Gene Section
Short Communication

AMBN (ameloblastin (enamel matrix protein))
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Published in Atlas Database: December 2011
DOI: 10.4267/2042/47320

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Identity
HGNC (Hugo): AMBN
Location: 4q13.3
Local order: AMBN is between the sequence tagged site markers D4S409 and D4S1558 (Karrman et al., 1997).

DNA/RNA
Note
The putative start codon location and exon-intron sizes differs among reports in literature.

Description
13 exons and 12 introns (Toyosawa et al., 2000; Macdougall et al., 2000) encompassing approximately 15005 bp.
Until 2011, 44 SNP were described (NCBI dbSNP).

The genomic organization of the human ameloblastin gene according to Mardh et al., 2001. The map is drawn to scale. Filled boxes represent exons and the thin lines indicate introns. Sequencing of AMBN intron 11 revealed an interrupted dinucleotide repeat (CA)_n.

Transcription
Alternatively spliced. Exon 6 can be excluded by the use of an alternative splice site (Macdougall et al., 2000). There are 2 validated alternative polyadenylation sites.

Protein
Description
The predicted protein has 447 aa (48.3 kDa). There are 3 protein isoforms. The human precursor protein contains a phosphorylation site for tyrosine kinase, a SH3 binding region, an O-linked glycosylation, and a heparin binding domain (Kobayashi et al., 2007; Krebsbach et al., 1996; Yamakoshi et al., 2001; Sonoda et al., 2009). Ameloblastin is cleaved after secretion into several lower-molecular-mass proteins that are developmentally expressed (Ravindranath et al., 2007).
Expression
Tomes processes of secretory ameloblasts (Krebsbach et al., 1996; Cerny et al., 1996; Fong et al., 1996), odontoblasts and pre-odontoblasts (Fong et al., 1996; Nagano et al., 2003). Outer enamel, and sheath space between rod and interrod enamel (Uchida et al., 1995; Macdougall et al., 2000). Early bone and cartilage extracellular matrices during embryogenesis (Spahr et al., 2006).

Localisation
Extracellular matrix.

Function
Tooth enamel biomineralization (Uchida et al., 1997). Interactions between the ameloblasts and the enamel extracellular matrix (Fukumoto et al., 2004). Dental epithelium cell adhesion (Sonoda et al., 2009). Early bone formation and repair (Iizuza et al., 2011; Tamburstuen et al., 2011).

Homology
Pig (sheathlin), cattle, rat, and mouse AMBN sequences showed a high amino acid sequence similarity.

Mutations
Somatic
AMBN gene mutations have been observed in several epithelial odontogenic tumor entities: unicystic ameloblastoma, solid ameloblastoma, adenomatoid odontogenic tumor, squamous odontogenic tumor, and calcifying epithelial odontogenic tumor (Toyosawa et al., 2000; Perdigão et al., 2004; Perdigão et al., 2009).

Implicated in
Odontogenic tumors
Disease
Odontogenic tumours arise from the residues of odontogenic epithelium and/or ectomesenchyme, as a result of disturbances in the development of teeth and associated structures.

Oncogenesis
AMBN gene is mutated in ameloblastomas and others odontogenic tumors (Toyosawa et al., 2000; Perdigão et al., 2004; Perdigão et al., 2009). Ambn-null mice develop odontogenic tumors of dental epithelium origin (Fukumoto et al., 2004). AMBN expression prevents odontogenic tumor development by suppressing cell proliferation and maintaining differentiation phenotype through Msx2, p21, and p27 (Sonoda et al., 2009). The absence of ameloblastin in epithelial odontogenic tumors has been considered a useful marker for the functional differentiation of secretory ameloblast (Takata et al., 2000).

Amelogenesis imperfecta
Disease
Amelogenesis imperfect is a common group of inherited defects such as hypoplastic or hypomineralized enamel. Autosomal dominant, autosomal recessive, and X-linked forms of amelogenesis imperfect are recognized.

Oncogenesis
Amelogenin and ameloblastin have an impaired secretion in ameloblasts of phenocopies human X-linked amelogenesis imperfect mice, which results in severe enamel bio-mineralization defects, loss of ameloblast phenotype, increased ameloblast apoptosis, and formation of multi-cellular masses (Barron et al., 2010). AMBN mutations in the coding region or splice sites were discarted to be responsible for autosomal dominant amelogenesis imperfecta (Mardh et al., 2001).

References


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