

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

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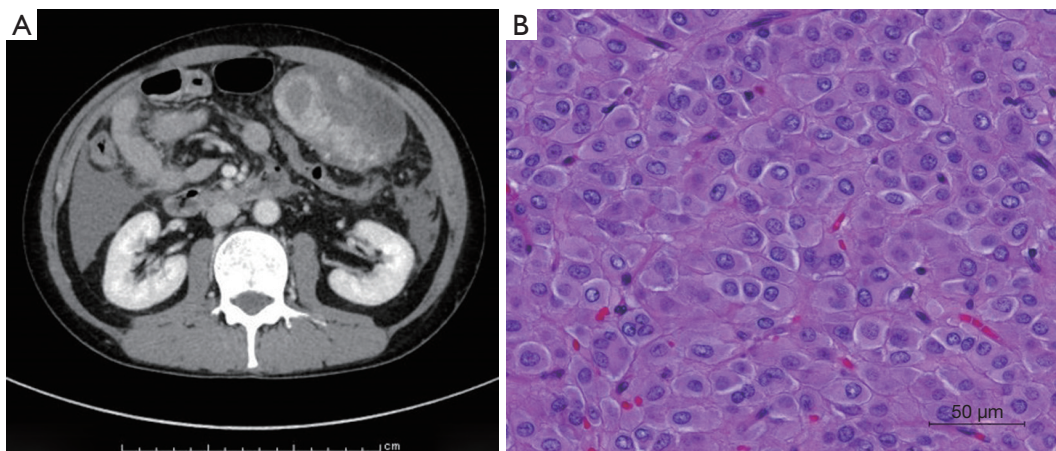
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A 44-year-old man visited a local hospital, complaining of abdominal pain. The patient presented with muscular guarding in the upper abdomen and moderate anemia (hemoglobin, 6.7 g/dL). Computed tomography revealed a 9.0-cm tumor adjacent to the stomach 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

Conflicts of Interest: The authors have no conflicts of interest to declare.

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from the patient for publication of this case report and any accompanying images.

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