Bloom syndrome

GENERAL INFORMATION

Description:

Defects in BLM are the cause of Bloom syndrome, an autosomal recessive disorder characterized by proportionate pre- and postnatal growth deficiency; sun-sensitive, telangiectatic, hypo- and hyperpigmented skin; predisposition to malignancy; and chromosomal instability.

Alternative names:

- BS
- BLS; Bloom-Torre-Machacek Syndrome; Syndrome Bloom;Syndrome Bloom-Torre-Machacek; Congenital Telangieactic Erythema

Classification:

- DNA breakage associated syndromes

Inheritance:

Autosomal recessive

OMIM:

- #210900 Bloom syndrome; BLM
- *604610 Bloom syndrome; BLM

Cross references:

Phenotype related immunodeficiencies:

- IDR factfile for Nijmegen-breakage syndrome
- IDR factfile for Ataxia-telengiectasia

Incidence:

Incidence unknown.

CLINICAL INFORMATION

Description:

Patients have small bodies, usually exhibit an erythematous (red skin produced by capillary congestion) "butterfly rash" that is sensitive to sunlight, excessive hyper- and hypopigmented skin lesions located anywhere on the body, and a high rate of bacterial infections due to immunodeficiency. These children are prone to cancer, chronic lung disease, and diabetes. Bloom syndrome is more common among Ashkenazi Jews than any other population.

Diagnosis:

Diagnostic laboratories:

Clinical:

- Bloom syndrome, ORPHANET
- Bloom syndrome, eMedicine

Genetic:

- Bloom Syndrome, GeneTest

Therapeutic options:

- Bloom syndrome, eMedicine

Research programs, clinical trials:

- Bloom syndrome, ORPHANET
- Jewish genetic disorders program, Chicago Center for Jewish Genetic Disorder
- Biological significance of the Bloom's syndrome protein

GENE INFORMATION

Names:

HUGO name: BLM
Alias(es): BS, RECQ2, RECQL3, Bloom's syndrome protein , RecQ protein-like 3 , DNA helicase, RecQ-like, type 2

Localization:

Reference sequences:

DNA: AC002312 (EMBL) , **cDNA:** U39817 (EMBL) , **Protein:** P54132 (SWISSPROT)

Chromosomal Location:

15q26.1

Maps:

BLM (Map View)

Variations / Mutations:

- BLMbase; Mutation registry for Bloom syndrome

Other gene-based resources:

Ensembl: ENSG00000197299, GENATLAS: BLM, GeneCard: BLM, UniGene: 169348, Entrez Gene: 641, euGenes: 641, GDB: 135698

PROTEIN INFORMATION

Description:

Protein function:

Participates in DNA replication and may participate in repair. Exhibits a Magnesium-dependent ATP-dependent DNA-helicase activity that unwinds single- and double-stranded DNA in a 3'-5' direction.

Subunit:

Part of the brca1-associated genome surveillance complex (basc), which contains brca1, msh2, msh6, mlh1, atm, blm, pms2 and the rad50-mre11-nbs1 protein complex. This association could be a dynamic process changing throughout the cell cycle and within subnuclear domains.

Subcellular location:

Nuclear.

Other features:

Other related resourses:

InterPro: IPR001410; DEAD, InterPro: IPR002464; DEAH_box, InterPro: IPR002121; HRDC, InterPro: IPR001650; Helicase_C, InterPro: IPR004589; RecQ, Pfam: PF00270; DEAD, Pfam: PF00271; helicase_C, Pfam: PF00570; HRDC, SMART: SM00487; DEXDc, SMART: SM00490; HELICc, SMART: SM00341; HRDC, PROSITE: PS00690; DEAH_ATP_HELICASE

Expression pattern for human:

Tissue	Exp. (%)	Clones
melanoma (cell line)	34.71	1:1599
leukopheresis	12.18	1:4557
B-cells	6.71	2:16533
testis, cell line	6.54	2:16978
eye	5.05	4:43934
blood, lymphocyte	4.90	1:11328
tonsil, enriched for germinal center B-cells	4.56	3:36522
blood	4.39	1:12646
lymph	3.45	4:64395
pool, lung+testis+B-cell	2.99	3:55714

OTHER RESOURCES

Societies:

General:

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

Disease specific:

- The Human Growth Foundation
- The Magic Foundation
- Chicago Center for Jewish Genetic Disorders
- Center for Jewish genetic disorders
- Xeroderma Pigmentosum Society, Inc