

## DCXR gene

dicarbonyl and L-xylulose reductase

### Normal Function

The *DCXR* gene provides instructions for making a protein called dicarbonyl and L-xylulose reductase (DCXR), which plays multiple roles in the body. One of its functions is to perform a chemical reaction that converts a sugar called L-xylulose to a molecule called xylitol. This reaction is one step in a process by which the body can use sugars for energy. There are two versions of L-xylulose reductase in the body, known as the major isoform and the minor isoform. The *DCXR* gene provides instructions for making the major isoform, which converts L-xylulose more efficiently than the minor isoform. It is unclear if the minor isoform is produced from the *DCXR* gene or another gene.

Another function of the DCXR protein is to break down toxic compounds called alpha-dicarbonyl compounds. These compounds, which are byproducts of certain cellular processes or are found in foods in the diet, must be broken down so they do not damage cells.

The DCXR protein is also one of several proteins that get attached to the surface of sperm cells as they mature. DCXR is involved in the interaction of a sperm cell with an egg cell during fertilization.

### Health Conditions Related to Genetic Changes

#### Essential pentosuria

At least two mutations in the *DCXR* gene cause a condition called essential pentosuria, which is found almost exclusively in individuals with Ashkenazi Jewish ancestry. Affected individuals have high levels of L-xylulose in their urine, but they have no associated health problems. The gene mutations involved in this condition lead to the production of altered DCXR proteins that are quickly broken down. Without this protein, L-xylulose is not efficiently converted to xylitol, and the excess sugar is released in the urine. Only the major isoform of L-xylulose reductase is affected by these mutations, but the minor isoform cannot compensate for the loss of DCXR's function in breaking down L-xylulose. It is thought that other processes are able to break down toxic alpha-dicarbonyl compounds, likely accounting for the lack of symptoms in people with essential pentosuria.

Males with essential pentosuria appear to have normal reproductive function, despite studies that show that a shortage of DCXR protein attached to sperm cells can be associated with the inability to have biological children (infertility).

## **Other Names for This Gene**

- carbonyl reductase 2
- carbonyl reductase II
- DCR
- dicarbonyl/L-xylulose reductase
- HCR2
- HCRII
- KIDCR
- kidney dicarbonyl reductase
- L-xylulose reductase
- L-xylulose reductase isoform 1
- L-xylulose reductase isoform 2
- P34H
- SDR20C1
- short chain dehydrogenase/reductase family 20C, member 1
- sperm surface protein P34H
- XR

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of DCXR ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51181\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=51181[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28DCXR%5BTIAB%5D%29+OR+%28dicarbonyl/L-xylulose+reductase%5BTIAB%5D%29%29+OR+%28%28L-xylulose+reductase%5BTIAB%5D%29+OR+%28P34H%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D%29>)

### Catalog of Genes and Diseases from OMIM

- DICARBONYL/L-XYLULOSE REDUCTASE; DCXR (<https://omim.org/entry/608347>)

## Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/51181>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DCXR\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DCXR[gene]))

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## **Genomic Location**

The *DCXR* gene is found on chromosome 17 (<https://medlineplus.gov/genetics/chromosome/17/>).

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