Beare-Stevenson cutis gyrata syndrome

Description

Beare-Stevenson cutis gyrata syndrome is a genetic disorder that typically features 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), a malformation of the airways (tracheal cartilaginous sleeve), 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. Approximately 25 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.

Learn more about the gene associated with Beare-Stevenson cutis gyrata syndrome

- FGFR2

Inheritance

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

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Beare-Stevenson cutis gyrata syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1852406/)

Genetic and Rare Diseases Information Center

- Cutis gyrata-acanthosis nigricans-craniosynostosis syndrome (https://rarediseases.info.nih.gov/diseases/332/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Catalog of Genes and Diseases from OMIM

- BEARE-STEVENSON CUTIS GYRATA SYNDROME; BSTVS (https://omim.org/entry/123790)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28beare-stevenson+cutis+g

yrata+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)

References

- Chen L, Deng CX. Roles of FGF signaling in skeletal development and humangenetic diseases. Front Biosci. 2005 May 1;10:1961-76. doi: 10.2741/1671. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15769677)
- Eun SH, Ha KS, Je BK, Lee ES, Choi BM, Lee JH, Eun BL, Yoo KH. The firstKorean case of Beare-Stevenson syndrome with a Tyr375Cys mutation in thefibroblast growth factor receptor 2 gene. J Korean Med Sci. 2007 Apr;22(2):352-6. doi: 10.3346/jkms.2007.22.2.352. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17449949) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2693607/)
- Hall BD, Cadle RG, Golabi M, Morris CA, Cohen MM Jr. Beare-Stevenson cutisgyrata syndrome. Am J Med Genet. 1992 Sep 1;44(1):82-9. doi:10.1002/ajmg.1320440120. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/1519658)
- Izakovic J, Leitner S, Schachner LA. What syndrome is this? Beare-Stevensoncutis gyrata syndrome. Pediatr Dermatol. 2003 Jul-Aug;20(4):358-60. doi:10.1046/j.1525-1470.2003.20419.x. No abstract available. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12869163)
- McGaughran J, Sinnott S, Susman R, Buckley MF, Elakis G, Cox T, Roscioli T. Acase of Beare-Stevenson syndrome with a broad spectrum of features and a reviewof the FGFR2 Y375C mutation phenotype. Clin Dysmorphol. 2006 Apr;15(2):89-93.doi: 10.1097/01.mcd.0000194407.92676.9d. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/16531735)
- Przylepa KA, Paznekas W, Zhang M, Golabi M, Bias W, Bamshad MJ, Carey JC, HallBD, Stevenson R, Orlow S, Cohen MM Jr, Jabs EW. Fibroblast growth factor receptor2 mutations in Beare-Stevenson cutis gyrata syndrome. Nat Genet. 1996Aug;13(4):492-4. doi: 10.1038/ng0896-492. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8696350)
- Ron N, Leung S, Carney E, Gerber A, David KL. A Case of Beare-StevensonSyndrome with Unusual Manifestations. Am J Case Rep. 2016 Apr 15;17:254-8. doi:10.12659/ajcr.897177. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/27079505) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4835158/)
- Vargas RA, Maegawa GH, Taucher SC, Leite JC, Sanz P, Cifuentes J, Parra M, Munoz H, Maranduba CM, Passos-Bueno MR. Beare-Stevenson syndrome: Two SouthAmerican patients with FGFR2 analysis. Am J Med Genet A. 2003 Aug15;121A(1):41-6. doi: 10.1002/ajmg.a.20101. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12900900)
- Wang TJ, Huang CB, Tsai FJ, Wu JY, Lai RB, Hsiao M. Mutation in the FGFR2 genein a Taiwanese patient with Beare-Stevenson cutis gyrata syndrome. Clin

Genet.2002 Mar;61(3):218-21. doi: 10.1034/j.1399-0004.2002.610309.x. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12000365)

- Wenger T, Miller D, Evans K. FGFR Craniosynostosis SyndromesOverview. 1998 Oct 20 [updated 2020 Apr 30]. In: Adam MP, Feldman J, Mirzaa GM,Pagon RA, Wallace SE, Bean LJH, Gripp KW, Amemiya A, editors.GeneReviews(R) [Internet]. Seattle (WA): University of Washington,Seattle; 1993-2024. Available from http://www.ncbi.nlm.nih.gov/books/NBK1455/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/20301628)
- Wenger TL, Bhoj EJ, Wetmore RF, Mennuti MT, Bartlett SP, Mollen TJ,McDonald-McGinn DM, Zackai EH. Beare-Stevenson syndrome: two new patients,including a novel finding of tracheal cartilaginous sleeve. Am J Med Genet A.2015 Apr;167A(4):852-7. doi: 10.1002/ajmg.a.36985. Epub 2015 Feb 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/25706251)

Last updated June 1, 2020