

329 – Hereditary Multiple Exostoses with Leigh Edwards

Steven Bruce

Good evening, I have Lee Edwards and Jackie, his mother joining us in the virtual studio and waiting to tell us all about hereditary multiple exostoses slightly unusual show this in that neither Leanne or Jackie is actually a medical practitioner, but you will understand why we have them both in the studio in a moment. Lee is in fact a trustee of the charity HMPE the new generation, and he was recommended to us by osteopath Ed Adams. Sadly, I'd hoped we'd get Ed into the studio as well for this evening show, but unfortunately, other engagements. But Lee and Jackie is great to have you with us. Thank you for joining us. Well, thank you. Hello.

Leigh Edwards

So as I say, I'm sorry,

Steven Bruce

no, no, no, I was just gonna say Lee, perhaps you you better introduce yourself and explain a little bit about the charity and, and what it's all about.

Leigh Edwards

Yeah, so of course. So as mentioned, I am one of the trustees for the HMA charity, which is a web based charity, and has very much a presence on Facebook, in terms of what we do we offer face to face meetings. So we're able to talk to our members hear their stories and offer support as well. And that support runs either face to face or discussions over phone video link excetera. And we also run a series of workshops as well to support our members. Topics include surgery, pain relief, how it can impact on education. One of the big things, themes that we're looking at is around emotional well being, and particularly how it impacts on people social, emotional, and mental health. We also run workshops around physiotherapy, it's really how we support our members to cope with the condition, both for when it's already been diagnosed, but also for when each newly diagnosed as well. So those families know we provide a lot of support for our members.

Steven Bruce

Okay, and what about yourself, what's your own background? You're not a medical practitioner, as I said earlier on, so what do you do?

Leigh Edwards

Yeah, in terms of my occupation, I work for local authority, and I'm one of them. A manager within Children's Services. We support vulnerable young people. So that's my day to day job.

Steven Bruce

Right. Okay. Jackie, perhaps we should have some background on you because you're a bit of a surprise guest for me because I wasn't expecting you to join us. I'm delighted you are joining us.

Jackie

Thank you know, aye. Aye. Aye, I am obviously retired. And, unfortunately, I've not been able to work much during my life, I trained as a secretary shorthand typist, got on well, and then all of a sudden, you know, I obviously lost movement in my hands and whatever. And I've had to change went into banking. But unfortunately, I had to give it work when I was about 48 years of age, because I just couldn't cope anymore. And

Steven Bruce

a couple of years ago, then

Jackie

only. But in those days when I was working, at one time, I used to have to take so much time off work because I needed so much surgery. And unfortunately, I was asked to leave not because of my work, but because obviously I was absentee a lot. So now I'm just retired.

Steven Bruce

Right? Okay. Right. So having established about what you yourselves knew where you've come from into this, I suppose the first thing Leah is to explain to us what hereditary multiple exostoses or any of its other titles actually means. We

Leigh Edwards

I mean, in terms of titles, it has been known by several different titles, and that has included multiple hereditary exostosis. difficile. ecclesia,

Jackie

walsman, osteo, condomless. Yeah.

Leigh Edwards

And there's another one as well as in a hereditary multiple us ask us. Roma. Yes. Yes, thank you. So has been known by many different titles, but in reality, it is a rare bone disorder, in which people develop multiple cartilage cat body lumps at the end of many of the bones as well.

Steven Bruce

And there is there is there a particular type of bone only long bones? Or is it could it be anywhere?

Leigh Edwards

It predominantly says does affect long bones, however, again, in your your digits, then you can also have access OCS developed there as well. So it's not just limited to Low long bones, although they are the main part which is affected by the condition.

Steven Bruce

Right. Okay. And so when I was reading before we came on air suggested that the current name, the modern name is hereditary multiple osteochondroma, as your charity isn't called that is that just because you're fed up with the changing names that medicine often gives to complaints?

Leigh Edwards

I think so as well. So I think the general term it's got that the condition is known by has been haitch Ma, or hereditary hereditary, multiple extra cirrhosis. So that's the name that we've stuck with rarely.

Steven Bruce

I think we'll stick with it because it's easier to see than all the other ones that they've mentioned. It's one of the irritations of medicine that people like to give long, convoluted names to things and then change them halfway through the cycle or whatever it might be. Yeah, okay, so we've got some outgrowths of bone primarily on long bones.

Leigh Edwards

So in terms and how it impacts on our day to day lives, why is obviously bone deformity, the extra sociis compress on nerves and blood vessels. It also causes joints to lock has a lot of motion, chronic pain, fatigue, mobility, and also obviously, that has a psychological effect as well. And I've seen a lot of people who have the condition, also experience, low self esteem, etc. Alongside that,

Steven Bruce

right. Now, I'm guessing that there is a genetic component in this because you've already told me, Jackie, that you have had you implied you have problems with HMV. You told me earlier on that you both have it. So Jackie, you're the first person in your lineage to suffer.

Jackie

I am the first person Yes, I you know, obviously, I've got Lanius as I say, if in those days, they weren't tested, I mean, obviously now if that was you, if it was younger, I'd have been tested with which one had one of my parents often and she passed it on. Unfortunately, you know, they've passed away and I just don't have that information anymore. But unfortunately, when I was before I you know, when I was expecting before I started to have a family. I was told in those days there was nothing you know, you're safe to go ahead and have a family you wouldn't pass it on just just how long they were Yes.

Steven Bruce

How many children do you have? I only have the one. So it wouldn't be the case that all of your children would have inherited this or is it?

Jackie

A 50% chance. I chose not to have any, any more children. But as I say, it all depends. I mean, some people I know have had children, they but but if you've got it, you're more or less likely to pass it on now. People have had family, maybe two or three children, either one or just one of the children have been affected, and the other two have been alive. So there's no there's no direct answer to that it's just hit or miss.

Leigh Edwards

In terms of the genes, it is x t one in chromosome eight. And that is 70% of people with HMA have exp one, there is a 62 that is in chromosome 11. And 30% of people with HMA have that gene as well, just to say men were mentioned then, is that HMA does impact on the body's ability to produce to produce now is it heparan sulphate I think that's not the enzyme which is known as a tumour suppressor. So that impacts on on their body's ability obviously to produce that enzyme. Hence, then we have the x t one and the X t two 10% of surfers are spontaneous. So as my mum is expected, she is spontaneous. And not. I've got obviously I've got the condition from my mum. And I've also passed it on to my daughter as well.

