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Summary

GASTROINTESTINAL STROMAL TUMOR IN A PATIENT WITH NEUROFIBROMATOSIS TYPE I: A CASE STUDY

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Oesophagus

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A 57-year-old woman with neurofibromatosis type 1 was diagnosed with an incidental gastrointestinal stromal tumor discovered during evaluation for diverticulitis and treated with surgical resection without adjuvant therapy.

KEY POINTS

- CT imaging for sigmoid diverticulitis revealed an incidental mesenteric mass measuring 4.4 x 3.1 x 3.7 cm in the left lower quadrant.
- Pathology after small bowel resection showed gastrointestinal stromal tumor with two foci (5.4 cm and 0.6 cm), negative margins, low mitotic index (2/50 HPF), and no clinically relevant KIT mutations on NGS sequencing.
- Oncology recommended CT surveillance every 6 months without adjuvant therapy because the tumor developed in the setting of NF1.
- The patient deferred routine screening due to loss of insurance and inability to afford out-of-pocket costs of CT imaging.
- The authors note that surgical resection is the mainstay of treatment for NF1-associated GIST, while tyrosine kinase inhibitors are typically reserved for sporadic cases or high-risk features.

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