Genetic changes tied to rare brain bleeds in babies

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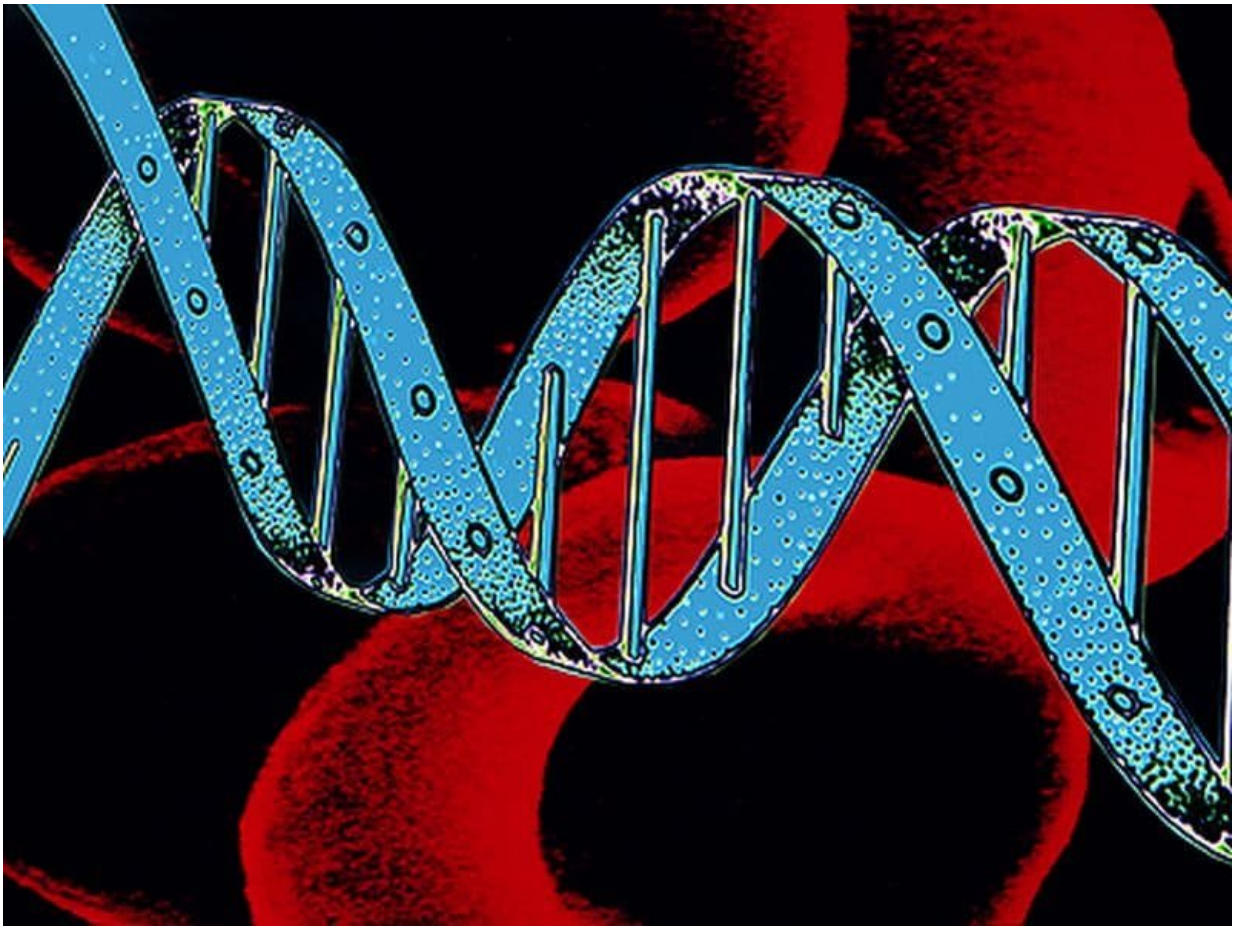

(HealthDay)—Researchers say they've identified genetic mutations

linked with a blood vessel defect that can lead to deadly brain bleeds in babies.

A rare hereditary condition, called vein of Galen malformation, causes high-pressured blood to be pumped from arteries into veins. The veins aren't meant to handle such pressure and can rupture, spilling blood into the brain.

"The effect is like trying to connect your backyard garden hose to a fire hydrant—the veins simply cannot withstand such high pressures from pumping arteries," explained study corresponding author Kristopher Kahle, of Yale University. He's an assistant professor of neurosurgery, pediatrics, and cellular and molecular physiology.

For the study, Kahle's team conducted genetic analyses of 55 children with vein of Galen malformation and their parents. The investigators identified a number of genetic mutations associated with the condition.

One mutation is in a gene called EPHB4, which plays a major role in the development of the vascular system, the study authors reported.

The researchers also discovered that many parents who carry mutations of this gene had skin lesions caused by vascular irregularities. This information might help doctors diagnose vein of Galen malformation before a baby suffers a brain bleed, Kahle and his colleagues said in a Yale news release.

Currently, the best treatment is a minimally invasive catheter procedure, but only a few facilities in the United States can perform the operation, the researchers said.

However, there are drugs to treat other conditions caused by genetic disruptions identified in the study. Those drugs might prove helpful in

treating children with vein of Galen malformation, Kahle said.

The findings were published Dec. 18 in the journal *Neuron*.

More information: The Weill Cornell Brain and Spine Center has more on vein of Galen malformation.