Researchers find wrinkles in human genome

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A team of international scientists, including researchers at the University of Alberta, have created a map of all the known human genomic variations discovered to this point.

The new map, recently published in the journal *Nature*, adds more detail to the HapMap project, which was an update of the original human genome map presented in 2003. Released in 2005, the HapMap project documented the genome sequences of 270 people of various ethnicities.

Researchers hope the new map will lead to a better understanding of the causes of many diseases, which will in turn lead to better treatments for them. Such is already the case for one of the variations, which was discovered on campus.

Dr. Martin Somerville and his research team discovered variation in a portion of chromosome 1 (1q21.1), which they've found in six Alberta and Saskatchewan families. Sometimes the variation does not correspond with any health defects, but other times it coincides with two distinct conditions: congenital heart defects and subtle changes in the eye lens. Other researchers have recently reported an association between this variation and mild to severe autism.

The primary heart defect that the variation signals is a blockage of the aorta. To complicate matters, the condition can also occur when the variation is not shown. The blockage can be managed surgically, but the surgery can lead to a variety of complications that, until recently, have been difficult for doctors to predict. However, when a patient has the

condition and also demonstrates the 1q21.1 variation, doctors are now able to predict what complications are more likely to happen following surgery.

"Since we've been able to identify this variation we've been in a much better position to care for these patients," said Somerville, who is a co-author of the paper, director of the Molecular Diagnostic Laboratory at the Stollery Children's Hospital and a U of A professor of medical genetics.

Somerville likened the human genome to a 2.4 million-page book; each paragraph is a gene, and each letter is a DNA base. He said the discovery of the 1,447 genomic variations that researchers have discovered so far is like finding missing or duplicated pages in the book. He believes many more discoveries will be made in the future.

"I'm really excited about the prospects for genomic research," Somerville said. "We're often able to describe clinically what's wrong with the patient, but with genomics research we're now moving toward finding the root causes of the trouble. And, of course, once we find the causes it is a great step closer to finding the cures."

Source: University of Alberta