Steven Bruce

Okay, and do you have more than one child?

Leigh Edwards

I know I just have the one child. Right.

Steven Bruce

So again, it's the same question. But you said there's a 50% chance that you'd pass it on. Yeah. Yeah. Okay, so it can be spontaneous. So how are people going to recognise this in someone who doesn't have some family history of HIV?

Leigh Edwards

I think often the majority of people with the condition it's identified at around the age of four, when obviously then they notice multiple bone growths. Because if you have just one bone growth, that's not as a single extra SOC, that is not usually due to HMA which having multiple extra sociis that leads to the diagnosis, but that tends to be often from the age of four onwards.

Steven Bruce

So these are exostoses, which are visible to the naked eye.

Leigh Edwards

Yes, yes. Okay. And that's where you see the lumps growing merrily on the joints. So I think often shoulders are one of the early indicators as well. I know that's why no mom repair she realised that I had the condition was when she realised I had an XR sociis growing on my shoulders.

Steven Bruce

When did you notice that first Jackie?

Jackie

I think Lee was about three. Yes, he was about four years of age I noticed it. And then obviously I you know, it was it was diagnosed. And you did have the hereditary multiple XR servicii.

Steven Bruce

Interesting for our audience, of course, because a lot of a lot of osteopath chiropractors will treat young children. There's every possibility that they could be the first people to come across this. I mean, a parent might bring a child with that to an osteopath chiropractor and say, Well, what's this? What's what's happening to my my child? I would suspect generally with lumps, they would go to their GP, first of all, but what's the what's your experience with GPS? How familiar are they with the condition? Or were they simply refer on to a an orthopaedic consultant?

Jackie

Well, they would Yes, but unfortunately, I have to say now quite often, GPS that I've seen in the past, just don't know anything about it. And I obviously have to explain everything to them. And then obviously, your your, your obviously prefer to go on to see an orthopaedic surgeon. But not It's not everybody, not all GPS know about it. It's quite well, I mean, as I say, but more wanting to watch 50 sounds just 50

Leigh Edwards

Currently, it's so the thinking is around one in 50,000 people has the condition

Steven Bruce

however, more common than I would have thought.

Leigh Edwards

So I was just going to say but I think further research is it is happening now. And the thing that should that number actually might be lower.

Steven Bruce

You mean that there will be fewer people with it or more? More heat, right? Okay. So is it is it possible is it conceivable that a young child might not have visible exostoses. So might have other problems as a result of what's going on?

Leigh Edwards

It does, because I think with the extra associates themselves, they think everyone has different levels of severity with a condition. And that's often why it can be hardest to diagnose, particularly with small child, because for some extra associates, they're more deep seated. So for example, if I look at my mum, her extra associates, are predominantly more deep seated within each of the joints, where the man is much more visible. So it's is quite difficult at that point to and obviously, how you would identify it is through, obviously, for the pain that child is experiencing.

Steven Bruce

Right. Okay. That being the case, with exostoses, which are not visible, Jackie, what was what was the first problem you encountered? How old were you?

Jackie

Well, the first problem I encountered, I was about 1516 months old, and I could walk but I just didn't want to walk, I used to scream. And everybody used to say, Oh, she's just being lazy. I went to all a children's hospital in Liverpool, and many a time member that was sent home and telling us that she was imagining it. It got got just got to a pitch where it was so bad, and just wouldn't walk at all. And so she took me back to soon orthopaedic surgeon, the same one as a senior after Yeah. And all of a sudden, he just said, turned on to my mother and just said, Well, when did you notice this mother? Obviously, whether he knew more about it, but made obviously it's going back about well over 70 years now. So as I say, you know, but I mean, mine are more deep, deep seated. So, I mean, obviously, you can see the odd few. But you know, as I say, more so different with this. Definitely.

Steven Bruce

Do you have any idea? How old might a child be before this is noticed? I mean, you've mentioned you were in your sort of first 18 months there. Lee, you said I think at about three or four you were you. It was noticed in new? Oh, some people? Can some people go much longer before these things manifest themselves?

Leigh Edwards

Yeah, very much. For me, I think, again, from a charity point of view, we have individuals who come for support, who are actually adults as well. And it's not until a later date that they've actually been diagnosed whilst they've had it. They've had the condition and lived with the condition. It was only in later life, it's actually had that diagnosis attached to it. So then that's why are the charities aims as well, is about raising awareness for the condition, both in terms of p of sufferers, but also medical professionals as well.

Steven Bruce

What Yeah, and I think that's very important. It's one of the things we tried to do with the show when we're dealing with conditions, which, you know, they're not necessarily within our remit to treat, but sometimes if we're the primary pet care practitioner, we're the first to see it. We need to be able to help people get the right care straightaway. One of our audience Kim was asked whether this condition is always painful in a child?

Jackie

I would say so, definitely. Yeah, I think my experience

Leigh Edwards

might obviously well, we can all relate to our own personal experiences. But as a family, I know that my daughter experienced discomfort in childhood as well, particularly in relation to her left ankle, and her ability to walk any distance as well. So what we found is that, whilst how it did save, it manifests itself in very different ways. But I think often what, for us that the early diagnosis has been through pain.

Steven Bruce

Okay. But in your case, you said the first noticeable exostosis was in your shoulder. Yeah, that presumably was painful or there was some discomfort with that, did it also restrict the movement of that shoulder?

Leigh Edwards

It does, yes. So again, for what you find is people with HMA, your body naturally compensates a certain movements. So for example, you might extend your arm or you rotate your joints, what you'll then find is that other joint muscles and then compensate to enable you to complete that movement as well.

Steven Bruce

Right? Okay. Does that mean that you end up potentially with problems in other areas of the body as a result of that compensation?

Leigh Edwards

Yes, definitely. Because obviously, your body learns to cope and what those coping mechanisms may not always be helpful, as well.

Steven Bruce

Yeah. So I think you know, that's why I think you were saying to me earlier on that you've both dealt with the problem in different ways. How have you approached, how have you gone about that leap?

