Reviewer's report

Title: Late onset Li-Fraumeni syndrome with bilateral breast cancer and other malignancies: case report and review of the literature

Version: 1 Date: 15 February 2012

Reviewer: Izilda Cardinalli

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- Major Compulsory Revisions:

1. The authors report a Li-Fraumeni-Syndrome (LFS) patient affected by four synchronous malignancies including a myxofibrosarcoma of the right upper arm, bilateral breast cancer and a periadrenal liposarcoma. The diagnosis of these tumors appears not to be in doubt when reading the text. However, histological images and microscopic descriptions were not provided. Photomicrographs of each neoplasm depicting the main histological findings would be helpful to support the evidence for multiple tumors of different histogenesis.


- Minor Essential Revisions:

1. In the Abstract, second paragraph, third line, the authors identified a heterozygous pathogenic TP53 germline mutation, and the analysis of both parents revealed that the mutation has arisen "de novo". However, in “Genetic counselling and molecular analysis”, first paragraph, it is noteworthy a frequent
occurrence of cancer in multiple family members at an early age. Authors are encouraged to explore the topic in more detail.

2. In Background section, third paragraph, second line, the authors describe that “The rare event of concomitant four primary malignancies led to delayed diagnosis of a periadrenal liposarcoma, which at first was classified as incidental tumor and is now crucial for the patient’s prognosis”. However, the periadrenal liposarcoma was the largest tumor at the moment of the diagnosis among the four concurrent neoplasms. Further down, in Case Presentation section, on Bilateral Breast Cancer topic, first paragraph, the authors comment that “Biopsy of the periadrenal tumor yielded an inconclusive histological result”. Why was the initial histological result inconclusive? Please provide further details and discuss as appropriate.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests with regard to this manuscript.