# Muenke syndrome

## Description

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 4 percent of all cases of craniosynostosis.

## Causes

A particular mutation in the *FGFR3* gene causes Muenke syndrome. The *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.

Learn more about the gene associated with Muenke syndrome

- FGFR3

### 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

- FGFR3-associated coronal synostosis
- Muenke nonsyndromic coronal craniosynostosis

## Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Muenke syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C1864436/)

Genetic and Rare Diseases Information Center

- Muenke syndrome (https://rarediseases.info.nih.gov/diseases/7097/index)

Patient Support and Advocacy Resources

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

Clinical Trials

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

Catalog of Genes and Diseases from OMIM

- MUENKE SYNDROME; MNKES (https://omim.org/entry/602849)

Scientific Articles on PubMed

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

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