

Mutational Analysis in Gastrointestinal Stromal Tumors - A Series of Three Cases

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Abstract

Background: Most of the Gastrointestinal Stromal Tumors (GISTs) are determined by mutations in C-KIT or PDGFRA genes. Mutation type influences the clinical evolution and treatment response.

Case reports: This study aims to present particular cases of GIST referred for molecular testing, diagnosed and treated in our institution.

Results: We present three cases in which genetic testing was performed. The first patient was diagnosed in a short-time interval with two GISTs, a gastric and a jejunal one, both with low risk of recurrence. To establish the relationship between the two, molecular analysis was performed and the conclusion was that we were facing synchronous, sporadic GISTs, a very rare instance. The second case was a patient with liver metastases, 3 years earlier having been diagnosed with a high risk duodenal GIST. The genetic test revealed a mutational status conferring imatinib sensitivity and therefore imatinib was continued. The third case was a patient with an early recurrence in within less than a year after surgery for gastric GIST. Molecular testing identified a mutation associated with a bad clinical outcome. The patient died 2 years later from diagnosis by disease progression.

Conclusion: Although rarely performed, genetic analysis provides useful information regarding the prognosis and the response to TKIs.

Key words: gastrointestinal stromal tumor, gene, mutation, exon, molecular analysis