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**GENETIC TEST:**  
**Craniosynostosis / Muenke syndrome (hot spot mutation - p.Pro250Arg)**

<b>FULL NAME:</b>	Craniosynostosis / Muenke syndrome (hot spot mutation - p.Pro250Arg)
<b>TEST TYPE:</b>	Clinical
<b>TEST SPECIALTY:</b>	Molecular Genetics
<b>TEST PURPOSE:</b>	Post-natal Diagnosis, Prenatal diagnosis
<b>SPECIMEN:</b>	Peripheral (whole) blood on EDTA, Chorionic villi, Amniotic fluid, DNA
<b>METHOD CATEGORY:</b>	Targeted variant analysis
<b>METHOD TECHNIQUE:</b>	Bi-directional Sanger Sequence analysis
<b>RIZIV CODE:</b>	565390-565401
<b>TURNAROUND TIME (MAXIMUM):</b>	21 days
<b>CREATED:</b>	21 Aug 2019 - 07:45
<b>CHANGED:</b>	09 Dec 2022 - 15:00

**URL:**

[https://www.chuliege.be/jcms/c\\_525327/fr/craniostenose](https://www.chuliege.be/jcms/c_525327/fr/craniostenose)

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## RELATED CONTENT

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### Related Diseases

- [Muenke syndrome](#)

### Related Laboratories

- [Centre de Génétique Humaine - CHU Sart-Tilman](#)

### Related Analytes

- [fibroblast growth factor receptor 3](#)

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