ELSEVIER

Cancer Genetics 204 (2011) 666–670

Cancer Genetics

A t(1;9)(q10;q10) translocation with additional 6q23 and 9q22 rearrangements in a case of chondromyxoid fibroma

Tahereh Dadfarnia [a], Gopalrao V.N. Velagaleti [a], Kelly D. Carmichael [b], Eduardo Eyzaguirre [a], Mahmoud A. Eltorky [a], Suimin Qiu [a,*]

[a] *Department of Pathology, University of Texas Medical Branch, Galveston, TX, USA;* [b] *Department of Orthopedics, University of Texas Medical Branch, Galveston, TX, USA*

Chondromyxoid fibroma (CMF) is a rare cartilaginous tumor of bone. It typically presents in the long tubular bones and to a lesser extent in the small bones of the hands and feet of young adults. To date, several cytogenetic abnormalities have been described in association with CMF. We studied a phalangeal CMF from a 13-year-old female by cytogenetic methods. We found a novel unbalanced translocation between the long arms of chromosomes 1 and 9, resulting in loss of 1p. In addition, rearrangements involving the 6q23 and 9q22 regions were also observed. To our knowledge, this is the first report in the literature describing this novel chromosomal translocation in CMF.

Keywords Chondromyxoid fibroma, cytogenetics, t(1;9) translocation

Chondromyxoid fibroma (CMF) is rare cartilaginous tumor of bone. It was first described in 1948 by Jaffe and Lichtenstein (1). It represents less than 1% of all bone tumors and less than 2% of benign bone tumors (2). Males are affected approximately twice as often as females, with a peak occurrence in the second and third decades of life. CMF is seen most often in the metaphyseal region of long bones, and approximately 25% of cases occur in flat bones, mainly the ilium. Other locations in descending order of frequency include the small tubular bones of the hands and feet, vertebrae, ribs, and femur. The juxtacortical region is an unusual location for this tumor, and this variant of CMF favors older individuals (3). Pain is the most common symptom; it is usually mild and sometimes is present for several years. Swelling is noted infrequently, more often in tumors of the hands and feet. In the small tubular bones, fusiform expansion of the entire bone is typical. Radiographically, CMF most often appears as a lucent, lytic, metaphyseal lesion with sharply demarcated, scalloped, or sclerotic borders (2). The classic histologic feature of CMF includes a lobular pattern with stellate or spindle-shaped cells in a myxoid background. Lobules demonstrate hypocellular centers and hypercellular peripheries (2). Despite its name, hyaline cartilage is present in only 19% of cases of chondromyxoid fibroma—more often in lesions affecting small bones of the hands and feet (3).

The recommended treatment is en bloc excision and removal of a margin of normal bone; however, curettage is also an acceptable method in cases of difficult access or if normal bone is in close apposition to vital structures. In cases treated with curettage, close surveillance is recommended due to the incidence of recurrence of CMF following incomplete resection. The recurrence rate in curettage with bone grafting is about 10–15%, mostly seen within 2 years. Patients under the age of 15 are especially prone to recurrence. Radiation therapy is generally avoided due to reported cases of malignant transformation (3–5).

Cytogenetic studies of CMF are limited, however, the most commonly identified chromosomal aberrations are on chromosome 6, specifically, with breakpoints at p25, q13, q23, q24, and q25 (6–19).

In this study, we report a case of CMF with an unbalanced translocation involving the long arms of chromosomes 1 and 9 in addition to structural rearrangements of 6q23 and 9q22.

Materials and methods

Clinical summary

A 13-year-old, Caucasian female presented with a 5-month history of pain and a mass in the region of the third proximal

Received June 14, 2011; received in revised form November 2, 2011; accepted November 16, 2011.

* Corresponding author.
E-mail address: sqiu@utmb.edu

2210-7762/$ - see front matter © 2011 Elsevier Inc. All rights reserved.
doi:10.1016/j.cancergen.2011.11.005

