Medical Xpress

Mouse model provides a new tool for investigators of human developmental disorder

April 21 2009

Wolf-Hirschhorn Syndrome (WHS) is a human disease caused by spontaneous genetic deletions. Children born with WHS have a characteristic set of facial features, including a wide flat nose bridge, downturned mouth, high forehead, and highly arched eyebrows. Other symptoms associated with this disease include heart defects, seizures, mental retardation, and skeletal abnormalities, and the severity of these symptoms varies between individual WHS patients.

While it was known that WHS is related to a genetic deletion in chromosome 4, the specific gene or genes affected were unknown. Now, a study by scientists at the European Molecular Biology Laboratory demonstrates that a gene called Fgfrl1 (Fibroblast growth factor receptor-like 1) plays a key role in WHS.

This report published in *Disease Models & Mechanisms* (DMM), dmm.biologists.org describes how they modify the Fgrfrl1 gene so that it loses function, then express the gene in mice. Fgfrl1 in humans is located on the short arm of chromosome 4 and mice born with the modified Fgfrl1 gene have a variety of physical features that are similar to characteristics seen in WHS patients. For example, the mice are born with heart defects due to thickening of the cardiac valves, and they have abnormal facial and skeletal structures compared to normal mice. The mutant mice also have deformities in throat cartilage structures, which may provide insight to the swallowing and speaking difficulties

experienced by many WHS patients.

This mouse model of WHS provides a valuable new tool for researchers studying this developmental disorder. It provides a new avenue for molecular research through study of Fgrf1 function, but also allows scientists to understand how structural defects might contribute to WHS symptoms, as is the case in the heart defects and swallowing difficulties.

The report, "Multiple congenital malformations of Wolf-Hirschhorn syndrome are recapitulated in Fgfrl1 null mice" was written by Catarina Catela, Daniel Bilbao-Cortes, Esfir Slonimsky, Paschalis Kratsios, Nadia Rosenthal and Pascal te Welscher of the European Molecular Biology Laboratory in Monterotondo, Italy. The report is published in the May/June issue of Disease Models & Mechanisms (DMM), a research journal published by The Company of Biologists, a non-profit based in Cambridge, UK.

Source: The Company of Biologists (news : web)

Citation: Mouse model provides a new tool for investigators of human developmental disorder (2009, April 21) retrieved 16 July 2024 from https://medicalxpress.com/news/2009-04-mouse-tool-human-developmental-disorder.html