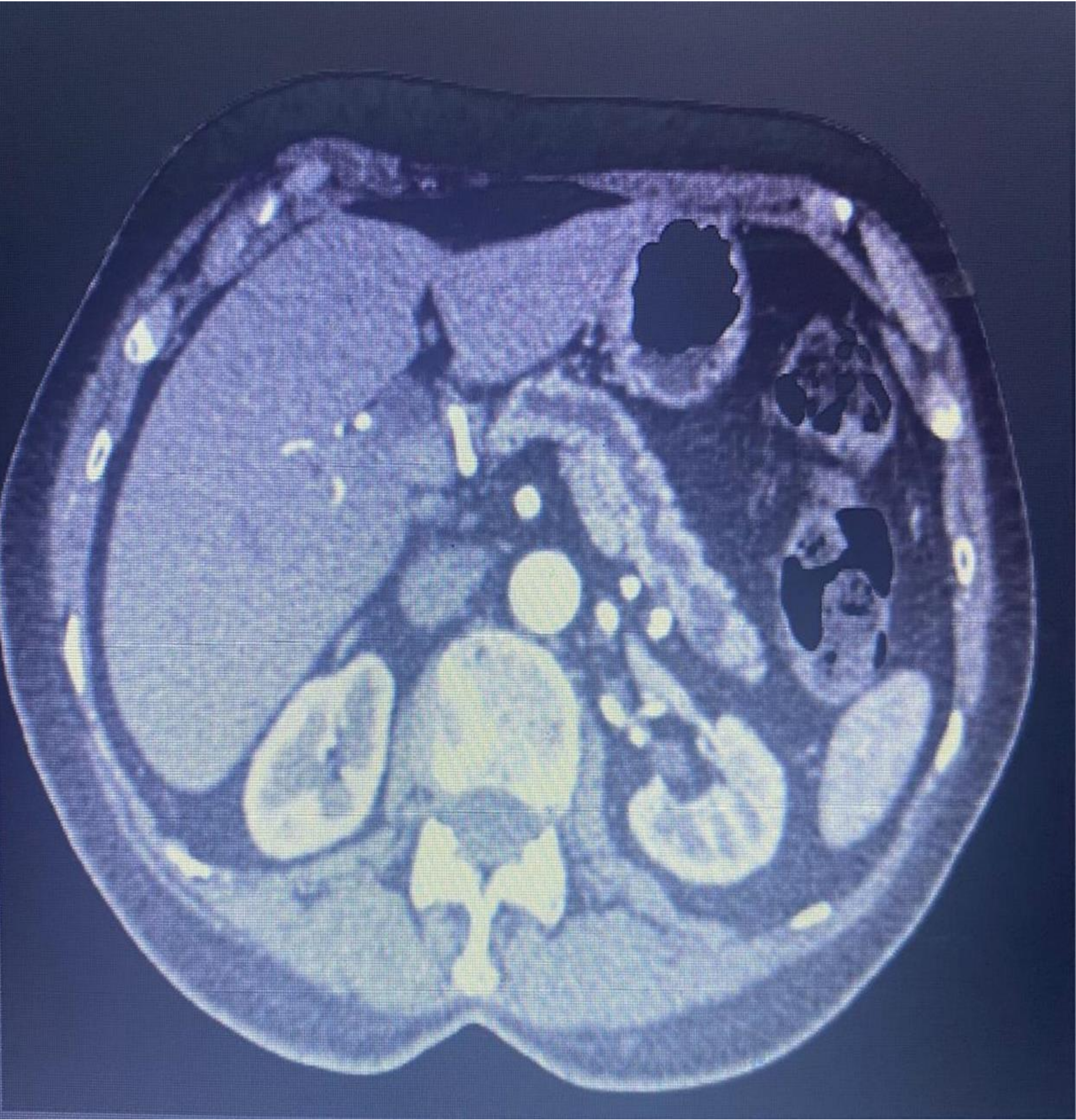




INTRODUCTION

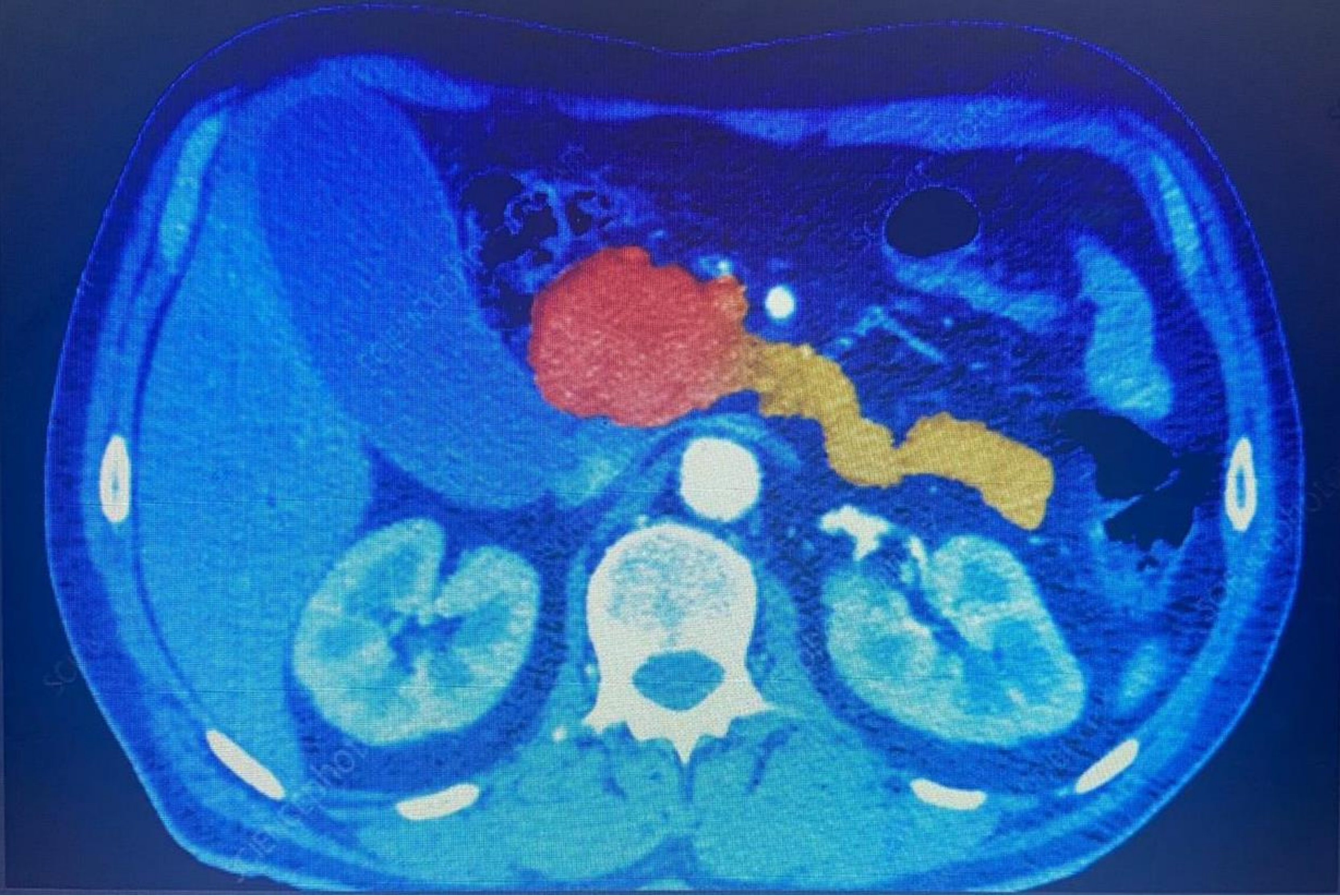
Li Freumeni syndrome is a rare hereditary autosomal dominant disorder that increases the risk of developing other types of cancers which often develops at an early age. LFS is due to TP53 gene mutation resulting in loss of its tumour suppressor function. People with Li-Freumeni syndrome are predisposed to a range of cancers including breast cancer, osteosarcoma, leukaemia and other cancers including skin and gastrointestinal. Here, we present a case of a patient with LFS that was diagnosed with osteosarcoma, breast cancer and subsequently recurrence of sarcoma



