Abstract:

Craniosynostoses are premature fusions of one or more cranial sutures. They affect all cranial sutures and are the main symptom of many genetic syndromes. Syndromes connected with craniosynostosis are serious disorders associated with skeleton abnormalities, limb malformations or mental disability. These syndroms are caused by different mutations in *FGFR1, FGFR2, FGFR3, TWIST1, EFNB1, RECQL4,* and *RAB23* genes. The aim of this review was to summarize contemporary knowledge of phenotype and genetic basis of these diseases.