**ID 6686798415**

**How did you find out you had Wilson disease? Context of the diagnosis, i.e., screening?**

[Audio starts 00:00]  
[Discussion about technical issues and introduction]  
  
Excuse me, I am not very well today. I am currently 61 years old and I was diagnosed with the disease at the age of… I must have been 14 or 15 years old. The problem that I had was that when I was younger I often had awful attacks of indigestion. Vomiting, with stomach pains, diarrhoea, and then I had joint arthritis and my mother took me to see all the doctors. She took me all the way to Paris, because I lived in [PI 02:28] to see what was wrong with me. Nobody really found what was causing the problem. Then at the age of... Yes, I must have been 14 years old, in the third year of high school, my writing started to be affected. My writing changed, and uncontrolled movements which started to become noticeable. I also had problems with my speech. I was hospitalised at [PI 03:06] and they did examinations. Initially, they thought that I had chorea because it was the same symptoms. They treated me for chorea, I don't remember how long I took the treatment for, but for a short time, and they made me return for further tests because they thought they'd found something. Actually, it turned out that they'd found Wilson disease after the second or third period of hospitalisation and examinations. So when the professor informed us that they'd found it, she did not tell us that it was a serious disease at all, that it should be treated for life. She told me 'We are going to give you Trolovol'. I don't remember how much she gave me initially as this was a long time ago and I haven't kept the paperwork from back then. In any case, I remember very clearly that she told me to take the treatment for 3 weeks and it would be resolved. She absolutely did not tell me that the treatment had to be taken for life, and above all, she did not say anything at all about diet. At the hospital, they gave me hot chocolate in the morning for breakfast, because at the time I was a kid and they did not pay any attention to diet at all. So, at around this time my parents moved from [PI 04:45] to [PI 04:45], I had my treatment for 3 weeks as she told me to, and there was no improvement. So, I stopped the treatment.  
  
(Just a moment, I will ask you a few short questions before we go any further. Approximately how old were you at the time of diagnosis?)  
14 or 15 years old. I was in the third year of high school.  
  
(Do you remember which type of doctor made the diagnosis?)  
Yes, she was a neurologist, (PI 05:27).

**Signs and symptoms [skipped]**

**How did you feel when you received the diagnosis?**

Well, nothing in particular. However, what I do remember about when I was hospitalised is that I was treated a bit like a circus freak, because I had the ring in my eye which was magnificent, apparently. So, there was a whole procession at the device, not a microscope but that thing they use to look at the eyes. There was a whole procession, without asking for my opinion or anything at all. It was the same, they took me into a room where there were, I don't know, maybe 200 interns to get me to walk, but without asking permission from my parents or from myself. They said 'Come along with us' and they made me walk around, as I had problems with balance and everything and they made their comments. Well, particularly at the age of 14, it was not very sensitive. I really remember that clearly, it is something that left a mark on me. However, the diagnosis, no, because as she did not really tell us about the severity of the disease, that it would be resolved after 3 weeks, I thought okay, I will be better in 3 weeks, that's it, we'll see. I was not particularly traumatised by the diagnosis.

**Which of the symptoms of Wilson disease did you struggle with before treatment?**

Well, it was mainly the neurological problems. I could not walk straight, the walking, and speech.

**How did you deal with these symptoms?**

Before the treatment? Well, when I had my arthritis I cycled to school. My parents did not have a lift and we lived on the third floor, so I managed as best I could. I had painful knees and my wrist joints hurt too. How did I manage? Well, I tried to manage as best I could and that's it. I am quite an optimistic and energetic person so... I tried to manage. I was well supported and I had help, so that's it.

