Mutations in the *FGFR2* gene in Mexican patients with Apert syndrome

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**ABSTRACT.** Apert syndrome (AS) is a frequentacrocephalosyndactyly, with autosomal dominant inheritance. AS has been associated with mutations in fibroblast growth factor receptor 2 (*FGFR2*), and approximately 99% of cases show 2 of the frequent mutations located in exon IIIa (Ser252Trp or Pro253Arg). The purpose of the present study was to describe the mutations in exon IIIa of *FGFR2* in Mexican AS patients and the relationships with clinical features. Exon IIIa of *FGFR2* from 6 AS patients was amplified by polymerase chain reaction. Mutations in exon IIIa of the *FGFR2* gene were identified by digestion with the restriction endonuclease Bstx1 and polyacrylamide gel electrophoresis. PCR fragments were cloned into the PCR 2.1 vector, and both DNA strands were sequenced using the T7 promoter and
M13 universal cloning region oligonucleotides. Sequence alignment was performed using the MEGA software version 5. The patients’ major clinical features included craniosynostosis, hypertelorism, proptosis, otitis media, midfacial hypoplasia, rhizomelic shortening, and hyperhidrosis. Mutation S252W was present in 4 patients, while the other 2 patients had P253R. In conclusion, either S252W or P253R mutations were present independently in AS patients; however, the 2 mutations were not found together. None of the clinical features were associated with any of the mutations, suggesting that other mutations may be involved in the development of this syndrome.

**Key words:** Apert syndrome; Craniosynostosis; FGFR2 mutations