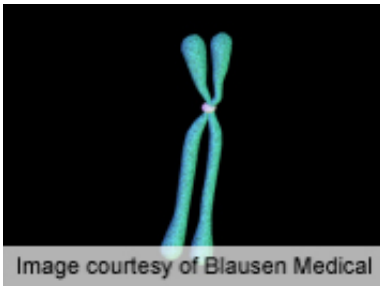


Compound inheritance ID'd in cases of congenital scoliosis

January 8 2015



(HealthDay)—In a case series of Han Chinese persons, compound inheritance of a rare null mutation and a hypomorphic allele accounted for a proportion of congenital scoliosis cases. These findings were published online Jan. 7 in the *New England Journal of Medicine*.

Nan Wu, M.D., from the Peking Union Medical College Hospital in China, and colleagues used comparative genomic hybridization, quantitative polymerase-chain-reaction analysis, and DNA sequencing to assess 161 Han Chinese persons with sporadic congenital scoliosis, 166 Han Chinese controls, and two pedigrees, whose family members had a 16p11.2 deletion. Tests of replication were conducted in 76 Han Chinese persons with congenital scoliosis and a multicenter series of 42 people with 16p11.2 deletions.

The researchers identified heterozygous *TBX6* null mutations in 11 percent of the 161 persons with sporadic congenital scoliosis; null mutations in *TBX6* were not observed in the controls. The null alleles included copy-number variants and single-nucleotide variants. In all 17 carriers of *TBX6* null mutations, the researchers identified a common *TBX6* haplotype as the second risk allele. This compound inheritance model was confirmed in replication studies involving additional persons with congenital scoliosis who carried a deletion affecting *TBX6*. The risk haplotype was suggested to be a hypomorphic allele in in-vitro functional assays.

"Compound inheritance of a rare null mutation and a hypomorphic allele of *TBX6* accounted for up to 11 percent of congenital scoliosis cases in the series that we analyzed," the authors write.

More information: [Abstract](#)
[Full Text](#)

Copyright © 2015 [HealthDay](#). All rights reserved.

Citation: Compound inheritance ID'd in cases of congenital scoliosis (2015, January 8) retrieved 10 April 2024 from

<https://medicalxpress.com/news/2015-01-compound-inheritance-idd-cases-congenital.html>

<p>This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.</p>
--