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Cancer Prone Disease Section

Mini Review

Congenital myofibromatosis

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Identity

Other names: Infantile myofibromatosis; Mesenchymal hamartomatosis; Hemangiopericytoma; Vascular leiomyoma of the newborn; Congenital generalized fibromatosis
Inheritance: Postulated as autosomal dominant (AD) with variable expression or autosomal recessive (AR).

Clinics

Myofibromatosis or infantile myofibromatosis (IM) is one of the more common fibromatoses that present during childhood. Presentation may occur as an adult or even prenatally.
These tumors grow and regress without known initiation factors, and the diagnostic classification depends solely upon the location of the tumors. Individuals with Solitary IM only have tumor involvement of the soft tissues.
However, those individuals with Multiple IM have tumors within bone tissue, and those with Generalized IM demonstrate visceral tumors. Soft tissue involvement may occur in all three, and bone involvement may also be present in generalized IM.

Neoplastic risk

Risk for neoplasm is considered to be very low. In those individuals who have multiple tumors, pathogenesis appears to be related to multifocal potential, not metastatic potential.

Treatment

Treatment is based solely upon clinical presentation. Those tumors causing secondary pathology via mass affect are commonly removed. Others may be watched due to their potential to regress.

Evolution

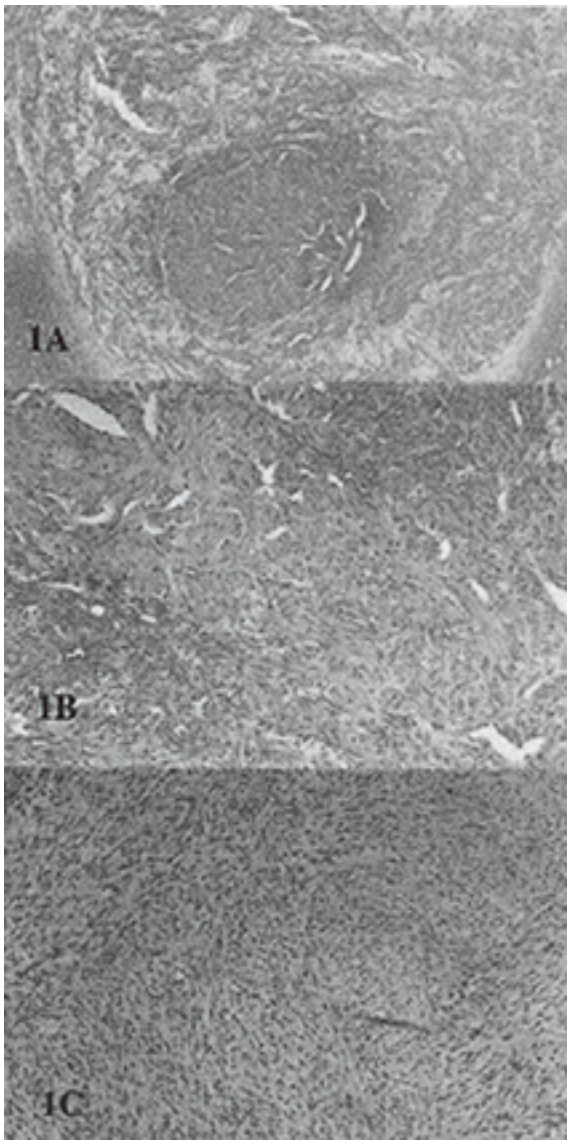
The evolution of the tumor is not well understood.
Pathologically, they are well circumscribed.
Histopathologically, hematoxylin and eosin (H and E) staining demonstrates growth in a zonal pattern with more primative appearing cells located centrally and spindle shaped cells peripherally. The spindle shaped cells resemble fibroblasts but are often arranged in a pattern similar to fascicles - thus resembling myocytes. As some tumors may grow rapidly, it is also common to see areas of central necrosis and calcification.

Prognosis

Prognosis is usually based upon the secondary complications caused by the tumors. Individuals with multiple tumors or visceral involvement tend to have more complications due to either number the increased number or increased possibility of poor location. In general, most individuals with uncomplicated presentations have a good prognosis.

Cytogenetics

Unknown.
Only two cytogenetic abnormalities in IM tissue have been reported: Monosomy 9q/trisomy 16q and an interstitial deletion on chromosome 6q. No comparison was made with the constitutive karyotype, and direct correlation was not able to be confirmed. It is presumed that the causative gene might allow for growth potential or affect cell cycle to account for the unique properties of both growth and regression of these tumors, but as of yet no gene has been identified.



Hematoxylin and eosin staining of infantile myofibromatosis (IM) biopsies.
A: Family I (III-9), showing zonal pattern of spindle shaped cells with central necrosis and calcification. The lesion was subcutaneous scalp mass obtained at 4 months of age, and the diagnosis of IM was confirmed by outside consultation (Dr. C. Coffin, U. of Utah).
B: Family II (IV-6), shoulder lesion obtained at 3 months of age, but present since birth. The sample demonstrates prominent vascularity.
C: Family II (III-5), temporal lesion, biopsed at age 28 years. Diagnoses initially considered included fibroblastic meningioma, Schwanoma-neurilemmona, and IM. The patient has generalized IM confirmed by multiple other biopsies of the deltoid, axilla, and shoulders. Note the architectural similarity of (B) and (C) despite their different origins.

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