

DDX11 gene

DEAD/H-box helicase 11

Normal Function

The *DDX11* gene provides instructions for making an enzyme called ChIR1, which functions as a helicase. Helicases are enzymes that attach (bind) to DNA and temporarily unwind the two spiral strands (double helix) of the DNA molecule so it can be copied (replicated) in preparation for cell division. ChIR1 is also involved in repairing any errors that are made when DNA is copied. In addition, ChIR1 is involved in other processes leading up to cell division. After replication, the DNA from each chromosome is arranged into two identical structures, called sister chromatids, which the ChIR1 enzyme helps to keep together until they are ready to separate into individual cells. This enzyme also ensures proper separation of chromatids during cell division. By helping repair errors in DNA and ensuring proper DNA replication, the ChIR1 enzyme plays a role in maintaining the stability of a cell's genetic information.

Health Conditions Related to Genetic Changes

Warsaw breakage syndrome

At least three mutations in the *DDX11* gene have been found to cause Warsaw breakage syndrome. This condition causes multiple abnormalities that may include impaired growth, distinctive facial features, hearing loss, and heart malformations. The mutations that cause Warsaw breakage syndrome severely reduce or completely eliminate ChIR1 enzyme activity. As a result, the enzyme cannot bind to DNA and cannot unwind the DNA strands to help with DNA replication and repair. A lack of functional ChIR1 impairs cell division and leads to an accumulation of DNA damage. This DNA damage can appear as breaks in the DNA, giving the condition its name. It is unclear how these problems in DNA maintenance lead to the specific abnormalities characteristic of Warsaw breakage syndrome.

Other Names for This Gene

- CHL1-related helicase gene-1
- CHL1-related protein 1
- CHLR1
- DDX11_HUMAN

- DEAD/H (Asp-Glu-Ala-Asp/His) box helicase 11
- DEAD/H (Asp-Glu-Ala-Asp/His) box polypeptide 11 (CHL1-like helicase homolog, *S. cerevisiae*)
- DEAD/H box protein 11
- hCHLR1
- keratinocyte growth factor-regulated gene 2 protein
- KRG-2
- KRG2
- probable ATP-dependent RNA helicase DDX11

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of DDX11 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1663\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=1663[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28DDX11%5BTIAB%5D%29+OR+%28CHLR1%5BTIAB%5D%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

- DEAD/H-BOX HELICASE 11; DDX11 (<https://omim.org/entry/601150>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/1663>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=DDX11\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=DDX11[gene]))

References

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Genomic Location

The *DDX11* gene is found on chromosome 12 (<https://medlineplus.gov/genetics/chromosome/12/>).

Last updated February 1, 2014