UNC93B deficiency

GENERAL INFORMATION

Description:

Herpes simplex encephalitis (HSE) is a rare complication of infection with herpes simplex virus-1 (HSV-1), which infects an estimated 80% of young adults worldwide. HSE susceptibility may be inherited as a monogenic trait resulting in the specific impairment of immunity to HSV-1. This notion of pathogen-specific mendelian immunodeficiency contrasts with the dominant paradigm, in which rare single-gene lesions confer vulnerability to multiple infections, whereas more common infections in otherwise healthy patients reflect polygenic predisposition. Onset may occur at any age but is most common in adults. This disease, which affects only a small minority of HSV1-infected individuals, could result from a genetic predisposition.
A mutation in the UNC-93B gene, inducing impaired production of interferon, an anti-infectious factor necessary to fight the herpetic virus infection in nervous tissue, has been identifiedin two children and may be responsible for the disease. The disease course is severe, with a mortality rate of 80% and severe sequelae among surviving patients.

Alternative names:

- UNC93BD
- Autosomal recessive UNC-93B deficiency
- HSV encephalitis, Herpes simplex encephalitis, Herpes simplex neuroinvasion, Herpetic encephalopathy Herpetic encephalopathy, idiopathic

Classification:

- Defects of innate immune system, receptors and signaling components

Inheritance:

Autosomal recessive

OMIM:

- #610551 Herpes simplex encephalitis, UNC93B-deficient
- *608204 Unc93, c. Elegans, homolog of, b1; UNC93B1

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for IFNγ1-receptor deficiency
- IDR factfile for IFNγ2-receptor deficiency
- IDR factfile for Interleukin-12 p40 deficiency
- IDR factfile for Interleukin-12 receptor beta 1 deficiency
- IDR factfile for STAT1 deficiency
- IDR factfile for TLR1 deficiency

Incidence:

The annual incidence varies between 1 in 250 000 and 1 in 500 000.

CLINICAL INFORMATION

Description:

Patients presents acute necrosing temporal encephalitis, after a primary or recurrent infection. Onset is rapid (less than 48 hours) with a fever of 40 C, headaches, and behavioural, language and memory problems. These initial manifestations are followed by numbness and coma, which may be accompanied by convulsions and paralysis.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Herpetic encephalopathy, ORPHANET, France
- Herpes simplex encephalitis, eMedicine, USA

Genetic:

- IDdiagnostics

Therapeutic options:

- Emergency treatment should involve intravenous administration of acyclovir, as soon as the diagnosis is suspected
- Herpes simplex encephalitis, eMedicine, USA

Research programs, clinical trials:

- Herpetic encephalopathy, ORPHANET, France
- HSEPID - Herpetic encephalitis in children : a new group of immune diseases

GENE INFORMATION

Names:

HUGO name: UNC93B1

Localization:

Reference sequences:

DNA: D0122 (IDRefSeq) , **cDNA:** BC033623 (EMBL) , **Protein:** Q05BS6 (SWISSPROT)

Chromosomal Location:

11q13

Maps:

UNC93B1 (Map View)

Variations / Mutations:

- UNC93B1base; Mutation registry for UNC93B deficiency

Other gene-based resources:

Ensembl: ENSG00000110057, GENATLAS: UNC93B1, GeneCard: UNC93B1, UniGene: 502989, Entrez Gene: 502989, euGenes: 81622, GDB: 11508132, HomoloGene: 41325

PROTEIN INFORMATION

Description:

Other features:

Expression pattern for human:

Tissue	Exp. (%)	Clones
ascites	25.03	38:47004
blood	9.31	24:79803
lymph_node	8.11	21:80139
uncharacterized_tissue	7.80	54:214464
tonsil	6.96	4:17800
colon	4.64	15:100063
mixed	4.20	39:287479
cervix	3.91	5:39633
placenta	2.93	18:190515
spleen	2.52	4:49068

Animal models:

Mouse:

MGD: ; Unc93b1

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+UNC93B deficiency