GSDMB (gasdermin B)

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Identity
Other names: GSDLML, PRO2521
HGNC (Hugo): GSDMB
Location: 17q12
Local order: ERBB2, GRB7, IKZF3, ZPBP2, GSDMB, ORMDL3, GSDMA, PSMD3, CSF3.

DNA/RNA

Description
GSDMB belongs to the novel gene family Gasdermin (GSDM) that consists of four genes in human: GSDMA, GSDMB, GSDMC and GSDMD (Tamura et al., 2007). The human GSDMB gene is located on chromosome 17q21 and consists of 12 exons, spanning 14 kb of genomic DNA.

Transcription
GSDMB has at least four alternatively spliced transcripts variants that range from 1578 to 1646 bp in length. These splicing variants, which lack exon 6, 7, and both, are named GSDMB transcript variant 1 (Delta exon 6), GSDMB transcript variant 2 (Delta exon 6 and exon 7), GSDMB transcript variant 3 (full length) and GSDMB transcript variant 4 (Delta exon 7). GSDMB has two different promoters, Alu-derived promoter and long-terminal-repeat (LTR)-derived promoter (Sin et al., 2006; Huh et al., 2008; Komiyama et al., 2010). These two promoters drive all GSDMB transcriptions except for those starting from exon1a and exon1b.

Pseudogene
No GSDMB pseudogene has been identified in the human genome.

Protein

Description
The full length of GSDMB gene (GSDMB transcript variant 3) encodes a 416 amino acid protein with estimated molecular weight of 47345.44 Da. GSDM family proteins have 9 conserved and unknown function motifs (motif I to IX). GSDMB lacks motif VIII where mouse alopecia mutation Rex-denuded occurs (Runkel et al., 2004; Tamura et al., 2007).

Expression
Expression of GSDMB was observed in esophagus and gastric epithelium (Saeki et al., 2009). In addition, GSDMB was frequently expressed in several cancer tissues and cancer-derived cell lines.

Localisation
Cytoplasm (Carl-McGrath et al., 2008).

Function
The function of GSDMB is largely unknown, because GSDMB is a novel protein without characteristic domains related to known function. Expression level of GSDMB seems to be changed along progression of several cancers. However, forced expression of GSDMB could neither promote malignant transformation of CHO cells nor confer colony formation ability or carcinogenicity to CHO cells in nude mice (Sun et al., 2008). Recent large-scale genome wide association studies (GWAS) revealed that GSDMB locus is strongly correlated with asthma (Moffatt et al., 2010).

Homology
GSDMB protein shares homology with other GSDM family members for N- and C- terminal sequences. Its
N-terminal sequence is also shared by DFNA5 (deafness, autosomal dominant 5) and DFNB59 (deafness, autosomal recessive 59, also known as pejvakin), although the extent of similarity to these proteins is lower than to the other GSDM family members.

GSDMB ortholog is identified in the chimpanzee, dog and cow, but not in the rat and mouse (Katoh and Katoh, 2004; Tamura et al., 2007).

### Mutations

**Note**

No human GSDMB mutations have been reported to date.

**Germinal**

Not known.

**Somatic**

Not known.

### Implicated in

**Various cancers**

**Note**

GSDMB is up-regulated in several cancers, such as gastric, colorectal, hepatic, cervical carcinomas, suggesting the possibility that it is used as a marker for cancer development and progression.

**Asthma**

**Note**

The genome wide association study (GWAS) demonstrated that GSDMB locus is strongly associated with asthma (Moffatt et al., 2010).

### References

Katoh M, Katoh M. Evolutionary recombination hotspot around GSDML-GSDM locus is closely linked to the oncogenic recombination hotspot around the PPP1R1B-ERBB2-GRB7 amplicon. Int J Oncol. 2004 Apr;24(4):757-63


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