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

Can you briefly introduce yourself, age, situation, and so on, without mentioning your surname of course?  
Yes. My name is [PI 01:21] and I am 28 years old. I live in the South of France, and I currently live my partner. I currently work full time in the civil nuclear sector.  
  
How did you first discover you had Wilson disease?  
I will tell you the story! I found out after my transplant. I have had a liver transplant, so therefore, I found out when my liver was sent to [PI 02:08] for analysis. Until that point, we did not necessarily have all the factors to confirm Wilson disease. So, we found out after my transplant and we found out from a geneticist when we took genetic blood samples from me and my family.  
  
When did you have the transplant?  
In November 2017.  
  
Was the transplant due to the Wilson disease that you weren't yet aware of, or did you have a liver transplant for a different reason?  
No, not at all. We actually realised, when I started to become unwell, and there are not many of us in France to have a transplant due to Wilson disease because usually, Wilson disease is identified very early, during childhood or as a teenager. I did not have any symptoms. I had no symptoms at all. As it is a genetic disease, I was ill from the moment I was conceived, as soon as I was born, and actually, it was realised that I was ill when my liver reached the cirrhosis stage. It was due to Wilson disease that I had accumulated copper in my liver for X number of years, and it had not been realised so I had not been able to take copper chelating agents. Therefore, my liver was too damaged due to the accumulation of copper.  
  
So, the impact of the Wilson disease occurred before you were even aware of it?  
Yes.

**Signs and symptoms**

Which symptoms did you notice, with hindsight?  
Over the long term, during childhood, I had hormonal imbalance, and we did not necessarily know why, so I was given treatments to speed up my hormonal development and to be able to take the pill and so on. That is apparently linked to Wilson disease. I was also under the care of a speech therapist when I was younger because I was... It was thought that I was dyslexic, but it was realised later on when I had a brain MRI that I had copper deposits. It had led to a delay in learning to read and write. Over the short term, shortly before my transplant the first symptoms that I had, my eyes went slightly yellow. Very slightly yellow. I was covered in spots all over my body, mainly on the chest. There were lots of small red spots. So, it was not necessarily acne, we didn't know what it was so I went to see a dermatologist. The treatments did not have any impact, and shortly before my transplant, the trigger factor was a drop in blood pressure. My blood pressure was 8.5 or 9. My blood pressure started to drop, and I had a lot, really a lot of retention in my legs. That's it.

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

Before I received the diagnosis, I was already searching quite a lot, because I stayed at the hospital for 8 months, and as I was young, I was 23 at that time and they were doing their best to keep me alive, to understand the whys and wherefores, I had quite a few complications before, during and after the transplant. When I heard that it was Wilson disease, I did not actually know what it was. I did not know anything about Wilson disease. What I am about to tell you is a bit strange, but for people who are ill, who are waiting for a transplant, and who have a transplant, it is kind of the beginning of a new life, a new start, a cure. As I did not necessarily feel ill before, I felt ill after the transplant, although of course in actual fact I was ill before. I kind of felt the opposite, in the sense that since I have had the transplant, I have felt ill. So, labelling what was wrong with me, it was totally unknown to me. I did not know what Wilson disease was, I did not know where it came from, I did not understand why I had it, so it was kind of the unknown but that said, we were able to diagnose why I had reached that stage.

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

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

Well, as I was saying, at that time I was 23 years old and living my best life, so I did not necessarily pay attention to them. Before we realised all this, I travelled to Spain and my sister had already noticed my eyes, that they were a bit yellow, etc. I remember that at that time, my mother was ill. She had cancer, and when I went to see my GP, my GP simply told me that it was the body reacting, it was the body reacting because my mother was ill. I trusted this doctor because he was my family doctor, and I said to myself that it was certainly... I thought that the body expresses itself a lot, with regard to how we are feeling and thinking and so on. So, I was not really worried at all, until I went to Spain, I felt dizzy several times and then I started to pay attention.  
  
