Craniofrontonasal syndrome

Craniofrontonasal syndrome (CFNS) is characterized by asymmetric facial features with hypertelorism and a broad bifid nose due to synostosis of the coronal suture. CFNS shows a unique X-linked inheritance pattern (most affected patients are female and obligate male carriers exhibit a mild manifestation or no typical features at all) associated with the ephrin-B1 gene (EFNB1) located in the Xq13.1 region.

In a study, we performed targeted, massively parallel sequencing using a next-generation sequencer, and identified a novel EFNB1 mutation, c.270_271delCA, in a Japanese female patient with craniosynostosis. Because subsequent Sanger sequencing identified no mutation in either parent, this mutation was determined to be de novo in origin. After obtaining molecular diagnosis, a retrospective clinical evaluation confirmed the clinical diagnosis of CFNS in this patient. Comprehensive molecular diagnosis using a next-generation sequencer would be beneficial for early diagnosis of the patients with undiagnosed craniosynostosis [1)]

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Yamamoto T, Igarashi N, Shimojima K, Sangu N, Sakamoto Y, Shimoji K, Niijima S. Use of targeted next-generation sequencing for molecular diagnosis of craniosynostosis: identification of a novel de novo mutation of EFNB1. Congenit Anom (Kyoto). 2015 Jul 24. doi: 10.1111/cga.12123. [Epub ahead of print] PubMed PMID: 26208246.