DISEASE:
Autosomal dominant hyper-IgE syndrome

NAME:	Autosomal dominant hyper-IgE syndrome
DESCRIPTION:	A very rare primary immunodeficiency disorder characterized by the clinical triad of high serum IgE (>2000 IU/ml), recurring staphylococcal skin abscesses, and recurrent pneumonia with formation of pneumatoceles.
ORPHACODE:	2314
SYNONYMS:	AD-HIES Autosomal dominant HIES Autosomal dominant hyperimmunoglobulin E syndrome Buckley syndrome Hyperimmunoglobulin E syndrome type 1 Hyperimmunoglobulin E-recurrent infection syndrome Job syndrome STAT3 deficiency
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	IL6ST STAT3
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RELATED CONTENT

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- Autoimmune disease, multisystem, infantile-onset (ADMIO) / Hyper-IgE recurrent infection syndrome
- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- interleukin 6 cytokine family signal transducer
- signal transducer and activator of transcription 3

Related Gene Panels

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