α2 isotype deficiency

GENERAL INFORMATION

Description:

Classification:

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

Inheritance:

Autosomal recessive

OMIM:

- *147000 IgA constant heavy chain 2; IGHA2

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for α1 isotype deficiency

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Most patients are healthy. Some patients develop symptoms later in life. Recurrent or chronic upper and lower respiratory tract infections leading to bronchiectasis or cor pulmonale are not common. G. lamblia infection of the gastrointestinal tract is common. Other gastrointestinal diseases, such as spruelike syndrome, ulcerative colitis, and Crohn disease may occur. Autoimmune and collagen vascular diseases such as rheumatoid arthritis, systemic lupus erythematosus without renal disease, autoimmune hepatitis, hemolytic anemia, and endocrinopathies have been described. Patients with undetectable IgA antibodies may develop anti-IgA antibodies of the IgE isotype after receiving blood products. Once sensitized, these patients are at risk for anaphylactic reactions if they receive blood products containing even small amounts of IgA antibodies. Increased excretion of monomeric IgM in the secretions of certain patients may compensate for the lack of IgA, rendering patients less vulnerable to mucosal infections. The association of an IgG subclass deficiency, such as IgG2/IgG4 or IgG3, may worsen the situation.

Diagnosis:

Additional Information:

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

Diagnostic laboratories:

Clinical:

- Immunoglobulin A deficiency, eMedicine
- 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 A 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), IMPAD
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: IGHA2
Alias(es): Immunoglobin heavy constant alpha 2, A2m marker, Immunoglobulin alpha 2, A2M marker, Ig alpha-2 chain C region

Localization:

Reference sequences:

DNA: J00221 (EMBL) , **cDNA:** AL389978 (GenBank) , **Protein:** P01877 (SWISSPROT)

Chromosomal Location:

14q32.33

Maps:

IGHA2 (Map View)

Other gene-based resources:

Ensembl: OTTHUMG00000029920, GENATLAS: IGHA2, GeneCard: IGHA2, Entrez Gene: 3494, euGenes: 3494, IMGT: IGHA2

PROTEIN INFORMATION

Description:

Protein function:

Ig alpha is the major immunoglobulin class in body secretions. It may serve both to defend against local infection and to prevent access of foreign antigens to the general immunologic system

Subunit:

Monomeric or polymeric

Other features:

Disulfide bond interchain (with light chain): 101

Disulfide bond interchain (with heavy chain): 109

Disulfide bond interchain (with heavy chain): 169

Disulfide bond intersubunit bond: 179

Disulfide bond with j chain: 339

Disulfide bonds: 26-85, 110-167, 134-191, 237-300

Other related resourses:

PIR: A2HU, PIR: B22360, PIR: C22360, 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

Other information sources:

- Primary Agammaglobulinemia by The Doctors' Medical Library