Selective deficiency in Ig class-switch recombination

GENERAL INFORMATION

Description:

Hyper-IgM syndrome is characterised by normal or elevated serum IgM levels associated with low or absent IgG, IgA, IgE levels and impaired Ig class-switch recombination (CSR). HIGM4 is a new entity of HIGM with the clinical phenotype similar with AID deficiency (HIGM2).

Alternative names:

- HIGM4
- Hyper-IgM syndrome type 4

Classification:

- Deficiencies predominantly affecting antibody production
  - Defects of class-switch recombination and somatic hypermutation (Hyper-IgM syndromes) affecting B cells

Inheritance:

Autosomal recessive

OMIM:

- *608184 Immunodeficiency with hyper-IgM, type 4

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for AID deficiency
- IDR factfile for X-linked hyper-IgM syndrome (CD40L deficiency)

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have recurrent bacterial infections from childhood and do not suffer from opportunistic infections. Lymphoid organ hyperplasia (spleen, liver, tonsil, lymph nodes) and autoimmune features, especially cytopenias.

Diagnosis:

Diagnostic laboratories:

Clinical:

- ORPHANET

Genetic:

- IDdiagnostics

Therapeutic options:

- (Intravenous) immunoglobulins started early to achieve residual IgG level>8g/l. This treatment leads to a decreased number of infections and diminishes or normalizes IgM levels. The lymphoid hyperplasia is not influenced by treatment. In case of enlarged lymphadenopathies there is need for surgical resection or biopsy.
- Hypogamaglobulinemia, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name:

Localization:

Chromosomal Location:

Other gene-based resources:

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

Other information sources:

- Immunodeficiencies