[0:00:00]

Interviewer: Okay, [name 1]. So, I’m expecting us to talk for about an hour maybe, but, you know, if we overrun, that’s fine by me but you see what works for you as well.

Respondent: Okay.

Interviewer: Alright. So, you were telling me that you’ve known that your mother had Huntington’s since about [2000-2005].

Respondent: Yes, yeah.

Interviewer: Was she living in [location 1] as well?

Respondent: Yes, yeah.

Interviewer: Alright, okay . And how did you find out about her diagnosis?

Respondent: She became symptomatic so she had the chorea, so we kind of… in those days we assumed that she’d got Parkinson’s, so her arms were particularly bad. So, yeah, just went for tests and it came up as having Huntington’s. I think it was quite early on in the development of understanding about Huntington’s in [2000-2005], so we had a geneticist that came and talked to us about it, explained about the sort of 50% thing and asked us what… because I’ve got two sisters and asked us what we wanted to do. And I’ve always been of the mind that I want to know, but my wife is a completely different… well, she was then, a completely different sort of mindset, so I didn’t want to sort of pressure her into us agreeing to me having the test then, so I agreed with her that I wouldn’t have the test then and we’d kind of just carry on with life and see how things went. But recently or within the last few years both my sisters have become quite severely symptomatic and my dad died, so I had quite a lot of pressures mentally, and one of the things that was preying on my mind was whether I’d got it or not. And we’d got to a point in our lives where actually knowing would help us make decisions, so I’m retiring at Christmas from work, knowing at the moment that I’m not symptomatic so I kind of assume that I’m going to have a good few years before things, whether they do or not, get to a point where I can’t enjoy life. So, I’m very lucky that I can do that. So, it was those sort of things that would help both of us make decisions that got me to going for the test. So, the way I’ve… since my mum’s diagnosis and being at 50% at risk, I’ve kind of assumed that I’ve got HD, rather than completely forgetting about it and hoping that I hadn’t, sort of thing. So, one of the questions that the geneticist asked was about how would you feel if your test was positive, and said I don’t think I’d feel any different to how I feel now, to before, because, as I say, I’ve assumed that it was. So, if it was negative, that would have been a huge positive thing, but unfortunately it wasn’t. Yeah, sorry, I’ve just waffled on there.

Interviewer: No, no. I need to know. You know, we need to set a bit of a context so that I can understand your situation. I’m quite interested that, you know, as a family, with your wife, first you decided not to test because she wasn’t inclined to know yet, and then you’ve decided then, okay, it’s time now because I need to take some life decisions and this is going to help us. Was she more open? Like was she happy to get the test, for you to get tested, or was it still a bit of…?

Respondent: Yeah, she was very supportive. So, yeah, and her mindset has probably changed a little bit as well, but yeah, so no, she was… obviously she wasn’t okay with the result, but…

Interviewer: Of course.

Respondent: She was supportive of me going and getting it done, yeah, yeah.

Interviewer: Okay. And how was the testing process? How did you… was it that you went to the GP first and the GP sent you to the geneticist, or how was it?

Respondent: How did it work? Yes, yes, I went to the GP first, and again no symptoms, explained the situation and asked them to refer me. There was also… too much information, but there were some… one of my sisters and I shared the same GP practice and then obviously you’re not allowed to sort of discuss issues with a GP that’s not… so I couldn’t discuss my sister’s problems with the GP, but I knew the GP was my sister’s GP, and she was a very good GP, she’s left the practice now, so one of the reasons was I actually asked that particular GP to refer me thinking that she might put two and two together and think, “Hang on a minute”. Because I knew she was treating my sister and having trouble finding out what was wrong with my sister, I think. So, I kind of thought that she might…

Interviewer: Your sisters were not tested then?

Respondent: Not at that time. They have, I think, well, my younger sister has, definitely. I don’t think my eldest sister has, but she is now… her family have brought the local HD counsellor in to help, so she’s getting… they’re getting some support from them, and my younger sister is as well actually. But I haven’t contacted them as yet. So, yeah, my younger sister has just had a test done and it’s positive. My elder sister is now, she’s bedridden and whether it’s the drugs she’s on or whether it’s the disease, she’s not overly… you can’t really have a conversation with her.

Interviewer: You don’t know if she’s tested but she is symptomatic.

Respondent: Very, yeah, yes. Yeah, yeah. So, yeah.

Interviewer: And your sisters are also in your county?

Respondent: Yes, yeah.

Interviewer: So, you’re saying you all attended the same GP, but of course you didn’t want to be… you wanted to focus on you but knowing the GP also had a connection with your sisters.

Respondent: Yeah, yeah.

