# Jeopardy in Tunisia

*A rare disease patient shares her fight to find the solution she desperately needed.*

My name is Hiba Tohmé, a 45-year-old young woman, mother of a 19-year-old boy. I led a quiet and serene, dynamic and hard-working life until that famous March 2020; when the world capsized following the pandemic, when a citizen curfew, travel bans, and the wearing of masks became the daily lot of billions of people around the world.

Lebanese by origin, Tunisia by residence, I did not experience the arrival of the coronavirus like other Tunisians, nor the excessive use of sanitizer gel, nor the purchase of masks... I struggled during March and April against a strange illness that overwhelmed me, without being able to name it, despite visits to a large number of doctors of various specialties. I vaguely remember deserted streets, hospitals that smelled of death; but what I recall most is being eaten away by pain, fever, cold sweats, and above all a feeling of pressing and sometimes latent morbidity that put my life in jeopardy.

No one could answer my insistent questions: ***when will this ordeal end?*** *What name should be given to this scourge? What will happen to my only son who passed the baccalaureate, during this year engraved forever in my memory?*

**The pain gnawed at me inside and out,** my abdomen kept swelling and my health deteriorated day by day with an impressive weight loss: I lost 27 pounds in a single month. Doctors kept ordering me the same tests that suspected tuberculosis. I went in vain from one laboratory to another in the hope of putting a word to this evil that was invading my whole body.

It was towards the end of May 2020, following the recommendation of two internists, Dr. Aissaoui and another renowned Professor Habib Houman, that an exploratory laparoscopy was prescribed on me to take tissue samples (liver and peritoneum) for analysis. This is how we were able to permanently remove the diagnosis of tuberculosis and I benefited from a protocol directed by Professor Houman who guided the analysis laboratories, asking them to dig the trail of histiocytes. Then begins a mad race between laboratories, institutes, hematologists, leading finally to the diagnosis of ECD with a *BRAF* V600E mutation. An announcement that provokes in me a succession of emotional states, going from worry and stupor to sadness, and finally l the acceptance of the diagnosis. Acceptance was mixed with an enormous doubt given the atypical profile of my ECD: systemic histiocytosis with peritoneal location accompanied by refractory ascites. **It was then that my life changed and took on a new meaning.**

It was while browsing the net that I came across Professor Julien Haroche, a specialist in internal medicine at the Pitié-Salpêtrière Hospital in Paris, and that I understood this disease and its effects on my body. This is the disease that had, I think, attacked the serosa (the outer lining of organs and body cavities of the abdomen and chest) since 2016 when I underwent a pericardiectomy with the removal of the pericardium. Unfortunately, the diagnosis had not been made and the origin of my chronic constrictive pericarditis was bequeathed to non-specific causes.

A visit to Professor Haroche confirmed the diagnosis and the prescription of vemurafenib (brand name Zelboraf) were made following the search for the positive *BRAF* mutation confirmed in Tunisia. This drug doubled my pain, caused my hair to fall out, and without eliminating the ascites, this liquid which swelled my peritoneum and which cost me repeated punctures (also known as a paracentesis, or an abdominal tap, a procedure that removes ascites (build-up of fluid) from your abdomen).

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My state of health not having improved, a second laparoscopy was prescribed by Pr. Haroche, revealing the absence of the *BRAF* V600E mutation with a strong expression of Phospho-Erk. [The vemurafenib previously prescribed is not effective for patients without the *BRAF* V600E mutation.]

In the meantime, I had the chance to know the ECD Global Alliance through which I was able to correspond at length with their Executive Director Jessica Corkran and Dr. Mohamed G. Atta [ECD Care Center Lead at John's Hopkins Hospital in Baltimore] who gave me great moral support. Moreover, following all our correspondence, Dr. Atta had me send my biopsies to the United States free of charge, which confirmed the diagnosis.

Cotellic was prescribed to me, a single sample box having been given to me free of charge, and it worked miracles and had completely dried up the ascites. **Unfortunately, the excessive cost of this medicine completely paralyzed me and I went for many months without medicine.**

Now, I am back on the Cotellic thanks to the support and help of the ECD Global Alliance. I am full of hope and I will never thank Jessica enough for her kindness and her relentlessness. She was able to surround me, understand me, and share with me my fears and my distress.

Despite everything, I always try to see life on the bright side to better get through this tough ordeal, something that was possible for me thanks to the unwavering support of my circle of family and friends. I would also like to particularly thank Pr. Habib Houman for his continuous presence and availability as well as Pr. Julien Haroche for his precious patience and unfailing professionalism.

**Thanks to the support of doctors from all over the world, my doubts about this disease have disappeared.** Erdheim and Chester are to become my companions and friends and I have to tame them. This is how my power of resilience was heightened and my faith in God was further accentuated. Knowing and understanding everything about what is happening to us allows us to manage our disease as well as possible, especially when it turns out to be extremely rare. Living with a rare disease has impacts on all levels: physical, moral, social, professional... Managing to make any sense of one's illness enables us to bring back some kind of energy and turn it from an enemy to an ally, by giving meaning to one's life.

It is vital to accept your illness and never give up by shouting loud and clear that you can cure it. I have become a specialist in this rare disease, unknown to everyone, including the most illustrious doctors.

Erdheim and Chester became two shadows that followed me everywhere and on which I wanted to shed light. I hope this testimony can help advance science and better care for people with Erdheim-Chester disease.