Cruzon's Syndrome – A Case Report

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Abstract

Fgfr2 Gene Related Cruzon's Syndrome is a Rare Autosomal Dominant Syndrome Caused by Pathogenic Variants in The Fgfr2 Gene, Which Plays Important Role in Craniofacial Development by Regulating Cell Proliferation, Differentiation, And Ossification of Cranial Sutures During Embryogensis. We Report A Newborn Male Child Born to A Secondary Consanguinity with Ttnb Which on Examination Had Craniosynostosis, Exophthalmos, Brahyshape Skull, Dysmorphic Facial Features, B/L Choanal Atresia, Protruded Mandible, Sacral Dimple, Right Sided Umbilical Hernia. Whole Exome Sequencing Identified Pathogenic Heterozygous Fgfr2 Variant, Confirming Cruzon's Syndrome. Early Diagnosis Aids Management and Counselling.