Leigh Edwards

In terms of what obviously, for how I now manage the condition, I think in 2007, was the last operation we'll see significant operation I had. Personally reasons for that was, I was, I've had multiple operations throughout, particularly in my earlier years, what I found is as I got older, not only is my recovery longer, but I found that the benefits were not as good as they were when I was younger, particularly around healing property, and you put your other bodies propensity to recover. So as a result, I then decided to move away from invasive procedures. And look at how else I could manage that the condition for myself. And so one of the things I use, I use exercise a lot, I do a lot of stretching, I do a lot of movements, to keep that to basically maintain my body, particularly on my core strength as well. And by by maintaining my core strength, I'm able to hold my posture better. Also, I use wellbeing apps, etc. So I think and also, I have been on the pain management course as well. Right? I found that to be of use. You

Steven Bruce

say the pain management course whose pain management course is that he runs that.

Leigh Edwards

That was inaudible.

Steven Bruce

Who was the governing body? Who was who set the course up was his NHS?

Leigh Edwards

Yes, it's NHS? Yes. Because I was referred to by my GP,

Steven Bruce

yes. Okay. And now, how do they teach you to cope with pain on that I asked, because I've heard about those courses, but I don't know any great detail about them.

Leigh Edwards

It is a it is an intensive course. And it's run over a six week period. And you have to devote two days per week. Fortunately, I was sympathetic employer, and he was able to release me from my day to day responsibilities. But what that course does enable and gives you coping strategies as well. It talks about the psychology of pain. It's also you also have access to a physiotherapist who is able to put together programmes of exercise bespoke to your where you are at as you also deal with the emotional impact of pain as well, around your well being. So also you have access to support a whole with a programme. So again, it's breaking down your day to day tasks, and how you might approach it differently. Okay, so,

Steven Bruce

no, go on, please. No, no,

Leigh Edwards

no, that's why you can no carry on. Right.

Steven Bruce

So you have had surgery? Jackie, I think you said you to me earlier on that you managed yours primarily through surgery, is that right?

Jackie

Yes. So as I say, my accessories are quite deep seated. You know, slight movement, they lock and I just cannot get it out. So unfortunately, I've gone down that way where I just had to have surgery, I've had well over 50 operations. i That's the way that I've coped with this, I still still get pain. But that's the way I've looked at it has helped me personally more than anything. So as I say, we're all individuals, we all treat things differently. I mean,

Steven Bruce

one of my, you've answered one of my questions already, because I was gonna say, Well, if you're going to deal with this by surgery, and by definition, it is multiple external services, how many? How many operations you'd likely to have to have? You said you had 50 or more.

Jackie

I've had over 50 Well, hopefully touching wards I you know, I'm coping at the moment. So I'm hoping I don't need any more surgery. Man through it all I've had to discuss, obviously, because my my left arm is very, very deformed. And I've had to have a replacement elbow with it. And then I've also had to have my both my hips replaced because it's just unbearable, you know, the taking the XR services away and there was there was still they were good, they've grow back. And they were still lacking. So to help me that's how I found it. You know, I've had replacement joints in that and that that that's helped me personally More than anything.

Leigh Edwards

I mean, just to add to that, because although extra associates grow throughout childhood, they also continue to do so in adulthood. But fortunately for myself, I've not experienced that, as far as I know that my mom has.

Steven Bruce

Yeah. Well, I guess yes, that's another question I have is that having had your hips or your elbow replaced? Clearly, if that's been the case, then they've taken a we've taken away one area where the ox exostoses can grow by replacing it with metal and plastic that they then delete, then continue to grow around the implants.

Jackie

At the moment, I know whether whether I can say that as a permanent I really don't know. But at the moment, no, I mean, it's a few years since I have my hip, you know, my hips replaced. Um, you know, I've not had an go for X rays and checkups. But nothing's been said about the exostoses coming back.

Steven Bruce

Is there a point? Is there a point in life at which they stop growing?

Jackie

Now that I can't?

Leigh Edwards

No, I think, I think the biggest, obviously, is when we realised the realisation was that they can continue to grow in adulthood. Because obviously, they were I was under the understanding, historically, that actually, once you're once you stop growing in the end of childhood, and interviews, now, that would stop the bone growth as well. But unfortunately, we've seen situations are just remember, for other members, where actually they have experienced continued bone grill?

Steven Bruce

Yes, well, I suppose that other title osteochondroma suggest that it's obviously not natural bone growth that's occurring here. So therefore, we could expect it to go on longer perhaps. Simon's asked a couple of questions. He asked one some time ago, both of you mentioned the idea that these these extra groups were deep seated. And he asked what you meant by that.

Leigh Edwards

Was I suppose it's actually growing, you're gonna explain because you obviously, impact more.

Jackie

I mean, as I said, it just goes to me, but I'll say mildly, you can see the visible lumps on his joints and but with me, you can't you can't see them. Or maybe the odd one will show up slightly. But as I say, mine just don't show us anywhere near as much as what these do.

Leigh Edwards

I think it's where we grow in the joints, actually. Whereas mine tends to be more surface baseball's are the best way I can describe it, whereas for years, they are much more with a joint itself. And therefore impact on how on how that jump moves except in performs.

Steven Bruce

Right. Okay. The other part of Simon's question was about what the operations are intended to achieve, whether they are intended to achieve pain relief or greater movement or whether to just remove the osteochondroma.

Leigh Edwards

Say, I think all of them really. Because obviously, it is initially a thrill, it is obviously to enable you to have that movement regain that movement or as much of movement is that as can be, but as a side effect of that will be hopefully reduction in pain as well. Yeah.

Steven Bruce

How was the surgery then? benefited you Jackie? Has it reduced the pain? I assume it must have done or you wouldn't have gone through three operations?

Jackie

Yeah, no, no, it has reduced the pain. I'm not saying I'm pain free anything but but it has helped the pain. And it's also given me more movement. Obviously, I'm restricted with my, you know, the hips being replaced, I can still do a reasonable amount. But for me, it was pain relief. And I did get some more movement, even when I was a child and more than anything than this day and age in this day. Yes, yes.

Steven Bruce

Yeah. Well, I presume that the means by which it was diagnosed in your case was by X ray. Yes, yes. Whereas my sure they did X rays on you as well. Lee, but yours were more visible. You said. So perhaps the the diagnosis was an easier path to follow?

