

Huntington's disease

Contents

As my father and my grandmother both died of **Huntington's disease** (hereditary degenerative disease with no treatment available), I had to admit during summer 2007, when I felt light involuntary leg movements at the age of 52, that I had probably inherited the fatal genetic abnormality. In December 2009, the movements intensified and spread to my whole body, leaving no room for doubt and raising fears of a rapid development. My father had died at age 66. I was 54. I had little time left to find a treatment. In fact, I already had found this treatment by chance at the end of 2007. Doctors don't believe at all in my discovery. Me..., not really more than them. But as years go by, I must recognize that the disease doesn't seem to develop in me as it did in my father and grandmother. It is this rather incredible story that I tell you here.

1. My current state of health.....	1
2. My very first symptoms	2
3. My intense symptoms of 2009	4
4. My symptoms today	6
5. Aggravating factors	8
6. I'm looking for a researcher.....	11
7. My genetic test	13

Since 2012, I send letters just about anywhere to try to interest doctors. But these letters are not at all successful and time flies. If you have any suggestion, thanks to contact me.

Best regards
Vincent

My current state of health

Let's begin with the most cheering news: **I'm still fine!** My movements no longer appear, except occasionally, much weaker than in 2009 and almost always related to some particular event (see the following pages). Since 2015, I have pulsatile tinnitus in my left ear, with a hearing loss of 25%. But the different examinations I had didn't show anything abnormal, and I finally got used to this half blocked ear. As for all the rest, the lights are green!

I'm still working and lucky enough to have a large amount of leisure hours that I fully enjoy. This year, I've released a major version of my four card game programs, with a lot of work behind (around 35 hours of programming per week). My capacity for reflecting and memorizing and concentrating is as good as always, and my morale is very high. As for my physical abilities, each week I ride my mountain bike for a total of 4 to 5 hours, I have 2 hours of light weight training and 3 kilometers of slow swimming at the pool (legs, arms, four strokes). What else could I ask for?

So no wonder why I had great summer holidays, as described by the little postcard below:

I had never visited the region of Martigues and the "Blue Coast" (Côte Bleue), to the west of Marseille (South of France). It's a beautiful area where I was surprised to be able to make numerous bike rides without too much effort. The forest paths often follow the small valleys between the hills, and are relatively flat. So, I managed to cover large expanses of forest and scrubland, and contemplate some unforgettable landscapes and viewpoints. Not to mention my daily games of bowls at the campsite, with the players coming from Marseille. So many good times!

And I remember a dream picnic in a little deserted creek on the bank of the Etang de Berre lake. With a swim in clear and cool water, hardly salty, in a nice warm sun, with no sound but the water lapping and the cicadas chirping. See by yourself:

My very first symptoms

The first clicks

The first strange symptom I had dates back to the night of November 23, 1985 (I was 30 years old at that time). It was just a "click" in my head, while I was about to fall asleep, lying motionless on my bed. I thought it odd. It was like a short high-pitched sound in my head, but not coming from the ear itself. Not a snap but instead a very little "electronic" noise, with a loud beginning and a short echo at the end.

I was not overly worried by that. Except that this click came again on the next day, always when I was falling asleep, and that I heard it in the same way on the following nights. Later, I didn't pay much attention to it, but I remember that during summer 1986, it was always a single click that would occur each night. And then soon it was two or three clicks, spaced by a few minutes, that seemed to make sure I would fall into deep sleep. It was not unpleasant after all. If laying on my bed I was still mentally active and not about to fall asleep, no click would occur. Next, if I turned and drifted off into daydreams more conducive to sleep, then, click, click, click, the sandman, by some clever mouse clicks, would seem to come and close my few windows still open.

These clicks never stopped until June 2007. And they have always increased, so far as to become much more numerous and much stronger. Towards the end, I would feel about fifteen clicks, sometimes very close together, and around ten times louder than the very first click of 1985. But I'd got used to them, all the more so since they would occur only when I was falling asleep and that it's always comforting to feel sleep coming on. What was more worrying was the question about the development of these unexplained clicks. I knew how they had developed over my last 20 years. And when extrapolating this development, I was forced to wonder: how was all this going to end?

In June 2007, the clicks stopped all of a sudden.

My first movements

The clicks went away suddenly and definitively. One night, I was lying after a very busy day, and dozing on my back I was waiting for the famous liberating clicks. And surprisingly it was not my usual clicks that appeared, but some little and jerky leg movements, both legs at the same time, very light but clear all the same. There were about ten of them. And then I fell asleep, as though nothing had happened.

But the following night, when the leg movements came back instead of the clicks, I had to admit that I had passed a milestone. At the age of 52, I had my first involuntary movements, and even if they would occur only when falling asleep, it was a bad sign for the future. As for the clicks, I never heard of them again. It was not either clicks or movements, sometimes the former, sometimes the latter, no. When the movements appeared, the clicks vanished definitively.

Passing this milestone was not a problem for me, inasmuch as my movements would not appear during the day but only before falling asleep. And my first click had taken 20 years to become a leg movement. I could hope many more years before it would change into something more bothersome. But in December 2007, while my little leg movements had become my inevitable ritual of the night, I was intrigued by a strange phenomenon. About once a month, my evening movements would not appear during two or three days. I was not very happy about it. As a developer, I didn't like it. It was a kind of "bug in the bug" that I had to explain.

So, I tried to find out. My movements seemed to always stop on the same day of the week, that was odd! I studied whether in my activities of that day there was some particularity that could explain the phenomenon. But no, my weekly activities were so regular that if my movements depended on the activity of this precise day, either they had to stop every week, or they should never stop at all. Then I had the idea of studying my meals. Was there a food that I would eat only on that day, but not every week? I remembered very well what my evening meal was the last time the movements stopped. And indeed, incredible as it may seem, there really was in this meal a food that I would only eat on that day, and around once a month. And this food was...