**How have your symptoms changed over time/after treatment initiation?**

Well, the first treatment that I took for 3 weeks, the Trolovol, I did not see any improvement because I did not take it for long enough. That is the major criticism that I would have for the doctor at the time, it is that they did not tell me that I had to take this treatment for life. So after 3 weeks, as I saw that it wasn't getting any better, stopped everything, I went off on holiday with my uncle in Italy because my parents had taken over a shop, and I ate... I ate everything, and so after that I went into the fourth year of high school at [PI 09:47] and I tried to hang on to be able to continue my studies, but I was no longer really able to write. So my schoolfriend wrote the lessons down for me with a carbon copy, at the time there were carbon copies! So I tried to hang on in order to continue my studies, but I was unable to learn. I was unable to write, I could see that I wasn't managing. So, I stopped my studies the next April, all the more so because it was a private school, so my parents were paying for it. So that's it. My schoolfriend who was writing my lessons down said 'Listen, [PI 10:38], if you can't do it, there is no point in persisting, stop and that's that," and so I stopped. Excuse me.  
  
[10:50]  
  
(No problem.)  
So I stopped, and in April I had to stop, and I still didn't have a diet nor a treatment, and my condition gradually deteriorated all the more, because the treatment had not been taken for long enough. I was not on a diet, that's it. So, I was starting to become... I could see myself starting to turn into a vegetable because I could no longer do anything. I couldn't go to school anymore, everything I touched fell on the ground, I helped my parents in the shop, but I felt a bit, how to put it? Like a vegetable, you know? That's it, like a parasite, that I was I unable to do anything, and that's it.  
  
(How did you come to take a treatment more seriously?)  
I will explain. I had a raging throat infection the following winter, and my mother called out the doctor. It was not even our family doctor, but another doctor who she found to treat my throat infection. When he examined me, my mother spoke to him about Wilson disease that they had discovered in [PI 12:18] and she could clearly see that I was not well, and that there was no improvement. She realised that. The doctor said 'But Wilson disease is a very serious thing that must be taken seriously! It must be treated with medication otherwise it can be fatal!'. Because he said that, we were a bit frightened. He advised my parents to enrol me in a healthcare centre to treat my disease seriously, with the diet and the medical supervision, and also, I could continue my studies. So, I was enrolled at [PI 13:09], it was not very far from [PI 13:15], it was near [PI 13:15]. I started to take the treatment there, I think I had 16 Trolovol pills per day initially, because I was really quite advanced, I think, and there was also the diet. Then within 6 months, I made amazing progress and so I was enrolled at school. I started the fourth year of high school there too, but we did not know what the progression would be like so I changed branches and opted for short studies. I chose a secretarial qualification, because initially I wanted to...  
  
(Your symptoms were actually quite well managed with the treatment, is that correct?)  
Yes, the treatment, and above all, the diet, too. The diet. I had a really drastic diet. At the moment, I am in the care of the hospital at [PI 14:17] at the neurological hospital where they discovered my disease, which has become the centre for... I don't know if you are familiar with [PI 14:27]? There is a specific department for Wilson disease, which I discovered by chance. It has become much less severe than what I'd had all my life.

**Do you still have symptoms now? How do you deal with the symptoms now?**

Yes. I still have a few micro-movements which interfere with me, but I live with it. I might be a bit clumsy sometimes, I sometimes drop things. It is mainly my left side which is affected neurologically, as opposed to the right side. Fortunately, I am right handed. Recently, I don't know if you are going to ask me about this later but they changed my treatment. I remained on Trolovol for over 30 years, I can't remember. Yes, because they changed that last year. I don't know if this is the reason or not, but I often have problems swallowing. I also see a speech therapist and she tells me that I must be extra careful with swallowing due to my neurological problems. I must not speak while I am eating, I must hold my head nice and straight. It is true that isn't an automatic reaction for me yet, because I only recently started seeing the speech therapist. That's what I have at the moment.

**Do you experience any other serious diseases?**

No.

**What challenges are you facing now living with Wilson disease?**

Well, as I said, apart from the trouble swallowing, when you are at the restaurant it is not very elegant, but now, at the age of 62, I have almost reached the end of my life so I think that I have lived quite well. My entourage, everyone knows that I have this problem. I do not really have any constraints.

**What do you see as the biggest challenge(s)?**

The aspects of Wilson disease which bothers me the most are the neurological problems that I have on the left side, with my clumsiness. Sometimes I am not able to do very accurate things, so I ask for help, and that's it. I live with it. I manage.  
  