Leigh Edwards

It was Yeah, definitely.

Jackie

Yeah, I mean, necessarily. I mean, I'm going back well over 70 years and obviously things aren't no you know, they were more like today in a gym in so advanced but I mean, as I say in those days, it was just unbelievable, you know, you'd had an x ray. And then you weren't you it was show it was sometimes it didn't sometimes one of mine haven't a few of them. Don't always show up. But leave Today But lately, I've noticed that they're doing more, you know, a CT scans and MRI, you know, MRI scans that then going back in my day and age, they have nothing like that.

Steven Bruce

Yeah, I wouldn't have thought that MRI was the first choice for these because of course, it's designed not strictly solely for soft tissue with X ray is the thing for determining bone growth, I would have thought and I wouldn't have thought CT was necessary. In this case, a CT has a lot more X rays inflicted on the body. Whereas I would have thought that it would be fairly obvious from a simple x ray that HIV was occurring.

Leigh Edwards

I think in terms of diagnosis, definitely, I think an x ray. However, I know that you've had signals where X rays were the extra service he hasn't shown, and that's required further investigation as well. Yeah.

Steven Bruce

Okay. This might sound a silly question. Pepper is asked what's the difference between multiple expert associates and someone just having a single exostosis? Well, obviously, the difference is that when multiple there's more than one, but could could someone simply have one in each me? Or is it always going to be lots and lots of

Leigh Edwards

it's always lots and lots? I think obviously, if something has just 1x SOC, that can be another diagnose the condition. But for us, it has to be that multiple elements to it to have the diagnosis.

Steven Bruce

And Christina has asked whether the excess thursties are ever malignant.

Jackie

Yes, that they can be malignant the certain parts of the body, a on the pelvis and the shoulders that they can cause malignancy. Going back on that every two years, I've got them on my pelvis, I have X rays just to make sure everything's all right. And normally, if there's more than 10, centre centres, it's centimetres or millimetres in, in the growth thing, they are always every time you have an extra

associate removed, just always check to see if he has come to us or not. But larger exostoses can be more prominent for cancer.

Steven Bruce

Okay. Do you know the incidence of cancer amongst people with HIV?

Leigh Edwards

I think it is rare. You're looking around two to 5% of sufferers, over a lifetime will experience where an X SOC can become cancerous. I think one of the things that we do support members about being aware is those rapidly growing and painful extra sociis And particularly in late adolescence or adulthood. Again, I think you might you my mum mentioned before, particularly with a cartilage cut thickness in excess of 10 millimetres, or usually the what the extra sociis were but who can be which can be prone to this.

Steven Bruce

Right. Kim was asked whether massage or acupuncture helps with either pain or movement or any other aspect of each me.

Jackie

I've not found it to be No, no, I found that not to help me at all. No, no.

Leigh Edwards

I've never tried acupuncture. And I'm not I know. But that being said, other members have and they've found that of use. I personally have found of massage to be helpful as well. But obviously, if your joints his luck, that's something to overheat.

Steven Bruce

Yeah, when we talk about joint locking is that something which resolved after a while or at once it's locked because you're gonna stay locked until you have some sort of surgical reduction

Leigh Edwards

can actually be both I mean, I know he's met in terms of for the last month for example, both my hips have locked both my elbows have blocked off my shoulders have locked up my hand and my wrist and my hand have locked in I have locked as well.

Steven Bruce

By which you mean completely immobile or just too painful to move. Yeah, well, yeah.

Leigh Edwards

So for example, it's a it only released itself this morning. So as three days my right shoulder I've been just been immobile with that, so that I just it was just too painful to even move the home. So it's really then I was having to take painkillers to help support me with the pain but also then to be able to them to go back to their said before is how I cope is using stretches, you know movements to eventually enable the joint to release itself. I'm very fortunately enough today it has released itself. And then the pain slowly subsides. But if that didn't unlock itself, that's when I'd be having to look at surgery to have that unlocked.

Steven Bruce

Yeah, there will be a lot of people watching this and myself, I would include myself in that, thinking that stretching a joint is not necessarily a logical way of preventing it from locking. Because you're stretching the soft tissue, you're not changing the the altered structure of the bone and the joints around the capsule in the image of the shoulder, the ball and socket.

Leigh Edwards

I think what I have found here from monitoring from our experience that has been is that the muscles by increasing my muscle, then it's been able to hold the joints as well. That's my whether that is a medical fact to that point. No. But that's my own personal experience has been that increasing my muscle density has definitely a net will help me to support myself with the condition.

Steven Bruce

Yes, we've, we've had a couple of questions about where the exostosis form, which are very similar. Lawrence has asked whether this is similar to EDS, diffuse idiopathic skeletal Hyperostosis. His short form is dish, Julia's asked whether these oxtr exostoses form at the muscle attachment points. Now, we discussed this before we went on air and you know, you're not medical people you're not familiar with dish. But from what you have described, this is quite distinct from dish. In though in that case, we're talking about osteo fights forming in the ligaments and tendons at joints, rather than in this case, actually, bone growth from the bone itself. So I think, again, I'm no expert myself, but it does seem as though there's two very difficult, very different conditions there. Don't know whether with the charity, you get any sort of exposure into the different ways in which it manifests itself amongst people? No, I

Leigh Edwards

mean, what we we haven't had that exposure. In all honesty. We haven't had anybody come to us with a condition that hasn't been diagnosed this HMA. And I suppose that's because actually, we are badgers ihme. Charity. Yeah. So we have that exposure.

Steven Bruce

From what you said earlier on, about your shoulder locking and the fact that your hands could lock another another joint new lock. Does this affect your ability, for example, to drive cars?

Leigh Edwards

Yeah. Yeah. So it does impact on my day to day ability to do things. So driving, it's just a no, I can't can't do that.

Steven Bruce

So just to clarify on that, is there? Are you legally prevented from holding a driving licence? Because if your joints were to lock at speed or something, you might not get to control the car? Or is it simply when a join lock, you wouldn't drive a car?

