Barth syndrome

GENERAL INFORMATION

Description:

Defects in tafazzin (TAZ) are the cause of Barth syndrome (BTHS). BTHS is a severe inherited disorder, often fatal in childhood, characterized by cardiac and skeletal myopathy, short stature and neutropenia. Presentation can be slowly progressive or sudden.

Alternative names:

- BTHS
- Cardioskeletal myopathy with neutropenia and abnormal mitochondria
- 3 methyl glutaconic aciduria, type II
- MGA, type II

Classification:

- Defects of phagocyte function

Inheritance:

X-linked

OMIM:

- #302060 Barth syndrome; BTHS
- #300069 Cardiomyopathy, dilated, 3A; CMD3A
- *300394 Tafazzin; TAZ

Incidence:

Incidence is not known.

CLINICAL INFORMATION

Description:

Patients have congestive heart failure, symptomatic before one year of age, neutropenia (chronic, cyclic, or intermittent), muscle hypoplasia and weakness, failure to thrive and growth retardation, 3-methyl-glutaconic aciduria, cardiolipin deficiency. The consequences of neutropenia may be severe (septicemia in newborns) or less dramatic with bacterial skin infections and oral aphtous lesions. Other important clinical problems are frequent diarrhea, recurrent aphtous ulcers, hypoglicemia, osteoporosis, chronic headache and body aches, especially during puberty, extreme fatigue, feeding problems, mild learning disabilities, high incidence of minor congenital malformations.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Barth syndrome, ORHANET
- Neutropenia, eMedicine

Genetic:

- 3-Methylglutaconic Aciduria Type 2, GeneTest

Therapeutic options:

- Treatment is supportive and multidisciplinary, patients are followed up by cardiologists and hematologists.
- Neutropenia, eMedicine

Research programs, clinical trials:

- European Initiative for Primary Immunodeficiencies 2001-2004, coord.Edvard Smith.

GENE INFORMATION

Names:

HUGO name: TAZ
Alias(es): BTHS, CMD3A , EFE , EFE2 , G4.5 , XAP-2 , tafazzin

Localization:

Reference sequences:

DNA: X92763 (EMBL) X92762 (EMBL) , **cDNA:** X58957 (EMBL) , **Protein:** Q16635 (SWISSPROT) Other Sequences

Chromosomal Location:

Xq28

Maps:

TAZ (Map View)

Markers:

SGC32232, DXS7010E, GDB:548776, REN89469, REN89476, REN89505, REN89506, REN89507

Variations / Mutations:

- TAZbase; Mutation registry for Barth syndrome

Other gene-based resources:

Ensembl: ENSG00000102125, GENATLAS: TAZ, GeneCard: TAZ, UniGene: 409911, Entrez Gene: 6901, euGenes: 6901, GDB: 120609

PROTEIN INFORMATION

Description:

Protein function:

Some isoforms may be involved in cardiolipin metabolism.

Subcellular location:

Isoforms with hydrophobic n-terminus are thought to be membrane anchored. The shortest forms, lacking the hydrophobic stretch, may be soluble cytoplasmic proteins.

Tissue specificity:

High levels in cardiac and skeletal muscle. Up to 10 isoforms can be present in different amounts in different tissues. Most isoforms are ubiquitous. Isoforms that lack the n-terminus are found in leukocytes and fibroblasts, but not in heart and skeletal muscle. Some forms appear restricted to cardiac and skeletal muscle or to leukocytes.

Similarity:

Belongs to the taffazin family.

Domains:

Hydrophilic domain: 124-194

Other features:

Other related resourses:

InterPro: IPR002123; Acyltransferase, InterPro: IPR000872; Tafazzin, Pfam: PF01553; Acyltransferase, SMART: SM00563; PlsC

Expression pattern for human:

Tissue	Exp. (%)	Clones
blood	14.67	8:46017
cervix	10.28	4:32826
prostate	6.99	8:96523
pancreas	6.24	5:67592
colon	6.15	6:82275
lymph_node	5.93	8:113869
muscle	5.57	6:90826
mixed	5.37	20:314320
brain	5.02	19:319574
bone	4.94	3:51252
blood	14.67	8:46017
cervix	10.28	4:32826
prostate	6.99	8:96523
pancreas	6.24	5:67592
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bone	4.94	3:51252

Animal models:

Mouse:

MGD: ; tafazzin

OTHER RESOURCES

Societies:

General:

- International Patient Organization for Primary Immunodeficiencies
- Immune Deficiency Foundation
- European Society for Immunodeficiencies

Disease specific:

- Barth Syndrome Foundation, USA
- Severe Chronic Neutropenia International Registry, Germany