Crouzon syndrome

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 people 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. People with Crouzon syndrome are usually of 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 probably overstimulate signaling by the FGFR2 protein, which causes the bones of the skull to fuse prematurely.

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.

Other Names for This Condition

- Craniofacial dysarthrosis
- Craniofacial Dysostosis
- Craniofacial dysostosis syndrome
- Craniofacial dysostosis, type 1; CFD1
- Crouzon craniofacial dysostosis
• Crouzon's Disease
• Crouzons Disease

**Diagnosis & Management**

**Genetic Testing Information**

• What is genetic testing? /primer/testing/genetic-testing

• Genetic Testing Registry: Crouzon syndrome

**Research Studies from ClinicalTrials.gov**

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22crouzon+syndrome%22+OR+%22craniofacial+abnormalities%22+OR+%22craniofacial+dysostosis%22

**Other Diagnosis and Management Resources**

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

• MedlinePlus Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm

**Additional Information & Resources**

**Health Information from MedlinePlus**

• Encyclopedia: Craniosynostosis
  https://medlineplus.gov/ency/article/001590.htm

• Health Topic: Craniofacial Abnormalities
  https://medlineplus.gov/craniofacialabnormalities.html

**Genetic and Rare Diseases Information Center**

• Crouzon syndrome
  https://rarediseases.info.nih.gov/diseases/6206/crouzon-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

- Boston Children's Hospital
  http://www.childrenshospital.org/conditions-and-treatments/conditions/c/crouzon-syndrome

- Collaboration for Craniofacial Development and Disorders, Johns Hopkins Medicine
  https://www.hopkinsmedicine.org/neurology_neurosurgery/centers_clinics/pediatric_neurosurgery/conditions/craniosynostosis/

- Headlines Craniofacial Support (UK)

- MalaCards: crouzon syndrome
  https://www.malacards.org/card/crouzon_syndrome

- Orphanet: Crouzon disease
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=207

- Seattle Children's Hospital and Regional Medical Center
  https://www.seattlechildrens.org/conditions/chromosomal-genetic-conditions/crouzon-syndrome

- UC Davis Children's Hospital

Patient Support and Advocacy Resources

- ACPA Family Services
  https://cleftline.org/

- Children's Craniofacial Association
  https://ccakids.org/

- National Organization for Rare Disorders (NORD)
  https://rarediseases.org/rare-diseases/crouzon-syndrome/

- 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=%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
Catalog of Genes and Diseases from OMIM

- CROUZON SYNDROME
  http://omim.org/entry/123500

Medical Genetics Database from MedGen

- Crouzon syndrome

Sources for This Summary

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  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8755573
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC38845/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15885794

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301628

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