Leigh Edwards

It's my pet, it's a personal choice, as I feel confident enough to be in control of the vehicle, if I've got to join up, and that's because of such a steep tuition, active movements, in all honesty,

Steven Bruce

which is obviously and that's a significant imposition on anyone, isn't it to be concerned about their ability to drive? It's a major impact on freedom, which I suppose leads us into mental health. And

I've got a question, which sounds almost as though it's not relevant. But it is from Anna, who says she had an elderly patient with spontaneous me a few years ago. And she had been told as a child that it had been caused by her mother being frightened by monkeys in the zoo when she was pregnant, which is a bit of a stretch by anybody's imagination. But having the condition really affected her self esteem for a very long time. And I think this is something that you mentioned earlier on. So could you talk us through the the impact that it does have on people's mental health?

Leigh Edwards

I think in terms of obviously, it's impacted, it's as physically insanely intense physical presence and how you display because obviously, by having the condition, it said before, you do have bone deformity, and so therefore, your use of shorter stature, your your left arm is often shorter than your right arm as well. And you physically present differently to and particularly this impacts on young people in school, because again, if you're not conforming you When are potentially, again, it's so I'm looking back on my own childhood thing I was the worst propensity for being bullied because you look different you presented differently. You're off school for significant periods of time as well. We're and also that and that then impacts negatively on one's self esteem as well.

Steven Bruce

One of the important things that goes on at school is sport for a lot of people. Lee, were you able to participate in sports to a reasonable level or at all?

Leigh Edwards

No, I think one of the things were definitely when I was at school, that age exercise you were discouraged from, and that was a direction we would get out remember being given. And so therefore, you you ensure you smell how can you then excluded oneself, you know, from any sporting activity. And why then is only later in adult life, that I found the exercise who actually was a real benefit and bonus for myself? Obviously, I'm not sort of I could never do contact sport or anything like that. But actually, I found that exercise was a way of me being able to cope and maintain the condition. Well, definitely. When I was younger, I was discouraged from any for any actual for any form of exercise as well.

Steven Bruce

Is that still medical guidance for young people?

Leigh Edwards

No, what I'm finding now has been entrusted with a charity is that actually exercise is encouraged much more than it was. So and we have a different outlook. And what I'm finding is that the young people who are part of our charity, don't haven't experienced the same thing as I did in that age, which is benefit. And also I can talk about my own daughter as well. And even if she never felt as though the condition as well, we're in a, in a in a bit of an inhibitor for her. Now, I don't think it was in all honesty, but that's where we've seen a change. But definitely, for other in certain members, we definitely see impacts on how on their self esteem, in terms of their emotional wellbeing. Definitely.

Steven Bruce

And, Jackie, you said you had to give up work because of this because you physically couldn't do the work again, that's that's going to have an impact on one's self esteem, isn't it? It's hard.

Jackie

So yeah, I'd say that's one of the things that's really upset me about having multiple exostoses is self confidence. I mean, I am quite badly deformed. You know, I wouldn't go swimming. Because in case people saw my joints and all the operation Scott, and I am very, very self conscious. I know I always wear long sleeves, long, trousers, long dresses, not just to conceal if you're on it's just, I'm not I'm not a confident person. Definitely not. No, I've just had no faith in myself and tried to do everyday management. I mean, without my husband, he doesn't have to do all the cooking. As I say it's had a bad impact, you know, impact on my mental health and the work my well being definitely as much as obviously going through all these operations.

Steven Bruce

And what mental health support Have you had?

Jackie

Well, I happen to have just coated myself I've never to be honest, I've never told anybody how I feel. I just you know, I just get on with this. I've not I've not sought any help whatsoever. No.

Steven Bruce

It was a tough city next year, what to be able to point you in the right direction I just thought for for that sort of like

what have you found there are particular types of mental health support which are useful ly in people approaching you in the charity.

Leigh Edwards

And, again, what we've found is very much a talking therapy base. And obviously, we can signpost, individuals, a lot of support online, through Cooper, etc. So there's a lot of support available for our members. And obviously we support our members as well around their well being. But one of the things that we often signpost as well, although you don't choose to use it. The wellbeing apps as well. about mindfulness, we found that to be a benefit and I say I use the mindfulness as last as well.

Steven Bruce

Yeah. I was just interested in know whether mindfulness or cognitive behavioural therapy or the other types of talking therapy would be useful, or even hypnotherapy in some cases which can be very useful, I think in restoring self esteem or achieving good self esteem. Again, maybe it's an individual thing.

Leigh Edwards

Yeah. But again, I think often from both myself and from members, we use that as a way of coping with the pain, or the condition brains.

Steven Bruce

Kim's asked if you know the cause of the pain, you know, whether it's excessive excess growth or lack of synovial fluid in crushing the joints. It says to

Leigh Edwards

be more from from personal experience about the girl with the and then obviously, for that bone, then again, that can apply pressure to nerves to blood vessels. Yeah. And then, so that's where we've found ourselves. Definitely.

Steven Bruce

Other Other than the bone growth itself, do you get swelling? edoema around the affected areas?

Jackie

I'm no, no, no, I'm not No, I've not experienced I don't know, whether you can or not, I don't know. But personally, I haven't experienced.

Steven Bruce

Interesting, I've just had comments in here, which has been flagged up for me and it's aimed at you, Jackie. Marion says, Well, no matter what you say you speak with extreme confidence. So you should be very pleased about that.

Jackie

That's nice to know.

Steven Bruce

Getting back to the the locking of the joints, Christina has said you get warning signs that your joints are going to lock? Or could that just be sudden, you're in the middle of some activity or other?

Jackie

It's just spontaneous, isn't? It just, you know, you could be doing anything and next things a lot doesn't. And you can compress it down to why it started to laugh. It's just one of those things.

Leigh Edwards

Ya know, it's a good question to ask them trying to just reflecting them is to, obviously it because I can relate to that quite clinicians experience, I think there was nothing, I could not put it down to one particular activity or in all honesty, the my shoulder locked, I was just actually in the kitchen as making tape when the partner and suddenly I could just, I could not move my shoulder, it just said, it literally froze on me. And I just had to go and sit down and there's nothing I could have done.