Interviewer: But anyway, so the GP was able to put it together and…

Respondent: Yeah, I assume so because my younger sister has had a lot of help. She was always… so my eldest sister is at a different GP practice. She lives locally but she’s a different GP practice, but it was the youngest one that was very much in denial and she was quite mentally affected, so she’d got a lot of, you know, she was quick to anger and very paranoid and lots of things like that. That was not my sister when she was younger, so there’s clearly something that changed. And she also was developing, with her it’s the feet, so quite a lot of movement in her feet. But her family, you know, tried to help and she couldn’t, she wouldn’t accept. So, again, it was a kind of a… I wanted to get the test done anyway, but I thought if I asked this particular GP, it might help my sister. But my sister’s only just recently (inaudible 0:09:45) to having the test done and it’s come back positive. I don’t know what the sort of CAG number or anything is, but I just know it’s positive. I don’t really… my youngest one I don’t really see that much. I struggled with her personality change and also that she was… it was difficult to deal with her when my dad passed away, so dealing with my dad’s estate was really difficult to do. So, I kind of distanced myself purposely, which sounds a bit selfish, but sometimes you’ve got to do these things, haven’t you? So, I’ve distanced myself from her because it upsets me too much to try and, you know, help. Yeah, I can’t.

Interviewer: It’s a different mechanism so, you know, I can’t… as you’re saying, her personality has changed and, you know, your father’s death must have been difficult as it is without any added struggles.

Respondent: Yeah, yeah.

[0:10:58]

Interviewer: Yeah, okay. So, [name 1], you were saying you went to the GP and you asked for the GP to refer you to the geneticist.

Respondent: Yeah.

Interviewer: And then what happened?

Respondent: Yeah, [location 2] genetics centre contacted me, a lady called… I can’t remember her name, last name [name 2], and set up a series. It was just after lockdown time, Covid, so we were still in sort of semi-restricted things, so we did a lot of Teams calls and Zoom calls. So, I think I went through a series of two, maybe three consultations. I did take a while to sort of, at one point, I think I had a first consultation and then thought about it for a while, not quite sure why. I think I just wanted… yeah, it was when I was still trying to get my dad’s estate sorted out. So, I got that done and then went back and probably had two interviews, like this conversation, one with myself and then one where my wife joined as well and we sort of just discussed everything and then came to the point where they agreed, you know, that it was appropriate for me to get the test. And then I went in, obviously had the test done, and then got called in for the results, so it was a person-to-person meeting with [name 2] and obviously got the disappointing news and then got information about things like HD-Enroll, etc., and the local HD support groups, etc. And have.. I think I had a follow-up within a few months, I think, probably three months, a follow-up. And then maybe six months and then a year, and we’ve just agreed that she’ll contact me or the centre will contact me on a yearly basis. And then I’ve also got enrolled on HD-Enroll. So, I went there last [month] for quite a long session and did sort of discussions and tests and questions, etc. And as I understand it, I just go back on a yearly basis and they track things, I guess, and obviously put me forward for…

Interviewer: Trials.

Respondent: Any research and trials, etc., yeah, yeah.

Interviewer: So, [name 1], when they gave you… I mean you went for the result, you said your wife was there as well when you went to get…

Respondent: She wasn’t when I went… I can’t think why she wasn’t. She wasn’t. I just went on my own. There was some reason why she couldn’t go. She would have gone, but she couldn’t go. I think she was poorly herself at the time, so yeah, I went on my own.

Interviewer: Yeah. And you got the result and then they gave you information. Was this like paper information, leaflet or…?

Respondent: Yeah, yeah, I had some paper information. It was a while ago now. And I got the… [name 2] wrote to me and went through everything that we’d gone through at the meeting.

Interviewer: Okay.

Respondent: And then I think initially she sent me a couple of emails with various things that were for interest or to help, and that was about it.

Interviewer: Was that helpful, like the information or…?

Respondent: Yes. Yeah, it was. I’m quite inquisitive, so I’d done lots of research myself anyway. I was aware of HD Association and what they did, etc. So, a lot of information was about that really. But yeah, I would imagine if I hadn’t done, it would help. It would certainly help signposting things to think about or to look into or things like that.

Interviewer: That’s good. I mean it can be so overwhelming to get the information at the appointment, so the fact that they sent information afterwards, you know, with the emails or the clinic letter for you to be able to go through it at your own pace and when you were feeling better in terms of absorbing all this information. Okay.

[0:16:22]

Respondent: Yeah. I think it’s good to have that… sorry, that… I think it was about three months to have that follow-up for that reason, so, you know, not to wait for a huge amount of time but to have a follow-up fairly soon afterwards so that it allows you to process what’s happened, and I maybe had more questions then, which I think I probably did, at that follow-up, because obviously even if you… it was still a shock even though you think you’re prepared for it, it’s still a shock.

