DISEASE:
Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency

NAME:	Autoimmune lymphoproliferative syndrome due to CTLA4 haploinsuffiency
DESCRIPTION:	A rare, primary immunodeficiency characterized by variable combination of enteropathy, hypogammaglobulinemia, recurrent respiratory infections, granulomatous lymphocytic interstitial lung disease, lymphocytic infiltration of non-lymphoid organs (intestine, lung, brain, bone marrow, kidney), autoimmune thrombocytopenia or neutropenia, autoimmune hemolytic anemia and lymphadenopathy.
ORPHACODE:	436159
SYNONYMS:	ALPS due to CTLA4 haploinsuffiency CHAI CTLA-4 haploinsufficiency with autoimmune infiltration disease
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CTLA4
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RELATED CONTENT

Related Genetic Tests

- Autoimmune lymphoproliferative syndrome, type V
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- cytotoxic T-lymphocyte associated protein 4

Related Gene Panels

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