Canned spinach!

My first six months of treatment

Spinach! Do *you* believe in this sort of treatment? Well, not me. In any case, not at first glance. If anything, I had fun believing in it. All right, I said to myself, I'm going to make some tests and I will see, but I must not delude myself, it cannot work. But, and I agree that I didn't make things in a very scientific way, it so happened that the first day I ate spinach at the evening meal, the night movements didn't appear. Now, after that, I should have carried out many experiments, with spinach, without spinach, etc. Yes..., but no. Me, I want above all to be all right. And I stupidly said to myself: *if it worked, then I eat them two or three times a week, and that's all*. And that's how I started my spinach treatment. Yes, it's true, my reasoning was not very rigorous, I admit.

In fact, the treatment didn't seem to fully work. One week, it was good, but the following week the movements would come back. So, I gradually increased the doses by adding one tablespoon of spinach to my two meals (lunch and diner). So, at noon for example, a fried egg with ham and... a spoon of spinach. And in the evening, a grilled meat with spaghetti and... a spoon of spinach. It was tolerable and seemed effective for I had practically no more movements at night.

Even so, without being really rigorous, I tried twice to stop eating spinach because it seemed to me that believing in such a treatment was not serious. The only thing was, I then couldn't sleep anymore! And it was no longer little movements occurring when I was falling asleep. It was sudden and brusque movements of my legs, arms and shoulders, appearing around every 3 or 4 minutes and literally preventing me from falling asleep. On my two tests (I should have made more, I know), I had no other option but to open a spinach can at about midnight, to warm it up in a saucepan, and eat a few spoonfuls of it. I can tell you that in the middle of the night, with no accompaniment and no seasoning and no real hunger, it was not delicious. But strange as it may seem, each time it proved to be the solution to stop my movements and allow me to go back to sleep.

So, I ate spinach twice a day for six months (in addition to a balanced diet, of course), with almost no more movements in the night. My summer holidays 2008 were the most pleasant ones. And above all, it's there that I had a brilliant idea that had not crossed my mind yet (I'm a bit slow!). When back home, I imperatively had to search on the internet which component was so important in spinach. I thought it was iron, as people were inclined to think in my younger days, at the time of the cartoon series *Popeye the Sailor Man*. Could Huntington's disease have something to do with an iron deficiency? I was longing to find it out.

My intense symptoms of 2009

My discovery of folic acid

When I got back from my holidays, in August 2008, I was a bit tired of eating spinach at each meal, and so I searched on the internet about this type of food and its components. And to my great surprise, it was not iron that spinach contained in large quantities, but **vitamin B9**, also called **folic acid**. Then I made a few searches about folic acid, and I learned that this vitamin had some connection with the good functioning of the brain. Good, that was encouraging. Next, I looked for the keywords "folic acid" and "Huntington", and found a page telling that *folic acid had been proposed as a treatment for Huntington's disease*. Stop! That was more than enough for me. I didn't try to check the reliability of the site in question. I didn't care, I thought I had found what in spinach might well have an effect on me. In any case, I had a track to follow.

So, I tried to buy the precious vitamin B9, and thanks to the advice of a kind chemist of my town, I could get tablets of 0.4 mg of folic acid, those prescribed to pregnant women to help the fetus' brain to develop properly. My first tests were conclusive. By taking a medium dose of this vitamin, I could get the same effect as with spinach, namely that my evening movements no longer appeared.

But it remained fragile. I don't know whether my "problem" was rapidly getting worse or if there were regular ups and downs, but I had the impression that the dosage I was taking was becoming less effective after one week or so. The movements were coming back little by little, which means that I was led to gradually increase the dosage, up to the maximum recommended dose. It was at that time that I found the tablets of 5 mg which seemed to be much more effective. I rapidly went up to the maximum dose (3 tablets of 5 mg per day), and then I went through a long period of stability of more than a year, with no more movements at bedtime, apart from one or two very weak ones occasionally. But I had the feeling of having found a soft and cheap treatment that maybe was going to allow me to live like that for several years. And yet, I was not totally sure of the real effectiveness of folic acid. I was waiting to see the development of my symptoms six months by six months.

The strong movements of December 2009

I hoped to reach the end of 2009 without mishap, but unfortunately at the beginning of December of the same year, some very strong movements appeared. I was quietly sitting in front of my computer, improving my programs as usual. When suddenly my left leg moved to the side by about twenty centimeters, without warning and with a brusque movement, a bit like the one produced by the doctor who tests the knee reflexes with his hammer. I had already had such movements during the night, that I considered as normal sleep movements. But now, it was in the daytime, and my left leg had just contracted violently, the movement apparently coming from the hip. It was not a trivial little movement, an insignificant little "tic". Almost my whole body had been shaken by the strength of the contraction. Fortunately, the movement didn't seem to be very frequent. It came back only two or three times in the afternoon, not more.

But the following days, again in the afternoon, at a time when facing the computer I'm the most physically inactive, the strong movement came back, and this time equally on one leg or the other. There were also variations. Sometimes, the movement would come from an area below the knee, then generating a foot contraction. And next, my shoulders started to move too. This time, it was rather spectacular. Either one shoulder or the other, or even both at the same time, with a very rough movement that seemed to come from the middle of the spine. I would have been eating peas, I would have sent them rolling all over the place!

But I was not too much worried yet. If my involuntary movements were violent and unexpected, they were none the less rare (one or two per hour, not more), and they didn't seem to be really disabling. However, the day my two forefingers began to move by themselves during nearly one minute, with a wide and regular movement quite impossible to stop, I started at this moment to realize that working on computer with keyboard and mouse was going to become difficult. And when, a few days later in the evening, arriving at work (where I am in contact with the public), my lower jaw began to move to the side, several times in a row with a horizontal movement so strong that I could no longer speak, I understood that I had to rapidly take a decision.

