Crouzon syndrome

Description

Crouzon syndrome is a genetic disorder characterized by the premature fusion of certain skull bones (craniosynostosis). This early fusion prevents the skull from growing normally and affects the shape of the head and face.

Many features of Crouzon syndrome result from the premature fusion of the skull bones. Abnormal growth of these bones leads to wide-set, bulging eyes and vision problems caused by shallow eye sockets; eyes that do not point in the same direction (strabismus) ; a beaked nose; and an underdeveloped upper jaw. In addition, people with Crouzon syndrome may have dental problems and hearing loss, which is sometimes accompanied by narrow ear canals. A few individuals with Crouzon syndrome have an opening in the lip and the roof of the mouth (cleft lip and palate). The severity of these signs and symptoms varies among affected people. Individuals with Crouzon syndrome usually have normal intelligence.

Frequency

Crouzon syndrome is seen in about 16 per million newborns. It is the most common craniosynostosis syndrome.

Causes

Mutations in the *FGFR2* gene cause Crouzon syndrome. This gene provides instructions for making a protein called fibroblast growth factor receptor 2. Among its multiple functions, this protein signals immature cells to become bone cells during embryonic development. Mutations in the *FGFR2* gene are thought to result in production of an FGFR2 protein with overactive signaling, which causes the bones of the skull to fuse prematurely.

Learn more about the gene associated with Crouzon 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.

Other Names for This Condition

- CFD1
- Craniofacial dysarthrosis
- Craniofacial dysostosis
- Craniofacial dysostosis syndrome
- Craniofacial dysostosis type 1
- Crouzon craniofacial dysostosis
- Crouzon disease
- Crouzon's disease

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Crouzon syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0010273/)

Genetic and Rare Diseases Information Center

- Crouzon syndrome (https://rarediseases.info.nih.gov/diseases/6206/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Crouzon syndrome%22)

Catalog of Genes and Diseases from OMIM

- CROUZON SYNDROME (https://omim.org/entry/123500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Craniofacial+Dysostosis%5BMAJR%5D%29+AND+%28Crouzon+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

References

- Azoury SC, Reddy S, Shukla V, Deng CX. Fibroblast Growth Factor Receptor 2(FGFR2) Mutation Related Syndromic Craniosynostosis. Int J Biol Sci.2017 Nov 2;13(12):1479-1488. doi: 10.7150/ijbs.22373. eCollection 2017. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/29230096) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC5723914/)
- Carinci F, Pezzetti F, Locci P, Becchetti E, Carls F, Avantaggiato A,Becchetti A, Carinci P, Baroni T, Bodo M. Apert and Crouzon syndromes: clinicalfindings, genes and extracellular matrix. J Craniofac Surg. 2005 May;16(3):361-8.doi: 10.1097/01.scs.0000157078.53871.11. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15915098)
- Conrady CD, Patel BC. Crouzon Syndrome. 2023 Aug 7. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; 2024 Jan-. Available fromhttp://www.ncbi.nlm.nih.gov/books/NBK518998/ Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/30085540)
- Galvin BD, Hart KC, Meyer AN, Webster MK, Donoghue DJ. Constitutive receptoractivation by Crouzon syndrome mutations in fibroblast growth factor receptor(FGFR)2 and FGFR2/Neu chimeras. Proc Natl Acad Sci U S A. 1996 Jul23; 93(15):7894-9. doi: 10.1073/pnas.93.15.7894. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/8755573) or Free article on PubMed Central (https://www.ncbi.nlm.nih.gov/pmc/articles/PMC38845/)
- Gray TL, Casey T, Selva D, Anderson PJ, David DJ. Ophthalmic sequelae ofCrouzon syndrome. Ophthalmology. 2005 Jun;112(6):1129-34. doi:10.1016/j.ophtha.2004.12.037. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/15885794)
- 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)

Last updated January 1, 2020