Syndromic Craniosynostosis Etiology

In terms of syndromic craniosynostosis not only do Fibroblast growth factor receptor 3 and TWIST transcription factor feature, but also FGFR1 and in particular FGFR2, which has been reported in 90% of the syndromic craniosynostoses such as Apert, Crouzon, Peiffer and Jackson–Weiss.

The mutations can be divided into mutations that lead to gain of function (in FGFR genes) and mutations that lead to loss of function (in TWIST genes).