

Among 101 infants tested in the Department of Pediatric Neurosurgery, French Referral Center for Craniosynostosis, Hôpital Femme Mère-Enfant Hospices Civils de [Lyon](#), University of Lyon, Department of Genetics, Lyon University Hospitals, INSERM U1028, CNRS UMR5292, Centre de Recherche en Neurosciences de Lyon, Department of Pediatric Cranio-Maxillo-Facial Surgery, Hôpital Femme Mère-Enfant, Université Claude Bernard Lyon 1, Lyon, and Department of Genetics, Robert Debré Hospital, Inserm 1132, Université de Paris Cité, Paris, France, 13 carried a total of 13 variants; that is, 12.9% of the infants carried a variant in genes known to be involved in craniosynostosis. Seven infants carried SMAD6 variants, 2 in [FGFR2](#), 1 in [TWIST1](#), one in [FREM1](#), one in [ALX4](#), and one in [TCF12](#). All variants were detected at the heterozygous level in genes associated with autosomal dominant craniosynostosis. Also, neurodevelopmental testing showed especially delayed acquisition of [language](#) in children with than without variants in [SMAD6](#). In conclusion, a high percentage of young children with isolated midline craniosynostosis, especially in isolated [trigonocephaly](#), carried [SMAD6](#) variants. The interpretation of the pathogenicity of the genes must take into account incomplete penetrance, usually observed in craniosynostosis. The results highlight the interest in molecular analysis in the context of isolated sagittal and/or metopic craniosynostosis to enhance an understanding of the pathophysiology of midline [craniosynostosis](#) ((Di Rocco F, Rossi M, Verlut I, Szathmari A, Beuriat PA, Chatron N, Chauvel-Picard J, Mottolese C, Monin P, Vinchon M, Guernouche S, Collet C. Clinical interest of molecular study in cases of isolated midline craniosynostosis. Eur J Hum Genet. 2023 Feb 3. doi: 10.1038/s41431-023-01295-y. Epub ahead of print. PMID: 36732661.)

From:

<https://neurosurgerywiki.com/wiki/> - **Neurosurgery Wiki**

Permanent link:

<https://neurosurgerywiki.com/wiki/doku.php?id=tcf12>

Last update: **2024/06/07 03:00**

