

[Open Peer Review on Qeios](#)

# Focal Dermal Hypoplasia

National Cancer Institute

## Source

National Cancer Institute. *Focal Dermal Hypoplasia*. NCI Thesaurus. Code C84715.

A genetic multisystem disorder caused by mutations in the PORCN gene. It is characterized by atrophy and hypoplasia of skin, eye defects, face defects, skeletal abnormalities, and limb malformations.