DISEASE:
Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency

NAME:	Mendelian susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency
DESCRIPTION:	Mendelian susceptibility to mycobacterial diseases (MSMD) due to complete interleukin-12 receptor subunit beta-1 (IL12RB1) deficiency is a genetic variant of MSMD (see this term) characterized by mild bacillus Calmette-Guérin (BCG) infections and recurrent Salmonella infections.
ORPHACODE:	319552
SYNONYMS:	MSMD due to complete IL12RB1 deficiency MSMD due to complete interleukin 12 receptor beta 1 deficiency Mendelian susceptibility to interleukin 12 receptor beta 1 deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	IL12RB1
CREATED:	13 May 2019 - 01:02
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RELATED CONTENT

Related Genetic Tests

- Immunodeficiency 30 / Susceptibility to mycobacterial diseases due to complete IL12RB1 deficiency
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- interleukin 12 receptor subunit beta 1

Related Gene Panels

- Immunogenetics (21 genes)
- Primary immune deficiencies (444 genes) - KUL