

## **PORCN gene**

porcupine O-acyltransferase

### **Normal Function**

The *PORCN* gene provides instructions for making a protein that belongs to a group of related proteins called the porcupine (Porc) family. Although the precise function of the PORCN protein is unknown, proteins in the Porc family are involved in the process of transferring a molecule called palmitoleic acid to Wnt proteins. Wnt proteins participate in chemical signaling pathways in the body and play critical roles in development before birth. Members of the Porc family are located in the endoplasmic reticulum, which is a structure inside the cell that is involved in protein processing and transport. The transfer of palmitoleic acid to Wnt proteins facilitates the release of these proteins from the cell so they can regulate development of the skin, bones, and other structures. Researchers are working to determine the specific role of the PORCN protein within human cells.

### **Health Conditions Related to Genetic Changes**

#### Focal dermal hypoplasia

At least 29 mutations in the *PORCN* gene have been found to cause focal dermal hypoplasia. These mutations may alter the protein's structure, lead to the production of an abnormally short version of the protein, or delete the entire *PORCN* gene. All of these mutations appear to result in the absence of any functional PORCN protein. Researchers believe Wnt proteins cannot be released from the cell without the PORCN protein. When Wnt proteins are unable to leave the cell, they cannot participate in the chemical signaling pathways that are critical for normal development.

#### Coloboma

MedlinePlus Genetics provides information about Coloboma

### **Other Names for This Gene**

- DHOF
- FODH
- MG61
- MGC29687

- por
- PORC
- PORCN\_HUMAN
- porcupine
- porcupine homolog (Drosophila)
- porcupine isoform A
- porcupine isoform B
- porcupine isoform C
- porcupine isoform D
- porcupine isoform E
- PPN

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of PORCN ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64840\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=64840[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28PORCN%5BTIAB%5D%29+OR+%28%28PORC%5BTIAB%5D%29+OR+%28porcupine%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- PORCUPINE O-ACYLTRANSFERASE; PORCN (<https://omim.org/entry/300651>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/64840>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=PORCN\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=PORCN[gene]))

## **References**

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Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19292719>)

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## Genomic Location

The *PORCN* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

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