Did that quickly lead to the transplant?  
Yes. Actually, to tell you the whole story, I returned home, and I started to feel dizzy until it reached a stage where I measured my blood pressure. I measured my blood pressure and I had an episode of diarrhoea and vomiting that just wasn't getting better. I thought that it was an episode of diarrhoea and vomiting which wasn't getting better, but it was actually because my liver was releasing so many toxins that I was no longer taking anything in, and everything was very complicated to manage. I could no longer digest anything. I went for tests at A&E, and I had extensive blood tests, and then they realised that my PT levels, that is prothrombin, that is coagulation, which is made by the liver... Well, it is the liver which regulates. Regulates? Well, it actually fluidifies the blood, and we realised that my blood was no longer coagulating at all. So, actually, everything went very quickly from that point. I had a scan, it was realised that my liver had reached the cirrhosis stage.  
  
[10:18]  
  
I am not going to ask you to repeat yourself, but before you had the transplant, you had minor, manageable symptoms. You were thought to have mild dyslexia, you saw a speech therapist, you had yellowed eyes and so on. Did you do anything to treat that?  
Actually, when I was younger, the dyslexia, when you think of dyslexia because you have difficulty learning, you do not link it to a specific disease, or a rare disease. You think that it could happen to anyone. When I had hormonal imbalances, it was not necessarily linked to Wilson disease, because in itself, Wilson disease... Wilson disease is a rare disease, and not everyone is aware of it, so it is not something that immediately springs to mind, when you think of hormonal imbalance. You might think more about the thyroid or something, which is better known, but not necessarily Wilson disease. So, when I was younger, it was absolutely not linked to Wilson disease, and when my eyes became yellow, so a few months before diagnosis, I had not necessarily realised. It was my sister who kind of alerted me to that, but again, as I was saying, I was 23 years old and thought to myself, 'Yes, my eyes are a bit yellow, because I am eating a bit of greasy food at the moment, and drinking a bit of alcohol' so I was not really attentive to it. The whole liver transplant thing was totally unknown to me then. I never really paid any attention to all the liver diseases, that liver disease could cause yellowness in the eyes. I had not made the link at all. As I felt well, despite everything, I was not concerned. At that point, I was not concerned.

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

Did you continue to have symptoms after the transplant?  
It has now been 6 years since my transplant. I no longer have any Wilson disease symptoms at all. None at all, because I am no longer ill with Wilson disease, and that is why I had a transplant. I needed a whole liver in order to really totally get rid of all the Wilson disease, which was mainly located in the liver. So, I do not have any symptoms linked to Wilson disease anymore. I am only genetically ill, because I still have the gene for the illness. However, I no longer have any clinical symptoms at all. I have symptoms which are linked to the anti-rejection agents that I take following my transplant.

**How do you deal with the symptoms now? [skipped]**

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

No.

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

The question to ask me would be more along the lines of the difficulties that I have due to my liver transplant, which was due to Wilson disease, but that is a separate topic on its own.

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

How long was it between the first, quite brutal symptoms, such as loss of consciousness, until the transplant? Was it quite quick? How long was it, 1 month? Between the holiday in Spain and the transplant?  
Yes, it was about… To explain it fully, when it was realised that my liver had reached the cirrhosis stage, due to my age, the doctors tried to restart my liver, because I was young and they wanted to avoid the need for a transplant. So, they tried to restart my liver, except that my liver never did restart. So, July, August, September, October, 5 months. I waited for 5 months before having a transplant.  
  
