

Developmental brain disorders studied

U.S. scientists say a recent genetic study of developmental brain disorders might lead to a better understanding of a number of congenital diseases.

The University of California-Irvine study is the first to find mutations of the structural proteins in brain cells -- beta-actin -- are linked with such disorders as deafness and dystonia. The research also suggests genetic variants of the proteins may play a wider role with inherited human diseases.

The study's lead author, Vincent Procaccio, said the findings provide vital clues to the basis of some developmental disorders and make early diagnosis possible for diseases such as dystonia, allowing for greater treatment opportunities.

"These types of actin proteins are prevalent throughout the body and play a key role in processes that are an essential part of development," said Procaccio, an assistant professor of pediatrics. "To find that these mutations are involved with brain disorders seems to be the tip of an iceberg. Since beta-actin is involved with many developmental cell functions, it would appear that its genetic variants can be involved with a number of other congenital disorders."

Study results appeared in the June issue of the American Journal of Human Genetics.

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