Folic acid in high doses

I had already thought of what I should do if one day my symptoms were getting worse. But I had to make things quite clear in my head. What seemed to me most likely, and that I had constantly kept in mind in the past two years, was that spinach and folic acid had never had any effect. The disease had seemed to recede, but it might have only been a normal feature of its development. So, I had to get ready to forget all about this empirical treatment which had been nothing but a sweet illusion.

However, there was still something that I could prove to myself by experimentation: in fact, it was just that folic acid had no effect on me. If by taking high doses of this vitamin for one or two weeks, my symptoms kept going on without any change or improvement, then it would be a clear and conclusive sign that I could free my mind from this unfounded belief. I had read somewhere that a tablet of 5 mg had an effect for about 3 hours. There again without checking this assertion, I decided to take one tablet of folic acid every 3 hours for a fortnight. And that, night and day, because it seemed obvious to me that the chemistry of my brain did not stop during the night.

From the day after the beginning of my high-dose treatment, my symptoms changed. But they got worse! During the afternoon, my leg still moved to the side violently, but now, just before the movement, I could feel an intense sting on the hip or thigh area. For the foot movements, it was a sting on the middle of the calf, causing a sharp pain comparable to the one felt when freezing a verruca off with liquid nitrogen. A great pain then, very unpleasant, but nevertheless bearable. I was not really delighted with the new thing that was suddenly imposed on me, but at the same time, I found interesting that something had changed. So, I continued my treatment, without any modification.

The next day was just as surprising. My leg still moved, I still felt the very painful sting just before, but now in between there was a little delay of about half a second. First I had the sting, then the delay, and after that the movement. It was funny. All right, the delay before the movement was very short, but it was clear that the two effects were now separated, whereas the day before they were totally simultaneous. So, there was again a change in my symptoms, which was rather encouraging.

It was my third day of treatment that was the most unexpected. I was again in the same situation, working quietly on my computer. And suddenly I had the sting which was sharp and well localized to the hip or the calf. I waited, I waited... And then nothing... The movement didn't come. The movement didn't come any more! In the following days, I had stings everywhere, on the legs, the three big toes of one foot or the other, the sole of the feet, the shoulders and even the fingertips. But no more movements! In the following week, the stings became weaker and less frequent. I just had to move a little the stung area to remove the pain. And about ten days later, I had no more symptoms, no more stings, no more movements, no more clicks.

What would you have done in my place? Me, I didn't try to understand, I continued my high-dose treatment...

My symptoms today

My symptoms when I forget to take my treatment

So, at the end of 2009, I started to take folic acid in high doses, and I've not stopped since. I should have carried out experiments consisting in stopping my treatment to see if my involuntary movements would then come back. But on the one hand, I didn't really want to deliberately let my brain degenerate, and on the other hand, these experiments, as carried out without medical control, would have been worthless. And besides, I wanted first to check that the treatment was effective in the long term, and that I could bear high doses of folic acid. According to my reading, overdosing this vitamin was not dangerous. But I was not certain of it.

Two years later, in 2011, I was still fine. I gradually increased the dosage to one tablet every two hours, because I thought it prudent in case the effect of a 5 mg tablet didn't last as long as expected. And for the night, it was more convenient for me as I can easily put up with waking up every two hours. I even find it very pleasant. It allows me to go to the toilet, to check the temperature of my bedroom, to drink a little and often wake up from a dream with an endless loop from which I couldn't manage to free myself. I also enjoy my dreams much more, as when a dream is interrupted, it sometimes leaves some comical memories and funny impressions. And then I go back to sleep very easily, which is indisputably a big advantage.

If I didn't try to voluntarily stop my treatment, it just naturally happened when I forgot to take my tablets. Even though I organized myself with an alarm clock that rang or vibrated at the required time, inevitably there were some hiccups, some times when I forgot to set the alarm, or even during the night, I wake up at the right time, I drink a little glass of water with a drop of grenadine, and oops!... I forget the tablet! After all the trouble to wake up in the middle of the night... When I was late for my treatment (generally by around 2 hours), it almost always increased the little movements for the following two weeks. These were very slight foot movements, a hand contracting nearly imperceptibly, or a forefinger that goes up on its own. When falling asleep, there were also clearly more arm or leg movements, and sometimes a sting without movement.

Filming my symptoms

At the beginning of 2013, the sign of being late was strangely given to me by a twitch in my eyelid. I told to myself: *Ah, what's happening, did I take all my tablets?* When counting all my daily tablets and seeing that my alarm was set to early afternoon, I realized that I was 4 hours late. Nothing happened on the same day (apart from my eyelid keeping on twitching), but on the following week, my two forefingers started to move both at the same time, less strongly than in 2009 but still quite sharply, and several times in a row, with phases of about thirty seconds each. It's only three weeks later, when my movements had nearly vanished, that I exclaimed: *But what a stupid idiot, I should have filmed them!*

My eyelid twitched during 2 months, about twenty times a day. I could even make it twitch voluntarily, by taking a deep breath or yawning. But this may have had nothing to do with the other movements.

From then on, the idea of filming my symptoms began to form in my mind, and during the following months I prepared myself to make my first medical report. I fumbled around to learn how to film with my camera, to find the right framing and light (at least something acceptable), and I trained myself to film my motionless hand. In April, I was ready. But as for animal documentaries, you have to be patient. For it was not until September, after again having been late with my treatment, that my right hand agreed to come out of the wood. Oh, it was not easy! For the movements were very short. By the time I saw them arrive, I checked they were strong enough to be filmed, I ran to the required place, I switched on my camera and started recording, bam, it was over. Or just finishing. At last, after several failed attempts, I still managed to record the short video below, where you can see rather well the stimulated area that eventually pulls the forefinger. It's much weaker than what I experienced in 2009, but even so it's visible and above all difficult to feign. Try yourself. Me, I can't do the same thing voluntarily.

