Homozygous familial hypercholesterolaemia, Andrei's invisible enemy

The young Romanian's story: "For seven years I was left without any therapy. Then, in Italy, I started doing plasmapheresis, and thanks to lomitapide I was able to halve its frequency"

Rome – **Andrei is 7 years old** when, after a routine check, **he discovers he has total cholesterol at frighteningly high levels**, 1,100 mg/dL. **It is the effect of a silent genetic disease**, which acts very quickly and, if not treated promptly, it can cause serious and irreversible damage: **homozygous familial hypercholesterolaemia**. The child has no symptoms that could suspect the disease: the only visible sign is the xanthomas, similar to yellow/orange gods on the elbows, knees and Achilles tendons, but they appear only after 8 years, when the pathology has already been randomly identified.

We are in Romania, at the end of the 90s: from the moment of diagnosis, **Andrei has lived for seven years without following appropriate therapy**, and with his cholesterol levels (LDL around 1,000 units and HDL always very low, usually less than 30) this means being constantly at high risk for cardiovascular events such as heart attack or stroke. "My parents took me to those who were theoretically the **top experts in metabolic diseases**, who **told us that there was no cure, even abroad**", recalls Andrei.

"I went monthly to take blood samples, and each time my mother prayed the head physician to inquire and ask for an opinion from the colleagues he had met at the international congresses in which he attended. When I was 12, a doctor told us about a treatment used abroad, with which the blood is filtered from fats, but the primary doctor told us that this treatment – **plasmapheresis** – would not solve the problem because "it would have been like pouring clean water into a dirty bath tub". **Only later did we understand that it was in the interest of the state to hide the existence of these treatments because they are very expensive**, therefore unsustainable by our health system", continues the young man.

"At the time the doctors told me I was too young for the drugs: statins were available but the side effects on the children were not known and therefore they were not prescribed to me until the age of 12. At least, this is the version they told us. In the absence of specific treatments, from 7 to 14 years **my parents made me try any type of alternative cure**, naturist and homeopathic, but since it was a genetic disease **nothing worked: at the age of 14 I already had coronary arteries occluded at the 80%**", explains Andrei.

The first turning point was in 2004, when the boy arrived in Italy, where at the Policlinico Umberto I in Rome he underwent a complicated triple coronary bypass surgery and left carotid thrombendarteriectomy, a procedure for the surgical reopening of blocked blood vessels and the removal of arteriosclerotic plaques. From that moment on, **for 10 years, he starts doing plasmapheresis weekly**, with total cholesterol values that are around 250-300 mg/dL, and HDL not higher than 30.

"I arrived in Italy thanks to the kindness of Mr. **Pierangelo Casa**, an entrepreneur from Pavia who contacted **Prof. Claudia Stefanutti** and asked her to welcome me to her center at Umberto I in Rome, to start the treatment I needed so much", continues Andrei. "He was my angel, and now that he's gone he has become a true angel in heaven. Remembering him is a way to express my gratitude, because I feel that I will never be able to repay the people through whom God saved my

life".

The second turning point occurs in 2015, when the treatment with the **lomitapide drug** begins, a small molecule that is administered orally once a day. **Thanks to this treatment, after one year, Andrei was able to halve the frequency of plasmapheresis**, from once a week to once every 15 days.

Today Andrei is 29 years old, takes 20 mg of lomitapide per day and his total cholesterol values are around 120-180 mg/dL, with the HDL having increased to 50. "I started to notice the difference already when I started taking 15 mg. **Some side effects such as tiredness, nausea and diarrhea also occurred, but over time they have diminished in intensity**: I still experience them but in a milder way, and they also depend on the foods I eat", he underlines.

"It was a great physical but above all psychological improvement, because moving after ten years from the weekly apheretic treatment to the fortnightly one represented **a revolutionary change**. Not only do my veins have more time to heal, but I have more time to devote to work and my passions, without having to think about the fact that one day a week I have a fixed appointment with the hospital. For me, finding a cure that saved my life is a miracle for which I thank God and the doctors who welcomed me here in Italy", concludes Andrei. "Having to do plasmapheresis with less frequency may seem like only a bonus, however it is **a bonus that has improved my quality of life from all points of view**".

But in recent months, especially in the acute phase of **COVID-19** infection, did hospitals continue to ensure treatment for patients with this condition? We asked **Domenico Della Gatta, president of the Italian National Association for Familial Hypercholesterolaemia (ANIF)**. "The first thought goes to those who in these years have devoted their lives to research, in the absolute certainty that the more research is done in this country then the sooner important goals will be achieved, unthinkable by the medicine of the past", commented Della Gatta. "In hospitals where traditionally each of us turned to for their own treatment, **the required and necessary treatments and therapies continued**. In particular, I focus on the Policlinico Umberto I in Rome, where everything took place (and takes place) in the best of the structure's clinical traditions. No downtime, no shortage of plasmapheretic treatment (at the Department directed by Prof. Claudia Stefanutti): **the only novelty reflected by the COVID-19 crisis is a significant drop in our periodic patients** (weekly or fortnightly)", continues Della Gatta.

"It should be stressed that most of our patients are currently using public transport and the limitations imposed by the government have weighed negatively on their daily lives. Many others, however, are unaware victims of the pandemic psychosis, so faced with a real risk of getting an infection in the hospital or subway, in the end they prefer to give up the scheduled therapy session. And all this despite being perfectly aware that **the lack of adequate therapeutic treatment can entail devastating and irreversible contraindications for each of them**", concludes the president of ANIF. "The final hope, therefore, would be to be able to imagine – from here in the future – the structural and organic involvement of the Units that traditionally take care of psychological assistance to patients in the hospital, so that they give them the necessary support also in order that they can find the path of trust and can rediscover a different hope for their future".

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