Interviewer: Of course. It’s never the news you want to hear, and there’s only so much you can…

Respondent: No.

Interviewer: prepare. Okay. And you’ve started taking part in Enroll after you got your result.

Respondent: Yeah, yeah. It took a while to get onto that, but yeah, I finally got onto it.

Interviewer: And are you seeing… who are you seeing now? Like you’ve mentioned you know about the Huntington’s advisor, for example, you know they are there, they exist, but you’re symptomatic, so who do you see now in terms of professionals?

Respondent: Well, I’m not symptomatic yet, as far as I know. So, I’m not seeing any professionals specifically for HD. I spoke to, again, the… I can’t think of [name 2]’s last name, but the lady at the centre in [location 2] recommended that I try to or recommended that I met with my GP, which I have done. As I said, the really good GP lady has gone now, she left the practice, so I’ve kind of set up a… it was supposed to be a regular meeting with the GP specifically to sort of go through the HD stuff, but I didn’t get the feeling that… it was… I know you shouldn’t really do that, but it was kind of me telling them what HD was and how it affected people. So, I’m not actually… we agreed that I’d go into the GP once a year specifically almost to be monitored and checked I’m alright, etc. I’ve not done that. I only went in once. I have been into the GP since, so it will be on my notes, I guess, but I did get a feeling that, as much as they were happy to, as a GP, look after me or treat me, certainly the GP I spoke to had very little knowledge about HD.

Interviewer: Okay. So, at the moment when you got your test result, you thought, in conversation with the geneticist, that it would be good if you kept regular contact with your GP.

Respondent: Yeah.

Interviewer: The good GP, as you said, has left.

Respondent: Left, yeah.

Interviewer: You have a different GP now, which you haven’t really established much of a connection with yet.

Respondent: Yeah, yeah.

Interviewer: And the geneticist thought it would be good if you go yearly to sort of educate your GP, but you just haven’t quite gathered the energy for that yet.

Respondent: Yeah, yeah. Yeah, I think that’s about right. In fact, the geneticist, did she use the word advocacy for HD? “Go to the GP and be an advocate for HD”, so yeah. I mean I had thought about joining the… they’ve got a patient group, I had thought about doing that, but I guess whilst I’m not symptomatic, I guess a bit of me thinks, well, let’s just let things carry on until I feel I need something, you know, something extra.

[0:20:57]

Interviewer: Have you been to the GP at all after you got your positive result one or two years ago?

Respondent: Yeah, the GP then, it’s quite a large practice so the lead GP wrote and said, “We were sorry to hear that you’d got a positive test. Would you like to come in and discuss it?” so I did go in once, and that’s when a GP that I’d been treated with before suggested that we have a regular… well, he asked me, “What do you want to do next?” so I said, “Well, maybe an annual kind of a check-up really”. But they’ve not proactively followed that up and I’ve not done myself, but I do go in quite regularly. I’ve got other… so I’ve got [type of arthritis], so I go in at least once a year for a blood test for that. And I’ve been in recently for, given my age, I’ve had a PSA test done recently, and various things. And I’d tried to keep fit, so I’ve just fallen off my bike, so I keep going in with bumps and bruises and things as well, so yeah, it’s not… I’m not a typical man that doesn’t go to the GP. I will go.

Interviewer: Yeah, yeah. You were just saying that Huntington’s-wise, you’ve not felt there was anything to be discussed now.

Respondent: No.

Interviewer: Yeah, okay.

Respondent: And I don’t really want to waste their time either because I know they’re… I work with the [REDACTED EMPLOYER] so I’m not, you know, I know they are quite stretched.

Interviewer: What do you do, [name 1]?

Respondent: I do [construction work]. I don’t work for a trust, I work for a national organisation.

Interviewer: No, I mean I’ve not used your… I don’t know, I used to work in [location 3] and, yeah, we would call maintenance plenty, plenty of times to fix our small office that was falling, just crumbling, basically.

Respondent: Yeah.

Interviewer: So, thank you for your service, and the [REDACTED EMPLOYER] needs you. Okay. Good. Now I understand you’re like… if you go there, you know, when you go for your [type of arthritis], for example, to get your bloods done or if you have any injury, you go for your injury, and with Huntington’s it’s a bit like, well, you don’t feel like the clock has started ticking or something.

Respondent: Yeah, yeah.

Interviewer: So, it’s a bit like, well, if I go there with generic questions, they’re likely not to know of it.

Respondent: Yeah, yeah.

Interviewer: Else you go with a question and the answer.