How my brain seems to recover

When, after being late with my treatment, I experience a phase of hand movements like the one of the above video, it lasts about four weeks. One hand or the other may be affected (rarely both at the same time), and while the movement concerns the forefinger more frequently, it sometimes concerns the middle finger or the thumb too. Never several fingers at the same time, and when the middle finger is affected, the activated area is clearly the top of the hand, and no longer the interval between thumb and forefinger. The tremor appears several times in a week, in the morning or the afternoon (sometimes during the night, but rarely). I may be doing the washing up and suddenly feel my finger moving for a few seconds. It comes back several times during around half an hour, and then it disappears.

As time goes by, the movement weakens. It's always the same finger of the same hand that is concerned, but the finger moves less and less. The last week, I can just feel the area in question trembling for a few seconds, but the finger itself does no longer move. And what is encouraging, it's that once the phase is complete, I may stay several months with no problem of that kind. It's like if my brain had managed to remove the damage causing my symptoms. From what I feel, folic acid doesn't act directly on my symptoms. It prevents damage from happening, damage that later, sometimes after several days, would cause the different symptoms felt.

So, one can understand my reluctance to the idea of suddenly stopping my treatment, if only for 48 hours. Because if being 4 hours late produces in me unpleasant effects for about a month, one can easily imagine that being two days late would cause problems that may last twelve times longer, that is a whole year! One year of after-effects, some of which may not be reversible. I don't know. There may be thresholds beyond which some neurological damage will be irreparable. There may also be cascading dysfunctions, some of which might be irremediable.

Now, let's make an assumption. Let's assume that folic acid really stops the disease from developing, that I never took any of it in tablet form, and that my brain is today deeply damaged. Then, what effects could taking high doses of folic acid have on my health? Logically, none! No visible effect, in any case. The damage will still be there, then the symptoms too, and the possibility of stopping the very slow development of this disease will not change anything in my current state.

This is why I support researchers in their search of a treatment to reduce the symptoms of seriously ill patients. They must be able to help these last ones to recover at least some of their cognitive abilities, and allow them to have a dignified end of life. But I can't help encouraging physicians to look for a treatment which attacks the causes of the damage done by the disease, and not only the symptoms that follow. According to my observations, it must be possible to act before the damage is done. Of course, I'm not sure of my analysis, and all my claims need to be verified. But I am convinced that researchers should not ignore the possibility of a preventive treatment that my personal case seems to reveal.

It won't be easy. All the more so since there might be aggravating factors.

Aggravating factors

Strenuous physical activity

I am fortunate to be in a privileged position to watch my symptoms and see the way they develop over time. I have a very quiet and regular life. I sit in front of my computer for long periods (sometimes more than 8 hours a day), writing my programs peacefully. And even if I am very focused on what I'm doing, I easily spot the slightest involuntary movement of my body. It's not the same when I train at the swimming pool or ride my bike. During these more active periods, there must be some symptoms that escape my notice. But overall, I am most probably in the best position to describe the slow development of the disease, or its non-development. Besides, I keep a *Symptoms diary* where I carefully record the dates of occurrence of the movements, their intensity and precise description, as well as all the events that may have had an effect on them, like when I'm late taking my tablets, for example.

To be sure to get my beauty sleep, almost every day I have a little nap of one hour around midday. I then don't sleep deeply, but even so there is a short phase of relaxation that has certain similarities to falling asleep. As soon as 2010, it became obvious to me that each time I had a strenuous physical activity in the morning (bike ride or weight training), my nap was more agitated. I had at least a brusque leg or arm movement, rather powerful. I often had a contraction of the lips and around the mouth, and sometimes of the tongue. I could also have my both eyes blinking strongly at the same time. Or the forefinger (sometimes even the little finger) of one hand that would go up on its own, with strength. My movements may considerably vary in their suddenness and amplitude. Those of a nap after a sports morning were clearly sharper than those of a normal nap.

So, I made the experiment of taking double doses of folic acid during my sports activities, and rather than opting for two tablets of 5 mg every two hours, I tried the rate of 1 tablet every hour. The result was significant. It could probably be verified by a doctor. When I increase the doses of vitamin during my physical activity, I still have a few movements during the following nap, but they are much weaker. The leg hardly moves, the finger may go up but very slowly, smoothly. Some naps may be done without any movement.

During my 2011 holidays, when as usual I move around a lot with 3 to 6 hours of bike ride every day, I noticed that my symptoms were increasing in the evening, even before going to bed. I was quietly sitting in an armchair, reading a book or consulting my region maps to prepare the ride of the following day. And then, I could have several little foot or hand movements. The foot slightly moved at the ankle, and the hand seemed to retract, all fingers at the same time. The phenomenon, absolutely not annoying, could still occur about ten times in the evening. So, I decided to take 1 tablet every hour in the daytime, and a few days later, the evening movements had disappeared.

The two following years, I increased in this way the dose of folic acid during my holidays and my days of physical activities. And as during the rest of year it seemed to me that the little movements were coming back in the end from time to time, I thought in 2013 that it was wiser to change to the following treatment:

- **During the day, 1 tablet of 5 mg every hour**
- **During the night, 2 tablets of 5 mg every two hours**

It may seem to be a heavy and dangerous treatment, but when you know how serious is Huntington's disease, which ends with the total inability to do anything, the person being stuck in a wheelchair, eyes haggard and empty, you don't hesitate to take risks.

The adrenaline rushes

It also very quickly became apparent to me that if I had a strong emotion (watching a soccer match on TV for instance), my movements were more numerous on the following days. Same thing when trying to ride up to the top of a big hill. Or when doing a timed sprint at the swimming pool, or when letting myself race with another swimmer. The nights and the naps of the following days always seemed to be disturbed by sharper and more numerous movements. So, I've gradually eliminated the situations with a very high increase of heart rate, by recording matches and watching them later more quietly, or by avoiding excessive physical effort. I also avoid running after a bus, or getting upset after a message from a dissatisfied user who, having lost a few belote games in a row, sends me insults and accuses my program to cheat against him.

