

## DISEASE:

### Severe combined immunodeficiency due to complete RAG1/2 deficiency

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| NAME:        | Severe combined immunodeficiency due to complete RAG1/2 deficiency                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| DESCRIPTION: | Severe combined immunodeficiency due to complete RAG1/2 deficiency is a rare, genetic T-B- severe combined immunodeficiency disorder due to null mutations in recombination activating gene (RAG) 1 and/or RAG2 resulting in less than 1% of wild type V(D)J recombination activity. Patients present with neonatal onset of life-threatening, severe, recurrent infections by opportunistic fungal, viral and bacterial micro-organisms, as well as skin rashes, chronic diarrhea, failure to thrive and fever. Immunologic observations include profound T- and B-cell lymphopenia, normal NK counts and low or absent serum immunoglobulins; some patients may have eosinophilia. |
| ORPHACODE:   | 331206                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| SYNONYMS:    | SCID due to complete RAG1/2 deficiency                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                               |
| XREF(S):     | <a href="#">Orphanet</a><br><a href="#">ICD-10</a><br><a href="#">OMIM</a>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |
| ANALYTE(S):  | <a href="#">RAG1</a><br><a href="#">RAG2</a>                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| CREATED:     | 13 May 2019 - 01:02                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |
| CHANGED:     | 22 Jun 2023 - 16:14                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                  |

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- [Ichthyosis \(gene panel\)](#)

### Related Laboratories

- [Centrum Menselijke Erfelijkheid - KUL](#)

### Related Analytes

- [recombination activating 1](#)
- [recombination activating 2](#)

### Related Gene Panels

- [Ichthyosis and erythroderma \(98 genes\) - KUL](#)

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