Condition: Craniosynostosis

Inheritance:

Most syndromes autosomal dominant.

Genetic etiology:

Genetically heterogeneous; many disorders due to mutation in various fibroblast growth factor receptor genes, including *FGFR1* (Pfeiffer syndrome), *FGFR2* (Apert, Crouzon, Beare-Stevenson, Pfeiffer , Jackson-Weiss syndromes), *FGFR3* (Crouzon, Muenke, isolated coronal synostosis syndromes).

Frequency:

Overall 1/2,000 – 1/2,500 live births.

Clinical features:

Craniosynostosis consists of premature fusion of one of more of the sagittal, coronal, lambdoidal, or metopic sutures of the skull. There is consequent abnormal skull shape, and, in many cases other anomalies, such as shallow orbits (Crouzon syndrome), syndactyly (Apert syndrome), etc.

Management:

Craniofacial surgery; supportive care.

Genetic counseling:

Most inherited as autosomal dominant with some cases due to new mutation; genetic testing available for many of the responsible genes.

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