



## Warsaw breakage syndrome

Warsaw breakage syndrome is a condition that can cause multiple abnormalities. People with Warsaw breakage syndrome have intellectual disability that varies from mild to severe. They also have impaired growth from birth leading to short stature and a small head size (microcephaly). Affected individuals have distinctive facial features that may include a small forehead, a short nose, a small lower jaw, a flat area between the nose and mouth (philtrum), and prominent cheeks. Other common features include hearing loss caused by nerve damage in the inner ear (sensorineural hearing loss) and heart malformations.

### Frequency

Warsaw breakage syndrome is a rare condition; at least four cases have been described in the medical literature.

### Causes

Mutations in the *DDX11* gene cause Warsaw breakage syndrome. The *DDX11* gene provides instructions for making an enzyme called ChIR1. This enzyme 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. This unwinding is necessary for copying (replicating) DNA in preparation for cell division, and for repairing damaged DNA and any errors that are made when DNA is copied. In addition, after DNA is copied, ChIR1 plays a role in ensuring proper separation of each chromosome during cell division. By helping repair errors in DNA and ensuring proper DNA replication, the ChIR1 enzyme is involved in maintaining the stability of a cell's genetic information.

*DDX11* gene mutations 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.

### Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## Other Names for This Condition

- WABS

## Diagnosis & Management

### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Warsaw breakage syndrome  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C3150658/>

### Other Diagnosis and Management Resources

- Centers for Disease Control and Prevention: Hearing Loss in Children  
<https://www.cdc.gov/ncbddd/hearingloss/>
- MedlinePlus Encyclopedia: Hearing Loss--Infants  
<https://medlineplus.gov/ency/article/007322.htm>

## Additional Information & Resources

### Health Information from MedlinePlus

- Encyclopedia: Delayed Growth  
<https://medlineplus.gov/ency/article/003021.htm>
- Encyclopedia: Hearing Loss--Infants  
<https://medlineplus.gov/ency/article/007322.htm>
- Encyclopedia: Intellectual Disability  
<https://medlineplus.gov/ency/article/001523.htm>
- Health Topic: Growth Disorders  
<https://medlineplus.gov/growthdisorders.html>

### Additional NIH Resources

- National Heart, Lung, and Blood Institute: What Are Congenital Heart Defects?  
<https://www.nhlbi.nih.gov/health-topics/congenital-heart-defects>

### Educational Resources

- Boys Town National Research Hospital: Types of Hearing Loss  
<https://www.boystownhospital.org/knowledgeCenter/articles/hearing/Pages/TypesofHearing.aspx>
- Centers for Disease Control and Prevention: Hearing Loss in Children  
<https://www.cdc.gov/ncbddd/hearingloss/>
- Centers for Disease Control and Prevention: Intellectual Disability  
[https://www.cdc.gov/ncbddd/actearly/pdf/parents\\_pdfs/IntellectualDisability.pdf](https://www.cdc.gov/ncbddd/actearly/pdf/parents_pdfs/IntellectualDisability.pdf)

- Cincinnati Children's Hospital: Short Stature  
<https://www.cincinnatichildrens.org/health/s/short-stature>
- Johns Hopkins Children's Center: Short Stature  
<https://www.hopkinsmedicine.org/health/conditions-and-diseases/short-stature>
- Kennedy Krieger Institute: Intellectual Disability  
<https://www.kennedykrieger.org/patient-care/conditions/intellectual-disability>
- MalaCards: warsaw breakage syndrome  
[https://www.malacards.org/card/warsaw\\_breakage\\_syndrome](https://www.malacards.org/card/warsaw_breakage_syndrome)
- Orphanet: Warsaw breakage syndrome  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=280558](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=280558)

#### Patient Support and Advocacy Resources

- American Association on Intellectual and Developmental Disabilities (AAIDD)  
<http://aaid.org/>
- American Heart Association  
<https://www.heart.org/en/>
- Little People UK  
<https://littlepeopleuk.org/>
- The MAGIC Foundation  
<https://www.magicfoundation.org/>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28warsaw+breakage+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

#### Catalog of Genes and Diseases from OMIM

- WARSAW BREAKAGE SYNDROME  
<http://omim.org/entry/613398>

## Sources for This Summary

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<https://ghr.nlm.nih.gov/condition/warsaw-breakage-syndrome>

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