best way forward, what do you think?'

I think healthcare has definitely improved in the last 20-25 years especially with the advent of the specialist Turner clinics. They're extremely important because general practitioners don't really have the experience and resources to deal with the specialist problems that Turner's women have. My GP is very good she knows what she doesn't know and is happy to refer me on if she thinks it's appropriate. The Turner's clinic in Edinburgh is very good, they are very supportive and they very much tend to discuss



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things for example 'how do you want to proceed?' and if there's any problems they are more than happy to deal with them and also they are very thorough that all the things that need checked, do get done.

My experience has been very different from that of the girls who are diagnosed much younger. I knew that there was something different because I was much smaller than everybody else in my class and my mother was concerned but the doctors said 'oh no there's nothing wrong with her. She's just little'. So in some ways it was no great surprise when I was diagnosed as having something wrong because we had always kind of guessed that there was something not quite right with my growth etc. But the ones who are brought up with it I think do much better because that's just how things are. It's not a shock to them. They get growth hormone, so their height is much more nearer the average of other women. I mean they still have all the other challenges that women face such as body image issues, but every woman has those. I don't think they're any different in that respect to any other women.

I knew that academically I would be no different, I would still do well at school etc, but I didn't expect to get married or have a partner and obviously that has happened. I think that I had no role models at that time. I hadn't met any older women with Turner Syndrome and I wish I had because I think that it would have been very helpful to see that their lives were normal.

I think that the girls growing up with Turner's now have a much more average experience, although it's not average to go and see your doctor every 6 months or whatever and have growth hormone injections daily. But physically things are much more average for example their height, when they go through puberty and I think that does help because it just makes them feel more like everybody else, less different although in some ways they know they are different because they're having the injections. They also know that they'll go through puberty the same time as everybody else, they won't be much shorter than everybody else and I think that is helpful.

HRT for Turner Syndrome is practically dependent on what age the woman is or girl is. I wasn't diagnosed till I was 16 whereas the girls who are diagnosed younger are now getting, starting hormone replacement therapy at 12 or 13. I think it was presented to me as a wonder drug, I mean they never said that it would cure my infertility or anything and I understood that, but it was presented as this will make you more normal and I don't think there was much appreciation of some of the issues around it that anyone on HRT can have, such as mood swings, irregular bleeding, things like that I think there was more understanding of that now, that it isn't just take these tablets and everything will be fine.

When I was diagnosed I was given a couple of booklets, they were actually Canadian ones which explained matters and I was given the address of the Child Growth Foundation because at that time the Turner Syndrome Support Society was still part of the Child Growth Foundation. That's now changed and the Turner Syndrome Support Society has broken off. I think it was important that we had our own group because we are such a large group with specific issues out with childhood. I think it was helpful to have our own group that we run ourselves or the parents could get involved with.

My doctors have been very good and I get a lot of support from the Society and also with the Turner's women. There tends to be very strong friendships with the Turner's women there's often a bond there even if we don't have any other things in common. I think that we have so many other experiences in common that it's a close group.



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After I received the initial booklets at that time there wasn't really an internet. So I was quite dependent on the booklets that I was given and the information from the Child Growth Foundation but now there's a lot of information on the web which obviously you have got to be careful and filter but most of the national societies including the British one are excellent and they also provide written booklets if you don't have internet access. So there is a lot of information out there available.

I frequently have had to provide health professionals with information because a lot of them have no experience of dealing with Turner Syndrome, for example, I've had to say well I'll need bone density scans, oh and diabetes and thyroid can be a problem I'll need regular blood tests for that. But especially with the specialist clinic I think that is improving also. I think doctors attitudes are changing. They're taking the issues much more seriously and the issues have been much more recognised for example at 16 I was sent out the room while my mother had a discussion with the doctor about what was happening, that would just not happen nowadays, I do think the whole attitude of the medical professionals towards us has changed.

I think it's important to be informed before you see your health professional if possible and say what you think, so you can have a proper discussion about your healthcare and make an informed choice about what's happening. Obviously you have to be guided by their opinions but if you know what to expect beforehand. I think it's helpful and also I think the girls growing up, I think it's important that they are included in decisions about their care as appropriate to their age.

I think what would have improved my condition management is what is known now, was known 20 years ago because things have improved a lot since then but I think there's still a lot more that can be done with teaching the younger women coping strategies for example navigation, use of GPS if need be, things like that. Just training for how to read facial expressions would be helpful and a reminder to them that although they are normal they do need the support of the health care professionals.

I think the first advice that I would give a woman or parents who have just faced diagnosis with Turner Syndrome is not to panic. Life will go on as normal, it will just be a little bit different because you will face a different set of challenges and issues to most people but there isn't anything usually that can't be overcome or learnt to be coped with.

My condition management for the future, the main challenge I face is obviously the menopause because I am now 44 and I'm really one of the first women who has sort of been through HRT from a relatively young age. There's lots of issues about what's the best way to approach menopausal management. At this moment we really don't know it's very much an individual thing and trying to decide between the woman and the doctor how to proceed for example, when do you stop HRT? Do you reduce the hormones before menopause. How do you do that without causing other issues? So that's the challenge for the future and I think that my experience will make it easier for the younger

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