

RAF1 variant in a patient with Noonan syndrome with multiple lentigines and craniosynostosis

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We report the case of a 14 years and 8 months girl, who is the first child of nonconsanguineous parents, with short stature, obstructive hypertrophic cardiomyopathy, multiple facial lentigines, high and wide forehead, downslanting palpebral fissures, low-set ears, short neck, and pectus excavatum; all features suggestive of Noonan syndrome with multiple lentigines (NSML). In addition, the patient exhibited craniosynostosis. Molecular analysis of rats sarcoma (RAS)/mitogen-activated protein kinase (MAPK) pathway genes with high-resolution melting curve analysis followed by sequencing showed a RAF1 amino acid substitution of valine to glycine at position 263 (p.V263G). The present report provides clinical data regarding the first association of a RAF1 variant and craniosynostosis in a patient with clinical diagnosis of NSML.