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**DISEASE:**  
**Congenital factor XII deficiency**

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| <b>NAME:</b>        | Congenital factor XII deficiency                                                                                                                                                                                                                                                                                                                                                 |
| <b>DESCRIPTION:</b> | A rare, autosomal recessive systemic dysfunction of the hemostatic pathway, that is due to a defect in the coagulation factor XII (FXII or Hageman factor), and is either asymptomatic or characterized by a prolonged activated partial thromboplastin time and an increased risk for thromboembolism. FXII deficiency is strongly associated with primary recurrent abortions. |
| <b>ORPHACODE:</b>   | 330                                                                                                                                                                                                                                                                                                                                                                              |
| <b>SYNONYMS:</b>    | Congenital Hageman factor deficiency                                                                                                                                                                                                                                                                                                                                             |
| <b>XREF(S):</b>     | <u>Orphanet</u><br><u>ICD-10</u><br><u>OMIM</u>                                                                                                                                                                                                                                                                                                                                  |
| <b>ANALYTE(S):</b>  | <u>F12</u>                                                                                                                                                                                                                                                                                                                                                                       |
| <b>CREATED:</b>     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                              |
| <b>CHANGED:</b>     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                              |

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## RELATED CONTENT

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### Related Genetic Tests

- [Trombosis - Hemostasis \(gene panel\)](#)

### Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

### Related Analytes

- [coagulation factor XII](#)

### Related Gene Panels

- [Trombosis - Hemostasis \(107 genes\) - KUL](#)

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