

## DCAF17 gene

DDB1 and CUL4 associated factor 17

### Normal Function

The *DCAF17* gene provides instructions for making a protein whose function is unknown. The gene is active (expressed) in several organs and tissues in the body, including the brain, skin, and liver. Within cells, the protein produced from this gene is found in the nucleolus, which is a small region inside the nucleus where cell structures called ribosomes are assembled. It is not clear whether the DCAF17 protein plays a role in this process.

### Health Conditions Related to Genetic Changes

#### Woodhouse-Sakati syndrome

At least 11 *DCAF17* gene mutations have been identified in people with Woodhouse-Sakati syndrome, a disorder that affects the body's network of hormone-producing glands (the endocrine system) and the nervous system. This condition is characterized by delayed or absent puberty, hair loss, abnormal movements, hearing loss, and intellectual disability.

Most of the *DCAF17* gene mutations that cause Woodhouse-Sakati syndrome result in a protein that is abnormally short and breaks down quickly or whose usual function is impaired. Loss of DCAF17 protein function likely accounts for the features of Woodhouse-Sakati syndrome, although it is unclear how a shortage of this protein leads to hormone abnormalities and the other signs and symptoms of this condition.

### Other Names for This Gene

- C2orf37
- FLJ13096

### Additional Information & Resources

#### Tests Listed in the Genetic Testing Registry

- Tests of DCAF17 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80067\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=80067[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DCAF17%5BTIAB%5D%29+OR+%28DDB1+and+CUL4+associated+factor+17%5BTIAB%5D%29%29+OR+%28C2orf37%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+OR+%28Woodhouse-Sakati%29+AND+english%5Bla%5D+AND+human%5Bmh%5D%29%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- DDB1- AND CUL4-ASSOCIATED FACTOR 17; DCAF17 (<https://omim.org/entry/612515>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/80067>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DCAF17\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DCAF17[gene]))

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## Genomic Location

The *DCAF17* gene is found on chromosome 2 (<https://medlineplus.gov/genetics/chromosome/2/>).

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