Steven Bruce

It's hard for most of us to understand that the sort of impact that that might have. Because as you said, Jackie, you don't do any cooking. There are activities like cooking, which could be potentially quite dangerous, if that can happen. So suddenly, when you're in the middle of dealing with hot liquids, or, or whatever. As I said to him,

Jackie

I was just saying one of the reasons why I do is because obviously, through this, it's made me quite weak. I can hardly lift anything. So obviously I couldn't lift a pound with water or I can't even hold a knife to peel vegetables. So as I say, my husband does all I can do I can do a bit do housework and things like that. But anything strenuous. I just have I just can't do it. No.

Leigh Edwards

It does. You don't realise often how it does impacting your day to day lived experience. Because when as last week when my wrist locked, I could feel I could actually feel where the extra SOC was applying pressure to the blood vessel. And then obviously, I could feel a tingling sensation in my hand. My hand became quite numb. And also I experienced with those carried by bloodshot and assures you that yes, yes. So then I couldn't do day to day tasks with that. So again, even shopping or doing anything like that, it just what it just wasn't able to do.

Steven Bruce

You've mentioned cancer overseas as being a you said a two or 3% risk of developing cancer as a result of Ah, me. I presume that's two or 3% greater than the incidence in the normal population. And what about other complications as a result of this?

I'm thinking particularly I mean, you haven't mentioned exostoses in the neck and there are clearly critical blood vessels in the neck which could be affected.

Leigh Edwards

I've not never, never, never heard of that. I've heard of members were in the back and the ribs but never in the neck or on the head injury as well. Right.

Steven Bruce

That's quite reassuring for a lot of people isn't going to recover.

Leigh Edwards

No exact Yeah, no, I've never because we recently had had a one of our HMB conferences. We had two consultants, one for older hay and the other from the from the Agnes. Agnes. Robert challenge and rubber germs was an IT.

Jackie

Specialist. Yeah, Oswestry

Leigh Edwards

and baseball there. And both of them had not identified as having experienced surfers who have had in the neck or on the head. Right.

Steven Bruce

Kim sort of developed that a little further and asked whether it affects the functioning of any of the other organs because of the deformity, whether that would be kidneys or heart or anything else. I don't think we mean other than through malignancy sort of cancerous growth.

Leigh Edwards

No, no.

Steven Bruce

I think I think the question was possibly prompted when you were saying that the exostoses could put pressure on blood vessels, and then immediately people start thinking, well, could it compromise the circulation in other any other way? Is it likely that you could compromise the circulation completely? For example, in one of the limbs? No,

Jackie

I've never heard of it. You know,

Leigh Edwards

I'm not aware of

Steven Bruce

that. Okay. I had a question from Charlotte coming in earlier on about whether there are other environmental factors which could aggravate this or affect it, and she's talking about diet or weight, and said that exercise has helped you and is that the case for all of HIV sufferers?

Leigh Edwards

I think in terms of what can be triggers, what we find a particularly cold weather, or change of weather or fat, cold weather moment, old weather also damp as well, damp weather that tends to impact negatively on the condition in terms of weight. I couldn't comment on that, because I think both of them and I've always make, I've always maintained, I think without piling condition as well, is that that's been at the forefront of our thinking in terms of how we can try and maintain our own well being Yes. From that as well from a health perspective.

Steven Bruce

I did say at the beginning, Lee that. It was an osteopath, Ed who had pointed us in your direction to get information about this. have you actually seen an osteopath? Did you go and see a because of the problem or have been treated by another physical practitioner? Physical Therapist other than a physiotherapist?

Leigh Edwards

Yeah, I haven't. No. And I think we're obviously for read the link came in here we have our chair, which was Helen small on Helen. Well, she's our chair, she doesn't actually have HMA. So that's where the link came from. Whereas as a welder mature, stay I also have the condition as well. And so all our trustees have the condition.

Steven Bruce

Yeah, and I suspect you don't know an awful lot necessarily about what osteopaths and chiropractors do. And of course, one of the questions that's come in is, you know, how can we in physical therapists help? physiotherapist or, of course, also physio physical therapists. But you've mentioned so far that they've prescribed specific exercises for you. Has there been any hands on treatment that's helped whether it's been well, we talked about massage earlier on osteo is and physio LCSW. In Cairo is a famous for walloping joints around us, you know? I have that's not how we express it publicly, generally.

Leigh Edwards

I personally haven't done down. No,

Steven Bruce

yeah. But you also said earlier on that there's one in 50,000 People might have this condition. That means there's a hell of a lot of people in the population who potentially have this. How many people are aware of your charity or part of the charity?

Leigh Edwards

I mean, in terms of membership, we have in excess of 500 members. But that is not only national, but that is international as well. So we have a number of members, not a great number in America, but we have a lot ended abroad as well, particularly in Europe.

Steven Bruce

Right? And what about all those people who aren't members of your charity? How are they getting by? You know, do you have any idea?

Leigh Edwards

No, we don't obviously, it's the unknown elements unless they make an attack. I mean, when we say membership again, it's just Oh, you look for him becoming a member for HMA is What is your connection to HMA? And because we aren't we tend to be more Well, obviously, we're based in terms of how we are our outreach to our members. So it gives us flexibility in supporting members but again, it's we are a dispose expectation is we Because is offer are for newly diagnosed is to find is an ace will seek us out because obviously we do have a Facebook presence. That's the way we pick up majority of our memberships, we tend to have about two, two new requests for membership each week.

Steven Bruce

Right? Okay. Are those people have found you simply through your Facebook presence? Or are health care professionals pointing them in the right direction? Now, do you think

Leigh Edwards

majority is where that habit condition diagnosed. And they think, Oh, I've got it, I need to find out more about it and start to Google, start a Facebook search, etc. And that's how we come into contact. We don't get a lot of referrals from the medical. But I think that again, is about for us as a charity. One thing is why I'm here today as well. It's about raising awareness as well. Yes. Yeah. You don't know what you don't know.

Steven Bruce

No. And I'm hoping that this will prove useful to other people who have the condition because there are, you know, I don't know how many people yet are watching this show, but there will be several 100 watching it, and they may well come across people in in their practices. Another question from Christina, who's asked whether this is equally prevalent in different races? You know, that for male female balance as well, I mean,

Leigh Edwards

what we find is, I can't comment on race. I don't know, I don't have that information. However, in terms of gender, I think looking at our membership anyway, we're looking at around 6040 60% being male having the condition.

