Science

Linkage Study Identifies Novel Autism Loci

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NEW YORK (Reuters Health) - In a large-scale high-density linkage study, US researchers have identified five genome-wide loci that appear to influence susceptibility to autism.

"There have been many linkage studies of autism, as well as numerous candidate gene scans for association," lead researcher Dr. Dan E. Arking told Reuters Health. "The study we are reporting is novel in that we are looking at ~500,000 SNP markers throughout the genome (a genome-wide association study) in an extremely large dataset, ~800 families with multiple affected members."

The study results were presented Saturday at the American Society of Human Genetics annual meeting in Philadelphia.

According to the meeting abstract, the researchers found four loci with a significant association with autism - 6q25.2, 15q25.3, 17p12, and 17q11.2. The fifth loci, 6q27, showed an even stronger association with the disorder.

With the exception of 15q25.3 and 17q11.2, there was little overlap between the current loci and ones previously tied to autism.

Coupled with the results of a related association study, the findings from this linkage study suggest that "there are unlikely to be genes with large effects that broadly influence risk for autism," Dr. Arking noted.

The linkage results suggest that there are rare variants with a fairly strong impact, but only in a subset of families, whereas in the association study, a susceptibility gene was found with a weaker, but broader impact, he explained.

Regarding future research, Dr. Arking said that "detailed characterization of the identified regions is required to identify candidate genes within the region that would then be screened for mutations."