I installed on my computer a little program that spots the discourteous messages and automatically responds to them in a very polite way. This saves me from entering into endless arguments. Moreover, my belote program now includes the best answer: you can give an advantage to North-South! Which goes to show that you can always find a solution to a given problem. All you have to do is search.

At the end of the 2011 season, I also decided to stop playing in bridge tournaments. I used to play only one afternoon per week, but except on very rare cases (nothing but bad hands, nearly no hand played as declarer or only too easy contracts), I always felt a big surge of adrenaline. I just had to bid a more or less difficult slam, or to double a contract bid by players of a higher level than me, and my heart started to beat fast under the excitement and the desire to win. It was not unpleasant, but I had the impression that starting from the next day, I was more restless. And as it happened every week, it became difficult for me to observe anything about my treatment. This is why in the summer of 2012, I replaced my weekly bridge tournament with a short bike ride, with no stress or great effort. And then I felt much better for it.

During my 2014 holidays, I signed up for a bowls tournament organized in the holiday village where I was staying. I was going to play with a very kind pensioner who could only roll the bowls. So, I was going to have to throw them, and as I try my hand at throwing but I'm not a "thrower" at all, I thought we were going to be eliminated in the first round. But we went to the final! Of course, my heart was beating hard during this last game that we unfortunately lost. We had a wonderful evening, but I was a bit worried about the consequences of these long hours of play with a lot of adrenaline. And I wasn't wrong. Two or three days later, during the night, I was waken up by a new symptom that I called *electrocution* because it really gave me that impression. I was peacefully sleeping on my front, as I usually do. When suddenly I pushed up with my hands, my chest raising off the bed, the back of the neck extended upward. My entire body was constantly agitated by a continuous but non painful spasm that lasted nearly 2 seconds. I was awake and conscious since I was afraid of falling out of the bed, but I could no longer do anything. It was a sensation quite close to the strong leg or arm movements, but over a much longer time and concerning all limbs at the same time. At last, the convulsion stopped. I got up, took a double dose of folic acid and went back to bed. The rest of the night was quiet.

The electrocution phase, as I call it, came back on the following days, during two other nights, but always diminishing. The last spasm lasted only about a second, with much lower agitation.

The experiment of 2015

In September 2015, I tried an experiment. As my electrocution symptom had only come back once in June, but very slightly, I had the idea of playing again in bridge tournaments, to see if I could prove the link between adrenaline and my night spasm. I also had the secret hope of managing to keep calm and cool when playing a contract, by the sole power of my mind. Alas, this was not at all the case! In the first tournament, yes, I did well, but it was mainly because my partner had done most of the job. But at the second and third sessions, the power of my mind didn't resist more than five minutes! And I felt adrenaline rush and quicken my pulse during the three hours of the tournament. Strangely, I didn't get the electrocution symptom I was expecting. However, I had so many leg and arm and shoulder movements during the following nights, I've been waken up by such painful stings in the toes, with instant contraction of the lower leg, that I ended up canceling all the tournaments I intended to play with my different partners. Quite obviously, for me, it was not a good idea to get back into regular high-stress and high-adrenaline situations.

But one of my partners was still on vacation, and I couldn't cancel in time a tournament I was to play with her one month later. So, I took part in this last tournament rather casually, and regretfully of course, as I was sad to have to deprive myself of this exciting activity in such a warm club. Once again, I realized how incapable I am of staying totally relaxed during a tournament, maybe because we could have finished first, and as usual I felt the effects of adrenaline throughout the whole afternoon. Well, during the night of the following day, at 5 o'clock in the morning, I again experienced an electrocution of the same type as that of my bowls tournament of 2014. According to what I wrote in my diary, I had the impression that the spasm lasted 4 seconds, always with the fear of falling out of the bed, the convulsion being so violent. The rest of the night was calm, and the phenomenon has not occurred since that famous day of November 2015.

Analysis errors

Of course, I know that one can be mistaken when analyzing events and linking some of them while they have no relation of cause and effect. I know it all the more so since I receive numerous messages from Belote players in which they maintain that they are 100% sure to be disadvantaged during the card dealing. And they tell me that they have made many experiments on my program, they have carefully counted all the points scored, they have played a large number of games. They are certain of what they say, they have a blind belief in it: their opponents get all the aces and all the melds, they are quite convinced of it. And that is untrue. But they initially have a preconceived idea which will then influence all their analysis. They will not count properly, not memorize correctly, or just be mistaken about the number of occurrences necessary for a test to have any reliability.

*We are not always aware of the rigor required to demonstrate a piece of knowledge. One can easily understand this by running an automatic test on my Belote or Bridge programs. After around fifty deals, the East-West team may be 15 deals ahead of North-South. **What was I saying?!**, some will say. All right, they make another test. And then again, East-West is 10 deals ahead. **Is that not evidence enough?!**, they will continue. Well, no, not at all. It doesn't mean anything. The number of tests is far too small and their duration far too short. Much longer tests are needed to analyze a random phenomenon like dealing cards. Only a test of more than 10,000 deals can be used to check that the win rate of each team always nears 50%.*

So, I know very well that I can make a mistake analyzing that an event A (an adrenaline rush) is connected to an event B (an electrocution symptom). Yes, we can accept that the connection between these two events seems to be confirmed by the fact that when eliminating A (adrenaline), event B (electrocution) no longer appears. But it's not a real scientific proof. It may be only an analysis error.