CASE REPORT

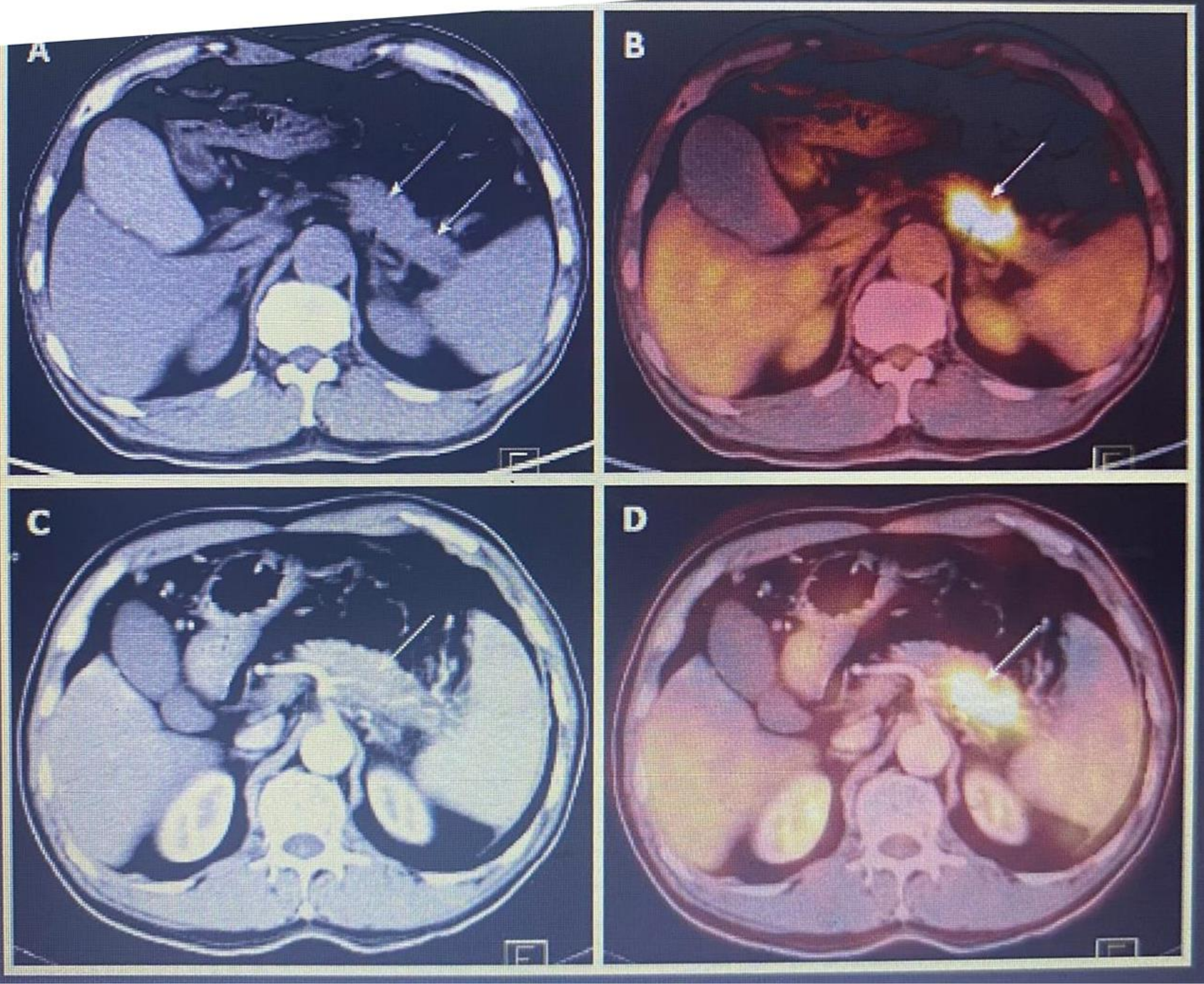
This patient a 44-year-old female with a diagnosis of LFS that was confirmed by a genetic test triggered by the patient's diagnosis of osteosrcoma of femur in 1999 with right below knee amputation. She had bilateral mastectomy due to breast cancer in 2009 and 2011. She was on yearly MRI and had colonic polypectomy in 2018.She was seen in RAC clinic in 2019 as her MRI showed bilateral pelvic mass measuring 7cm on right side and 6cm on left side, CA125 was 12 She had abnormal uterine bleeding, tumor markers and endometrial biosy were normal, diagnosis ofadenomyosis was made. MRI Pelvis showed multiple metastaic disease with large lesion centered on right adrenal, widespread liver mets involving all segments..MDT outcome revealed metastaic disease. Patient developed leiomyosarcoma of head of pancreas with liver metastasis confirmed by CTAP and was commenced on palliative chemotherapy till may 2022.She had serial CT sacns.CTAP in feb 2023 2023 showed slow progression, with retroperitoneal mass increased, bulky uterus with irregular cavity and enlarged ovaries, slow measurable progression of liver and lung metastasis.She presented on 5/june/2024 with abdominal pain/constipation/generally unwell treated for dehydration /AKI/hypercalcemia/hyponatremia/reaccumulation of ascites, IPC drain was placed for management of ascites and care in community

She had, 7 children, 3 had LFS and 1 had sarcoma removed at age of 3 years.



DISCUSSION

Li-Freumeni Syndrome is diagnosed based on clinical criteria and/or genetic testing for the mutation in TP53 gene. Women have 100% lifetime risk of developing cancer compared to 73% incidence in males. Our patient was diagnosed with osteosarcoma from an early age. Following diagnosis of Li-Freumeni Syndrome, she had annual surveillance with MRI. Despite surveillance, unfortunately she was diagnosed with recurrence of sarcoma.



CONCLUSION

It is important to suspect hereditary disorder in patients who develop early or multiple tumours because early detection of genetic syndromes could influence diagnosis and treatment

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