Does Commonly Occurring Mutations In Fgfr3 Gene Affect Maxillary Morphology?

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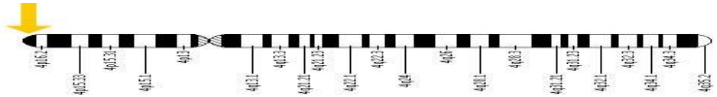
Introduction:

The fibroblast growth factors (FGFs) are a family of polypeptide growth factors involved in a variety of cellular and developmental responses in many different cell types and organisms.

Mutations in the fibroblast growth factor receptor (FGFR) genes 1, 2, and 3 are causal in a number of craniofacial dysostosis syndromes featuring cranio-synostosis with basi-cranial and mid-facial deformity

More than 97% of achondroplacia cases were reported to be caused by one of two mutations G1138A and G1138C, in axon 10 of the FGFR3.

Methodology: The genomic DNA of 30 subjects having Prognathic Maxilla, 30 individuals having Retrognathic Maxilla were compared with that of 30 having normal maxilla.



PCR Protocol:

- Genomic DNA extraction from serum using HiPura™ Forensic Sample Genomic DNA Purification Kit.
- Standardization of PCR conditions for FGFR3 Forward and Reverse primers-A programmable gradient Thermo cycler (Corbett)
- Agarose Gel Electrophoresis - to detect the PCR products., digested with MspI and SfcI (Thermo Scientific) in separate tubes and visualized under UV Trans-illuminator.

Results & Conclusions: None of the analyzed DNA samples of the selected subjects showed any presence of the targeted mutation sites. The targeted mutation sites, G1138A and G1138C are not associated with the maxillary growth of the selected subjects analyzed in the present study.

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