Steven Bruce

Okay. That's interesting, I was expecting you to say it the other way around, because there is a widely held? Well, I think there's a widely held preconception that women are more interested in healthcare generally, and so therefore might be more likely to approach you. So perhaps that is reflective of the gender balance of the condition, what has to change, I mean, how much support you get from the government and the charity.

Leigh Edwards

We don't get any civil support from governments. We are self funded, and we raise our finances through the support of our members. So it's about obviously then we look to our membership for contributions from that, I suppose in our activities, but everything has to be self funded.

Steven Bruce

And from your experience, if someone with each me finds their way through the health care pathway through their GP and finds himself in front of an orthopaedic consultant, what is likely to be that orthopaedic consultants advice to them? Now, as opposed to, for example, when Jackie was first diagnosed?

Leigh Edwards

I would, in all honesty, I would say, mixed. That's from my own personal experience from my daughter. But also as well, from the feedback I get from members, I think, for certain consultants, are they okay with the condition, whereas some aren't? I'll be honest with that random, we see that impacts on that individual's experience. But what we find definitely is that from our membership, and when we talk about this, is that every member's experience can be quite different. Yes, so there's no yeah.

Steven Bruce

I guess it's it's kind of helpful that she has a grandmother and a father who have the condition and help her help her to cope. Can you tell us how it's affected your daughter?

Leigh Edwards

My daughter, has been less affected than both myself and my mother think it's fair to say whilst I have extra sociis In all my joints, and so do you? Yes. My daughter doesn't. She has in her left ankle. She's also got them in her knees and in her, Oh, gosh, in her wrists and in her elbow. But then, and then not in the frequency that both my mom and I have in. So it hasn't had such as you only had to have two operations so far. My daughter is 24 and is a qualified primary school teacher. So in terms of impacting on her, Yes, she does get pain, but she's able to manage that pain as well. And it hasn't stopped at a pathway and aspiration.

Steven Bruce

And is it likely then that that reduced incidence of exostoses will continue he won't sudden may develop a whole host more in the way that you both have?

Leigh Edwards

I would hope not. But I think you say, Well, I mean, again, whilst my mum has experienced additional growth in adulthood, I haven't. So I'm hopeful that being same pathway for my daughter, I think the challenge as well as going forward from adults will be if in the future she chooses to have children, and what the options will be for her in terms of insurance, so I think obviously, one of the options of is around pre Implantation Genetic Diagnosis as well. So I know that one of our trustees was able to talk about that at one of our recent conferences. So that's that path, the pathway they have chosen to go down. And I think that's obviously up to my daughter when she chooses to go down that pathway or not.

Steven Bruce

Yeah, interesting. Are you able to just give a bit more information on that? Because clearly, you said earlier on that it's a 50% chance of passing it on? Is there any way according to what the trust, the trustee, you mentioned, has discovered that you can predict in your daughter's case, whether she would pass it on or not? Because otherwise, it doesn't really help her much, does it?

Leigh Edwards

No, I think what obviously, then, really, it's the genetic testing, because obviously, we have our identify the genes, what has happened in that case, is that they were able to test the embryos. And therefore, I think they had five embryos, three of which had the condition to deadens. So therefore they were they moved forward on both under two embryos, and then obviously, they were successful. And she's now pregnant. And that's that pathway now to discussion with doors, we'll have to have her herself. Or she could choose to conceive. And again, that will be for her to make that decision. But knowing full well that there is that 50% chance that condition can be passed on.

Steven Bruce

Yes, indeed. And, Jackie, you obviously you've been more effective than Lee Lee, it sounds as though you're more effective than your daughter. I have been asked by Christina, whether there is a chance that this condition burns itself out after a few generations, Is that likely to be the case? Or could your daughter's children be as badly affected? As you Jackie?

Jackie

At the moment, I can't really answer that. I don't think so. But, you know, I just don't have don't have that knowledge. You can't answer that question. Yeah.

Steven Bruce

Okay. And what about, again, another question from the audience, Julius asked whether there is any gene therapy in development, which might help to overcome the problem.

Leigh Edwards

I think, as obviously, we've got the two genes which have been identified, and I think other genes may be responsible. But we don't know that at the moment. And there's no mind. I'm not aware of any further research that is going on around that topic.

Steven Bruce

Yeah, imagine that. Given your role in the charity, you probably wouldn't be aware if there was some research going on. There are probably more more prevalent conditions which are absorbing people's time at the moment, I guess. Which means you need to as you've said, you need more visibility, and you need more traction with the research community perhaps. Yeah, I would have been interested and I don't know if you have any anecdotal information about this, I'd have been interested to know whether the sorts of therapy that osteopaths and chiropractors provide can give any greater relief simply than providing exercises. And I hope I'm not offending any of my physiotherapy friends, or any of physiotherapists in the audience by saying that it seems to be a trend, particularly in physiotherapy, hands off treatment these days and simply to prescribe exercises. Massage has helped in your case, but actually where the joint mobilisation can be useful would be informative for us. I think. You haven't heard of anything going on in that regard?

Leigh Edwards

I haven't know.

Steven Bruce

Well, maybe we'll have to get some information from Ed. Ed, who referred you to us has he had a role in treating people with HIV do unit

Leigh Edwards

at Delta actually, in all honesty. It's not it's not a topic that has that but definitely it's something we can explore you know, as a charity and look to to have or to run workshops as we have with physio, etc. So it's definitely something for us to consider. So it's good idea. Thank you.

Steven Bruce

Where is the where is the focus at the moment of the charity on helping HIV sufferers? And I know people who To use the word suffer as these days, but actually, it seems like a fair one, given the amount of pain, particularly you, Jackie have gone through.

Leigh Edwards

I think one of our focuses historically, we are called the new generation was about supporting those families who are newly diagnosed. I think one of the switches for us as a charity has been, is that we also know that we do have an ageing population. And I would definitely include myself in this not you yourself. But I'm one of the switches now is about how we cope with this lifelong condition. It's about then what we put in place, or earlier on in life, and how we can carry that forward. Because often what I get if I look at myself, I'm 54. And if I look at other members who have a similar age, who haven't, who didn't start this pathway of trying to manage the condition, and they I look at how that their experiences now to my experiences, and they are a lot more pain than I am. And it impacts more on their day to day life as well. So it's about really how we have that early intervention, how we see this as a lifelong condition, because unfortunately, it's never going to go away. It's about how you cope with it individually.

