

Examining GIST Patients' Understanding of Somatic Tumor Testing and Personalized Medicine

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The identification and classification of an immense number of genes has created opportunities for advancements in the realm of personalized medicine, most notably in the field of oncology. In 1998, Hirota and colleagues discovered that most gastrointestinal stromal tumors (GISTs) harbor mutations in the proto-oncogene *KIT*, which lead to the use of the targeted therapy, imatinib. Recently, specific mutations in *KIT*, *PDGFRA*, and *BRAF* identified through tumor mutation analysis have been correlated with response to therapy, treatment dose, recurrence risk, and prognosis. Little is known about the understanding and attitudes of patients towards these new medical advances. Adults with GIST were surveyed to assess tumor mutational analysis uptake, understanding and attitudes. Out of the 193 survey responses, 53.1% of participants received GIST tumor mutation analysis. 44.7% of participants reported that a health care provider initiated tumor mutation analysis, while 26.2% of respondents initiated the process of tumor analysis themselves. The majority of participants in this study were able to correctly identify the benefits of tumor mutation analysis; however, many participants incorrectly reported that tumor mutation analysis could be useful in defining familial risk. Genetic counselors may be beneficial resources to patients undergoing tumor mutation analysis, as they are health care providers skilled in disseminating genetic information and addressing patient misconceptions. More work is needed to identify effective methods of communicating complex genomic concepts to patients.