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

I did not find out, but my parents did, because I was diagnosed at 4 years old. My mother is a nurse, and she noticed something was not right. We went to doctors and the diagnosis was determined. We have been knowing it for a while, because this was in 2006.

**Signs and symptoms**

**There was not antecedents, blood diseases in your family?**  
No.

**If you know about details, how did your mother noticed that something was not right? To which doctor did she go to for diagnosis? Were there any specific symptoms?**  
As I was 4 years old, I cannot really tell. From what I remember, I had issues on the level of… Since this is about the gastrointestinal system, I think it had to do with that. I think we went to the GP and we conducted a series of tests and they noticed that my liver markers were not functioning. One thing leading to another, we were referred to gastroenterologists and the diagnosis was given.

**How did you feel when you received the diagnosis? [skipped]**

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

I do not recall at all. From what I remember, I had stomach ache, maybe. I think this was one of the symptoms and the diagnosis was given very rapidly. I was treated very rapidly. With this disease, there is no symptoms or pain, it evolves under the radar. I do not have striking memories or symptoms, pain… I think I had stomach ache, but nothing memorable.

**How did you deal with these symptoms? How have your symptoms changed over time/after treatment initiation?**

They did not evolve, as I did not have any. The only symptoms I had were on the tests’ results. I did not have any clinical signs, any manifestations… On the blood level, we knew that the liver did not work, but I never had any symptoms.

(This is true until now?)  
Yes, I never have any.

**How do you deal with the symptoms now?**

(You are not dealing with any symptoms since you do not have any, but is there anything that you are supposed to do so that no symptom appear, apart from taking your treatment? Did you had to adjust your lifestyle, or are you taking your treatment and living a normal life?)  
I have some until last November, in 2023, because I had a liver transplant. My Wilson disease was circumscribed to the liver only. Since I do not have my original level, I do not have the disease anymore as such. Since I got transplanted, we stopped the treatment for Wilson disease.

(You got transplanted. Before that, did you have symptoms or not at all and everything was well?)  
Not at all.  
This was based on results alone…  
Even at the lowest… The disease degenerated and this is why doctors wanted me to get transplant, but even at my lowest point, I never had any symptoms.  
(This must be strange for you. Everything was well but you go to the doctor, you are given results, the results were not good, and you were fine?)  
[00:10:08.13]  
Yes, exactly. It is typical to this disease, because we have no physical impact, it is only the body not functioning inside.  
(This must be strange. You are told that nothing is going well but you feel…)  
Yes, we are not aware of it.

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

Chronic disease, no, but I have an ovary disease. This is however unrelated. Wilson disease was really the only severe disease I had.  
(This ovary disease is not related?)  
No, this is independent.

**If yes, how do you feel this other disease impacts your attitude to Wilson disease? [skipped]**

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

Yes and no. As I was diagnosed very early on, I do not remember not taking medications or not having this disease. I grew up with it and developed with it. As such, it was not difficult to be sick, but taking medications was limiting, knowing that these medications have to be taken a while before eating, we have to stop eating… They depend on meals. We have to wait for 2 hours after or one hour prior, to take the treatment. This was quite limiting. We also have a diet to stick to, low in copper. This was also limiting, but I got used to eating like that. Most food do not have that much copper, so if you are careful, life can be normal.

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

(It was mainly the precise times you needed to take the treatment in and the low copper diet?)  
Yes.  
(Apart from that, you said no symptoms… No impact on mood?)Personally I did not feel that, but maybe that at the end my mother felt that I was not feeling well. We are not really aware of it, but relatives can notice that. She told the doctor that sometimes she could tell that I was not very… I do not know how to explain that.   
(Not very nice…)Yes, exactly.  
(Did you have any issues of access to treatment? Pharmacy available, and so on?)  
The first medications I used to take were hospital delivery only. We had to go to a hospital centre to get the medications, which was more limited than going to the pharmacy, but if you organise yourself well it is okay. The last one I was taking was pharmacy delivery, but you needed an authorisation that the pharmacy had to ask the company and so on, because it was a rare and complicated medication to get. We always needed to plan ahead, I could not just pop in the pharmacy if I had none left. We always needed to plan ahead. However, if you manage this well, there is no difficulty.

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

What about taking a medication every day without feeling sick, was this difficult? Did you tend to not stick to it during teenage years or…  
Yes, once during my teenage years and once lately, which led to the transplant. This is also the disease’s evolution, but there is a part of that too.  
Did you feel a difference when you stopped the treatment?  
Not at all. We do not feel anything.  
If I may, what made you discontinue the medication during your teenage years and then lead to transplant?  
We do not feel sick, when you have this disease. Our life is normal, and we only take medications because we know why, but not really, since nothing really affects us. This is very complicated. The first time it upset by hepatic markers quite a lot, and the second time, it was unstable. In September we notice nothing was going well anymore and so in November I got a transplant.

