Skin: Clear cell hidradenoma of the skin (CCH)
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Identity

Other names: Nodular Hidradenomas; Eccrine Acrospiromas.

Disease
Clear Cell Hidradenomas of the skin (CCH) are benign sweat gland tumors of eccrine duct origin often presenting as solitary, intradermal nodules. They are usually circumscribed, nonencapsulated tumors composed of polyhedral or fusiform cells with clear or eosinophilic cytoplasm. In some tumors, epidermoid differentiation may also be encountered. CCHs rarely undergo malignant transformation.

Cytogenetics
Cytogenetics molecular
t(11;19)(q21;p13) translocation.

Genes involved and Proteins
CREB regulated transcription coactivator 1 (CRTC1)
Location: 19p13.11
Note: Alias: MECT1, WAMTP1, TORC1.
DNA/RNA
DNA: spans about 94 kb and includes 14 to 16 exons.
RNA: two variants of 2342 bp and 2505 bp in size.
Protein
634 amino acids; 67300 Da.

Mastermind-like 2 (MAML2)
Location: 11q21

DNA/RNA
DNA: spans about 365 kb and includes 5 exons. RNA: a major transcript of 7.5 kb.
Protein
1153 aa; 125000 Da; conserved N-terminal basic domain (aa 29-92) which binds to the ankyrin repeat domain of Notch receptors; two acidic domains (aa 263-360 and 1124-1153) and a C-terminal transcriptional activation domain.

Result of the chromosomal anomaly

Hybrid Gene
Detection protocol
1- RT-PCR using total RNA from frozen tumor tissue or paraffin-embedded tumor tissue: amplification of the CRTC1-MAML2 fusion transcript by nested RT-PCR using the first-round primers CRTC1, 5'-AGGAGGTTGAGGAGGAGGAG-3', and MAML2, 5'-TGTTGGCAGGAGATAGGTTAACTACCTG-3' (product size 221 bp), and the second-round primers CRTC1, 5'-GAGAAGATCGCGCTGCAC-3', and MAML2, 5'-GTAACTACCTGTTTCTTTCTCAAGG-3' (product size 127 bp).
2- Dual-color FISH on metaphase chromosomes: The CRTC1-MAML2 fusion gene may be detected by dual-color FISH, using BAC RP11-697H10 (MAML2) and cosm id LLNLR-255A4 (MECT1) as probes.


Schematic representation of the MAML2 protein and the CRTC1-MAML2 fusion protein. Reprinted partially from Publication: Exp Cell Res 292, see above.
References


This article should be referenced as such: