Topic D313Y and other "controversial" variants from the patient's point of view.

Hello everybody,

my name is Christian Lotze, I am the husband of a Fabry patient: Manuela Lotze. I stand here for a group of patients and relatives who are fighting for the so-called controversial mutations (like D313Y)

First of all I would like to explicitly thank Natascha and Berthold. The two of them support our group very strongly and help us with a lot of commitment for the affected patients.

We have heard several times that this topic is too much in the focus from the point of view of other patients. On the one hand I can understand that, on the other hand I ask for understanding! On the one hand, we are dealing with patients who have been arbitrarily deprived of therapy and thus of their livelihood. On the other hand, this problem has existed for a long time. It is also about a very high number of undiagnosed patients who could be helped, who currently lead a very limited and painful life.

If you think back to your journey to diagnosis, most of you have had a long road as well. For our fellow sufferers, the path is now completely blocked and they are sent back into the vicious circle of the doctors' odyssey without ever being able to be treated causally!

Therefore it is important for us to inform. Not only on the subject of controversial mutations, but also on Fabry in general. We have seen too often that even doctors do not know everything. That is why everyone should be as well informed as possible about the disease and not believe everything that is said. Especially with the controversial mutations!

Unfortunately, patients also occasionally spread false information, here we rather suspect ignorance.

What also disturbs us very much is that treatment is often started only after irreversible organ damage has occurred. This is not comprehensible at all.

Therefore, we have compiled information about Fabry disease in general and the controversial variants in particular. You can find the information at a143t.org and at www.fabrienne.online. (You are also welcome to contact us personally!).

Due to health reasons, we are looking for further comrades-in-arms or successors for the future, who are committed to the diagnosis and treatment of patients with a GLA gene defect of any kind. It costs us a lot of strength to learn of so much suffering and to look into human abysses, of doctors who have obviously sold their souls.

Thank you!