MedicalXpress

IGF2 variant affects prenatal and postnatal growth

July 9 2015

(HealthDay)—An *IGF2* variant (c.191C—>A, p.Ser64Ter) affects postnatal as well as prenatal growth among those who have inherited the variant through paternal transmission, according to a report published online July 8 in the *New England Journal of Medicine*.

Matthias Begemann, Ph.D., from the Rhine-Westphalia Institute of Technology in Aachen, Germany, and colleagues describe an *IGF2* variant with evidence of pathogenicity in a multigenerational family. Noting previous research that implicates *IGF1* and *IGF1R* in intrauterine and postnatal growth restriction, the team sought to determine effects of mutations of *IGF2*, encoding insulin-like growth factor (IGF) II.

Four family members were found to have severe growth restriction. The researchers found that only family members who had inherited the variant through paternal transmission were affected by the phenotype,

consistent with maternal imprinting status of *IGF2*. Affected family members had severe growth restriction, suggesting that IGF-II affects postnatal growth, as well as prenatal growth. Affected family members also had dysmorphic features, which was consistent with a role of deficient IGF-II levels in the cause of Silver-Russell syndrome.

"In conclusion, the identification of an *IGF2* mutation in patients with growth restriction indicates that IGF-II not only is a mediator of intrauterine development but also contributes to postnatal growth and has pleiotropic effects," the authors write. "Our findings also suggest that treatment with recombinant human growth hormone could be considered."

More information: Abstract
Full Text