Cartilage-hair hypoplasia

GENERAL INFORMATION

Description:

McKusick Type Metaphyseal Chondrodysplasia is a rare progressive inherited disorder which include neutropenia, together with T-and B-cell lymphopenia. The disease is inherited as an autosomal recessive trait, the gene being located on chromosome 9. CHH occurs particularly in Finland and older Amish in America.

Alternative names:

- CHH
- Metaphyseal chondrodysplasia, McKusick type

Classification:

- Other well-defined immunodeficiency syndromes

Inheritance:

Autosomal recessive

OMIM:

- #250250 Cartilage-hair hypoplasia; CHH
- *157660 Mitochondrial RNA-processing endoribonuclease, RNA component of; RMRP

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Cartilage hair hypoplasia affects bone metaphyses causing small stature from birth. It is associated with fine, slow-growing hair, and sometimes immune deficiencies. Other symptoms include short hands and possibly short, deformed limbs (varus). X-rays evidence metaphyseal lesions, especially in the knees, and large, round epiphyses during childhood. The outcome of the disorder depends on immune deficiency and its possible association with Hirschprung's disease. Short stature is common and has very early onset, while immune deficiencies are absent in some forms.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Cartilage-hair hypoplasia, eMedicine
- Metaphyseal chondrodysplasia, recessive type, ORPHANET

Genetic:

- RMRP, IDdiagnostics
- Laboratory of Metabolism and Molecular Pediatrics - University Children's Hospital(Zurich), EDDNAL
- Laboratory of Molecular Genetics - HUCH (Helsinki), EDDNAL

Therapeutic options:

- CHH patients usually have infection problems, especially with varicella, which may be treated with acyclovir. Live viral vaccines should be avoided, such as oral polio and measles, mumps, and rubella (MMR). Other infections are treated with appropriate antimicrobials. Also palliative bone reconstruction procedures may be performed in patients with CHH, in case is needed. IVIG of 400-600 mg/kg/mo or a dose that maintains trough serum IgG levels greater than 500 mg/dL is desirable.
- Cartilage-hair hypoplasia, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies

GENE INFORMATION

Names:

HUGO name: RMRP
Alias(es): CHH, HGNC:1931, RMRPR, RNA component of mitochondrial RNA processing endoribonuclease, cartilage-hair hypoplasia

Localization:

Chromosomal Location:

9p21-p12

Maps:

RMRP (Map View)

Other gene-based resources:

GENATLAS: RMRP, GeneCard: RMRP, Entrez Gene: 6023, euGenes: 6023

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
- European Society for Immunodeficiencies
- NIH/National Institute of Allergy and Infectious Diseases