Images in Clinical Medicine

Gastrointestinal stromal tumor with a platelet-derived growth factor receptor-alpha mutation

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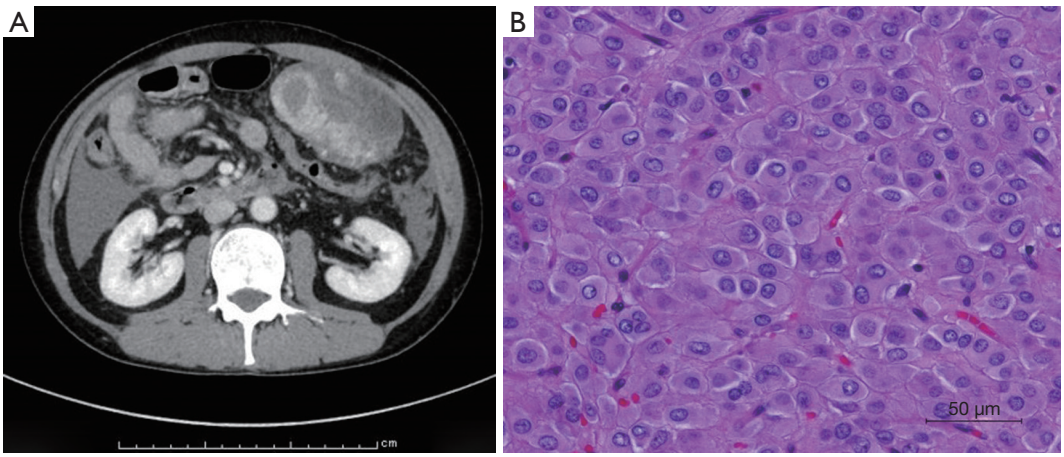
A 44-year-old man visited a local hospital, complaining of abdominal pain. The patient presented with muscular guarding in the upper abdomen and moderate anemia (hemoglobin, 6.7 g/dL). Computed tomography revealed a 9.0-cm tumor adjacent to the stomach and massive ascites (Panel A). The patient was diagnosed with hemoperitoneum caused by tumor rupture, and underwent laparotomy. Pathological examination revealed that the excised tumor was microscopically composed of cytoplasm-rich polygonal cells that were immunohistochemically positive for CD34 but negative for KIT (Panel B). Gene analyses revealed that the tumor had a deletion mutation in exon 18 (del 842-845) of *platelet-derived growth factor receptor alpha* (*PDGFRA*). Based on this finding, confirmative diagnosis of gastrointestinal stromal tumor (GIST) of the stomach was made. Despite tumor rupture, which is a significant risk factor for recurrence, the patient was postoperatively followed up without adjuvant treatment with imatinib because the *PDGFRA*-mutated GIST was reportedly resistant to the tyrosine kinase inhibitor (1). Eight years after the surgery, the patient is alive with no evidence of disease recurrence.

Clinical points

- KIT-negative, epithelioid-type GISTs of the stomach should be examined for *PDGFRA* mutations.
- *PDGFRA*-mutated GISTs are clinically indolent in general.
- Tumor gene analysis should be considered before starting adjuvant imatinib therapy.



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Footnote

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Ethical Statement: The authors are accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved. All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee(s) and with the Helsinki Declaration (as revised in 2013). Written informed consent was obtained from the patient for publication of this case report and any accompanying images.

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