

Gene Section

Mini Review

CRTC1 (CREB regulated transcription coactivator 1)

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Identity

Hugo: CRTC1

Other names: KIAA0616; FLJ14027; WAMTP1; MECT1; TORC1

Location: 19p13.11

DNA/RNA

Description

Spans about 94 kb and includes 14 to 16 exons.

Transcription

Two RNA variants of 2505 and 2342 bp, respectively.

Protein

Note: 634 amino acids; 67300 Da.

Description

Transcriptional coactivator for CREB1.

Expression

Expressed in a restricted number of tissues including fetal brain and liver and adult heart, skeletal muscle, liver and salivary gland.

Localisation

Nucleus.

Function

Interacts through its N-terminal CREB-binding domain with the bZIP domain of CREB1; Binds to CREB1 as a homotetramer. Transcriptional coactivator for CREB1, which activates transcription through both consensus and variant cAMP response element (CRE) sites, leading to activation of CREB1 target genes. Does not appear to modulate CREB1 DNA-binding activity but enhances the interaction of CREB1 with TAF4/TAFII-130.

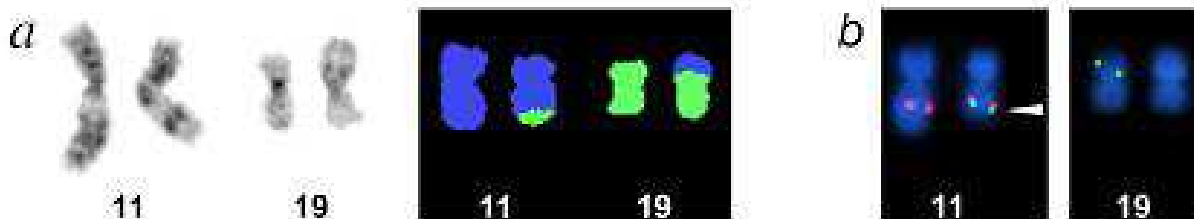
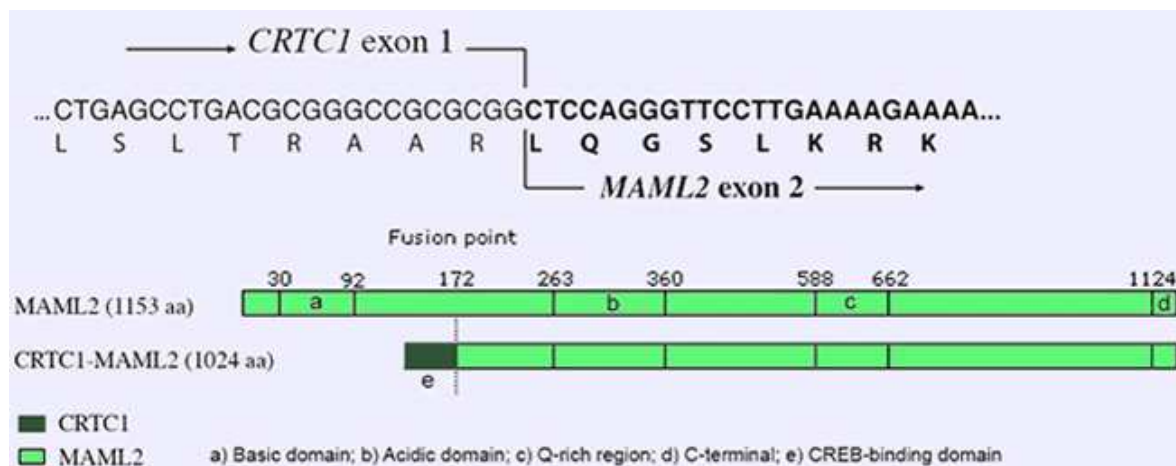


Fig1a (a) Partial G-banded and SKY karyotypes showing the t(11;19) in a mucoepidermoid carcinoma (MEC) case. (b) Dual-color FISH experiment of the MEC using BAC 697H10 (MAML2, red signal) and cosmid LLNLR 255A4 (CRTC1, green signal) as probes. Note the presence of the CRTC1-MAML2 fusion gene on the der(11) (fused red-green signals marked by an arrowhead).

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Implicated in

Mucoepidermoid carcinoma (most common type of malignant salivary gland tumor; second most frequent lung tumor of bronchial gland origin); benign Warthin's tumor; clear cell hidradenoma of the skin (CCH; a.k.a. nodular hidradenomas or eccrine acrospiromas). All sharing a t(11; 19) (q21; p13).

Cytogenetics

t(11;19)(q21;p13).

Hybrid/Mutated Gene

CRTC1-MAML2. Exon 1 of MECT1 fused to exons 2-5 of MAML2.

Abnormal Protein

MECT1-MAML2; in the fusion protein the first 171 aa including the basic domain of MAML2 are replaced by 42 aa of MECT1; there are no sequence similarities in the N-terminal domains of MAML2 and MECT1; the fusion protein activates transcription of the Notch target gene HES1 independently of both Notch ligand and CSL.

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