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Solid Tumour Section

Mini Review

Soft tissue tumors: Lipoblastoma

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Classification

Two forms of the tumor lesion have been described: encapsulated circumscribed type (lipoblastoma), and a noncapsulated diffuse infiltrative type (lipoblastomatosis).

Clinics and pathology

Disease

Benign uncommon soft-tissue tumor of embryonal fat.

Embryonic origin

Develops from embryonic remnants of white fat tissue.

Epidemiology

Primarily occurs in young children (<3 years) prevalently male.

Clinics

Presents in superficial tissues of arms and legs (deeper in lipoblastomatosis), though mediastinum, retroperitoneum, trunk, head and neck may be affected.

Pathology

Lobulated tissue composed of immature fat cells separated by fibro-vascular septa and areas with a myxoid matrix. The lobules contain lipoblasts in different stages of differentiation, ranging from primitive, spindle-shaped cells to lipoblasts simulating mature fat cells. Differential diagnosis, particularly in older children or in diffuse lipoblastoma, includes myxoid liposarcoma and atypical lipoma and may be based on distinct cytogenetic abnormalities.

Treatment

Surgical excision.

Prognosis

These tumors have an excellent prognosis but local recurrence is possible expecially in diffuse lesions.

Cytogenetics

Cytogenetics Morphological

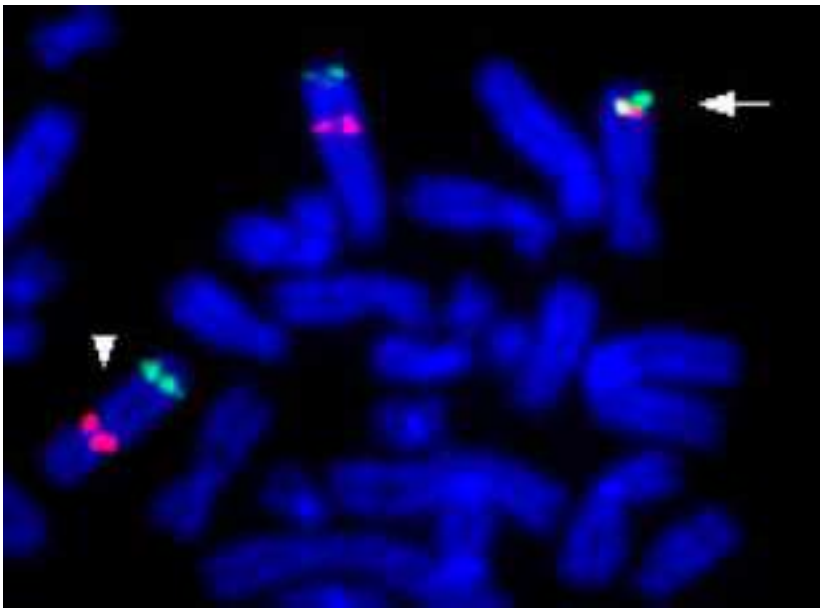
Pseudodiploid karyotype with clonal chromosomal rearrangements involving the 8q11-13 region. Gain of chromosome 8 is reported.

Cytogenetics Molecular

Detectable by metaphase and/or interphase FISH using specific PLAG1 probes.

Probes

RP11-140I16, BAC227k20, YAC164H5, RP11-299N14, YAC947h7.



Dual-color FISH analysis in a case of lipoblastoma with complex structural rearrangement: RP11-140I16 (PLAG1) (red) was cohybrized with RP11-299N14 (HAS2) (green). Arrow indicates PLAG1-HAS2 fusion signal on the der(8), arrowhead indicates the normal chromosome 8. The BAC clones were provided by Prof. M.Rocchi.

Genes involved and proteins

PLAG1

Location

8q12.1

DNA / RNA

7313 bp mRNA

Protein

PLAG1, (together with PLAGL1 and PLAGL2), belongs to a subfamily of C2H2 zinc finger transcription factors that activate transcription.

HAS2

Location

8q24.12

COL1A2

Location

7q22.1

Result of the chromosomal anomaly

Hybrid Gene

Note

HAS2-PLAG1, COL1A2-PLAG1 Chromosomal rearrangements in tumor tissue determines PLAG1 transcriptional up-regulation.

Description

The 8q12 rearrangement results in a promoter-swapping event, whereby the PLAG1 promoter element is replaced by promoting regions from other genes, notably hyaluronic acid synthase 2 (HAS2) or collagen 1 a 2 (COL1A2).

5' HAS2 - 3' PLAG1 fused after an 8q intrachromosomal rearrangement that determined the juxtaposition of band 8q12.1 to 8q24.1. The breakpoint of HAS2 gene is in intron 1, whereas its coding sequence starts at the first codon of exon 2.

5' COL1A2 - 3' PLAG1 has been described in one case of t(7;8)(p22;q13)

Transcript

Alternative splicing variants which included or lacked PLAG1 exon 2.

Fusion Protein

Description

HAS2-PLAG1 and COL1A2-PLAG1 both encode a full-lenght PLAG1 protein.

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