(Does the disease impact your mood?)  
No.  
  
(What about managing the disease on a daily basis? Is it not too difficult to have to take a treatment every day for life? Are you okay with that? How many pills do you take daily?)  
I used to take Trolovol in the morning at breakfast, like they did at the medical centre where I stayed for 2 years. That was fine. However, they have changed my treatment now and I have to take in the morning half an hour before eating, and in the evening 2 hours after eating, so there are constraints. It is mainly in the morning. What I do is, I put it next to my bed and when I wake up in the night or early in the morning, I try to take the pill as I wake up but it kind of disrupts the night. That is the constraint that I have, because if I have to wait for half an hour or an hour to eat breakfast after getting up, when I go to work and everything, it isn't convenient.  
  
(Is it a challenge to take your treatment every day, keeping in mind the fact that sometimes you may not have any symptoms? It is sometimes difficult to take a treatment when you do not feel ill.)  
No, no! Yes, because as I said, initially I took 16, then they reduced the ration and then it went down to 8, then 4, then when I left the centre they put me on 2 pills. I took 2 Trolovol pills every day all my life. No, it became automatic, it was not a burden for me at all.

**How has this changed over time (or was it different at other times?)**

(Okay, so you continued the treatment, you took Trolovol for 30 years and there were no problems, did the symptoms reappear?)  
Yes they did, that's why they changed it. I had side effects, in my neck. The first one that I had in [PI 20:12] because I was treated by my GP, but he is not specialised in Wilson disease.  
  
[20:18]  
(Yes.)  
As he is going to retire, and it is very difficult to find a GP, I wondered what I was going to do about my prescriptions and everything. So I turned to [PI 20:35] and really by chance, I discovered the Wilson disease centre at the neurological hospital in [PI 20:42]. Sorry, I have gone off topic!

**What challenges do your relatives face if they also have Wilson disease?**

(Do any of your relatives also have Wilson disease?)  
None at all, I am a unique case in my family.

**What aspect of Wilson disease concerns you the most?**

As things stand, the doctor says it won't have an impact, but I caught the COVID last November, I had the tests that [PI 21:36] hospital prescribed for me and there were 2 liver markers which were slightly higher than the norm. The doctor told me not to worry, that it was the COVID which impacted the results, and that I would have to do the tests again in 2 months. I did them again last week, and those 2 markers are still higher than normal. I looked it up on the internet, which you should never do! It is the liver, they refer to liver cancer and things like that. My doctor said that there is no cause for concern at the moment, and that there is no need to do any more tests at the current stage. However, I still a bit worried, because my test results were always perfect all my life, I never had any higher markers for anything at all. I need to look at it, how are the indicators calculated, is it due to the disease? In any case, with the last 2, they had gone above. I will see him in April, we will discuss it face to face.  
  
(Aside from this, are you concerned about any other aspects of the disease, or are you not worried because things have gone well over the past 30 or 40 years?)  
No, usually... I am really lucky, because I know... Well, I don't know directly, but I have been in contact with a patient in Morrocco, via messages. She is a girl who I found on the internet, and she had all her siblings who had Wilson disease. I think she already lost a sister when I met her, and she is very ill, and she recently lost her brother and sister due to this, because in Morrocco they don't have the appropriate care to treat the disease. So I think to myself that I am very lucky because I have reached the age of 61 with this disease and I have had a good life. I am not to be pitied, compared to some other people.

**According to data from national health insurance databases in France and South Korea, only 44% and 35% of patients diagnosed with Wilson disease respectively, receive standard of care treatment, i.e., a copper chelator (D-penicillamine or trientine) or zinc salts.**

**Are you surprised by these findings?**

Yes. Why don't they take it? Do you know?

**What could be the reasons for this?**

Maybe because they haven't been informed as I was at the beginning. They weren't given enough information about the importance of the treatment. All the more so because my mother was not in favour of chemical medications, she preferred alternative medicine, so that's it. That might be why, but I am surprised, the figures are high.