During these 5 months, what was life like? Was it endless vomiting and diarrhoea, and dizzy spells?  
It was very difficult, because as soon as it was found that I had reached the cirrhosis stage, I was immediately admitted to hospital for a liver transplant. I was immediately admitted to hospital, so I went from a life where I was 23 years old, I was enjoying life, I was carefree and thinking about having fun, to [PI 13:14], you are a... We are waiting for a transplant organ for you and your life is in danger. So, it is true that it was brutal for me, for me and for my family and friends. Because we quickly switched from one world to another, but actually, I think that my brain understood that it was ill. Everything deteriorated very quickly, I started to fill up with ascites in the stomach, I started to gain a huge amount of weight because I was filling up with ascites and water. My clinical results really started to deteriorate. My liver workup too. Everything deteriorated very quickly over the course of a few weeks, as though my brain... It was like a denial of pregnancy before that, that sounds silly but it was as though my brain had understood that I was really ill and from that point, my clinical condition deteriorated very quickly. However, due to my age, they tried to hold off for as long as possible in the hope that my liver would restart with cortisone boluses and so on, but unfortunately it never restarted because it was too damaged.  
  
If I understand correctly, now that you have had the transplant there is nothing to worry about for the rest of your life, with regards to Wilson disease? It has been removed and you are now in a similar situation to your sister currently?  
No. Well, yes. I am ill genetically, but only genetically. I just have to do in-depth tests if I want to have children in the future. I will automatically give a bad allele to my child. My partner will just need to not have the same thing on his side. That's it.

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

Did any of your family members test positive for Wilson disease?  
My mother and father both had the faulty gene. The faulty gene for Wilson disease, but they each only had 1 of the 2 alleles affected, or faulty. You need both alleles in order to be ill. I picked up both alleles, the 2 bad alleles! One each from my mother and father, which meant I had the disease. I have an older sister who only received the bad allele from my mother, but not the one from my father, so she is asymptomatic.  
  
This is not in my questionnaire, I am just trying to learn, so, your sister is asymptomatic, does that mean that she is monitored because she may develop symptoms one day, or will she never have anything? Will she be all right because she only has one allele?  
No, not at all. Yes, that's right, in order to have the disease, in order to trigger the disease you need to have the 2 alleles for the ill gene, and as she only has 1 she will never develop the disease.

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

Actually, I have learned a lot about Wilson disease, and I also learned about everything that the transplant would mean. I found out a lot about Wilson disease in a very short time, because I did not actually know anything at all about this disease before. I also contacted the Wilson centre of reference in [PI 19:47] and I am part of associations where they supported me with regard to Wilson disease, and explained the mechanisms, where it comes from and so on, and I realised... I may be going off topic a bit here, but this just to explain the aspect of Wilson disease which I am most concerned about, it is that Wilson disease can affect the liver, but it can also affect the brain only, part of the brain. I have met people who have Wilson disease, only in the brain, and the symptoms and the lifestyle is really more complicated than what I am going through at the moment. I actually almost think that I am lucky that my disease affected my liver more than my brain. Because when it is the brain, you can't say, 'We'll change it' or 'We'll do a transplant'. That is not possible! They take treatments which are almost more of a burden than a transplant, with side effects which are more or less acceptable depending on the person. However, that's it, when I saw that the disease could also affect the brain and when I saw the damage that it could do to a person, that frightened me, and I thought that my situation was not as bad as it could have been.  
  
[21:02]  
  
Did you do all this research into Wilson disease during the 5 months before the transplant when you felt really ill, or was that after the transplant when you were no longer ill, in a way?  
No, I started my research maybe a year after my transplant. To tell you the whole story, I stayed in the hospital for 8 months, because I had post-transplant complications. They were linked to the transplant itself, not even to the disease. When I was hospitalised, I had a single obsession and that was to recover as quickly as possible in terms of my body. Because I was bedbound a lot of the time, I have a huge scar on my stomach, I lost a lot of weight and so I needed to recover. That was my only obsession. So, anything emotional, psychological, the disease, the whys and wherefores, I did not necessarily think about any of that. I think that my brain protected me, too. It was really when I went back to a so-called normal life, post-transplant, when I started to ask myself questions about the disease and all that. So, it was really several months after my transplant.  
  
