Muenke syndrome

Muenke syndrome is a condition characterized by the premature closure of certain bones of the skull (craniosynostosis) during development, which affects the shape of the head and face.

Many people with this disorder have a premature fusion of skull bones along the coronal suture, the growth line that goes over the head from ear to ear. Other parts of the skull may also be malformed. These changes can result in an abnormally shaped head, wide-set eyes, and flattened cheekbones. About 5 percent of affected individuals have an enlarged head (macrocephaly). People with Muenke syndrome may also have mild abnormalities of the hands or feet, and hearing loss has been observed in some cases. Most people with this condition have normal intellect, but developmental delay and learning problems are possible.

The signs and symptoms of Muenke syndrome vary among affected people, and some features overlap with those seen in other craniosynostosis syndromes. A small percentage of people with the gene mutation associated with Muenke syndrome do not have any of the characteristic features of the disorder.

Frequency

Muenke syndrome occurs in about 1 in 30,000 newborns. This condition accounts for an estimated 8 percent of all cases of craniosynostosis.

Causes

A particular mutation in the \textit{FGFR3} gene causes Muenke syndrome. The \textit{FGFR3} gene provides instructions for making a protein that is involved in the development and maintenance of bone and brain tissue. The mutation associated with Muenke syndrome causes the FGFR3 protein to be overly active, which interferes with normal bone growth and allows the bones of the skull to fuse before they should.

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

- FGFR3-associated coronal synostosis
- Muenke nonsyndromic coronal craniosynostosis
Diagnosis & Management

Genetic Testing Information

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

• Genetic Testing Registry: Muenke syndrome

Research Studies from ClinicalTrials.gov

• ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22craniosynostoses%22+OR+%22muenke+syndrome%22

Other Diagnosis and Management Resources

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

• GeneReview: Muenke Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK1415

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

• NIH Clinical Study of Muenke Syndrome
  https://muenkesyndrome.nhgri.nih.gov/

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

• Muenke 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

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

- Centers for Disease Control and Prevention: Craniosynostosis
  https://www.cdc.gov/ncbddd/birthdefects/craniosynostosis.html

- Children's Hospital of Philadelphia
  https://www.chop.edu/conditions-diseases/muenke-syndrome

- Children's National Health System: Craniosynostosis

- Great Ormond Street Hospital for Children (UK)
  https://www.gosh.nhs.uk/conditions-and-treatments/conditions-we-treat/muenke-syndrome

- MalaCards: muenke syndrome
  https://www.malacards.org/card/muenke_syndrome

- Orphanet: Muenke syndrome
  https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=53271

- Seattle Children’s Hospital
  https://www.seattlechildrens.org/conditions/chromosomal-genetic-conditions/muenke-syndrome

- UC Davis Children’s Hospital: Craniofacial anomalies – Craniosynostosis

Patient Support and Advocacy Resources

- AboutFace
  https://www.aboutface.ca/

- AmeriFace
  http://www.ameriface.org/

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

- National Association of the Deaf
  https://www.nad.org/

- Resource list from the University of Kansas Medical Center: Facial Anomalies / Craniofacial Conditions
  http://www.kumc.edu/gec/support/craniofa.html
Clinical Information from GeneReviews

- **FGFR-Related Craniosynostosis Syndromes**
  https://www.ncbi.nlm.nih.gov/books/NBK1455
- **Muenke Syndrome**
  https://www.ncbi.nlm.nih.gov/books/NBK1415

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28muenke+syndrome%5BTIAB%29%29+OR+%28fgfr3-associated+coronal+synostosis%5BTIAB%29%29+OR+%28%28Muenke%5BTIAB%29+AND+%28craniosynostos*%5BTIAB%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

Catalog of Genes and Diseases from OMIM

- **MUENKE SYNDROME**
  http://omim.org/entry/602849

Medical Genetics Database from MedGen

- Muenke syndrome

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/22872262
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4101189/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18000976

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301588

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/26740388
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/20301628

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14963686

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/10696568

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