



## Beare-Stevenson cutis gyrata syndrome

Beare-Stevenson cutis gyrata syndrome is a genetic disorder characterized by skin abnormalities and the premature fusion of certain bones of the skull (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face.

Many of the characteristic facial features of Beare-Stevenson cutis gyrata syndrome result from the premature fusion of the skull bones. The head is unable to grow normally, which leads to a cloverleaf-shaped skull, wide-set and bulging eyes, ear abnormalities, and an underdeveloped upper jaw. Early fusion of the skull bones also affects the growth of the brain, causing delayed development and intellectual disability.

A skin abnormality called cutis gyrata is also characteristic of this disorder. The skin has a furrowed and wrinkled appearance, particularly on the face, near the ears, and on the palms and soles of the feet. Additionally, thick, dark, velvety areas of skin (acanthosis nigricans) are sometimes found on the hands and feet and in the genital region.

Additional signs and symptoms of Beare-Stevenson cutis gyrata syndrome can include a blockage of the nasal passages (choanal atresia), overgrowth of the umbilical stump (tissue that normally falls off shortly after birth, leaving the belly button), and abnormalities of the genitalia and anus. The medical complications associated with this condition are often life-threatening in infancy or early childhood.

### Frequency

Beare-Stevenson cutis gyrata syndrome is a rare genetic disorder; its incidence is unknown. Fewer than 20 people with this condition have been reported worldwide.

### Causes

Mutations in the *FGFR2* gene cause Beare-Stevenson cutis gyrata syndrome. This gene produces a protein called fibroblast growth factor receptor 2, which plays an important role in signaling a cell to respond to its environment, perhaps by dividing or maturing. A mutation in the *FGFR2* gene alters the protein and promotes prolonged signaling, which is thought to interfere with skeletal and skin development.

Some individuals with Beare-Stevenson cutis gyrata syndrome do not have identified mutations in the *FGFR2* gene. In these cases, the cause of the condition is unknown.

### Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. All reported cases have

resulted from new mutations in the gene, and occurred in people with no history of the disorder in their family.

### **Other Names for This Condition**

- cutis gyrata syndrome of Beare and Stevenson
- cutis gyrata syndrome of Beare-Stevenson

### **Diagnosis & Management**

#### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetic-testing](#)
- Genetic Testing Registry: Cutis Gyrata syndrome of Beare and Stevenson  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1852406/>

#### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22craniosynostoses%22+OR+%22beare-stevenson+cutis+gyrata+syndrome%22>

#### Other Diagnosis and Management Resources

- GeneReview: FGFR-Related Craniosynostosis Syndromes  
<https://www.ncbi.nlm.nih.gov/books/NBK1455>
- MedlinePlus Encyclopedia: Acanthosis Nigricans  
<https://medlineplus.gov/ency/article/000852.htm>
- MedlinePlus Encyclopedia: Craniosynostosis  
<https://medlineplus.gov/ency/article/001590.htm>

### **Additional Information & Resources**

#### Health Information from MedlinePlus

- Encyclopedia: Acanthosis Nigricans  
<https://medlineplus.gov/ency/article/000852.htm>
- Encyclopedia: Craniosynostosis  
<https://medlineplus.gov/ency/article/001590.htm>
- Health Topic: Craniofacial Abnormalities  
<https://medlineplus.gov/craniofacialabnormalities.html>

#### Genetic and Rare Diseases Information Center

- Beare-Stevenson cutis gyrata syndrome  
<https://rarediseases.info.nih.gov/diseases/332/beare-stevenson-cutis-gyrata-syndrome>

### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Craniosynostosis Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Craniosynostosis-Information-Page>

### Educational Resources

- Collaboration for Craniofacial Development and Disorders, Johns Hopkins University  
[https://www.hopkinsmedicine.org/neurology\\_neurosurgery/centers\\_clinics/pediatric\\_neurosurgery/conditions/craniosynostosis/](https://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/craniosynostosis/)
- MalaCards: beare-stevenson cutis gyrata syndrome  
[https://www.malacards.org/card/beare\\_stevenson\\_cutis\\_gyrata\\_syndrome](https://www.malacards.org/card/beare_stevenson_cutis_gyrata_syndrome)
- Orphanet: Craniosynostosis  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1531](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1531)
- Orphanet: Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=1555](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1555)

### Patient Support and Advocacy Resources

- AmeriFace  
<http://www.ameriface.org/>
- Childrens' Craniofacial Association  
<https://ccakids.org/>
- Resource list from the University of Kansas Medical Center  
<http://www.kumc.edu/gec/support/craniofa.html>

### Clinical Information from GeneReviews

- FGFR-Related Craniosynostosis Syndromes  
<https://www.ncbi.nlm.nih.gov/books/NBK1455>

### Scientific Articles on PubMed

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

### Catalog of Genes and Diseases from OMIM

- BEARE-STEVENSON CUTIS GYRATA SYNDROME  
<http://omim.org/entry/123790>

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Reprinted from Genetics Home Reference:

<https://ghr.nlm.nih.gov/condition/beare-stevenson-cutis-gyrata-syndrome>

Reviewed: February 2008

Published: June 25, 2019

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services