You mentioned associations etc. so you kind of got involved in Wilson disease once you no longer had it?  
Yes.  
  
Did you meet patients who were suffering from the consequences of Wilson disease, or even who, to go further, who died?  
I don't know anyone who died due to Wilson disease. I know people who died while they were waiting for a transplant, unfortunately. Because I am a member of associations for liver transplants for all types of diseases. I am also in a Wilson disease association. To tell you the truth, with regard to Wilson disease, via social media, because I try to communicate and share my experiences, so there are people who contacted me. I also met 2 girls who had transplants due to Wilson disease, and we were able to talk. One thing led to another and I reached this centre of reference in [PI 23:08], the Wilson centre of reference, where I was able to talk with people who are part of the association. So, we talk that way, we also communicate daily via social media. That's it.  
  
That's interesting, so you basically no longer have the disease but you still have, you feel that you need to get involved and share your experience with others, that is something that you want to do, if I understood correctly?  
Yes. Yes, because... It took me a long time to take in and accept everything that I had been through, because I was a bit... Why me? Why so young? So, at first I was not in denial but I thought that I would quickly set it aside and get back to my life as it was before. Except that I thought to myself that I must not see it as a failure or think why me, but that thanks to this I learned this, I experienced this, and as I see it, I learned a lot about myself, about life, about the value of life, that health is a luxury whether you are 20 years old, or 30 or 50. I thought to myself that even though I experienced this, Wilson disease remains a rare disease, we hear about it increasingly often because I am interested in it, but if someone is not interested in it they won't necessarily hear about it. I kind of want to be a voice, and if I can share my experience with people who are hospitalised or waiting, explain my journey... Because when I was hospitalised and had questions about Wilson disease, I did not necessarily get many answers. I really had to fight in order to get answers, and I thought that if I can provide answers myself, and reassure or provide answers to certain people, then I will do that willingly. So, that's it I do not consider it to be a duty, I see it as a natural thing to do. To communicate and help if necessary.

**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?**

I had already heard of this. It both surprises and doesn't surprise me. It surprises me in the sense that I think that we are fortunate in that a treatment for this disease exists. A treatment which has varying degrees of efficacy for people, but it does address the consequences of the disease. So, I think that these people are fortunate to have a treatment, which does have varying levels of side effects, so... It does not surprise me because I have met people who took this treatment, and when I see their side effects, I think that people don't want to have these side effects. Well, it is not that I understand it, because if I was in their shoes, I would take my treatment in the same way that I take my treatment every day for my transplant. It does cause side effects, but it also saves my life every day. We each have our own outlook. For me, regardless of the symptoms, the treatment, whether that is a copper chelating agent or anti-rejection agents, they save the person and enable them to have a better life, on a daily basis, or to live... To learn to live with the side effects. So I did know this, but it surprises me because it is something that I would not dream of doing myself, but some people think that they can manage to live without it, or they don't want to live with the side effects so they won't take it. That is each person's decision, unfortunately.

**What could be the reasons for this? [skipped]**

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

You did not receive treatment before the transplant. Are you currently taking treatment? Did that change on diagnosis, has it changed since, or has it always been the same one?  
Yes. Before my transplant, I did not receive any treatment, I had a normal life like everyone else. At the age of 23, you think that you are indestructible. So, I did not take any treatment, because I thought that I was in good health, even though with hindsight we have realised that there were certain symptoms which were linked to that, but we would never have made a diagnosis based on those.  
  
Aside from the liver transplant, have you never had any treatment for Wilson disease?  
Never. Never, so I know that before I had my transplant the doctors were discussing whether they should give me copper chelating agents, will it maybe free up the liver a bit? Except that as it reached the cirrhosis stage, they thought that it would no longer have any effect at all, because the liver was destroying itself. So, they did think about it but I never received a single drug for Wilson disease, no.  
  
