

Li-Fraumeni Syndrome and the Benefits of Annual Comprehensive Screening: A Pancreatic Adenocarcinoma Case

Li-Fraumeni Syndrome (LFS), an autosomal dominant disorder commonly associated with a *TP53* mutation, located on chromosome 17p13.1, manifests as a variety of malignancies starting from an early age. Due to the rapid onset of changing presentations, comprehensive annual screenings and examinations are essential to prevent the growth of future malignancies.

We present a 65-year-old Caucasian male with a complex medical history including a history of LFS marked by prior colon cancer with resection, prostate cancer with prostatectomy, malignant fibrous histiocytoma resulting in a right hip disarticulation, along with basal cell skin cancer, non-Hodgkin B-cell lymphoma, and laryngeal dysplasia. During his annual MRI screening, he was found to have a 3 mm dilation of the distal portion of the main pancreatic duct without clear visualization of a mass. While it was found to be unchanged since the year prior, the concern for an obstructing intrapapillary mucinous neoplasm warranted gastroenterology consultation. He is a never smoker and denied significant alcohol intake, and experienced no symptoms relevant to pancreatic cancer during this time. Endoscopic ultrasound disclosed a 15 mm hypoechoic lesion in the pancreatic body with well-defined borders. Fine needle aspiration showed atypical cells, with no definitive malignancy. The patient agreed to a diagnostic laparoscopy despite definitive diagnosis, and ultimately underwent an open distal pancreatectomy with splenic flexure mobilization and splenectomy due to bleeding from the splenic vein branch. Final pathology demonstrated a 2.5 cm moderately differentiated ductal adenocarcinoma with invasion of peripancreatic soft tissues, with 0 out of 21 positive lymph nodes and negative margins. A follow-up MRI confirmed no evidence of local recurrence or metastatic disease, and Ca 19-9 has remained stable between 8U/mL to 15U/mL since 2021.

This case highlights the significance of regular screenings for individuals diagnosed with LFS to proactively detect, prevent, and remove potential malignancies. Rigorous screening for this patient allowed him to avoid drastic consequences of pancreatic cancer, which is known to be very aggressive and lethal. We continue to underscore the importance of comprehensive exams for patients with LFS annually, if not bi-annually, to prevent unwanted outcomes.