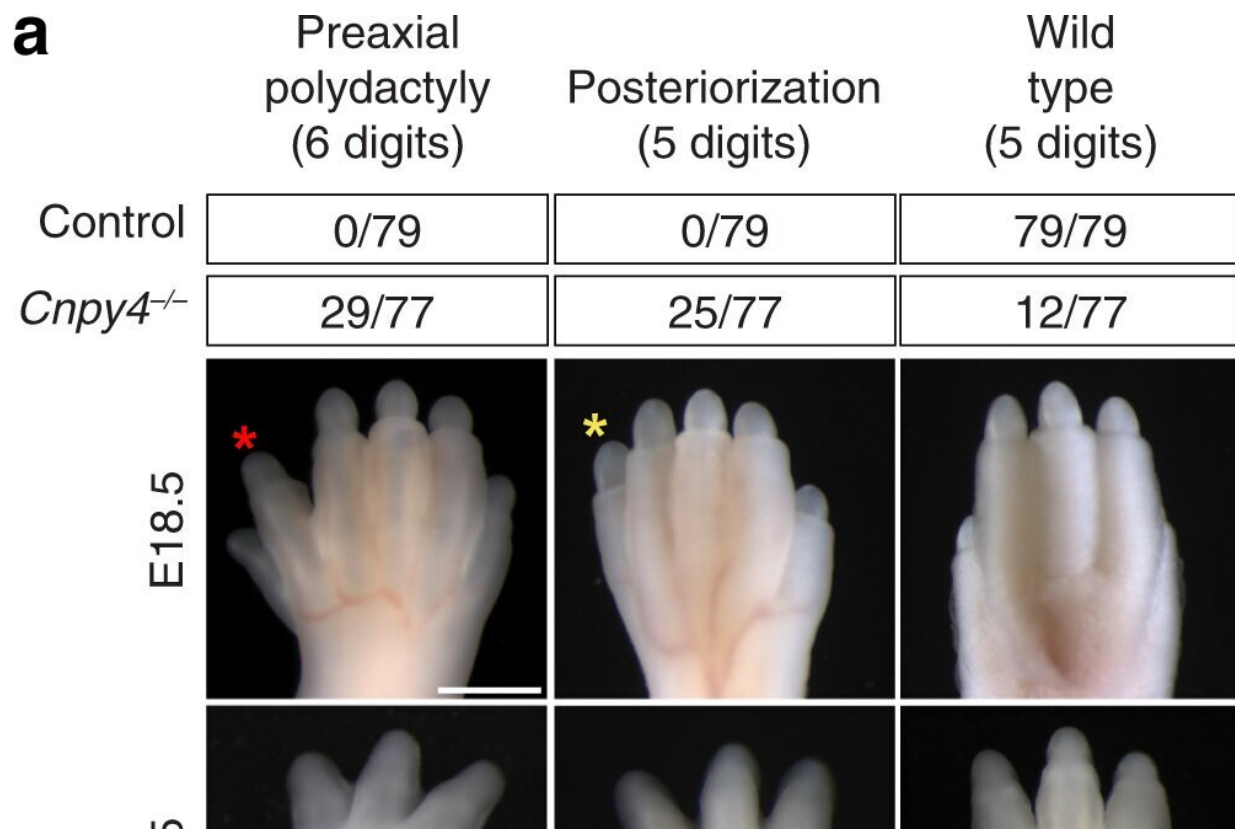


Scientists discover a gene that plays a critical role in embryo development

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Developmental defects in *Cnpy4*^{-/-} hindlimbs. Credit: Megan Lo et al, *Nature Communications* (2022). DOI: 10.1038/s41467-022-30186-x

A new study co-led by a Cedars-Sinai investigator has identified a gene that plays an important role in a biological pathway involved in embryo

development. The gene's impact at the cellular level could explain why some babies are born with physical abnormalities and why some adults develop diseases such as cancer. The findings are published in *Nature Communications*.

Human embryos develop by way of many complex [cellular processes](#). Chemical reactions known as "signaling pathways" activate molecules in a cell to guide its functions. For this study, investigators sought to better understand the so-called Hedgehog signaling pathway, which regulates growth of the human embryo but also is active in adulthood. They discovered that a gene, *Cnpy4*, can affect the ability of a cell to carry out the Hedgehog pathway.

"There are a handful of signaling pathways that are critical regulators of all biological processes that occur during development and are also involved in disease, so we want to understand how these pathways function," said Ophir Klein, MD, Ph.D., co-senior author of the study and executive director of Cedars-Sinai Guerin Children's. "What we've identified here is a gene that is a very important modulator of the critical Hedgehog pathway."

The investigators observed that laboratory mouse embryos with a mutation in the *Cnpy4* gene were born with polydactyly, a birth defect involving extra fingers or toes. Some of the mice also had abnormalities in their spines and ribs. Prior studies have shown that alterations to the Hedgehog pathway can cause these congenital malformations.

The team discovered that the *Cnpy4* gene affects levels of lipids found on the cellular membrane, or the area that separates the interior of the cell from its external environment. The changes in the levels of lipids affect an important protein called Smoothed that is a vital component of the Hedgehog pathway. Mice that lacked *Cnpy4* had elevated levels of accessible cholesterol on their cellular membranes, which caused

Smoothened to signal more strongly. This led to hyperactivity of the pathway and the uncontrolled generation of new cells.

"This pathway is one of a handful that are central to [human development](#)," said Klein, who holds the David and Meredith Kaplan Distinguished Chair in Children's Health at Cedars-Sinai. "And because the same pathways are affected in various disorders, we think they could be a target for therapies for cancers that we know are related to Hedgehog signaling."

Klein said developing drugs that affect the function of the *Cnpy4* gene may provide potential new treatment options for diseases, including cancer.

More information: Megan Lo et al, CNPY4 inhibits the Hedgehog pathway by modulating membrane sterol lipids, *Nature Communications* (2022). [DOI: 10.1038/s41467-022-30186-x](https://doi.org/10.1038/s41467-022-30186-x)

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