Partial γ3 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:

- *147120 Immunoglobulin Gm3; IGHG3

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for γ1 isotype deficiency
- IDR factfile for γ2 isotype deficiency
- IDR factfile for partial 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. IgG3 deficiency can be associated with recurrent sinopulmonary infections with viruses and M. catarrhalis, asthma and sinusitis.

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 symptomatic patients should be treated. Antibiotic therapy in case there are recurrent infections followed by (intravenous) Ig if infections are not controlled. (Intravenous) Ig has benefits in asthma due to IgG3 deficiency and in chronic sinusitis.
- 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: IGHG3
Alias(es): Immunoglobin heavy constant gamma 3, Immunoglobulin gamma 3, Ig gamma-3 chain C region, Heavy chain disease protein, HDC

Localization:

Reference sequences:

DNA: D78345 (GenBank) , **cDNA:** J00231 (EMBL) , **Protein:** P01860 (SWISSPROT)

Chromosomal Location:

14q32.33

Maps:

IGHG3 (Map View)

Other gene-based resources:

Ensembl: ENSG00000130076, GENATLAS: IGHG3, GeneCard: IGHG3, UniGene: 525646, Entrez Gene: 3502, euGenes: 3502, GDB: 119339

PROTEIN INFORMATION

Description:

Subunit:

Dimer linked by 12 disulfide bonds; it has an extra interchain disulfide bond at position 7 in addition to the 11 normally present in the hinge region.

Domains:

Hinge domain: 12-73

Ch2 domain: 74-183

Ch3 domain: 184-289

Other features:

Other related resourses:

PIR: G3HUWI, 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
breast, normal duct	33.09	76:346
ovary, surface epithelium	15.86	4:38
prostate, stroma	9.02	17:284
lymph node	8.44	7:125
spleen	5.73	275:7229
adipose	3.37	19:849
thymus	3.35	10:449
B-cells	2.36	259:16533
nasopharynx	2.10	9:646
genitourinary tract	1.94	49:3813

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