Antibody deficiency with normal immunoglobulin levels

GENERAL INFORMATION

Description:

The cause of the syndrome is unknown. It is related to IgG subclass deficiencies and there is a failure to respond to specific antigens. This entity, observed more in adults, clinically resembles common variable immunodeficiency (CVI). In small children, it is due to a maturational delay that resolves spontaneously.

Alternative names:

- Antibody deficiency with normal or elevated serum immunoglobulin levels
- Selective antibody deficiency with normal immunoglobulins

Classification:

- Deficiencies predominantly affecting antibody production
  - Other antibody deficiencies

Inheritance:

Unknown

OMIM:

- %240500 Common variable immunodeficiency

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Common variable immunodeficiency

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have a history of recurrent typical infections with normal and IgG subclasses and failure to respond to specific antigens (test immunization). Many patients are clinically normal, some have recurrent bacterial sinopulmonary infections (Haemophilus, Pneumococcus, Moraxella), complicated by chronic lung disease and bronchiectasis. Serum immunoglobulin levels are normal or elevated. There are low specific antibodies, to capsulated organisms, and poor responses to test immunization, and to polysaccharide antigens.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Common Variable Immunodeficiency (CVID), eMedicine

Therapeutic options:

- (Intravenous) immunoglobulins. Antibiotic therapy together with physiotherapy and postural drainage in case of lung damage. Ciprofloxacin has not been licensed for small children. Oral poliovaccine should not be given because there is a risk of paralytic disease.
- Common Variable Immunodeficiency (CVID), 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, University of Albama at Birmingham
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

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

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