

Crouzon Syndrome

Subjects: Genetics & Heredity

Contributor: Nicole Yin

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.

Keywords: genetic conditions

1. Introduction

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.

2. Frequency

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

3. 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.

3.1. The Gene Associated with Crouzon Syndrome

- FGFR2

4. 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.

5. 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
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