To correct my errors or confirm my observations, it is therefore imperative that a researcher establishes strict and serious experimental protocols, and that he (or she) carefully works on my case. Hence the title of my next page:

I'm looking for a researcher

I'm looking for a researcher

Research on caffeine

By an article in the Figaro of September 9, 2014, I heard of French researchers who studied the possible role of caffeine in the earliness of onset of Huntington's disease. I found their work interesting even though I have not drunk coffee since 1984. I used to drink coffee and tea when I was young, but never in the afternoon for my sleep was highly sensitive to caffeine. In 1984, I started computer programming and my pace of life slowed down considerably. It's at that time that I felt "big thumps" in my chest, as my heart seemed to stop for a second and give a big thump on the next beat. I could feel it mostly when sitting and working, or even during the nap, with then a feeling of moving horizontally on the bed at the time of the stronger beat. It was rather unpleasant. After consulting a cardiologist, I was told that it was caused by an occasional extra heartbeat (called *extrasystole*), and that it was probably due to caffeine. Even then I opted for the simplest and most radical solution to the problem: stop drinking coffee and tea.

But since the years 2000, I had started drinking decaffeinated coffee again, with a little milk. I used to drink one or two cups in the morning, and one in the early afternoon. In May and June 2015, I had small unusual movements of the right hand, almost every evening around 8 pm, when I stopped working to watch a little television. It was jerky movements affecting the top of the hand, near the thumb and the forefinger but different from those shown on my video. The fingers were not concerned, there was just a repetitive and marked tremor of the top of the hand. I wasn't too worried about it. I was just waiting to see how it would evolve.

During my two weeks vacation in July 2015, these movements disappeared completely. I attributed that to my holiday activities. But when I got back, after only two days spent at home, the hand movements reappeared as if by magic. I was still on vacation, I had not really changed my habits. The only thing that had changed was that I could drink decaf again. Because during these holidays, as I was renting, I had bought a small pot of decaf, but I had forgotten the sugar! As a result, I said to myself: *too bad, I'll do without decaf*. I come back home, I drink decaf again and then, the movements are back. I found that very odd. So, I visited websites about decaffeinated coffee, and much to my surprise, I learned that there was caffeine in some of these products. In smaller amounts, but there was some all the same. Immediately, true to my motto *Take heart! Run away!*, I stopped drinking decaf. And my hand movements of the evening, so characteristic, have not reappeared (not once) since August 2015.

A similar phenomenon occurred in December 2016. I had bought dark chocolate truffles for Christmas, and I could eat 2 or 3 of them every day, with delight. But after about two weeks, the little finger of my left hand started to regularly move. On the last days, it moved a few times every fifteen minutes or so, which was rather worrying. I said to myself: *It cannot possibly be the chocolates. There is no caffeine in chocolate!* Well, in fact, there is quite a lot, and particularly in dark chocolate. Like a shot, I threw all my truffles in the bin, and two days later, my little finger stopped moving. The more it goes, the more I think caffeine is a real poison to me.

There again, when eliminating event A, event B does no longer occur. It seems to show that A causes B, but it's not irrefutable proof. I leave the information here, for researchers working on caffeine and Huntington. I have contacted them several times, but how could they respond to me?

About the difficulty of studying my case

Because I am not an easy case: I'm all right! When I tell my neurologist, that I consult once a year, that everything is fine, he answers me with a smile: *People don't often come to tell me that, but I'm glad you do every year*. Then he tests my reflexes, makes me walk a lot. Maybe he sees that the disease is developing? But he doesn't want to tell me. As I'm still fit and my symptoms have not reached a severe stage, the disease has not yet started and the doctor is not allowed to make a diagnosis. It's then difficult to talk of a possible treatment since there is not even a real disease!

Besides, I'm not a subject easy to compare with others. I don't smoke, I don't drink alcohol and I never take medicines (apart from vitamin B9). Moreover, I have a stress-free way of life, with good control of my diet and physical activities. It's not very common, and it will most likely be difficult to analyze whether these behavioral factors play an important role in the development of the disease or in the effectiveness of any treatment.

Anyway, studying my case would be very complicated. How to observe my symptoms? They are too infrequent, I can't make them happen on the same day and time of a doctor's appointment. And I refuse to let them get worse because I'm afraid of no longer being able to get rid of them later. I strive to find ways to prevent symptoms from appearing, but if I manage it, how can I prove it? I had the idea of a short experimental protocol, which could perhaps show a result. But I'm probably wrong about the conclusions that could be drawn from it. Here is the idea:

Test idea

*The researcher prepares **groups of 4 tablets**, each group being numbered. These test groups contain either folic acid (active group) or an inactive product (placebo group). The researcher sends me the tablets, he is the only one to know which group is active or placebo.*

As soon as I get the test groups, at a time of my choosing, I use a test group (taking 1 tablet every hour) instead of my normal tablets of folic acid. In the following 2 or 3 weeks:

1. If I notice **no change** in my health, I label the group number as **active**. And I can continue the test without further delay.
2. If I notice much stronger **movements**, I label the group number as **placebo**. And, before continuing the test, I wait until I get back to a stable state with no movements.

That way, I never set myself a delay of more than 4 hours when taking my vitamin. I make sure I'm always able to get back to my initial state before the test. To prevent any aggravating factor from distorting the test, I take a test group only if I am in a stable state with no movements. If just after taking a test group an aggravating event occurs (like being late with my vitamin or an adrenaline rush), I label the group as unknown without result.

In the end, I must be able, after a test that may last one or two years, to give with no error the numbers of the placebo groups and show the active principle of folic acid on my movements.

A funding problem?

But who will fund such a test, or a more serious study of my case? Researchers are already all busy working on drugs that could soon improve the condition of severely affected people. And anyway, for any research, you need funding. Who will fund a research on a product like folic acid that has already fallen into the public domain?

