MedicalXpress

Researchers identify gene linked to vertebral defects in patient populations

May 15 2008

Stowers Institute researchers Karen Staehling-Hampton, Ph.D., Managing Director of Molecular Biology, and Olivier Pourquié, Ph.D., Investigator, collaborated with colleagues from around the world to show that genes known to cause spinal mutations in chick and mouse model systems also play an important role in human patients with congenital vertebral abnormalities.

The discovery was published today on the Web site of the American Journal of Human Genetics.

Working with samples from 31 patients at Boston Children's Hospital with various congenital vertebral defects, the team sequenced five genes thought to be involved in the malformations. In a patient of Puerto Rican descent, the team discovered a mutation in the MESP2 gene – a mutation that completely disrupted the function of the gene.

The affected patient had Spondylothoracic Dysostosis, also known as Jarcho-Levin Syndrome. Spondylothoracic Dysostosis is a rare genetic disorder characterized by distinctive malformations of the vertebrae and ribs, respiratory problems, and other abnormalities. Infants born with Spondylothoracic Dysostosis have short necks, limited neck motion, and are short in stature. Spondylothoracic Dysostosis is prevalent in the Puerto Rican population.

Sequencing of samples from additional Spondylothoracic Dysostosis patients of Puerto Rican descent demonstrated the same mutation in the

MESP2 gene.

“Spondylothoracic Dysostosis was first characterized in the Puerto Rican population 70 years ago,” said Dr. Staehling-Hampton, co-equal first author on the paper, “but the gene mutation causing the condition was not isolated until now. The MESP2 mutation can be detected by a simple assay, so identification of this mutation will allow people who have a family history of Spondylothoracic Dysostosis to determine if they carry the mutation and whether they are at risk of passing the disorder on to future generations.”

“After working for many years to study spinal formation in chicks and mice, it is rewarding to expand our investigation to clinical applications,” said Dr. Pourquié, senior author on the publication. “We will continue to work with colleagues to collect more samples from patients with congenital vertebral abnormalities and sequence more genes to look for causative mutations. Additionally, we will sequence MESP2 in a larger collection of DNA samples from Puerto Rico to determine the carrier frequency of this MESP2 mutation in the general Puerto Rican population.”

Source: Stowers Institute for Medical Research

Citation: Researchers identify gene linked to vertebral defects in patient populations (2008, May 15) retrieved 23 April 2024 from https://medicalxpress.com/news/2008-05-gene-linked-vertebral-defects-patient.html

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.