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Association between *SMN2* methylation and disease severity in Chinese children with spinal muscular atrophy

Key words: CpG island, methylation, *SMN2*, spinal muscular atrophy

Research Summary

This article mainly focused on the DNA methylation levels of *SMN2* from 35 SMA children, and evaluated the correlation between the methylation modification and childhood onset of SMA progression.

- ***SMN2* methylation level**
- ***SMN2* transcript level**
- ***SMN2* alternative gene splicing**

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Innovation points

- ***SMN2* methylation, particularly at nt -871 and nt -290, regulates SMA clinical phenotypes by possibly regulating *SMN2* transcription.**

- ***SMN2* nt +938 potentially affects transcription by regulating the alternative splicing.**

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