Even now, as you said, you are ill but only genetically at the moment, so even after the transplant or just before you never received any copper chelating agents?  
No.  
  
Never received, let me see, I have a list of treatments, Trientine, or Trolovol, nothing at all?  
No, no.

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

I go to hospital approximately every 5 months. Actually, after my transplant it was every 3 days, every week, every month, every 2 months. Now, 6 years post-transplant, I go approximately every 5 months. I have a blood test approximately every 2 or 3 months, and my doctor receives directly. He calls if there is a problem. I have learned to read blood test results now, so I know when it is okay and when it isn't. Let's say, with the disease, hospitalisation and so on, I think that you get to know your own body and I know when it is okay and when it isn't. So I go to the hospital approximately every 5 months, I have a blood test and an ultrasound, I am weighed, my blood pressure is measured, they ask me a few questions. I see my surgeon, he asks me about my daily life, what is going well, what isn't, what needs to be adjusted. Then based on the results, she looks at adjusting my treatment. That's it.  
  
Who is responsible for your treatment? Is it a combination of the GP, surgeon, and possibly...  
No, it is only at the hospital. Only at the hospital, it is at a public hospital in [PI 28:27]. It is the surgeon who did my transplant who has been treating me since. It is either my surgeon, or a hepatologist, that's it, and my GP... I do see my GP when I am ill or something, and my GP sees me for my disease if necessary, but for anything like prescriptions or follow-up after the liver transplant, it is solely at the public hospital.

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

I have seen several different doctors, because the one I had, my surgeon, retired shortly afterwards. However, the one who took over from him, I met them too. It was complicated, because I think that French public hospitals are going through a crisis. I think that we all know that. They lack resources, they lack staff, and there are far too many patients for very few surgeons, very few nurses and very few healthcare assistants and so on. So, I viewed my hospitalisation very negatively due to the doctors, because we were just numbers, in a way. It was very difficult for me to feel like a number and not a person. Time passed and you build relationships which means that it goes better, but as a patient, it is very difficult to accept that you are just a number.  
  
[30:10]  
  
What about after the transplant, how would you describe the relationship with the doctor who you see every 5 months? The surgeon who you see every 5 months?  
The surgeon who I currently see is a woman, so I knew her but she did not do my transplant. She was part of the team at that time, when I had my transplant. Things are going well. I am not saying that things are better because she is a woman, far from it, but she is someone who pays more attention to my daily life, the impact of my transplant on my daily life, on my wish to become a mother, my wish to have a baby, the side effects, and she does not solely focus on 'Is the liver in working order? Yes or no? Goodbye'. It is all the little, routine things that she takes into consideration and it is a great relief to me to know that I am being listened to and heard, aside from my liver.

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

So, again, as I said, I had Wilson disease but without knowing it, so I… What I might change about Wilson disease is that I think that we need to talk about it more. I think that we need to talk about this disease more, because I think that it affects quite a few people, and it is still too poorly known by a lot of people. Few people are familiar with this disease. The impact that it has on daily life, and even the patients. Often, when you have a transplant it is called... Taking my own case, I have had a transplant, or even when you have Wilson disease, the infamous chronic diseases, or invisible diseases, you are not taken as seriously. No, look, you can walk! You are not disabled! You are not in a wheelchair, so you are healthy! I think that it is one of these invisible diseases which are just as much of a burden and just as significant as people who are in a wheelchair or people who have mobility issues. I think that we are not sufficiently taken into consideration in life and in society, in the world of employment, and I think that we should... This is just a wish, I know that there is a long way still to go, but I think that invisible diseases such as Wilson disease and liver transplants, or transplants of any kind... I have had a transplant and I currently have a good life, but I am living with my disease, I am living with my transplant, and I manage my side effects. I do not talk about them because I adapt and I live with what I have, but people think that everything is fine, [PI 33:44] you are in good health, you work and you do sports. If you work and you do sports, you must be in good health. So yes, currently, with everything that I have been through , I am not going to complain because I know where I have come from and I know where I am today, but I fight to get through every day unlike someone else who doesn't have anything wrong with them. I think that these invisible diseases are not sufficiently put forward, and diseases like Wilson disease which are considered rare. I don't think we talk about them enough. I think that these rare diseases need to be better publicised, to talk about them more, to be better diagnosed and treated, too. So, that's it. That would be the feedback that I would give.

