X-linked hypogammaglobulinemia with growth hormone deficiency

GENERAL INFORMATION

Description:

The clinical syndrome of X-linked hypoglammaglobulinemia and isolated growth hormone deficiency (XLA/GHD) is characterized by reduced serum of Ig and normal cell-mediated immunity. Linkage analysis demonstrated the most likely location of the gene (or genes) to be the midportion of Xq between DXS3 and DXS94 that includes the gene for XLA.

Alternative names:

- Agammaglobulinemia and isolated growth hormone deficiency, X-linked
- Fleisher syndrome
- Isolated growth hormone deficiency, type III
- IGHD III
- Growth hormone deficiency with hypogammaglobulinemia

Classification:

- Deficiencies predominantly affecting antibody production
  - Agammaglobulinemia

Inheritance:

X-linked

OMIM:

- #307200 Hypogammaglobulinemia and isolated growth hormone deficiency, X-linked

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for X-linked agammaglobulinemia

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients with X-linked hypoglammaglobulinemia and isolated growth hormone deficiency have recurrent sinopulmonary infections, short stature and retarded bone age during childhood. The adults have delayed onset of puberty. The immunodeficiency is characterized by absent specific antibody production in vivo and impaired immunoglobulin production in vitro. Patients have deficient growth hormone responses to insulin and arginine or levodopa.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Agammaglobulinemia, eMedicine
- Agammaglobulinemia X-linked, ORPHANET

Therapeutic options:

- (Intravenous) immunoglobulins, antibiotic therapy together with physiotherapy and postural drainage in case of lung damage. Oral poliovaccine should not be given because of the risk of paralytic disease.
- Agammaglobulinemia, eMedicine
- Hypogammaglobulinemia, eMedicine

Research programs, clinical trials:

- Improved Healthcare for Patients with Primary Antibody Deficiencies through new Strategies Elucidating their Pathophysiology (IMPAD), IMPAD
- European Initiative for Primary Immunodeficiencies
- Immune Regulation in Patients with Common Variable Immunodeficiency and Related Syndromes, ClinicalTrials.gov

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