

DISEASE:
Omenn syndrome

NAME:	Omenn syndrome
DESCRIPTION:	Omenn syndrome (OS) is an inflammatory condition characterized by erythroderma, desquamation, alopecia, chronic diarrhea, failure to thrive, lymphadenopathy, and hepatosplenomegaly, associated with severe combined immunodeficiency (SCID; see this term).
ORPHACODE:	39041
SYNOMYS:	Combined immunodeficiency with hypereosinophilia
XREF(S):	Orphanet MedDRA ICD-10 OMIM

ANALYTE(S):	ADA <u>RAG1</u> <u>RAG2</u> <u>CHD7</u> <u>DCLRE1C</u> <u>IL2RG</u> <u>LIG4</u> <u>RMRP</u> <u>IL7R</u>
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

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- [interleukin 7 receptor](#)
- [DNA ligase 4](#)
- [recombination activating 1](#)
- [recombination activating 2](#)
- [RNA component of mitochondrial RNA processing endoribonuclease](#)

Related Gene Panels

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 - Hypogonadotropic hypogonadism (33 genes) - VUB
 - Ichthyosis and erythroderma (98 genes) - KUL
 - Primary immune deficiencies (444 genes) - KUL
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