

CDG One-to-One Interviews, a series of interviews with some of the brightest minds, who make a difference in the lives of CDG Children and Adults. This is an initiative led by the **Portuguese Association for CDG (APCDG, [www.apcdg.com](http://www.apcdg.com))**, coordinated by Dr Vanessa Ferreira, under the scope of the CDG & Allies PPAIN (more information [HERE](#))



## **Pf Dr David Cassiman: A Physician and Researcher dedicated to improving the lives of patients affected by metabolic disorders**

27<sup>th</sup> November 2016, By Rita Francisco (CDG community social manager. Email: [sindromecdg@gmail.com](mailto:sindromecdg@gmail.com)).

### **Introduction**

My name is Rita Francisco, CDG Patient Community Liaison and researcher at the Portuguese Association for CDG ([APCDG](http://www.apcdg.com)). Today, I have the pleasure to talk with Pf David Cassiman, a respected and admired professional among the CDG community and related rare diseases. His remarkable and relentless work in the CDG field is impressive. Welcome Professor—it is such a rewarding experience for us to you at CDG One-to-One!

### **David Cassiman: a diversified career path combined with close collaborations with CDG patients and patient groups**

**Q1: Rita Francisco:** As a clinician, one of your main focus are the liver and metabolic diseases. What drew you to this field?

**Pf Cassiman:** Honestly, that was mainly chance. I always found the liver and liver diseases most interesting, and was stimulated to explore that further by the head of Hepatology at that time. The gradual switch from liver diseases to metabolic diseases was induced by a growing necessity – the group of metabolic patients being referred for liver transplantation was increasing and no one in the department of Hepatology was focusing on that particular segment. I guess I jumped to both occasions, as I was naturally drawn to the medical, social and scientific aspects involved.

**Q2: Rita Francisco:** You are the working group leader for CDG and liver under the scope of the unique international patient-led CDG network named CDG & Allies PPAIN (more information [HERE](#)). Could you tell us why?

**Pf Cassiman:** The extent of liver involvement in CDG seems to be studied only superficially so far, while there are very good reasons to explore this further. All plasma proteins are glycosylated in the liver. A better view on liver function and dysfunction in CDG opens possibilities for available treatments and is a necessity for treatments that are being developed.

**Q3: Rita Francisco:** What are the 3 major lessons you have learnt from working in tight collaboration with CDG patient families and groups?

**Pf David Cassiman:** I am impressed with the drive and almost globalized cohesion of CDG patients and families and their good spirits. They are empowered, well informed and they have taken charge of the science and care themselves. The CDG community is a heart-warming example of how, with joint effort and some guidance, patients with rare diseases can impact on their treatment, care and visibility.

**Q4: Rita Francisco:** Some CDG types, may present liver involvement. How do you think hepatologists can help CDG patients? What are your short and long term plans for this project?

**Pf David Cassiman:** When the underlying cause of liver disease (in this case: CDG) can't be treated (yet), the diseased liver still can be treated or at least major complications of liver disease can be treated or prevented. Just like the neurological disease or brain development in CDG can't be treated (yet), but e.g. epilepsy can be. That needs to be clear to physicians taking care of CDG patients.

The goals of the liver-CDG project are to raise awareness for liver involvement in CDG, to define liver-related end-points for clinical trials in CDG, to get a more robust view on the natural history of liver involvement in CDG and to gain insight into the impact of liver disease in CDG patients' lives.

### Becoming a FOSTER expert on orphan drugs

**Q5: Rita Francisco:** You have led a project named **FOSTER (Fund for Orphan drugS Towards Ethical Reimbursement)**. What are the main challenges to get a sustainable situation concerning the price and reimbursement of orphan drugs?

**Pf David Cassiman:**

1. Rare examples of abuse in the parallel world of orphan drugs must be dealt with urgently, as these deplorable cases are poisoning society's view of the whole field.
2. The national budgets for orphan drugs should be separated from the general national drug budgets, as the financial comparison between drugs for orphan diseases and drugs for more common diseases will always be in favor of the latter.
3. The quality of clinical trials in orphan diseases is a cause for concern. That should be addressed.
4. The idea supporting special status and financial incentives for orphan drug development, is based on the fundamental idea that patients with rare diseases are entitled to equal access to adequate treatment, as patients with more common diseases. Let's not forget about that. One in 20 people on this planet will have a rare disease at some point in their life. You could be next.

**Q6 Rita Francisco:** Can you share 3 innovative strategies to incorporate patient perspective and accelerate orphan drug development for CDG?

**Pf David Cassiman:**

1. Design and validate patient-reported outcomes for CDG, so they are ready to be used in clinical trials.
2. Start an international patient registry, to get an overview of the disease spectrum and number of patients for each CDG subtype.
3. Collect detailed long-term follow-up data for as many CDG subtypes and patients as possible, to get a grip on disease evolution. This will allow comparison of 'natural history' with the evolution of CDG patients on experimental treatments.

## Personal life and hobbies

**Q7 Rita Francisco:** How did your father, Pf Dr Jean-Jacques Cassiman, a renowned professor in human genetics, influence your professional career?

**Pf David Cassiman:** Frankly, he was wise enough not to. Of course we are all defined by the example given by our parents but he's my father, not my career advisor. I was genuinely shocked to find out at some point 10 years ago that I ended up in a very similar field as the one he used to be in. And I hadn't seen it coming. It was then I fully realised that genetics is not just a science or a profession, it's a hard reality. On the other hand, I like to remind people that half of my genetic make-up is my mother's, not to mention my daily formation and training...

**Q8 Rita Francisco:** What's your secret to find a good balance between your busy work and family life?

**Pf Cassiman:** There's no secret, to my knowledge. Finding a decent work/life balance is simply a daily struggle that we need, to avoid looking young forever.

**Q9 Rita Francisco:** What are your main hobbies? And how do they help you in your challenging career?

**Pf David Cassiman:** I like to fool around with flowers, plants and trees in the garden. I like to read (only fiction) and I like to pretend I can play piano. Please don't ask my kids about the latter. In any case, all three 'hobbies' are essentially ways to void my mind, which is crucial in between work and the many work-related concerns.

**Rita Francisco:** Pf Cassiman, you are a very talented and inspiring professional. It is a privilege to have access to your insights as well as to have you as a member of the CDG Community. Thank you so much for accepting our challenge and joining us at **CDG One-to-One**. Last but not least, a warm thank you to all our readers. Keep following our interviews!

## Who is David Cassiman?



Dr Cassiman is a Medical Doctor at UZ Leuven, with a focus on liver and metabolic diseases. Moreover, Dr Cassiman completed his PhD in 2001 and has been a half-time fundamental clinical researcher at Research Foundation Flanders (FWO) Vlaanderen for ten years. Additionally, David Cassiman has a teaching position. The variety of his career posts can only be matched by the range of his research interests and projects, which translates into an extensive and remarkable list of publications in several relevant scientific magazines. He hopes his knowledge and work will ultimately make a difference in the lives of CDG patients.