**What experiences with Wilson disease treatment do you have?**

(You took Trolovol initially, what are you currently taking?)  
Cuprior.  
  
(Are those the only 2 treatments that you have taken during your life, for Wilson disease?)  
Yes. I took Trolovol until last year, and they switched me to Cuprior last September.  
  
(Do you not take any OTC medications? For example, zinc or other supplements?)  
No, because it is forbidden to take zinc at the same time as Cuprior. It is written on the label, when I went to the pharmacy, I asked for vitamins and she looked carefully at the contents to avoid anything that doesn't go with Cuprior.

**How often and where do you see your physician (within or outside the reference centre/ centre of excellence)?**

In the past, I never went, I just had regular tests and everything was fine. That was when I was under the care of my GP. He did my prescriptions. I didn't even go and see him, he sent them by email, and as my test results were good so we carried on as we were. Now that I am being treated at [PI 26:59] I have an appointment every 6 months.

**What does the relationship with your phyician look like?**

(How would you describe your relationship with your doctor? Is the doctor who you see every 6 months a neurologist or a GP?)  
The one I had before was a GP, the one who is going to retire.  
  
(So currently your GP is the person who you see every 6 months, is that correct?)  
No. No, no, I am now under the care of... Because the other one is going to retire, so this is what I was saying. I looked for another doctor and by chance, I found the Wilson disease experts in [PI 27:35] and I am now being treated for my disease by the doctors in [PI 27:41]. They are experts, and they are the ones who I see every 6 months. They are the ones who changed my treatment, because my doctor was a GP, he was not specialised in the disease at all.  
  
(How long have you been under the care of [PI 28:00]?)  
It will be... I discovered this last year, so it is very recent. Since September.  
  
(Did you have a good relationship with your GP before you changed?)  
Yes. Of course, yes. He is still my GP, I send him my test results. Of course. He had been treating me for years, there were absolutely no problems.  
  
(What about the current doctors? Do you have a good relationship with them? Is there a good level of trust between the patient and doctors?)  
Yes. Yes, yes.  
  
(Are the doctors who you see in the specialist centre neurologists?)  
There are gastroenterologists, I think.

**Can you discuss your concerns or problems with your doctor?**

**[not asked]**

**Who is supporting you and what kind of support do you get to manage your Wilson disease and your treatment (e.g., by reminding you about taking the drug, taking you for the appointment or picking up the drugs from the pharmacy or hospital, making decisions about treatment)?**

I am autonomous.  
  
(I did not ask you at the beginning, do you live alone or with your family?)  
No, I have a partner, he has been with me for 20 years.  
  
(Does your partner ever help you with your Wilson disease or not at all, do you manage by yourself?)  
When I can't open something, or there is something I can't do with my hands, I call him, he comes along and does it for me, there are no problems! He helps me a lot, he is amazing!  
  
(What about in terms of taking your treatment, not forgetting, etc.?)  
No, I manage that by myself.  
  
(Are you a member of any patient associations or anything at all?)  
I am a member of the [PI 29:59] association, that's all. I am under the care of [PI 30:03] and they had a patient day with the doctors. It was really amazing, I thought that was such a good idea, to meet other patients. It was really amazing, it was the association [inaudible 30:30].

**Which situation in your life led to stopping your treatment for Wilson disease?**

**How did it happen?**

[30:34]  
(Over the past 30 or 40 years, have you ever stopped taking your treatment at all for a while, or have you really never stopped, and always taken it?)  
Well, I wanted to try to stop. I wanted to stop taking Trolovol and take zinc instead, earlier you asked me about zinc. Because I had heard that zinc was helpful for copper and everything. I bought nutritional zinc, what I mean is that it was not medication, it was pure zinc or whatever, I don't know what they call it. A nutritional supplement, that's the word! I stopped, but for a really short time, maybe a month at the most. I wanted to do tests to see the results of the zinc and when I saw there was absolutely no copper in my urine at all, I thought oh no! Not good! I must start taking Trolovol again because this isn't good, if it isn't in my urine it is being deposited in my body and that isn't good. I did not talk to my doctor about it, I don't even know if he is aware. You are the first person I have told about this! I made an attempt to stop.

