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Cilia structure plays a major role in determining susceptibility to neural tube defects

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Research published online in *The FASEB Journal* shows that the improper methylation of a protein called "Septin2," which regulates the structure of cilia, was associated with an increased risk of having a neural tube defect (NTD) and confirms that cilia are important factors in determining susceptibility of NTDs.

"NTDs are devastating birth defects that compromise multiple aspects of a child's development," said Richard H. Finnell, Ph.D., DABMGG, a researcher involved in the work at the Department of Pediatrics, Dell Medical School, University of Texas at Austin (Austin, Texas). "Preventative methods, while remarkably effective, are not perfect. Efforts must be taken to understand leading developmental pathways that are amenable to modifications that offer hope for correcting the deficits secondary to failure of the neural tube to close properly during early embryogenesis."

Finnell and colleagues used genetically modified mice in which a gene involved in folic acid transport, called "slc19a1," was conditionally inactivated. The deactivated gene allowed the scientists to analyze whether the folic acid transport mechanism functions properly in certain cells of the developing embryo. They found that embryos without a functional slc19a1 gene had neural tube defects. The scientists then used pharmacological methods in these mice to reveal the methylation defects of the cilia protein.

"Despite their impressive name, primary cilia are sometimes not accorded the high stature they should have in both embryonic development and in adult organs," said Thoru Pederson, Ph.D., Editor-in-Chief of *The FASEB Journal*. "The same might be said of the field of protein methylation. This work brings the two together in a most interesting case."

More information: Manami Toriyama et al, Folate-dependent methylation of septins governs ciliogenesis during neural tube closure, *The FASEB Journal* (2017). DOI: 10.1096/fj.201700092R

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