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Behavior of advanced gastrointestinal stromal tumor in a patient with von-Recklinghausen disease: Case report

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Gastrointestinal stromal tumors (GISTs) represent a malignant gastrointestinal tumor of neurofibromatosis type1 (NF1) Von Recklinghausen disease. In the current case, we report a 27-year-old woman with NF1, who presented with a lower abdominal mass, symptomatic anaemia, and significant weight loss. We employed multiple approaches to assess the tumor behavior, including computed tomography (CT) scan, surgical tumor resection, histological and immunohistochemical analysis and gene sequencing. Additionally, the patient was given Imatinib mesylate (Gleevec) as adjuvant therapy. CT scan delineated a large thick wall cavity lesion connecting to the small bowel segment. Resection of the tumor yielded a mass of 17 cm × 13 cm with achievement of safety margins. The diagnosis was GIST, confirmed by immunohistochemical expression of CD117, CD34, and Bcl-2. Sequencing revealed no mutations in either KIT or platelet-derived growth factor receptor- α , genes which are mutated in over 85% of sporadic GIST cases. Further, there was no evidence of recurrence, metastasis or metachronous GIST for over three years in our patient. From our analyses, we believe selective genotyping is advisable for high risk patients to predict potential tumor behavior.

Biography

Samer Sawalhi has completed his Bachelor degree from Baghdad University, and then he joined the general surgical program, after that he worked as a surgical specialist in King Hussein Cancer Center. In 2010, He was appointed as Assistant Professor-surgical department and permanent researcher in the Center of Genetics and Inherited Diseases in Taibah University-Saudi Arabia. He is interested in Onco-genetic mutation in surgical cancer patients. He has published 10 papers in reputed journals and serving as an Editorial Board Member in 5 Journals. Recently, he is doing Minimal invasive thoracic surgery fellowship at Dalhousie University-Thoracic surgery department.

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