Respondent: Yeah, well, again, I feel like that would be the case because, again, like I say, I’m quite inquisitive so I will, I get the HDA's newsletter through and I read that, so I look at any new developments, so I do research about various things, and do the things you shouldn’t do and every time I can’t remember something, I just suddenly think, “Oh, this is it, I’m going to start,” or you read something about something else and think, “Oh, yeah, that might be an issue,” but hopefully I keep a rounded view on it and keep a positive mindset around the symptoms, etc.

Interviewer: So, when that happens, [name 1], when you’re like, “This is it, you know, it’s starting,” what do you do then?

[0:25:07]

Respondent: I kind of just personally assess. So, I’ve done, with work I’ve done like CBT [cognitive behavioural therapy] and things like that, so I kind of rationalise the thought. So, it’s probably no different to anybody else, but I’m [learning disability] . One of my [learning disability] traits is names. I can’t remember names and I can forget names of people… I can forget people’s names who I’ve been friends with for years, but I’ve always had that, well, obviously with being [learning disability], I’ve always had it. So, when that happens, so quite often that will happen with a work colleague and I kind of, over the years, you develop little techniques, deflection techniques to get someone else to introduce somebody. So, I can walk into a room and be next to somebody that I’ve walked into the room with and go to meet somebody that we don’t know and introduce them and I can forget their name. And as I say, so when things like that happen, you know, “Oh, is this… maybe is this me-[learning disability] me, or is this something [HD] that’s starting?” So, it’s kind of just… I mean sort of self-effacing or self-critical and that sort of CBT, stoic mindset and thinking, “Hang on a minute, don’t be silly. This has always happened. It’s not”. And then my wife helps, so she’s been great. So, we have discussions about it, you know, on a regular basis, so she’s been really good. So, yeah, it helps to bounce stuff off. And same again, I’ve got as a kid and I’ve always waggled my feet, I remember my dad used to go crazy with me because I’d sit in a chair and just waggle my feet all the time, so I catch myself…

Interviewer: You’re restless.

Respondent: Pardon?

Interviewer: You’re naturally restless.

Respondent: Yeah, restless feet. So, I catch myself doing that now and I think, again, it’s the same thing, “Is this the chorea?” Yeah, so I guess when I do things like that and I spot it, I put myself at ease with it all and think, “Yeah, you’re fine. You’re alright”. But also trying to… it’s only a mental note I make, and I probably should diarise things more, but I kind of make a mental note to think, well, how many times as something happened over the last however long and look at it like that just to try and sort of evaluate whether anything is getting worse rather than it just being as it was before, if that makes sense. And I’m thinking actually that was the one thing the GP, and I’ve not done it, when I met him, he sort of said, “Oh, you should start diarising little events so you understand them and also you’ve got a record to see if anything’s getting worse,” and I haven’t done that and I really should do that.

Interviewer: Mmm. Who would you go to if you were really worried with something about your Huntington’s?

Respondent: Other than sort of family, other than, well, yeah, I guess if I got to the point where I got really worried about things, I would contact the support element of the HDA. I wouldn’t… my GP wouldn’t be my first point of call. Potentially, the geneticist at [location 2], if feel like I’d contact her. But yeah, I think now I think I would probably go to the person that provides the support locally for part of the HD Association.

Interviewer: Have you ever linked with them before?

Respondent: No, I haven’t, no. I must admit, I have… sorry, go on.

Interviewer: I was going to say, you know they exist, right?

Respondent: Oh, yes, yeah, yeah, yeah. And I know that… I think it’s a lady which covers the whole of [location 4], I know that both my sisters’ family have been in touch with them to help with my sisters. And as I understand it, they [HDA advisors] have been some quite good help with them. And to be honest with you, the biggest thing that kind of concerns me now is my children, and that’s probably the one negative side of it that affects me in a negative way, you know, I’m quite stoic, quite positive with personal issues and problems and illness, I kind of just get on with it and look at ways of dealing with it as best I can, but it does obviously, it does worry me about my kids because they’ve not had tests, so they’re obviously just at risk at the moment, and they’re both male. My eldest is [20-30] and my youngest is [20-30], so they’re both kind of coming to an age, and because, including my mum, all our family seems to have sort of become symptomatic towards middle age and older, but I sort of thought, oh, you just carry on and you get it in old age, sort of thing. But I do worry that my kids are coming up to 30 now and I think, well, can they develop it? Will they develop it early on? So, that’s the biggest worry. And I suppose I might… that’s something I had thought about contacting the support.

[0:32:39]

Interviewer: Do they know you’re positive, [name 1], your children?

Respondent: My children? Yes.

Interviewer: They know.

