# Regulatory factor X-associated protein deficiency

## GENERAL INFORMATION

### Description:

Defects in RFXAP are a cause of hereditary MHC class II deficiency, also known as Bare Lymphocyte Syndrome (BLS) or HLA class II-deficient combined immunodeficiency). RFXAP is linked with BLS complementation group D. The disease is very rare; only approximately 70 patients from 57 unrelated families have been reported worldwide.

### Alternative names:

- RFXAP deficiency
- Bare Lymphocyte Syndrome type 2 (BLS)
- RFXAP (complementation group D)

### Classification:

- Combined B and T cell immunodeficiencies
  - Major histocompatibility complex class II deficiency

### Inheritance:

Autosomal recessive

### OMIM:

- #209920 Bare lymphocyte syndrome, type II
- *601861 Regulatory factor X-associated protein; RFXAP

### Cross references:

### Incidence:

Incidence is not known yet.

## CLINICAL INFORMATION

### Description:

The children have extreme susceptibility to bacterial, viral, and fungal infections in the first year of life and usually results in death by age of four. It affects children born to cosanguineous families of nothern African or Mediterranean origin. Patients have liver diseases associated with chronic Cryptosporidium infection. Recurrent bronchopulmonary infections have been observed in all patients. The infectious agents include viruses (CMV, respiratory syncytial virus, enterovirus), bacteria (Steptococcus, Haemophilus, Proteus, Pseudomonas), Pneumocystis carinii, and Candida albicans. Neurological manifestation due to viral infections have been diagnosed in a number of patients. Hematologic manifestations are characterized by neutropenia and severe autoimmune cytopenia.

### Diagnosis:

### Diagnostic laboratories:

**Clinical:**

- Defective expression of HLA class 2, ORPHANET

## Therapeutic options:

- Patients can be cured with bone marrow transplantations of haematopoietic stem cells. Other recommendations include treatment of infections with antibacterials, antifungals, and antivirals, intravenous gamma-globulin infusion, irradiation of all blood products.
- Bone marrow transplant, UCSF Medical Center
- Stem Cell Transplant, National Marrow Donor Program (NMDP)

## Research programs, clinical trials:

- Pilot Study of Allogeneic Bone Marrow Transplantation Plus Cyclosporine and Mycophenolate Mofetil to Induce Mixed Hematopoietic Chimerism in Patients With Primary T-Cell Immunodeficiency Disorders, ClinicalTrial.gov
- Beckman Research Institute
- University of Califonia
- ORPHANET
- Leiden University Medical Center

# GENE INFORMATION

## Names:

**HUGO name:** RFXAP
**Alias(es):** regulatory factor X-associated protein, Regulatory factor X-associated protein (RFX DNA-binding complex 36 kDa subunit) (RFX-associated protein)

## Localization:

**Reference sequences:**

**DNA:** AL159973 (EMBL) , **cDNA:** Y12812 (EMBL) , **Protein:** O00287 (SWISSPROT)

**Chromosomal Location:**

13q14

**Maps:**

RFXAP (Map View)

## Variations / Mutations:

- ; RFXAPbase: Mutation registry for Regulatory factor X-associated protein deficiency

## Other gene-based resources:

Ensembl: ENSG00000133111, GENATLAS: RFXAP, GeneCard: RFXAP, UniGene: 24422, Entrez Gene: 5994, euGenes: 5994, GDB: 9475355

# PROTEIN INFORMATION

## Description:

**Protein function:**

Part of the RFX complex that binds to the x-box of MHC II promoters

**Subunit:**

RFX consists of at least 3 different subunits; RFXAP, RFX5 and RFX-B/RFXANK; with each subunit representing a separate complementation group. RFX forms cooperative DNA binding complexes with x2bp and cbf/nf-y. RFX associates with CIITA to form an active transcriptional complex

**Subcellular location:**

Nuclear

**Post-translational modification:**

Phosphorylated

## Domains:

**Poly-ala domain: 38-41**

**Poly-lys domain: 171-178**

**Nuclear localization signal domain: 163-178**

## Other features:

## Expression pattern for human:

| Tissue | Exp. (%) | Clones |
|---|---|---|
| hepatocellular carcinoma | 21.14 | 8:13916 |
| retina | 14.50 | 3:7607 |
| 2 pooled high-grade transitional cell tumors | 9.55 | 1:3850 |
| fibrosarcoma | 9.32 | 1:3946 |
| lymphoma, follicular mixed small and large cell | 7.89 | 2:9317 |
| testis | 7.06 | 1:5207 |
| from acute myelogenous leukemia | 6.14 | 1:5984 |
| cochlea | 4.05 | 1:9090 |
| placenta | 3.46 | 1:10629 |
| B-cell, chronic lymphotic leukemia | 3.20 | 1:11493 |

## Animal models:

**Fly:**

euGenes: ; Rfx

**C. elegans:**

euGenes: ; daf-19

## 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:

- Birth disorders information directory