



## **GENETICS**

### **The French CF Registry: A Very Valuable Resource!**

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Every person living with CF is unique! The mutations causing CF may be different, and other genes (such as those that predispose someone to develop asthma) that influence whether CF is severe or mild differ from one person to another.

It is very important to study every person living with CF and compare results. Patient registers give us information on what works and what doesn't, and why. If one clinic gets particularly good (or bad) outcomes, is it because they are doing something that is different, or perhaps because the mutations in their patients are different from the rest of their country.

France has a truly excellent CF patient register that is facilitated by Dr. Ingrid Dugueperoux, who works in Brest in the Normandy region, but covers the whole country. Ingrid is the recipient of a post-doctoral fellowship from the French CF Association, *Vaincre la Mucoviscidose*.

This register has details of almost every patient in France, with a full record not only of clinical data, but also age, employment and other points that allow the analysis of success in social and community terms, as well as medical outcomes. There are almost 4000 persons living with CF who are listed in the registry.

Ingrid has confirmed, using data from all of France, that the mutation causing CF plays a major part in determining the severity of each symptom, and the basis on which the disease was diagnosed. It has always been known that persons who have the common mutation DeltaF508 often have problems with pancreatic function and require careful diet management and enzyme supplements. If a child is diagnosed with CF because of diarrhoea, they are more likely to carry this mutation, but if the diagnosis results from lung disease, it is less common. In France, mild mutations often lead to late diagnosis, particularly if the diagnosis is based on sterility or nasal polyps.

Well done, Ingrid Dugueperoux and colleagues, for providing such a detailed and interesting catalogue of the outcomes of the many mutations in the CFTR gene that cause CF.

**Editor's Note:** For a copy of "Clinical manifestations at diagnosis: a study of CFTR mutations based on the French Cystic Fibrosis Registry" report (I. Dugu  peroux, M. De Braekeleer), email [editor@cfww.org](mailto:editor@cfww.org)

