Common variable immunodeficiency of unknown origin

GENERAL INFORMATION

Description:

Common variable immunodeficiency is a heterogeneous group of diseases of unknown etiology. Patients have low serum immunoglobulin concentrations, defective specific antibody production and an increased susceptibility to bacterial infections of the respiratory and and gastrointestinal tracts. Most CVID patients have normal numbers of circulating T cells and surface immunoglobulin-positive B cells. CVID may present at any age from childhood to old age. The peak of presentation is in early childhood and early adulthood.

Alternative names:

- CVID
- common variable hypogammaglobulinemia
- hypogammaglobulinemia
- adult-onset agammaglobulinemia
- late-onset hypogammaglobulinemia
- acquired agammaglobulinemia
- immunoglobulin deficiency, late-onset

Classification:

- Deficiencies predominantly affecting antibody production
  - Common variable immunodeficiency

Inheritance:

Complex

OMIM:

- %240500 Common variable immunodeficiency

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for IgA deficiency

Incidence:

1: 50,000

CLINICAL INFORMATION

Description:

Patients with CVID present recurrent bacterial infection, autoimmune disease like thrombocytopenia, haemolytic anaemia and organ specific autoimmunity. Nodular lymphoid hyperplasia of bowel is unique to CVID. In severe form of CVID, granulomatous disease with lymphadenopathy is common. The major complications of CVID are due to delay in diagnosis. Patients may have bronchiectasis, chronic sinusitis, unusual infections, such as Campylobacter cholangitis, and Mycoplasma/ Ureaplasma arthritis. There is an increase risk of lymphoma and gastric carcinoma. Thymomas (benign or malignant) can also occur and often give rise to myasthenia gravis and haematological problems.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Common Variable Immunodeficiency (CVID), eMedicine

Therapeutic options:

- (Intravenous) immunoglobulins and antibiotic therapy together with physiotherapy and postural drainage in case of lung damage. Oral poliovaccine should not be given because there is risk of paralytic disease. The granulomatous disease responds well to steroids. Splenectomy may be necessary for hyperplenism.
- Common Variable Immunodeficiency (CVID), eMedicine
- Hypogammaglobulinemia, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies
- Immune System and Gut Abnormalities in Patients with Common Variable Immunodeficiency with and without Gastrointestinal Symptoms, ClinicalTrials.gov
- 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 Cente
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

GENE INFORMATION

Names:

HUGO name:

Localization:

Maps:

(Map View)

Other gene-based resources:

Ensembl: , GENATLAS: , GeneCard: , UniGene: , LocusLink: , euGenes: , GDB:

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones

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