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## Craniosynostosis is a feature of CHD7-related CHARGE Syndrome

CHARGE syndrome is a rare genetic disorder characterized by the presence of multiple malformations involving the craniofacial district, in particular eyes, ears, heart, and genitourinary tract in association with delayed growth and development. Its phenotypic spectrum is highly variable. The syndrome is caused in most cases by heterozygous variants in CHD7 and, more rarely, SEMA3E. Craniofacial alterations involving eyes, ears, nose, and mouth as well as the upper airways are typical of this condition, however, to date, craniosynostosis is not considered part of the clinical spectrum. We report a newborn affected by CHARGE syndrome confirmed by the identification of the de novo heterozygous c.6157C>T, p(Arg2053*) CHD7 variant, presenting with bi-coronal craniosynostosis and found two additional subjects in the literature with different craniosynostoses and distinct CHD7 alterations. We suggest the inclusion of CHD7-related CHARGE syndrome within the group of rare causes of syndromic craniosynostoses.

**Recent Publication**

1. Fortugno P, Monetta R, Cinquina V, Rigon C, Boaretto F, De Luca C, Zoppi N, Di Leandro L, De Domenico E, Di Daniele A, Ippoliti R, Angelucci F, Di Cesare E, De Paulis R, Salviati L, Colombi M, Brancati F, Ritelli M. Truncating variants in the penultimate exon of TGFBR1 escaping nonsense-mediated mRNA decay cause Loeys-Dietz syndrome. Eur J Hum Genet. 2023 Jan 4. doi: 10.1038/s41431-022-01279-4. Epub ahead of print. PMID: 36599937.

2. Maria Asif, Emrah Kaygusuz, Marwan Shinawi, Anna Nickelsen, Tzung-Chien Hsieh, Prerana Wagle, Birgit Budde, Jennifer Hochscherf, Uzma Abdullah, Stefan Höning, Christian Nienberg, Dirk Lindenblatt, Angelika A. Noegel, Janine Altmüller, Holger Thiele, Susanne Motameny, Nicole Fleischer, Idan Segal, Lynn Pais, Sigrid Tinschert, Nadra G. Samra, Juliann M. Savatt, Natasha L. Rudy, Chiara De Luca, Paola Fortugno, Susan M. White, Peter Krawitz, Anna C.E. Hurst, Karsten Niefind, Joachim Jose, Francesco Brancati, Peter Nürnberg, Muhammad Sajid Hussain. De novo variants of CSNK2B cause a new intellectual disability-craniodigital syndrome by disrupting the canonical Wnt signaling pathway. Human Genetics and Genomics Advances, 2022, 100111, ISSN 2666-2477, doi.org/10.1016/j.xhgg.2022.100111.

3. Fortugno P, Monetta R, Belli M, Botti E, Angelucci F, Palmerini MG, Annarita NS, De Luca C, Ceccarini M, Salvatore M, Bianchi L, Macioce P, Teson M, Ricci F, Network IUD, Macchiarelli G, Didona B, Costanzo A, Castiglia D, Brancati F. RIPK4 regulates cell-cell adhesion in epidermal development and homeostasis. Hum Mol Genet. 2022 Feb 26:ddac046. doi: 10.1093/hmg/ddac046. Epub ahead of pri`nt. PMID: 35220430.

**Biography**

Chiara De Luca is a Medical Geneticist currently attending her last year's Ph.D. course at the University of L'Aquila (Italy) on the genetic diagnosis of rare diseases. She has worked as a senior assistant at the Department of Human Genetics at KU Leuven, Belgium for six months. She works on the definition of the genetic bases of human genetic disorders, and she has 11 publications in the field.

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