λ5 surrogate light-chain deficiency

GENERAL INFORMATION

Description:

Mutations that impair early B cell development result in profound antibody deficiency, characterized by a paucity of mature B cells and the early onset of recurrent pyogenic infections. Defects in λ5 can cause a block in B cell development at the transition between the pro-B cell and the pre-B cell stage.

Classification:

- Deficiencies predominantly affecting antibody production
  - Agammaglobulinemia

Inheritance:

Autosomal recessive

OMIM:

- *146770 Immunoglobulin lambda-like polypeptide 1; IGLL1

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for X-linked agammaglobulinemia
- IDR factfile for X-linked hypogammaglobulinemia with growth hormone deficiency
- IDR factfile for BLNK deficiency
- IDR factfile for Igα deficiency
- IDR factfile for μ heavy-chain deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients with mutation in #5 have recurrent otitis, hypoglammaglobulinemia and absent B cells. Patients can develop hemophilus meningitis complicated by arthritis.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Agammaglobulinemia, eMedicine

Therapeutic options:

- (Intravenous) immunoglobulins and antibiotic therapy. Oral poliovaccine should not be given because of the risk of paralytic disease.
- Hypogammaglobulinemia, eMedicine

Research programs, clinical trials:

- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD), IMPAD
- European Initiative for Primary Immunodeficiencies
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: IGLL1
Alias(es): IGL1, IGL5, IGLL, IGO, IGVPB, immunoglobulin lambda-like polypeptide 1, pre-B-cell specific, Immunoglobulin lambda-like polypeptide 1 precursor , Immunoglobulin-related 14.1 protein, Immunoglobulin omega polypeptide

Localization:

Reference sequences:

DNA: IGLL1_DNA (IDRefSeq) , **cDNA:** M27749 (EMBL) , **Protein:** P15814 (SWISSPROT)

Chromosomal Location:

22q11.22

Maps:

IGLL1 (Map View)

Variations / Mutations:

- IGLL1base; Mutation registry for #5 surrogate light-chain deficiency

Other gene-based resources:

Ensembl: ENSG00000128322, GENATLAS: IGLL1, GeneCard: IGLL1, UniGene: 348935, Entrez Gene: 3543, euGenes: 3543

PROTEIN INFORMATION

Description:

Subunit:

Associates non-covalently with vpreb1

Domains:

J region (by similarity to lambda light-chain) domain: 97-108

C region (by similarity to lambda light-chain) domain: 109-213

Other features:

Signal peptide: 1-37

Immunoglobulin lambda-like polypeptide 1: 38-213

Other related resourses:

PIR: A33911, 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
colonic mucosa with ulcerative colitus	37.51	1:1218
cord blood	17.67	3:7759
bone marrow	16.11	7:19854
thymus, pooled	14.42	1:3169
leukopheresis	10.03	1:4557
testis	2.64	5:86533
heart	1.24	1:36928
kidney	0.39	1:117548

Animal models:

Mouse:

MGD: ; Igl-5

C. elegans:

euGenes: ; Y106G6D.5

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