Chromosomal microarray analysis proves accurate

March 29 2007

Two years and more than 2,000 samples after researchers at Baylor College of Medicine started to use a new gene-chip technology called chromosomal microarray analysis to look for potential genetic abnormalities in children, they find that it is remarkably sensitive in detecting abnormalities in individual chromosomes, according to a report that appears online today in the journal Public Library of Science ONE.

"Chromosomal microarray analysis is far better at picking up these abnormalities than other common methods," said Dr. Arthur Beaudet, chair of the department of molecular and human genetics at BCM and an author of the paper. "It allows us to identify the source of abnormalities in many cases."

He said the technique does not help with identifying the problem in all children, but there is a five to 12 percent chance that it will identify an abnormality in children with various disabilities where the previous chromosomal testing did not. This study involved two versions of the test, and the newer version was more successful in identifying chromosomal abnormalities.

The targeted microarray analysis used here is essentially a gene chip method of assessing the makeup of chromosomes in a particular part of the human genome (genetic blueprint).

In areas of the genome, there are portions of the genetic material that show imbalances because they are duplicated or deleted abnormally with

gains and losses of genetic information.

"If these changes are big enough, they can cause significant disabilities," said Beaudet.

The technique used is also much more efficient, making it possible to do the equivalent of a couple of thousand of more common testing methods in one test.

"If you tried to do these individually, it would be cost prohibitive," said Beaudet. Currently, chromosomal microarray analysis costs from $950 to $1,500.

In the current study, Beaudet and his colleagues analyzed the results from 2,513 samples taken from patients referred to the laboratory because they had physical or mental features that suggested one of these repetitions or deletions as a genetic cause. The array technique identified the chromosomal problem (too much or too little genetic material in a particular spot) in 8.5 percent of the total group of patients studied. This high resolution genome analysis promise to transform the practice of clinical genetics," said Beaudet.

More important, the technique found abnormalities in 5.2 percent of patients for whom more traditional techniques had not identified the genetic cause.

"We are constantly improving the test," said Beaudet. Eventually, he said, he hopes to develop tests that will look at greater areas of the genome. However, he said, there are many areas of the genome where all normal people have gains or losses of genetic material compared to the average genome.

"It is difficult to determine if it is causing a problem or not," said

Beaudet. Comparing a child's genome to that of the parents can often determine if this is a normal familial variation or a disease-causing one.

Source: Baylor College of Medicine