UNDERSTANDING CS, TTD, AND A-T

Dear parent or guardian of a person with CS, TTD, or A-T:

I am a researcher based in California. I've been trying to understand how the symptoms of CS, TTD, and A-T develop. A new part of my research involves looking for problems that are shared by rare and common diseases.

We're hoping to find biological processes that are shared between rare and common disorders. If we succeed, we'll increase the chances that a treatment for a common disorder will work for a rare one. We're starting with CS, TTD, and A-T, and are comparing with problems caused by malnutrition. Malnutrition causes many problems, and many people with CS, TTD, and A-T are malnourished.

In addition to poor growth, malnourished children have problems that also happen in CS, TTD, and A-T. They include kidney disease, problems with brain white matter, and problems with the immune system. We want to know if there are patterns in medical records that give hints about later development of myelin abnormalities, kidney disease, tremors, cancer, etc. We can also compare individual patients before and after the start of tube feeding, and compare tube-fed and non-tube-fed patients.

We realize that gene mutations are the ultimate causes of these problems in syndrome patients. However, a mutation is just the beginning of the disease process. We think that there are a finite number of ways that something like myelin problems can develop, and we want to know if they develop the same way in malnutrition and in CS or TTD. The same is true for immune system problems. If some biological processes are the same, regardless being caused by a gene mutation or malnutrition, it's possible that treatments created for the common disorder will also help someone with one of the rare ones.

Purpose of the study: how you can help

We're asking families for complete medical records from their children (or as complete as you can get). Researchers at the Forgotten Diseases Research Foundation will enter information from the records into a database. We'll use the information in different ways:

- We'll see how each syndrome develops over time
- We'll look for common changes over time shared by the syndromes
- We'll look for changes shared by malnourished children.

We're also looking for small blood samples for running basic laboratory tests. These samples will be especially important from the youngest patients, though we're grateful for any volunteers.

Our goals for this study are to:

- Look for patterns specific to each disorder (e.g. does high IgM increase risk of cancer in A-T?)
- Look for patterns shared by two or more rare disorders (e.g. is there a pattern in blood tests prior to development of kidney disease?)
- Look for patterns shared by at least one of the rare diseases and malnutrition due to poverty (e.g. is there a pattern in any test prior to development of myelin abnormalities?)

Participation is voluntary

You aren't under any obligation to take part in this study, and you will be welcome at any session discussing it, whether or not you participate.

If you have any questions, you can contact me at vnatale@forgottendiseases.org or by phone at 408-529-5755.