Steven Bruce

Yeah. And Jackie, do you feel the the condition has now settled down for you? Or is it? Will it just continue to cause you pain and cause you new problems over the years?

Jackie

Well, at the moment, although I'm still in a lot of pain, I can't say anything is lacking. It's just a gradual pain all the time. And I'm just hoping fingers crossed this. Just it doesn't, you know, get any worse. But at the moment, I can cope with it quite well. But as I say we you know, you just don't know, this time in a couple of months, it could be needing more surgery, but I'm just hoping that I'm not but at the moment, I personally found surgery has helped me more than anything. Definitely.

Steven Bruce

Right. Okay. There was a little bit of confusion in your mind earlier on Jackie, about whether a 10 millimetre or a 10 centimetre exostosis was significant. I imagined 10 millimetre is when they were start to be interest, be interested in the medical community? How, how large can one of these things grow? Do you know? Do you have any idea?

Jackie

I haven't. I don't really know. I just can't answer that one. I really don't know. As I say,

Steven Bruce

I'm just a curiosity. Somebody did ask. And this is a very cheeky question. Please leave feel free to say no bugger off. Someone did ask if you prepare to show that shoulder exostosis of yours. So we have an idea of what it is we might be looking at. Because as we look at you both on the screen here, we wouldn't see there's anything different about either of you? I do feel free to say no. Or just say, you know, I wouldn't describe it. It's a

Leigh Edwards

to be honest. I don't know. It's not it's not as I'm trying to think about it. Because for my shoulder there. It's more deep seated. I mean, I'm happy

Jackie

to share my own. No, no, I'm Tracy Scott. As you show the shape of

Leigh Edwards

your own. Yeah, I suppose. Can you see that? Oh, okay. Yeah. Can you see that? Okay, so that's my left arm. Yeah. And that's, that's as far as I can extend it.

Steven Bruce

Oh, gosh. That is restricted, isn't it? Yeah.

Leigh Edwards

Yeah. That's they can't extend to any of them nice and simple and pressure on that. And that that's as far as it will extend. If you notice, now, when I turn my See, I'm not turning my actual rip. My whole whole arm is moving.

Steven Bruce

Yes. So it's not coming from the elbow. It's coming from the shoulder. Yeah,

Leigh Edwards

yes, it's quite old. And that movement is and that's what I said before is that your body then compensates to initiate that same movement, whereas other people can move and move obviously from me notice that my arm moves

Steven Bruce

and what what efforts have been made to mobilise your left elbow then

Leigh Edwards

I've had no efforts, reason being bar occasionally locking. I've not had a lot of pain from my elbow. Well, therefore, I think if it's not broke, don't fix it type of approach.

Steven Bruce

When I say we've got just under 400 people watching at the moment, who will be saying well, it clearly is broke because you ought to be able to rotate From the elbow, not just from the shoulder. And they're all wondering, well, what would happen if we got hold of the elbow and just tried to gently get it moving? Would it help in any way at all? But I guess I'd have to look at X rays to see exactly what's stopping it from moving first of all.

Leigh Edwards

Exactly, yeah. Because so that's, that's just on one particular joints, we can see how that then restricts in that one. One area.

Steven Bruce

Yes. Another question, possibly the last one, Anita has asked whether you know if this is affected by or influenced by auto immune conditions, things like rheumatoid arthritis and so on.

Leigh Edwards

I think often what you find is that arthritis runs alongside it. I know, for example, when, gosh, I remember finding out that I did have that arthritis. Now, in my right elbow, when I'm looking for an x ray, and but I don't have you haven't got arthritis, have you not been diagnosed? I know that I was when I went to see a consultant. So a double I don't know, which goes it's chicken and egg, which came first so that I couldn't answer,

Steven Bruce

right. There is a distinction here, which is not fair to ask of you, because auto immune conditions like rheumatoid arthritis are different probably from the sort of arthritis you're describing. So maybe we should look that up ourselves. If it influences our diagnosis or treatment. Anyway. Interesting. Another good one from Kim. Are you at increased risk of fractures as a result of this?

Jackie

Yes, yes, you are. Yes. Yeah. I, I've had two falls and obviously have fractured both of my hips to falling. As I say, yes. So I will say I would say yes, yes.

Steven Bruce

Okay. And do you think that increased risk is because your joints are affected, so you're more likely to fall and therefore you are more likely to fracture? Or because the bones are in some way altered in structure or weaker, and therefore they'll fracture if you've just hit them normally?

Jackie

To be honest, I can't really answer that. I'm not? No, sorry, I can't answer that one.

Steven Bruce

The way I'm thinking is that if you can't exercise normally, because of the way your joints are affected, then the bones won't strengthen the way bones should because they're not getting the stresses that would normally go through them. But equally, you are going to be more likely to fracture simply because you're more likely to fall, which is a different cause. Yeah. As I said, we've had 400 People just shy of 400 people watching us this evening. And we had sheduled, a full 90 minutes for the show. But I think you know, you, we've learned a hell of a lot from you, Lee, and from you, Jackie, possibly, you know, as much as we're going to this evening. We have had an enormous number of people just I think in awe of the fact that you're both prepared to sit in front of the camera and talk to us like this. And Jackie, particularly you people saying, you know, we've got words like amazing and humble and, you know, brave and caring, and all the rest of it are coming out from the audience. And so you and I can only echo that it's just it is amazing. This is a condition which I had no knew nothing about before we started our conversation on this. Well, obviously, our conversation started a little while ago, when we planned the talk. I imagine that most of the people watching have very little if any experience of it or any knowledge of it. And I'm hoping that you know

what you shared might well just help to make people more aware of what can be done for those who are in need of your charity services. And maybe maybe some osteopath, chiropractors are pre prepared to get involved in seeing what we could do to complement what the physiotherapist are doing what the mental health agencies are doing as well to make people's lives better. So again, thank you for everybody for joining us this evening. And best of luck to your daughter, whatever she decides to do in terms of having children Lee, it's very unfortunate for you all that you've got the condition, but you're clearly coping very well with it. And yeah, it's great that you shared all that information. So thank you.

DRAFT TRANSCRIPT