Respondent: Yeah, again, I mean they’re both a bit like me really, they’re quite pragmatic and although we don’t really talk too much about it, they are… they’ve been supportive for me and they generally, from what I can see, have a very similar outlook to the one that I’ve got, for themselves. Certainly, the youngest one has more than the eldest. But I’m sure it will… and they’ve got partners. My eldest one’s just got married, so again they’re kind of coming to that time of life when they’re thinking about children, as we’ve discussed with them. So, I guess it will play on their minds and they won’t know what to do. We don’t really discuss it, we don’t hide it, but we don’t discuss it. You know, I kind of left it up to them if they want to come and talk to me about it. But yeah…

Interviewer: But it’s not something you’re going to be talking every day.

Respondent: No, yes.

Interviewer: It lives with you constantly, but it’s not something you would always be banging on the same nail.

Respondent: Yeah, yeah.

Interviewer: Okay. I guess you’re saying how can you maybe… although you don’t have to always be discussing it, should you sometimes be discussing it and how should you guide the conversation? So, you’re saying probably the Huntington’s charity could help you have better conversations maybe with your children.

Respondent: Yeah, yeah.

Interviewer: Okay.

Respondent: Yeah, so that is something I’ve thought about doing, but I haven’t got… you know, I’m not really… I haven’t done as yet. That would be the kind of initial conversation that I’d have with her [HDA advisor] then, it would be around a bit more understanding about what the two boys can do going forward. And they’ve researched it and I’ve researched it myself around testing embryos, etc., for the gene and things like that. And I suppose they haven’t… they would tell me, so they haven’t, I know they haven’t had the tests, so they both are just at risk at the moment. So, yeah, yeah, so that would be… that’s the only thing that really worries me now because I kind of think, well, I’ve got to a point where, as I say, I’m retiring in [less than a year] months’ time, so, you know, I’ve had a good life so far and I’m going to have a nice time while I’m lucky that, you know, I can enjoy life for at least ten years and I’ll just keep doing that. If I get to 70 and I’m alright, I’ll think, right, I’ve got another ten years to go and just take it on the chin when…

Interviewer: Those are good problems to have, that’s the…

Respondent: Yeah, yeah.

[0:36:19]

Interviewer: So, with work, did you disclose at work or, you know, with your insurance and things like that? What…?

Respondent: Until I got the test done, we’d never… so I don’t have life insurance because I’ve got the [REDACTED EMPLOYER] pension. We’ve got, my wife has worked in [healthcare] as well, so we’ve both got similar sort of pensions that have some form of life assurance attached to it. So, we’ve never applied for insurance in that respect, and certainly not since we’ve been tested, since I’ve been tested. We’ve just got some new or we’re getting some new things like travel insurance and declaring it on there now, because I’m [chronic lung disease] as well. So, I’m [chronic lung disease]. I’ve declared the [type of arthritis] and declare the fact that I’ve got HD as a positive as well. So, you know, we can… it costs a little bit more, but insurance is available. But certainly that was one of the reasons when we were younger, in fact I was probably about… no, I was bit older actually, I was in my mid-30s, we’d already got the kids when my mum was diagnosed, but it was certainly one of the reasons for not getting the test done was just around things like long-term loans or mortgages and things like that that we were worried about. But, you know, I know now, although it probably costs more, I think things have got a lot better in that respect than they were in around about [2000-2005]. It was quite difficult to get certain types of insurance, but yeah, no, since I’ve got the positive result, I declare it or we declare it on anything we’ve got where you have to declare it.

Interviewer: Have you ever said anything at work, like to occupational health, sorry.

Respondent: Yeah, so again, it’s a really good company that’s part of the [REDACTED EMPLOYER], and I’ve got a really good, fortunately, got a really good line manager. So, I actually spoke to him about it before going for a test. After my dad died I was struggling mentally anyway and I used the… so we’ve got a colleague support system at work, so you get a certain number of counselling. So, I had counselling at work, so work’s supportive and my line manager is very supportive. So, he’s aware that I’ve been tested positive, and again I’ve kind of said to him, you know, “If you notice something, if you start noticing something regularly that’s a bit odd, other than me normally being odd, you know, let me know”. I know he would do, but yeah, no, he’s been really good. I was very lucky. Good manager.

Interviewer: That’s excellent. Yeah, no, I mean I think to have that extra pair of eyes, again, for you it’s a bit harder, you know, you have to put more effort into going is this the normal me or is this the HD me?

Respondent: Yeah, yeah.

Interviewer: So, to have that extra pair of eyes that are more impartial to say, “Well, you know, [name 1], we’ve noticed this is happening while it wasn’t before,” so it just gives you that extra reassurance that I’m fine, you know.

Respondent: Yeah.

Interviewer: Okay, alright. [name 1], is there… because you’re quite early in your journey with your personal journey with Huntington’s, you know, you’ve only recently, fairly recently found out you were positive, of course you’ve lived through it with your mother and your sisters, of course. Is there anything you wanted to be in place that it’s not, or anything that you would like or it would have been helpful if this was happening or we had this extra support or…?