Or is it a problem of medical education, with doctors much more oriented towards the cure of people in bad health? In an article in the International New York Times dated December 29, 2014, several American researchers report the case of a man whose family is affected by an early onset of Alzheimer's disease. As this gentleman was still fine at the age of 62, he thought he had escaped the genetic mutation of the disease. Yet, he decides to have the test and finds that he has the same mutation as the other members of his family, who were all ill very early. The researchers are interested in his case for they want to understand why he didn't develop the disease. But they recognize that their research is not simple because it goes against their medical education. The range of possibilities is endless, they must study the genetic and environmental factors, and in the end they admit that it is not really easier to understand why people are in good health than to understand why they are not. But they have confidence in their research and the gentlemen in question says he wants to help them.

I am in about the same situation. I wonder why I am still in good health. And I'm looking for a doctor or a researcher who would ask the same question. Maybe in a year I'll be ill and I will start degenerating like the other patients affected by this disease. But even if that were the case, considering that I am probably going to live longer than my father who died at 66, wouldn't it be wise to look for the possible reasons for my longer survival? A few more years to live pleasantly, even under treatment, isn't it worth it? And are there not, behind my observations, some of which are very likely erroneous, one or two truths that could lead to a better understanding of the disease?

So, who? Who will study this avenue of treatment that my story seems to bring to light? And who, if his (or her) research is successful, will get the Nobel Prize in Medicine for his (or her) discovery of a preventive treatment for Huntington's disease?

Who?

My genetic test

How my genetic test was sabotaged

Since the 90's, there is a genetic test to predict Huntington's disease. At the time, I had a blood test at the hospital where my father was looked after, in order to help research. I was not supposed to get any result, but I was insistent and the doctor who took the test accepted to tell me orally that I was positive. He even suggested that I should be put on medication, but as I was still very well and there was no real treatment, I declined.

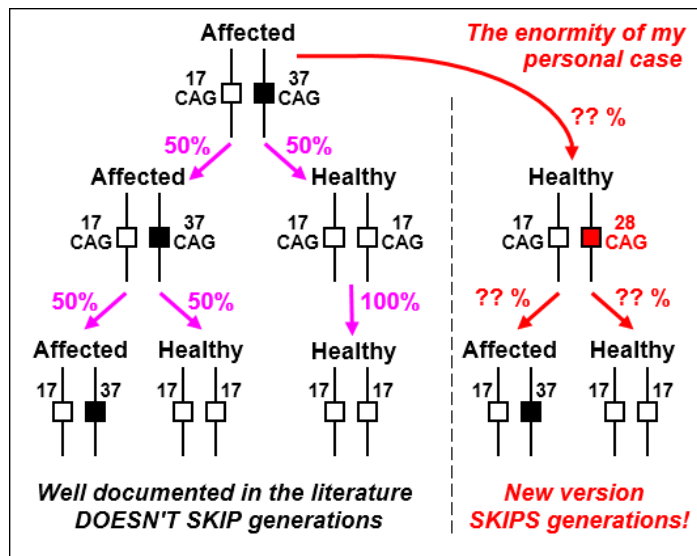
In 2011, while I had been taking high doses of folic acid for two years and all my involuntary movements had vanished, I wished to take the famous test in a more official way. Unfortunately, I quickly understood that considering I was advanced in years, I would never get the right result. To avoid alarming the patient who has his first symptoms and whose health is going to deteriorate soon, the doctor comes to an agreement with the laboratory to give an "intermediate" result. He tells the patient that he cannot himself develop the disease, but that there is a slight risk that his intermediate abnormality will worsen when transmitted to his offspring. Thus, the patient is reassured and can live his last years without worries. While his relatives are still aware that the disease may strangely resurface on the next generation. I say *strangely*, because Huntington's disease is "autosomal dominant" and doesn't skip generations. But the patient is rarely shocked by this blatant incoherence. And if he is and contests the negative result given orally, the doctor sends him a written copy of the laboratory analysis, showing clearly the intermediate result. The laboratory is beyond reproach since it made the test and its interpretation according to specific guidelines (for example, those of the *European Journal of Human Genetics (2013) 21, 480-486*). It can also claim that every year it successfully joins the quality control schemes for Huntington disease made by the *European Molecular Genetics Quality Network (EMQN)*. As for the doctor, he takes great care to never comment the result in writing. He just writes that he sends the laboratory documents, but never writes himself whether the result is positive or negative, so as to be beyond reproach him as well. It's the standard technique used when one doesn't want to give the right result to a genetic test.

A medical error

I think I've been the victim of a medical error. I took my test in the same hospital where I gave my blood sample twenty years ago. So, the geneticist who received me probably knew the value of my genetic abnormality. He made his first consultation with the preconceived idea that I was already very ill. He spoke to me as if I was a mentally handicapped person, asserting that I had nothing, and that if I had involuntary movements, it was precisely a sign that I didn't have the disease. At a certain point, he even turned towards his intern to tell him "*He still smiles*", showing him that I was not yet in the phase where the patient looks dazed and doesn't smile anymore. The doctor was speaking in front of me as if I wasn't there, most likely thinking that I had already lost my cognitive abilities. It was with this misconception about me that he took the decision from the outset to give me a negative result. He even told me that, with what he defined as *mild tics*, I could live without problems until my retirement. At that time I was 56 years old, I was sporty, non-smoker, physically and mentally on top form. This geneticist seemed still convinced that I didn't have many years to live in good health. He didn't carry out the basic checks on my mental state. It was a real medical error.

A year later, I sent to this doctor a letter by certified mail, to inform him that for three years my movements had disappeared thanks to high doses of folic acid, and that it was for that reason that I wanted to get in writing the real positive result of my test. The doctor could then have admitted having given me a softened oral result, for the sole purpose of reassuring me. And he could have agreed to give me the true value of my genetic abnormality, as my case might seem interesting to study. But he did nothing of that kind. He sent me the copy of the laboratory analysis, showing the intermediate result of 28 CAG repeats, which would have us believe that I cannot myself develop the disease, while being able to transmit it to my descendants. This result absolutely incoherent and unacceptable, the doctor himself has of course never confirmed it in writing, thus applying his professional guidelines without further consideration.

