IMP Press Release

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Genes for "Michelin Tire Baby" Syndrome Found

Scientists from the Research Institute of Molecular Pathology (IMP) in Vienna, Austria, and the University of Leuven in Belgium report the discovery of two genes that cause the "Michelin Tire Baby" Syndrome. The study is published by the American Journal of Human Genetics today.

First described in 1969, children with this rare syndrome are intellectually disabled, have flat faces, and circumferential skin creases reminiscent of the mascot for the Michelin Tire Company. Long suspected to have a genetic origin, the cause of this disease has nonetheless remained unknown.

Dr. David Keays, Group Leader at the IMP, and Prof. Hilde Van Esch, head of the Laboratory for Genetics and Cognition at KU Leuven, report in a study published online by the American Journal of Human Genetics that the genes responsible are *TUBB* and *MAPRE2*. These genes make proteins that are important for the cells' internal scaffold - the microtubule cytoskeleton. Cells rely on microtubules to divide, migrate, and for transporting cargo.

The TUBB gene makes tubulins, the building blocks of the scaffold, whereas MAPRE2 plays an important role in its organisation. Experiments that were conducted in collaboration with Professor Nicholas Cowan from New York University (NYU) showed that the formation and the dynamics of the microtubule cytoskeleton are altered in patients with TUBB mutations.

"We now know the genes that are responsible for the disease" says David Keays "but it's still a mystery how they cause such unusual symptoms". Future studies will look into this "in the hope that this knowledge will translate into better diagnosis and treatment for those patients with rare genetic diseases".

Original Publication

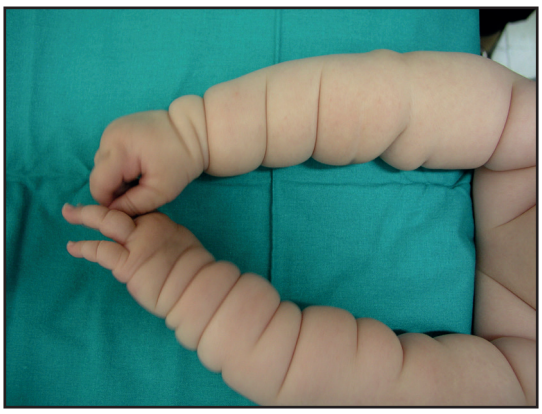
Isrie et al.: Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. The American Journal of Human Genetics 97, 1–11, December 3, 2015.

Illustration

An image can be downloaded from the IMP Website at
http://www.imp.ac.at/pressefoto-michelinbaby
and used free of charge in connection with this press release.

Caption

The image shows a so-called "Michelin Tire Baby" with the characteristic circumferential skin creases due to the folding of excess skin. Credit: KU Leuven – Hilde Van Esch



The arms of a so-called "Michelin Tire Baby" show characteristic circumferential skin creases. Credit: KU Leuven – Hilde Van Esch

About the IMP

The Research Institute of Molecular Pathology (IMP) in Vienna is a basic biomedical research institute largely sponsored by Boehringer Ingelheim. With over 200 scientists from 37 nations, the IMP is committed to scientific discovery of fundamental molecular and cellular mechanisms underlying complex biological phenomena. Research areas include cell and molecular biology, neurobiology, disease mechanisms and computational biology.