Respondent: Well, I don’t know really. I kind of think from the geneticist’s point of view, but they have said, you know, if there’s anything you… they’ve always said, “Always contact us,” and they’re keeping it a regular meeting, I guess from a clinical point of view, I don’t know what you would do, but I guess in my head it would be nice if you could go and just have like a regular assessment. And I know the HD-Enroll, I’ll kind of get that, I assume, but just generally check your cognitive ability, etc., so check the symptoms. But I don’t know how that would… so in my head that would be really good. If I could go once a year, every six months or once a year and have, you know, like I do…I guess it’s like I do for my [chronic lung disease] and like I do for the [type of arthritis] kind of thing, have a blood test, you know, and check your breathing, so those things, if you could have something where, in an ideal world, if you could have something where you went every six months, you went to a specialist who understood the disease and they tested you for symptoms and they checked you against your previous tests and could say to you, “Everything’s fine,” or, “There’s been some change with this. You might want to think about this with your GP”. So, I think, again, I realise time and money, isn’t it, but I think that, for me, would be a great way to sort of go forward with the management and treatment of my personal journey or symptoms. So, you know, all those things. Sorry, go on.

[0:44:24]

Interviewer: No, no, finish.

Respondent: Yeah, no, I think it’s just having all those… because, again, there are so many symptoms or symptomatic things that you can have with HD, just having them checked. So, something I do, again, when I spoke to the GP about it, they said, “Oh, it’s probably like something reflux,” but every now and again, if I’m gulping water, I kind of choke on it or it feels like it gets stuck. So, again, straight away I think, “Oh, God, I’ve got some sort of… you know, I can’t swallow any more”. (laughs) And I mentioned that to the GP and they were just, “Oh, I’m sure it’s just a reaction to cold water and you spasm,” which I’m sure it is, but if you could have all those things checked and tested, it’d give you the reassurance. And also, I guess, I suppose in many ways it would also help with future research because you are actually tracking everybody’s progression through the disease, so it would give clinicians also a better idea of how things do develop and what things get worse and what things could be treated at different stages, I guess. But certainly for me, it would put my mind at ease if I had that regular checkup, if you like.

Interviewer: It’s interesting how you get review for your [chronic lung disease] and you get a review for your [type of arthritis], but then you have a lifelong hereditary disease that potentially affects the whole family, but then they leave it to you to decide…

Respondent: Yeah.

Interviewer: When and from whom do you need help.

Respondent: Yeah.

Interviewer: It’s peculiar.

Respondent: (laughs) Yes, yeah.

Interviewer: I’m not sure how [chronic lung disease] and [type of arthritis] are more… are a higher priority to be checked, but…

Respondent: Well, they’re probably not for me, but… and again, I put my pragmatic head on, there are more people with [chronic lung disease], more people with [type of arthritis], so you’re getting… it’s a numbers thing, isn’t it? You know, so it’s… you get better value from treating… monitoring as well as anything else. It’s easier to train people to treat an issue or a problem that’s on a mass scale than it is to… you know, why would you… how… the cost of treating… educating all GPs, and their brain wouldn’t be big enough, to understand all problems and diseases, etc., to the same extent as how to treat [chronic lung disease] or how to treat [type of arthritis] or something, or things like that, you know. So, that’s always going to be a problem, but it would be good, I guess, for them to be able to say, well, just as easily as understanding that, you know, if you’ve got cancer, you go to an oncologist and then you would go to them on a regular basis and they would test various things. Yeah, so you do feel a little bit kind of left to your own devices, which, being the sort of person that I am, I don’t necessarily… it’s not something I’m mindful of or it doesn’t bother me because I’ll go and do, you know, I’ll go and do the research myself. You know, I will check myself and I will, I follow… try to follow dietary advice and exercise, you know, so I am quite positive, proactive around my own health most of the time. So, it does worry me, I guess it worries me, like both my sisters are not like that, so it does worry me that people might… they would fall through the cracks really, I guess, if that makes sense.

[0:49:56]

Interviewer: Yes, yes, exactly. No, it does. And as you’re saying, of course the GP can’t know every disease, particularly the rare ones, it’s not value for money, but if you have cancer, you go to the oncologist. So, do you have a specialist Huntington’s team in your area, do you know?

Respondent: Well, they’ve got a good geneticist in [location 2]. Whether they’re particularly HD specialist, I’m not sure. But the clinician that I’ve been involved with has been very good. Yeah, and that’s somewhere in [location 2]. I’ve forgotten again. (inaudible 0:50:54), yeah.

Interviewer: When you went for your research appointment, was there a neurologist there or who did the tests, for the Enroll?

Respondent: No, the lady that did my enrolment was… was she a neurologist? She was a nurse. Again, I’ve forgotten.