**What would be your dream treatment?**

What about your own personal treatment, what would you change about that? You mentioned earlier that you felt as though you were just a number, and so on. Are there any other aspects like that?  
Yes, when I was discharged from the hospital… I stayed in the hospital for 8 months, and I think that I currently have repercussions due to this poor treatment, because I left hospital after 8 months, I was in bed for 8 months, I was totally destroyed on a psychological level. At that time, I was fortunate to be very well supported and assisted, but I had no psychological follow up at all. I had no follow up at all. I had to go and look for...  
  
Refer yourself, look for...  
Sorry?  
  
No, I was just completing your sentence, you were saying that you had to go by yourself to look for, or fight for a psychologist?  
Yes, that's it. I dropped from 58 kilos to 45 kilos, so I had no muscle left, I was no longer able to walk straight and I had to fight my doctor to have physiotherapy sessions, theotherapy sessions, and I went to see some doctors who did not always take me seriously. No, no, you are going to start walking again and it will improve! I currently have pains in my stomach on a daily basis, because maybe my scar was not correctly cared for, and I have pains in my knee because I might not have done the correct exercises. That was because I was kind of... I am not placing the blame on the hospital because they saved my life, there are amazing surgeons and healthcare professionals, and I thank them. When I go there, I thank them. However, the liver is cured, see you next time, goodbye! That was hard for me, because there is the mind, too. There is mind which took indirect hard knocks, and the body needs to recover too. We kind of get left to our own devices with that, and that is the aspect that I really regret, because I currently have repercussions due to this, and that is the other reason why I talk about it, and I want to help and share in order to say listen, I had this, maybe that's not the way it should have been done. Today, with hindsight on my experience, if I can share this with you and help you with your pre or post-transplant period then I want to do that. I don't know if I am clear.

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

No. As I said, I cannot tell you about all the treatments in detail, or everything that people with Wilson disease have, their side effects, because I unfortunately haven't experienced all of that. Well, I have not experienced it so I cannot talk about it.

**Any other comments?**

Did you feel comfortable during the interview? Were the questions clear?  
Yes, no problem. I will admit that I have a lot of work on at the moment so I had not necessarily prepared the interview. It is true that I have never done any interviews before so I was not the most comfortable with giving my answers.   
  
There is no prep needed, it is spontaneous, your experience.  
I think it's a good thing that you are doing this, interviewed people with this disease, who have had a transplant, or no transplant, different people with different experiences. Again, if it can help certain pharmaceutical or other companies, or help certain patients, I think that it is important. I will be here if you have any other questions, don't hesitate to contact me, I will be very happy to help.  
  
I hear you. Finally, and I am not going to ask you to coordinate anything at all, but do you know anyone who has Wilson disease who is not treated or who is not taking their treatment, and who could take part in this study? If you say yes, I won't take the person's contact details, I'll tell the research company that you said yes and they will contact you and look at it with you, if necessary.  
No, I don't know anyone. I know people who have Wilson disease and who are taking copper chelating agents, and who take their treatment. I also know people who have had a liver transplant due to Wilson disease and who also take their treatment. I do not know... I have heard about people, but I do not personally know anyone who does not take their treatment.  
  
Thank you, you were very clear. The company which recruited you will be in touch again with regards to the payment of your compensation. Have a nice evening!  
Thank you very much, have a good day! Goodbye!  
  
Goodbye!  
  
[39:47]