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## Canadian team maps chromosome

Unique achievement, 15 years in making, may hold key to diseases such as autism

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MEDICAL REPORTER

Canadian researchers have completed a detailed map of Chromosome 7 — the country's main contribution to the Human Genome Project — discovering dozens of disease genes along the way, including those linked to autism.

The map, published today in the journal *Science*, marks a 15-year effort by Canadian scientists who collaborated with 90 researchers in 10 countries.

The project grew out of Toronto's Hospital for Sick Children back in the 1980s as famed geneticist Lap-Chee Tsui started hunting the cystic-fibrosis gene.

Dr. Tsui helped discover the CF gene in 1989 on Chromosome 7, which has since been nicknamed "Canada's chromosome."

"It's absolutely exciting to have a whole picture in front of us now," said lead author Stephen Scherer, a senior scientist at the sick children's hospital who was a graduate student in Dr. Tsui's lab when the work began.

"We didn't have databases then, we kept the information in our

notebooks."

People inherit 23 pairs of chromosomes from their parents and several have been mapped. But the Canadian effort is considered unique because the scientists have not only sequenced the 158 million nucleotides — chemicals — that make up the chromosome; they have explained many of their functions.

Drawing on their own analyses and all of what is medically known about the genetic material contained in Chromosome 7, the scientists have made the map "medically legible" to doctors, not only biology decoders.

If doctors discover a mutation in a region of Chromosome 7, for example, this map offers them some sense of what behaviour or trait that region might control.

"It is like a road map," Dr. Scherer said, "but it not only gets you to every town and city, it tells you where all the good restaurants are."

Chromosome 7 might be considered among the most human of chromosomes. It contains the so-called "giant gene," a massive sequence that helps build the



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**'It's absolutely exciting to have a whole picture in front of us now,' Stephen Scherer, a lead researcher on Chromosome 7, said yesterday.**

human brain.

Another of its genes prevents humans from being fertilized by another species.

And it also contains genes that help form human hands and feet and the flexible elastin lining that coats humans' guts, lungs and arteries.

In all, it's estimated that Chromosome 7 contains 1,455 genes of the 28,000 in the human genome. As many as 360 of its genes are involved in diseases such as cystic fibrosis and leukemia, and Williams Syndrome, a disorder that results

in mental impairments, distinctive facial features and cardiovascular ailments.

Researchers have also discovered that Chromosome 7 harbours a previously unknown gene linked to autism.

Genes linked to the neurodevelopmental disorder — and there may turn out to be more than 20 — are now of crucial interest to scientists because autism rates have jumped dramatically in the past two decades.

Dr. Scherer, who is also an associate professor of molecular genet-

ics at the University of Toronto, said that researching Chromosome 7 has yielded a clearer understanding of how the human genome operates.

Genes themselves, for example, do not have to carry defects to cause disease.

Some mutations that cause a gene to malfunction lie a fair distance outside the gene.

The mutation that results in "lobster claw," the limb deformity that produces a split foot or hand, is located some way from the gene involved in the formation of these limbs, Dr. Scherer said.

For this reason, researchers are now anxious to study Chromosome 7's "gene deserts," vast chemical stretches that contain no genes, but somehow play a complex role in regulating neighbouring genes.

Dr. Scherer noted that much of the credit for compiling this final version of the map goes to the young team that has spent countless hours crunching and analyzing data using state-of-the-art computer programs, instead of in traditional labs.

Martin Godbout, president and chief executive of Genome Canada, praised the Chromosome 7 effort, calling it "an unprecedented Canadian contribution to the Human Genome Project."