IgA deficiency

GENERAL INFORMATION

Description:

Selective IgA deficiency is the most common primary immunodeficiencies. B-lymphocytes are present there but are unable to change into the IgA-producing plasma cells. Because all the other antibodies are normal or near-normal, the condition is called selective IgA deficiency. Haplotype analysis, linkage disequilibrium, and homozygosity mapping indicated that HLA-DQ/DR is the major IGAD1 locus, and suggest the autoimmune pathogenesis of IgAD/CVID. It has been associated with other chromosomal abnormalities involving chromosome 18.

Alternative names:

- IgAD
- Selective IgA deficiency
- Selective deficiency of immunoglobulin A
- Selective deficiency of Gamma-A-globulin

Classification:

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

Inheritance:

Complex

OMIM:

- %137100 Immunoglobulin a deficiency susceptibility 1; IGAD1

Incidence:

Incidence varies between countries and racial group. Between 1: 400-800 individuals will be affected.

CLINICAL INFORMATION

Description:

Many individuals with IgA deficiency are asymptomatic. Patients with IgA deficiency have an increased incidence of upper respiratory tract infections, allergies and autoimmune disease like SLE, rheumatoid arthritis, and juvenile chronic arthritis. Others have persistent or recurrent infections and some develop CVI over time. Also, there is a possibility of transfusion reaction due to anti-IgA antibodies. Malignancy may be increased.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Immunoglobulin A deficiency, eMedicine

Therapeutic options:

- Treatment is directed at the presenting disease. Antibiotic therapy (10-14 days) together with physiotherapy and postural drainage in case of lung damage. Intravenous immunoglobulins (200-600 mg/kg/month at intervals of 2-3 weeks) with a low IgA content in case it is needed. It should be avoid IgA-containing products. Associated autoimmune diseases should be treated conventionally.
- 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
- The Genetics of IgA Deficiency and Common Variable Immune Deficiency, Comprehensive Cancer Center
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name: IGAD1
Alias(es): immunoglobulin A (IgA) deficiency susceptibility 1

Localization:

Chromosomal Location:

6p21.3

Maps:

IGAD1 (Map View)

Other gene-based resources:

GeneCard: IGAD1, Entrez Gene: 10986, euGenes: 10986, GDB: 6929077

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

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