Sequencing the Genome For a Very Good Cause

An interview with **Stephen Scherer**, Geneticist
University of Toronto

My job is to direct the Human Genome Centre here at SickKids Hospital, and what we do is study the genetic blueprint. We sequence DNA and try to understand what it means in disease and in health.

Very early in my career, I was involved in the Human Genome Project, and we were mapping and sequencing human chromosome 7s. So that's about five percent of the human genome.

Probably our biggest discovery came in 2004 when we found a new type of genetic variation in human DNA, and we called it copy number variation. What that means is, we found that some genes are present in only one copy, instead of the typical two copies, or three copies, or in some cases no copies in an individual's DNA. These copy number variations can lead to things like autism and schizophrenia. It opened up a whole new window, and we've now found these CNVs, or copy number variations, are found in every different type of human genetic disease that has been documented so far.

What does your work mean to your personally?

I walk through the Hospital for Sick Children every morning before I come to the laboratory, and I do that to remind myself why I'm doing the research. And I see the families, I see the kids. And so the questions that we're addressing now are applied in a way. We're trying to answer questions for the doctors and for the families that come to this hospital. And what we're trying

to do now is sequence the DNA of tens of thousands of Canadians, both healthy individuals and people who are sick. I spend a lot of my day thinking about where science should go in genetics; where it shouldn't go, which is just as important; and then how to raise the funds and the teams to make it happen.