Interviewer: No, that’s alright.

Respondent: But she wasn’t a… she certainly wasn’t a consultant. Whether she was a neurological specialist nurse, if there’s such a thing, I don’t know.

Interviewer: You said she was not a consultant.

Respondent: She wasn’t a consultant.

Interviewer: She was not a consultant, okay, yeah.

Respondent: No.

Interviewer: Yeah, okay. So, you know there’s a geneticist, a good geneticist, but you don’t know if there’s like a neurologist or neuropsychiatrist that knows about, like really knows, as expert, knows Huntington’s?

Respondent: No, yeah, I’ve not been made aware of anybody.

Interviewer: Yeah, that’s fine. Yeah. I’m not saying you should, I was just curious in terms of if there was a specialist team up there. But then again, as you were saying, if you did notice that you were concerned that the symptoms were starting, you would link with the Huntington’s advisor, and they would know, of course.

Respondent: Yes, yeah.

Interviewer: So, I’m not saying you should know, I was just asking.

Respondent: Yeah, I guess, yeah, so there is a process there, so, yeah, I would get in touch with them [HDA] and then they would signpost me, yeah. But I don’t know personally.

Interviewer: Yeah. [name 1], can I ask you a bit about your experiences with your mother and your sisters, which of course are more advanced? My interest here in the question is if they were well supported by the healthcare system or with your view, as son and sibling, if you thought there was need for improvement. It’s a bit of a generic question, but since I’m trying to understand how professionals could work together better and since there’s so many different people involved, do you have some thoughts that you would like to…?

Respondent: Yeah, so, well my mum’s been passed a long time ago, and I think, as I said, as my understanding about the… it’s my understanding about the clinical world’s understanding of HD, I think [2000-2005] was quite an early, you know, developments were quite sort of… hadn’t been, well, out there at that point, so I think… and my mum and dad weren’t very open to… my mum and dad were very, very old-fashioned, very stoic, you know, they’d gone through the war and all that sort of business. They were quite old parents for me and they were old-fashioned even being old parents. So, they wouldn’t talk about… I was amazed that my sisters were… we were told what mum had got really, because they just would not talk about illnesses or problems that you had, but it was just their kind of generation. So, I don’t really know how well mum was supported in that sort of era and because of how they were. Certainly, actually, thinking about it, I’m assuming… I think it would be like a genetic nurse came and spoke to us with mum at mum and dad’s house, but again, that was kind of, “Here’s a leaflet about how it can affect you and off you go. Give me a call if you want testing,” kind of thing. So, yeah, so that was a bit… whilst it was good to understand what was wrong with mum and how that… what implications that had on us, it was kind of, well, this is what your mum’s got, actually it can affect you, let us know if you want to know, and that was it, gone, finished.

[0:56:38]

And I guess that, again, all three of us decided not to have a test at that point, so I guess from a clinician’s point of view it’s difficult to do any follow-up if someone doesn’t want to have the test done or doesn’t want to understand about things. You can’t really force it upon them, can you? So, yeah, from that point of view, I don’t know if mum had the right level of treatment. And then going forward, certainly my mum was at the same GP that I’m at now, and there was, from what I can see, very little understanding then of her condition really. And then I can’t really say for my eldest sister so much because obviously her children have looked after her, but again, from the bits I know, even knowing my mum’s diagnosis, it’s always seemed difficult for… not being present at some of the conversations my nieces and nephews have had with clinicians, both sisters, particularly my elder sister, I think they’ve found it hard to get clinicians to consider Huntington’s as being the main problem with… sorry, is your throat bad?

Interviewer: Sorry. I’ll mute myself so that you don’t hear me coughing, but I can still hear you.

Respondent: Oh, yeah. Okay. Yeah, so thinking about it, the clinicians that, certainly for my eldest sister, were kind of almost dismissive, kind of like the GP has been with me. And I know some of that will be, you know, “Don’t worry about it. You’re okay. It’s fine,” but it does feel a little bit, you know, you are constantly asking the question, “Is this because of Huntington’s?” And the answer you always get is, “No”. Their first answer, any clinician I’ve spoken to about it, and from what I’ve heard of my nieces and nephews, that’s the first answer that you get, “No, no, it’s not Huntington’s, it’s this or that”. So, my eldest sister has psychotic episodes. She was quite manic. So, they were asked the question, “No, it’s not,” because she’d not have the test done and they used to say, “Well, my grandma had Huntington’s and it’s on the death certificate, she died of Huntington’s, could this…?” “No, no, it’s not Huntington’s. It might be, well, we’ll have a look at this or we’ll have a look at that”. And then, you know, then she developed, I don’t know the name of it, but the stiffness, so when she started getting… she’d go…

Interviewer: Very rigid.

