

Clinical Medical Image

Title: Apert Syndrome: Molecularly Confirmed C.758C>G (P.Pro253Arg) in *FGFR2*

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Figure: A-G Complete picture of Apert syndrome.

Introduction

A 5-day-old girl was referred to our clinic for evaluation of congenital malformations. She was identified with a pathogenic mutation c.758C>G (p.Pro253Arg) in *FGFR2* gene using targeted exome sequencing. The de novo mutation was confirmed with Sanger sequencing in the patient and her parents. She showed occipital plagiocephaly with frontal bossing (Figure A and B). Skull frontal and lateral radiography revealed fusion of most of the sutures except coronal suture, with convolutional markings (Figure D and E). She had complete cleft palate (Figure C). Her fused bilateral hands showed type II syndactyly with complete syndactyly between the ring and the little fingers (Figure F1-F3). Both toes were simple syndactyly with side-to-side fusion of skin (Figure G1-G4).

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