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Sonic hedgehog signaling affected by promoter hypermethylation induces aberrant *Gli2* expression in Spina bifida

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GLI2 is a key mediator of the Sonic hedgehog (Shh) signaling pathway and plays an important role in neural tube development during vertebrate embryogenesis; however, the role of *gli2* in human folate-related neural tube defects remains unclear. In this study, we compared the methylation status and polymorphisms of *gli2* between spina bifida patients and a control group to explore the underlying mechanisms related to folate deficiency in spina bifida. No single nucleotide polymorphism was distributed significantly differently between the two groups, although *gli2* methylation levels were significantly increased in spina bifida samples, accompanied by aberrant *GLI2* expression. Moreover, a significant negative correlation was found between the folate level of brain tissue and the *gli2* methylation status (r=−0.41, P=0.014), while *gli2* hypermethylation increased the risk of spina bifida with an odds ratio of 12.45 (95% confidence interval: 2.71–57.22, P=0.001). We also used a cell model to illustrate the effect of *gli2* expression and the accessibility of chromatin affected by methylation. High *gli2* and gli1 mRNA expression was detected in 5-Aza-treated cells, while *gli2* hypermethylation resulted in chromatin inaccessibility and a reduced association with nuclear proteins containing transcriptional factors. More meaningful to the pathway, the effect gene of the Shh pathway, gli1, was found to have less expression along with decreased expression of *gli2* in the cell model. Aberrant high methylation resulted in the low expression of *gli2* in spina bifida, which was affected by the change in chromatin status and the capacity of transcription factor binding.

Biography

Xiaolin Lu has completed her MM at the Capital Institute of Pediatrics in 2007, and became a young researcher at Beijing Municipal Key Laboratory of Child Development and Nutriomics, Capital Institute of Pediatrics.The main work of her research is about the interaction of genetics and epigenetics in the mechanism of birth defects. Since 2013, she has published about 2 articles on genetic and epigenetic alternations in birth defects as the first author and joint first author.

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