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Gastrointestinal stromal tumors: A histopathological and immunohistochemical study.

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Introduction

Gastrointestinal stromal tumors (GISTs) represent the most common mesenchymal neoplasms of the gastrointestinal (GI) tract, accounting for approximately 1–3% of all GI malignancies. They originate from the interstitial cells of Cajal or their precursors and exhibit distinct histopathological and immunohistochemical characteristics that aid in diagnosis and prognostication.[1].

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Immunohistochemistry (IHC) plays a crucial role in the diagnosis of GISTs. The majority (>95%) express CD117 (c-KIT), a transmembrane tyrosine kinase receptor, which is a hallmark marker for GIST [3]. DOG1 (Discovered on GIST-1) is another sensitive and specific marker, particularly useful in CD117-negative cases . CD34 is positive in approximately 70–80% of cases, while SMA (smooth muscle actin), S100, and desmin are variably expressed [3]

The discovery of c-KIT and PDGFRA mutations has revolutionized the understanding and management of GISTs. These mutations lead to constitutive activation of tyrosine kinase signaling pathways, driving tumorigenesis. Molecular testing for these mutations is recommended, especially in cases considered for targeted therapy. Imatinib mesylate, a tyrosine kinase inhibitor, remains the first-line treatment for advanced or metastatic GISTs
and has significantly improved survival rates.
Histopathological assessment also helps identify tumor necrosis, cellular atypia, and mucosal ulceration, which are additional features associated with aggressive behavior. Furthermore, IHC can help distinguish GISTs from other mimickers such as leiomyomas, schwannomas, and desmoid tumors, which lack CD117 and expressio n [4].

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Conclusion

GISTs are distinct mesenchymal tumors of the GI tract with specific histologic and immunohistochemical profiles. Histopathology in combination with IHC, particularly CD117 and DOG1, is essential for accurate diagnosis. Molecular characterization further supports personalized treatment strategies, underscoring the importance of an integrated diagnostic approach.

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