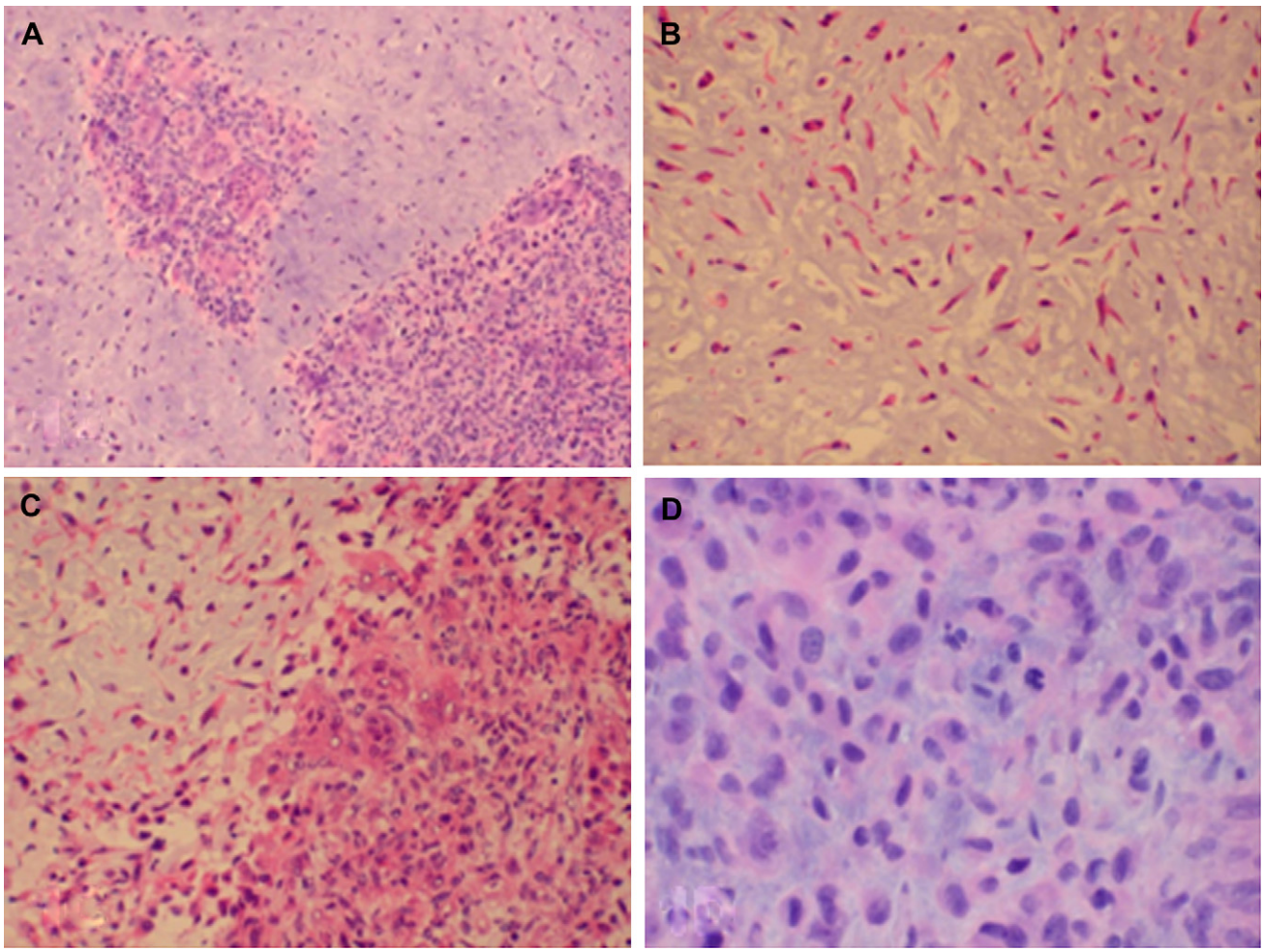

Figure 1 Histologically, the tumor consists of lobulated parenchyma with a central hypocellular area and a peripheral hypercellular area in the background of a myxoid matrix (1A). The predominant cell populations are spindle or stellate cells with elongated nuclei and eosinophilic cytoplasms (1B). Osteoclast-like giant cells are present peripheral to the hypercellular nodule (1C). Mild nuclear atypia with mitosis are identified (1D).

phalanx of the left hand. No history of trauma or complaints suggestive of infection was elicited. The patient reported increasing size of the lesion and worsening of hand pain over a period of 3 months. The left hand radiographs showed a multiloculated, expansile, and radiolucent lesion involving the proximal third phalanx of the left hand, with some reactive sclerosis at the periphery without calcification (Supplemental Figure).

The patient underwent elective surgery. An open procedure with tumor curettage was performed, and a tan-white, cartilaginous, translucent tissue fragment measuring 4×3 cm in aggregate was removed. Post-curettage, the remaining bony cortex was found to be paper-thin. The initial impression by frozen section at that time was CMF, and the patient was treated by curettage and bone grafting. A small portion of the tumor was sent to the cytogenetics laboratory for chromosome analysis. The permanent sections confirmed the diagnosis. The histologic feature of the curetted tumor was similar to that previously described in the literature.

As the name indicates, CMF shows a variety of histological features. When a CMF is curetted, it tends to fracture through the fibrous septa, making the latter inconspicuous in curetted material. Therefore, it may be observed only as a focal rim around isolated fragments of lobules. These septa contain large venules, muscular arteries, multinucleated giant cells, and, occasionally, osteoid. The tumor consists of lobules of hypocellular pale blue myxoid matrix containing spindle and stellate cells (Figures 1A and 1B). Moving toward the periphery of lobules, the tumor appears as hypercellular areas that contain similar rounded-to-spindle cells mixed with variable numbers of osteoclast-like giant cells (Figure 1C). In our case, occasional mitoses were seen within the fibrous septa (2 mitoses/10 hpf) (Figure 1D). No areas of necrosis or calcification were identified.

Cytogenetic analysis

Cytogenetic analysis was performed on the tumor biopsy specimen. Culture initiation, maintenance, and harvest were performed using standard methods. The tumor cells were cultured for a period of about 9 days before harvesting. Chromosomes were G-banded using pancreatin (20,21) and then analyzed using a Cytovision image analysis system