How my result ridicules the laws of genetics!



*The principle of an intermediate abnormality is likely to be applied also for other hereditary genetic diseases, as Huntington's disease is cited as an example for the quality of its communication protocols. But for the average patient, it is inconceivable that a deliberately biased result can be given to such a crucial genetic test, and what's more with written evidence from a laboratory. It's a profound attack on people's freedom. Because what doctor can be sure at 100% that there are no possibilities of preventing the fatal outcome? Is it up to the doctor to decide what the patient's life will be, hiding the true value of his genetic abnormality? Does not lying to the patient mean condemning him to death? Because it's leading him to live his life without worrying about possible aggravating factors, without paying attention to his first symptoms, without perhaps being able to discover by chance ways to slow down the progression of his illness. It's not up to doctors to decide that there is no treatment! They should just say that there is no **known** treatment. And if the patient makes the express request, reveal the truth about his genes, so that in the end he remains master of his destiny.*

The different steps I took

I have since tried to assert my right to obtain the true result of my genetic test, so that researchers can take an interest in my case. But the situation seems at a standstill. All the laboratories, whether in France or in the world, have undoubtedly authorizations to ask for this test, and they are probably made aware as soon as a person has already been tested, in order to always give the same result. I tried to complain to the officials of a medical defense organization. But they told me that the intermediate result was clear, that they didn't have to intervene, and that in any case zero risk did not exist. Even lawyers seem completely powerless in the face of my request. And for a very good reason, it's the law itself which authorizes the principle of false results for genetic tests. Here is an extract of an old article in the French Public Health Code, which deals with the topic:

Extract from Article R145-15-14

....

Exceptionally, for legitimate reasons and in the interest of the patient, when the latter has symptoms, the prescribing physician assesses the appropriateness of not communicating the results of the examination of genetic characteristics to the person concerned, or to the person with parental authority if the patient is a minor, or to his legal representative if the patient is an adult under guardianship.

This old article made it clear that the prescribing physician was allowed to lie about the test result. Because if the text said "*do not communicate the result of the examination*", it was hard to imagine how the doctor could say "*No, finally, I don't give you the result*" without alarming the patient more than is reasonable. It seemed logical then to think that he preferred to give a false or softened result. This surprising practice has become explicitly contrary to the right to transparency introduced by the Law of 4 March 2002. The article was therefore repealed in 2003 and replaced by Article R1131-19 of the Public Health Code, written in a much more vague form, so as not to disclose to the citizen that the doctor was allowed to lie about the examination of his genetic characteristics:

....

The prescribing physician communicates the results of the examination of genetic characteristics to the person concerned, or (if applicable) to the persons mentioned in the second paragraph of Article L 1131-1, during an individual medical consultation.

It's the law that allows genetic tests. And when it comes to predicting for sure a serious illness, with all the ethical issues it raises, it is normal to place severe restrictions on it. The law apparently gives the doctor full powers to decide whether or not to give the true result to the person concerned. It is certainly a wise precaution. But no one seems to have foreseen the case of a doomed patient who has been reassured by a negative result, and who ends up showing unexpected resistance to the disease. How can medicine then be interested in his case, since it categorically denied that the person could one day become ill? And isn't this giving the doctor too heavy a responsibility? If he is wrong in his decision not to give the right result. If he is subject to conflicts of interest that drive him to hide from the rest of the medical community this case of unexpected resistance. Has the patient any recourse against this decision? He cannot even turn to another specialist, since they all are obliged to respect the diagnosis of the first doctor, for fear of disclosing the deception of the false result. In what other field of medicine do we have no right to an independent second opinion?

Non-commercial research

I think, but this is only my opinion, that we see here one of the failings of a medical research organized like a commercial firm, with millions invested and profitability objectives comparable to those of industry and commerce. In this context, researchers do their best to find treatments for various diseases. But it would be wrong to say that they are doing all what is humanly possible, and that they are really exploring every avenue of treatment. They have neither the time nor the means for that. It is necessary, in addition, that other researchers work in certainly less profitable and less rewarding directions, more focused on the prevention and the observation of the various forms of evolution of the different diseases. Besides, these researchers exist. We sometimes read their articles in the medical pages of major newspapers. These researchers are not paid by the pharmaceutical industry and have no hope of finding the miracle product that will make their fortune. No, they try to group data, to understand what genetic, environmental or behavioral factors may affect the onset or development of a particular disease.

This non-commercial research is important too. Look at my personal case. Even if one can question my word, there are at least 3 indubitable points: (1) my genetic abnormality; (2) the fact that I'm still fine, which can be proved by a simple medical examination; and (3) the illness of my father, who died at age 66, and that of my grandmother who died at age 65 and a half. The latter spent the last years of her life confined to a wheelchair, unable to move and do anything. As far as I can remember, I've never understood a single word of what she was saying, so much her mouth, lips and tongue were shaken by uncontrolled movements. Doctors know that. If we put these three elements together, it is obvious that at 62, I show a much greater resistance to the disease than my two relatives. It is imperative to understand why. It is essential for the other patients, who could perhaps, by a small change in their habits of life, succeed in maintaining their good state of health for a few more years. Who said it was insignificant, that it didn't deserve any attention? I personally don't agree with that!

Well now, I don't really know how the law can ever solve the problem. Doctors will probably let me die of old age, this disease or any other cause without ever, never record my case in medical annals, never ask questions, never carry out experiments, never try to check my various observations. All that because a geneticist was wrong and decided by mistake to give a false result to my genetic test.

And yet, if we put the interest of the other patients above all, there must be a solution. Ladies and gentlemen lawyers, it's up to you to be imaginative!