Atlas of Genetics and Cytogenetics in Oncology and Haematology

OPEN ACCESS JOURNAL INIST-CNRS

Solid Tumour Section

Short Communication

Skin: Cylindroma

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Published in Atlas Database: April 2012

Online updated version : http://AtlasGeneticsOncology.org/Tumors/SkinCylindromaID6377.html
DOI: 10.4267/2042/47544

Identity

Other names
Dermal cylindroma
Dermal eccrine cylindroma and (less specific) cylindroma

Clinics and pathology

Disease

Dermal cylindroma

Note
Dermal cylindroma is a common benign adnexal tumor which occurs mainly in the head and neck region, especially on the scalp and face (Weedon, 2002; McNiff et al., 2006).

Clinics

There is a strong predilection for middle-aged and elderly females. Dermal cylindromas may be found as solitary or multiple lesions, the latter being associated with the autosomal dominant Brooke-Spiegler syndrome (familial cylindromatosis; OMIM 605041) which is caused by mutations in the tumor suppressor gene CYLD (Bignell et al., 2000).

Pathology

Microscopically, dermal cylindromas are composed of multiple tumor lobules arranged in a jigsaw pattern. The lobules are typically surrounded by a rim of PAS-positive basement membrane material and are composed of undifferentiated basaloid tumor cells. The histogenesis of dermal cylindroma is controversial. It is still uncertain whether they originate from apocrine or eccrine sweat glands or if they as recently suggested may be derived from hair follicle epithelium (reviewed in Weedon, 2002; Klein et al., 2005; Massoumi et al., 2006; Massoumi and Paus, 2007).

Cytogenetics

Cytogenetics Morphological

t(6;9)(q22-23;p23-24) translocation.

Genes involved and proteins

MYB (v-myb myeloblastosis viral oncogene homolog (avian))

Location
6q23

Note
Leucine zipper transcription factor. MYB in solid tumors was recently reviewed by Stenman et al., 2010.

DNA / RNA
RefSeq DNA sequence: NC_000006.11, NT_025741.15.

Protein
The MYB protein plays an essential role in the regulation of hematopoiesis and may play a role in tumorigenesis.
Alternative splicing results in multiple transcript variants.

NFIB (nuclear factor I/B)

Location
9p23-p22

Note
CCAAT-box-binding transcription factor.

DNA / RNA
RefSeq DNA sequence: NC_000009.11, NT_008413.18.

Result of the chromosomal anomaly

Hybrid Gene

Note
MYB-NFIB.

Description
RT-PCR analysis revealed that 6 of 11 dermal cylindromas were positive for the MYB-NFIB fusion; one case had a fusion of MYB exon 12 to NFIB exon 9 and 5 cases had a fusion of MYB exon 14 to NFIB exon 8c. The composition of the chimeric transcript variants identified was identical to that previously reported in adenoid cystic carcinoma, suggesting a similar molecular mechanism of activation of MYB in both tumor types (Persson et al., 2009; Brill et al., 2011; Fehr et al., 2011).

Detection
Described by Persson et al., 2009 and Fehr et al., 2011.

Fusion Protein

Description
Analysis of MYB expression by immunohistochemistry using a monoclonal anti-MYB antibody recognizing an N-terminal peptide of the protein revealed overexpression of MYB in 6 of 10 cylindromas. Fusion-negative tumors lacked MYB expression, indicating that a subgroup of cylindromas in fact are MYB-negative (Fehr et al., 2011).

To be noted

Note
The results from Fehr et al., 2011 indicate that MYB oncogene activation through gene fusion is an important pathogenetic mechanism that may contribute to cylindroma tumorigenesis. In this context it will be of interest to test also familial cases of cylindroma (Brooke Spiegler syndrome) for the MYB-NFIB fusion to determine whether MYB activation along with loss of CYLD1 simultaneously occur in these cases.

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This article should be referenced as such:

Fehr A, Stenman G. Skin: Cylindroma. Atlas Genet Cytogenet Oncol Haematol. 2012; 16(9):683-684.