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

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

There was nothing that concerned me as I accepted it. I never lived without it, so it might be more difficult for someone whose diagnosis came later than me. At 4 year old, you do not recall your first years. I have never known myself without it, so it was part of my life. It was not a weakness or something that made me panic. This was something here with me.  
Did you see transplant as relief, as stress? Did it frighten you?  
They did not talk about it for the first time. My parents, when I was small, were already told that it was a possibility. Often, if the treatment did not work, then it was a possibility. The treatment worked so… Lately, the doctor did not want to transplant me. I had to take treatment again at correct times, a good diet. It did not work well enough, and so they announced that it was time for transplant. It did not worry me that much. In early September, when they told me things were not going well, it was one of the first things I was told. I had been prepared for one or two months. When they told me it was unavoidable, I had already been told it was a possibility, so it was not a surprise.   
You said you had no symptoms related to the disease, but did you have any symptoms related to the treatment?  
Not at all.  
Did you ever met with other patients with Wilson disease?  
No, I did not. There is an association. My mother was a bit involved in it, but I was never interested. Since I was feeling normal and I did not want to get close to patients… This might be a bit stupid, but I did not feel sick, so why being close to sick persons, since I was not? Although I was. In the beginning, my mother was talking with a lady, quite old, who had this disease, but I did not personally have interactions with patients.

**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?**

Surprised that they did not take their treatment correctly? Since it was my case, I am not surprised.

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

At the beginning, we take it because it is a brand new situation, but I think that after a few years, we do not see… I am talking about my case without symptoms, because I think some patients might have symptoms. Patients who do not have any physical impact or problem in their lives, taking a medication is a bit limiting and so at some point, they might think, “Either I am cured, or my life is not changing”. I think that since you do not feel anything… It is not that it is not useful, it is useful, but we might think that without it, we can also feel better.  
[00:20:20]

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

Did you take the same treatment until the transplant, or did it change?  
No, I took several of them. I tested them all. Do you want me to say…  
I can list them and you tell me yes or no, or you may tell me what you took.  
I can list them to you.  
Please go ahead.  
First I took Trolovol. Do you want me to tell you of the adverse effects I had with the treatments? With this first one, it was quite serious. I do not know whether you want to…  
Let us first list all the treatment. Trolovol. Then the second one, the third one, and then I will ask you what happened with each.  
Okay. So I took Trolovol first, then Wilzin, then Trientine, and then it was Trientine too, but it was Cuprior. In total, I had four treatments.  
Was Cuprior the last one?  
Yes.  
What Trientine was the third one?  
Trientine, the base molecule. Then Cuprior was trientine but not under this…  
A generic or something like that?  
Yes, the name the company gave to trientine.  
Trolovol, Wilzin, Trientine, and Cuprior. No other “experimental” treatment taken within the context of a clinical trial or…?  
Not at all.  
Did you ever combine one of the four treatments together, or always sequentially?  
Yes, one after the other. We never combined molecules. Always one molecule at a time.  
Could you briefly talk about your experience, from what you remember about Trolovol?  
I was taking it, and after a year or a year and half, I started having muscle fatigue. I could not walk or talk or anything… Write. I was diagnosed with myasthenia. I had to be operated in Paris, sternotomy and thymectomy. From one day to the next, it stopped myasthenia. However, we realized it was one of the adverse effects of Trolovol, so…  
Trolovol was discontinued, and you switched to Wilzin?  
Yes, exactly.  
Can you tell me about Wilzin, if you recall it? When did you take it, at what age? 6 to 10? 6 to 15?  
I was in second year… I think I took it for 8 years.  
What was your overall experience and what made you switch to Trientine?  
It was working well. It was not that difficult. There was always a thing about taking it before or after meals, but in itself, no memorable aspect to this treatment.  
What made you switch to Trientine? Was this because it was a new medication? Did something specific happen?   
From one day to the next, we realised it was not working very well anymore. The professor switched my treatment, and they realised it was due to therapeutic failure because I was not taking my treatment very well. During this period, they switch to Trientine because for the first time, it was not working, me taking medications.  
You did not respond to Wilzin as well… In the beginning you did not notice, but on results, and they thought there was a lack of response, but this was when you were a teenager and you did not take your medications…  
Yes, exactly. I did not tell them, so they thought the medication was not working. They realised then, when my mother told them what happened. We did not switched back to Wilzin and we kept Trientine.  
What is your overall experience with Trientine?  
It was okay too. The first Trientine I was taking had to be stored in the fridge. It was a bit more complicated.  
When going on holidays or…  
Yes, or grocery shopping, you needed to take a cooler with ice. It was complicated. I did not have a choice.  
The logistics… You needed to plan ahead. Is this because you switched to Cuprior? Or does it also need to be stored in the fridge?  
No, knowing it was the same molecule, they switched to it because it was new, I do not know. It did not really make any difference. It is true that Cuprior does not have to be stored in the fridge. This is why they gave me this medication, I think. To make it simpler. It was the same molecule, so it did not change anything, except that it could be stored in room temperature.  
If I understand correctly, it was Cuprior until you turned 20 or 21, and then transplant, and now, nothing?  
Yes. They left me take it, because they did not know whether I still had the disease, whether it was genetic. During the first weeks after transplant, I was taking it, and then they analyzed my liver, my own, affect the transplant, and they notice I did not have any copper left in the liver, meaning that the medication had eliminated it well, so it was not necessary for me to take the treatment. I have not been taking anything since two months.

