γ4 isotype deficiency

GENERAL INFORMATION

Description:

Immunoglobulin G sublass deficiency is defined as a decrease of an IgG sublass greater than 2 standard deviations below the normal mean for age. One or more IgG subclasses and other Ig isotypes may be involved. This deficiency may be isolated or associated with other immunodeficiencies (IgA deficiency, ataxia-telangiectasia).

Classification:

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

Inheritance:

Autosomal recessive

OMIM:

- *147130 Immunoglobulin Gm4; IGHG4

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for γ1 isotype deficiency
- IDR factfile for γ2 isotype deficiency
- IDR factfile for partial γ3 isotype deficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

IgG sublass deficiency is clinically significant only if an impaired response to bacteria such as tetanus, diphteria, and pneumococcus occurs. IgG4 subclass deficiency is very common, affecting 10-15% of the general population. It may be isolated or associated with other IgG subclass deficiencies. The clinical significance of this disorder is still unknown.

Diagnosis:

Additional Information:

- IgA and IgG subclass deficiencies, eMedicine
- Hypogammaglobulinemia, eMedicine
- Agammaglobulinemias, Primary, Health Library

Diagnostic laboratories:

Clinical:

- IgA and IgG subclass deficiencies, eMedicine

Therapeutic options:

- Only symptomatics patients should be treated. Antibiotic therapy in case there are recurrent infections followed by intravenous immunoglobulin if infections are not controlled.
- Hypogammaglobulinemia, eMedicine
- IgA and IgG subclass deficiencies, 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
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: IGHG4
Alias(es): Immunoglobin heavy constant gamma 4, G4m marker, immunoglobulin gamma 4, Gm marker, Ig gamma-4 chain C region

Localization:

Reference sequences:

DNA: K01316 (GenBank) , **cDNA:** BC025985 (GenBank) , **Protein:** P01861 (SWISSPROT)

Chromosomal Location:

14q32.33

Maps:

IGHG4 (Map View)

Other gene-based resources:

Ensembl: OTTHUMG00000029942, GENATLAS: IGHG4, GeneCard: IGHG4, UniGene: 534324, Entrez Gene: 3503, euGenes: 3503, GDB: 119340, IMGT: IGHG4

PROTEIN INFORMATION

Description:

Domains:

Ch1 domain: 1-98

Hinge domain: 99-110

Ch2 domain: 111-220

Ch3 domain: 221-327

Other features:

Disulfide bond interchain (with a light chain): 14

Disulfide bond interchain (with a heavy chain): 106

Disulfide bond interchain (with a heavy chain): 109

Disulfide bonds: 27-83, 141-201, 247-305
Other related resourses:

PIR: G4HU, InterPro: IPR003006; Ig_MHC, InterPro: IPR003597; Ig_c1, Pfam: PF00047; ig, SMART: SM00410; IG_like, SMART: SM00407; IGc1, PROSITE: PS00290; IG_MHC

Expression pattern for human:

Tissue	Exp. (%)	Clones

Animal models:

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