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A Case of Hereditary Gastrointestinal Stromal Tumor (KIT Mutation p.Asp820Tyr) in a Portuguese Family and a Good Response to Imatinib – An Update

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Abstract
Gastrointestinal stromal tumors (GIST) are the most common mesenchymal tumors of the human gastrointestinal tract. They are derived from transformed neoplastic precursors of the interstitial cells of Cajal. They account for 0.1% to 3% of all gastrointestinal cancers. Up to 20% of cancers of the small bowel are GISTs. Less than 5% of cases are associated with hereditary predisposition, like Neurofibromatosis type I, Carney syndrome, and Familial GIST syndrome. The latter, is a rare autosomal dominant genetic disorder originated by germline gain-of-function mutations of KIT or PDGFRA. The study objectives were: to update the response to imatinib of family of our institution and review the families cases published in the literature with this syndrome. Review of the cases of Familial GIST syndrome published in literature and update of the only portuguese family be consultation of clinical processes. In the literature, we found 35 cases of unrelated families with this syndrome. This report is also an update on the only portuguese family with this syndrome and a good response to imatinib. The role of imatinib was not established in cases of familial GIST syndrome. In our opinion, it seems wise to use 400 mg/day for an indefinite period. The objective is disease control and hindering the development of additional lesions.
Key words: germline GIST gene mutation, gastrointestinal cancer, surgery, imatinib, stroma tumours