γ2 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:

- *147110 Immunoglobulin Gm2; IGHG2

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for γ1 isotype deficiency
- IDR factfile for partial γ3 isotype deficiency
- IDR factfile for γ4 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. Antipneumococcal antibodies for IgG2 should be checked if the titers for total Igs are normal and the patient is unable to produce antibodies to specific antigens. IgG2 deficiency may be associated with an inability to have a response to polysaccharides, leading to recurrent sinopulmonary infections with encapsulated bacteria such as H. influenza and S. pneumoniae. Asymptomatic cases of IgG2 deficiency have been reported, and some patients without IgG2 will not respond to polysaccharide antigens.

Diagnosis:

Diagnostic laboratories:

Clinical:

- IgA and IgG subclass deficiencies, 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.
- Immunoglobulin G deficiency, eMedicine
- 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: IGHG2
Alias(es): Immunoglobin heavy constant gamma 2 (G2m marker), immunoglobulin gamma 2 (Gm marker), Ig gamma-2 chain C region

Localization:

Reference sequences:

DNA: J00230 (GenBank) , **cDNA:** AJ294731 (GenBank) , **Protein:** P01859 (SWISSPROT) Other Sequences

Chromosomal Location:

14q32.33

Maps:

IGHG2 (Map View)

Other gene-based resources:

Ensembl: OTTHUMG0000002994, GENATLAS: IGHG2, GeneCard: IGHG2, UniGene: 510635, Entrez Gene: 3501, euGenes: 3501, GDB: 119338, IMGT: IGHG2

PROTEIN INFORMATION

Description:

Domains:

Ch1 domain: 1-98

Hinge domain: 99-110

Ch2 domain: 111-219

Ch3 domain: 220-326

Other features:

Disulfide bond interchain (with a light chain): 14

Disulfide bond interchain (with a heavy chain): 102

Disulfide bond interchain (with a heavy chain): 103

Disulfide bond interchain (with a heavy chain): 106

Disulfide bond interchain (with a heavy chain): 109

Disulfide bonds: 27-83, 140-200, 246-304
Other related resourses:

PIR: G2HU, 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:

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