**Have you ever considered not taking your treatment, and for what reason(s)?**

**[skipped]**

**For how long have stopped taking treatment?**

I did not want to go back to how I was at the age of 14. I tried it for a month, and the results were negative in my view, so I soon went back to my treatment.  
  
(Approximately when did you stop taking the treatment?)  
It must be 15 years ago, 20 years at the most.  
  
(So you never stopped taking the treatment again since then? You basically did a short test to see what would happen, at least once, if you did not take the treatment?)  
Yes, that is correct.

**Did you discuss the decision to stop taking treatment with anyone?**

No, no. I did it entirely on my own initiative.

**If you could change anything about the treatment and generally management of Wilson disease, what would it be?**

Firstly, as a doctor, I would avoid treating teenagers or anyone else like circus freaks when they discover a rare disease, without asking for permission from anyone. That's the first thing. I was telling you about a meeting we had in [PI 34:11] with other patients, when I spoke to other people they were in the same situation as me, they were treated like circus freaks without anyone asking for their permission at all. I do think that as a doctor, you should use a bit of psychology with the patients. I understand that they need to study and look and everything, but they should explain and ask for permission first. That's it.  
  
(What about taking daily medication, is that a major burden for you or has it become habit?)  
No. No, as I said, it is not a burden at all. The only thing with Cuprior is that there are constraints around mealtimes, particularly for the morning, because it disrupts the night a bit, and if I take it later I have to wait to eat my breakfast. That is the only drawback with the treatment that I am currently taking, but with Trolovol I never had any constraints. I took it automatically.

**Is there anything else you feel is important in your experience with Wilson disease that we have not yet covered?**

No, I think that we have covered everything.  
  
(Do you currently have any remaining symptoms? You said that you have slight tremors in your hands occasionally, particularly on the left side, but other than that, is everything fine?)  
[Inaudible 36:17] but other than that it's fine, you can see that my speech is okay, you can understand me well, the speech therapist told me that my voice is very easy to understand although I don't like my own voice, but compared to the past when I had real difficulty enunciating and speaking, it is totally different now. I can be considered cured in a way, but I do know that you are never cured. That symptom is cured, anyway.

**Any other comments?**

(Just a couple more questions and I will let you go. Did you feel comfortable during the interview that we just did? Were the questions clear to you?)

Yes, no problem. I was very comfortable, when I didn't understand the questions I asked you to repeat, but that is more a hearing problem than a comprehension problem. I have problems with my ears. With the loudspeaker it isn't great. No, I had no problems, it was very good. Thank you for showing an interest in Wilson disease, I think that's a really good thing for the future and for patients' wellbeing.  
  
(Did you feel as though you had enough time to answer the questions?)  
Yes. I am thirsty now, I need a drink because I have talked so much!   
  
(Just one final question, as you know it is important for the people organising this project to hear about the experiences of people who have Wilson disease. Do you know any other patients who have Wilson disease who are not treated, or who do not take their treatment and who could possibly take part in this study?)  
Not me personally.  
  
(That's perfect, I was going to ask you not to give me any contact details directly, to remain confidential, but just to find out if you know anyone?)  
No, nobody at all.  
  
(No problem. Thank you, it was interesting to speak to you. One minor point, the company who recruited you will be in touch relating to the payment of your fee for this 45 minute interview, and aside from that, unless you have any questions for me, I will let you go and have a good day and lovely weekend!)  
Thank you. Are you in England?  
  
(Yes, exactly.)  
What's the weather like in England? Is it raining?  
  
(No, it is cloudy and a bit cold right now. It is not very nice, I hope it is better for you in central France.)  
No, it is the same here. It is not very cold, it is 12 degrees but it is cloudy and foggy and damp.  
  
(We are nearly there, another 2 months and it will be warm!)  
Thank you!  
  
(Thank you very much anyway, and have a great day.)  
Thank you, have a nice day. Goodbye.  
(Goodbye.)  
[Audio ends 39:46)