Respondent: Yeah, yeah, and that was, “No, no, it’s not Huntington’s”.

Interviewer: Well, I guess how much value does a no have when it’s an ignorant no? When you know, when you know with a K, when you know that the no with an N comes from someone that doesn’t know, I mean do you really sleep better?

Respondent: No.

Interviewer: No, because it has no… it’s actually worse because, as you’re saying, it’s dismissive. The problem hasn’t disappeared, the disease hasn’t disappeared, they’re just sweeping it under the carpet.

Respondent: Yeah, yeah.

Interviewer: I am so sorry because I mean that is deeply, yeah, ignorant, because of course the psychiatric symptoms are one of the components and the one that most bothers families and people.

[1:01:43]

Respondent: Yeah.

Interviewer: And to have psychotic episodes and to be told, “No, no, this is not Huntington’s,” I mean I don’t even know what was crossing… I mean I’m a bit puzzled if they truly thought no or did they thought saying no would make the family feel better because, again, it’s not like a psychotic episode would disappear. So, the problem is still… God knows what… I mean what crosses people’s minds.

Respondent: But, yeah, and I don’t know why, but both sisters, from what I’ve gathered, have… it’s taken a long… so as far as I know, my eldest sister’s never had the test, it’s just that my nieces and nephews have actually contacted HDA and said, and obviously then can come in and help anyway because it’s in the family, so she’s at least at risk, but the fact now that she can hardly talk and she’s bedridden beggars belief, I guess. And then again, my other sister, whilst… I suppose if a clinician GP was to ask is there anything… is there a family history of anything that you think might be causing this, and you didn’t say Huntington’s, and I could well expect my youngest sister not saying Huntington’s, then I guess it would be difficult for them to… or why would they suggest that? I don’t know. Certainly, I think… I’ve got an alarm going off somewhere. I don’t know why.

Interviewer: What’s that? Oh, is it your phone? If you need to take it, take it.

Respondent: No, no, it’s my work’s phone and I don’t recognise the number anyway. Oh, it is on silent. That’s weird. I’ll throw that away. (laughs) Oh, heck, I’ve lost my train of thought. So, my youngest sister went through a long, long, long process and, I must admit, I didn’t raise it with her because of a lot of the difficulties I had with communicating with her. But she’s gone through a lot of issues, I think sort of neurological-related, and it’s taken a long time for them. It seems to have taken the clinicians a long time to come to the point of either… I don’t know whether they recommended that she had the test or whether she finally thought, “I need to have the test done.” But yeah, I think it seems to have… there’s lot of things that my sister’s had issues with that for me would… the first thing I’d think about is is this HD, Huntington’s, or is it something else? And you think, well, if Huntington’s is in the family and it can cause this issue then surely that would be the first thing that you’d think of.

Interviewer: Yeah.

Respondent: But because I’ve never had the conversation with my sister, I don’t know, because she might have been not relieving that… not making clinicians aware that it’s in the family. But she’s had all sorts of tests. Even she’s had problems with being light-sensitive. Now, I don’t know if that’s something that HD can affect, but, you know, I suspect it might be because…

Interviewer: Yeah, some people report that, yeah.

Respondent: Yeah, oh, well there you go.

Interviewer: Yeah, I mean it’s a disease that affects the brain, so the variety of symptoms and how people feel differently with it, it’s, yeah, while the chorea, you know, with the movements, people are like, “Oh, yeah, that’s it,” I think with this sort of sensitivity to light, it’s harder to make the connection that it could be. But there are people that… but I just mean this in terms of my clinical experience with different families. Like I’ve had patients saying, “I have sensitivity to light,” or, “I’ve grown to have sensitivity to light”. Now, if it’s directly connected or not, I don’t know, but I know there’s people that complain.

Respondent: Yeah, yeah.

[1:07:18]

Interviewer: But again, I think, as you’re saying, if people don’t want to be tested or if they don’t even raise it with the clinical teams that it could be that, it of course becomes difficult. But surely someone with a connection at some point so they’re still linked with the charity, which is a bit like if it walks like a dog, talks like a dog, maybe. But even if it’s not, you know, the support, referring to the right teams and so I believe you still should have care despite not having the diagnosis. If a person has a psychotic episode, if a person has depression, you still need to treat all those things. But, of course, it’s not making anyone’s life easier, only because it’s much easier to follow the process if you know which process you’re following, you know, which category do you fall into it, which is not very nice that we’re like categorising people, but within the system it sort of helps. Okay, [name 1], I’ve taken up a lot of your time.

Respondent: That’s fine.

Interviewer: I’m going to ask you a couple of boring questions that I ask everyone.

Respondent: Okay.

…

[redacted]

[End of Transcript]