



UNDERSTANDING CS, TTD, AND A-T: GOALS

Background and goals

In this study, we're going to analyze data in medical records. The idea is to see if we can find patterns that make predictions. For example, does better nutrition affect life expectancy or growth? What about health problems later in life? Do young children whose weight is good have fewer problems or milder problems when they're older? To answer these questions, we'll compare information from CS/TTD/A-T kids who have been tube-fed with those who haven't.

Also, we'll look for health problems, test results, and gene expression pathways shared by two or more syndromes. Do problems shared by diseases predict something or help us understand similarities between multiple syndromes? The goal is to find common ground between different diseases that can be used to help patients. We don't know if we'll find anything (or what we'll find) in this regard, but more data will help.

Overall, we hope to generate a lot of information from this project. For example, with luck (and enough data), we might be able to make some basic growth charts for the CS severity groups (mild, moderate, severe; some people call these groups type 1/classic, mild, and type II). Making charts depends on getting enough growth data — in fact, for every aspect of the project, the more data we get, the stronger the results will be.

We're also building a software tool that will help us analyze the data, but first we have to get it and enter it into spreadsheets. We've already started, and hope to have enough preliminary information to make a presentation at the family meeting in July.

What we need

We need medical records that are as complete as possible so that we can track what happens in patients over time. Medical centers usually send records free of charge if they're being used for a study. If not, we'll reimburse for costs. If you have any questions, call Dr. Valerie Natale at 408-529-5755 or email her at vnatale@forgottendiseases.org.

Thank you very much.