

ϵ isotype deficiency

GENERAL INFORMATION

Description:

Immunoglobulins of the IgE isotype are responsible for the immediate hypersensitivity reactions in diseases like hay fever, allergic asthma, and anaphalaxis. There are two forms of IgE, one which is secreted and one which is on the membrane lymphocytes.

Alternative names:

- IgE deficiency

Classification:

- Deficiencies predominantly affecting antibody production
 - Selective deficiency of IgG subclass, IgE and/or IgA class or subclass

Inheritance:

OMIM:

- *147180 Immunoglobulin: heavy epsilon chain; IGHE

Cross references:

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

IgE deficiency is associated with higher incidence of autoimmune diseases and nonallergic reactive airway disease.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Hypogammaglobulinemia, eMedicine

Therapeutic options:

- Only symptomatic patients should be treated. Antibiotic therapy in case there are recurrent infections followed by (intravenous) Ig if infections are not controlled.
- Hypogammaglobulinemia, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies
- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD), IMPAD

GENE INFORMATION

Names:

HUGO name: IGHE

Alias(es): Immunoglobulin heavy constant epsilon, immunoglobulin epsilon, Ig epsilon chain C region

Localization:

Reference sequences:

DNA: L00022 (EMBL) , **Protein:** P01854 (SWISSPROT)

Chromosomal Location:

14q32.33

Maps:

IGHE (Map View)

Other gene-based resources:

Ensemble: ENSG00000177154, GENATLAS: IGHE, GeneCard: IGHE, Entrez Gene: 3497, euGenes: 3497, GDB: 119335

PROTEIN INFORMATION

Description:

Structures (PDB):

1IGE IGHE

Other features:

Disulfide bond interchain (with a light chain):

14

Disulfide bond interchain (with a heavy chain): 121

Disulfide bond interchain (with a heavy chain): 209

Disulfide bonds: 15-105, 29-85, 135-193, 239-299, 345-405

Other related resources:

PIR: EHHU, InterPro: IPR003006; Ig_MHC, InterPro: IPR003597; Ig_c1, Pfam: PF00047; ig, SMART: SM00407; IGc1, PROSITE: PS00290; IG_MHC

Expression pattern for human:

Tissue	Exp. (%)	Clones
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Animal models:

Mouse:

MGD: ; Igh-7

Fly:

euGenes: ; Toll-7

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- March of Dimes Birth Defects Foundation
- NIH/National Institute of Allergy and Infectious Diseases
- European Society for Immunodeficiencies