Scientists identify gene vital to early embryonic cells forming a normal heart and skull

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New research from Cincinnati Children's Hospital Medical Center highlights the critical role a certain gene and its protein play during early embryonic development on formation of a normal heart and skull.

In a study posted online June 15 by the *Proceedings of the National Academy of Sciences*, a research team at Cincinnati Children's reports that too little of the gene/protein SHP2 interferes with the normal developmental activity of what are called neural crest cells. These cells, which occur very early in embryonic development, migrate to specific regions of the embryo. While doing so, the cells are supposed to differentiate and give rise to certain nerve tissues, craniofacial bones or smooth muscle tissue of the heart.

"Our findings show that a deficiency of SHP2 in neural crest cells results in a failure of cell differentiation at diverse sites in the developing embryo," said Jeffrey Robbins, Ph.D., co-director of the Heart Institute at Cincinnati Children's and senior investigator of the study. "This leads to anatomical and functional deficits so severe that it precludes viability of the developing fetus."

SHP2 is a tyrosine phosphatase - an enzyme that helps trigger a cascade of biochemical reactions in cells as they specify to form certain tissues.

Although the study was conducted using mouse embryos, the findings are significant in efforts to understand congenital malformations of the heart and craniofacial region in people. Especially relevant, the researchers said, is the insight gained into early molecular events during embryonic development that might help explain such birth defects.

Dr. Robbins said the findings from this study can be used to develop specific drugs that could target the affected pathway, leading to treatment of heart and craniofacial malformations. About 4 percent of human infants are born with congenital malformations. Abnormal heart development is the most common human birth defect, affecting about 1 percent of newborns. The researcher team also wants to explore the exact alterations in neural crest cell migration, expansion and differentiation that contribute to birth defects of other organ systems.

Source: Cincinnati Children's Hospital Medical Center (news : web)