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

After transplant, we are supposed to see the physician every week. Then, every two or three weeks. I am feeling extremely well since November, so now it is once a month.  
Before transplant, how often did you see your physician? Once every 6 months, once a year?  
Once a year, yes.

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

Is it the same physician who have been monitoring you for the last 10 years or more, or did you change? I suppose the transplant made you go to hospital more than to the GP, and so on…  
I was never monitored by my GP because this is a rare disease that not many people know. You need to go to the research hospital centre. This is in the gastroenterology department. In the beginning, I was monitored in paediatrics, by a paediatrician specialised in gastroenterology. When I turned 18, they send me to the adult department and I was monitored by another physician for two years, and then in September, I spent a lot of time in hospital because my health was starting to worsen seriously, and since then, I have the same physician who monitored me before and after transplant. She will monitor me until…  
She is a gastroenterologist at the hospital, is that correct?  
Yes.  
How would you describe your relationship with this physician?  
Extremely good.  
She has been knowing you for a while, she is nice…  
We did not know each other before September, but with everything I went through and the fact that I spent a lot of time in hospital between September, November and after that, I remained for months, so a closer relationship was built. It is my doctor, but we are getting along well.

**Can you discuss your concerns or problems with your doctor? [skipped]**

**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)?**

My mother was managing it with me when I was little, and during the teenage years, she tried leaving me to manage it alone but we have seen that it did not work out very well. She left me to manage it by myself, but in September, with what happened, she was present all the time. Since I feel better… We always talk about it, every day. My prepare my medications… My family is included in that, but I still manage by myself.  
[00:30:22]  
What about psychological support, without necessarily be a psychologist? Do you need some emotional support sometimes or…  
Not at all.

**Which situation in your life led to stopping your treatment for Wilson disease? How did it happen?**

(Transplant). This was when I stopped taking the treatment officially, apart from the several times I stopped taking it on my own…

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

This was not a real discontinuation, but irregular administration, times not really… When I needed to take it at specific times, I was not very precise.

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

It was just me. My mother found out and then everybody knew.

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

About management of the disease, or my own behaviour?  
Management, and what happened around you, treatments, but if your behaviour was dictated by what was bothering you in how it was managed, then it is related… Thinking about your whole journey, what would you have liked to changed, made it easier?  
Honestly, no. I was always very well monitored with very competent professionals. It was not a burden, even going to hospital, and being hospitalised because of it. Going to my consultations. Things were always very good.  
It seems the only thing, if I am not mistaken, and since you do not have any symptoms… What could have been managed better is this first treatment, Trolovol, which let to another operation? Were you resentful afterwards?  
No, this is more like, “It had to happen to me”. Since this is a very rare adverse effect, it happened to me and that is all there is to it. I always accepted situations… Even this was not that problematic. I accept, because we do not have a choice.

**What would be your dream treatment?**

The common point to all treatments is taking it with meals. Outside of meals, either one hour before, or two hours after eating. This is very limiting. If you manage it well, it is feasible, but it is very limiting.  
This is not a temporary treatment either…  
Yes, this is lifelong. You need to set an alarm on the phone not to forget taking the treatment two hours later. For people who take Trientine, although I do not know whether it still exists, since they switched me on Cuprior, but people taking this treatment have the limitation of storing in the fridge. Now, Cuprior being trientine, I do not think they prescribe trientine anymore. They switched me to a room temperature form…

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

We covered everything.