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Oncological Follow-up with 2-[^{18}F]-FDG PET/CT in Li-Fraumeni Syndrome

Li-Fraumeni Sendromunda 2-[^{18}F]-FDG PET/BT ile Onkolojik Takip

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Abstract

Li-Fraumeni syndrome is a rare disorder caused by abnormalities of the tumor-suppressor protein *P53* gene. We present the case of a 26-years-old female diagnosed with bilateral ductal carcinoma. The genetic panel for breast cancer gene 1 (BRCA1) and BRCA2 mutations was negative and positive heterozygous germline tumor protein *P53* gene mutations, considering Li-Fraumeni syndrome. A 2-[^{18}F]-fluorodeoxyglucose (FDG) positron emission tomography/computed tomography (PET/CT) was used for postsurgical staging to show the right lung hypermetabolic nodule. A lobectomy was accomplished, and histopathology reported pulmonary adenocarcinoma. A year later, oncological follow-up was conducted with 2-[^{18}F]-FDG PET/CT without evidence of abnormalities.

Keyword: Li-Fraumeni syndrome, positron emission tomography, fluorodeoxyglucose ^{18}F

Öz

Li-Fraumeni sendromu, tümör baskılayıcı protein *P53* genindeki anormalliklerin neden olduğu nadir bir hastalıktır. Burada, bilateral duktal karsinom tanısı almış 26 yaşında bir kadın olgu sunulmuştur. Meme kanseri geni 1 *(BRCA1)* ve *BRCA2* mutasyonları için genetik panelin negatif oluşu ve pozitif heterozigot germline tümör proteini *P53* gen mutasyonlarının varlığı, Li-Fraumeni sendromunu düşündürmüştür. Post-op evreleme için yapılan 2-[^{18}F]-florodeoksiglukoz (FDG) pozitron emisyon tomografisi/bilgisayarlı tomografide (PET/BT), sağ akciğerde hipermetabolik nodül saptanmıştır. Lobektomi yapılmış ve histopatolojisi pulmoner adenokarsinom olarak raporlanmıştır. Bir yıl sonra, herhangi bir anormallik kanıtı olmadığı görüldü ve 2-[^{18}F]-FDG PET/BT ile onkolojik takip yapıldı.

Anahtar kelimeler: Li-Fraumeni sendromu, pozitron emisyon tomografisi, florodeoksiglukoz ^{18}F

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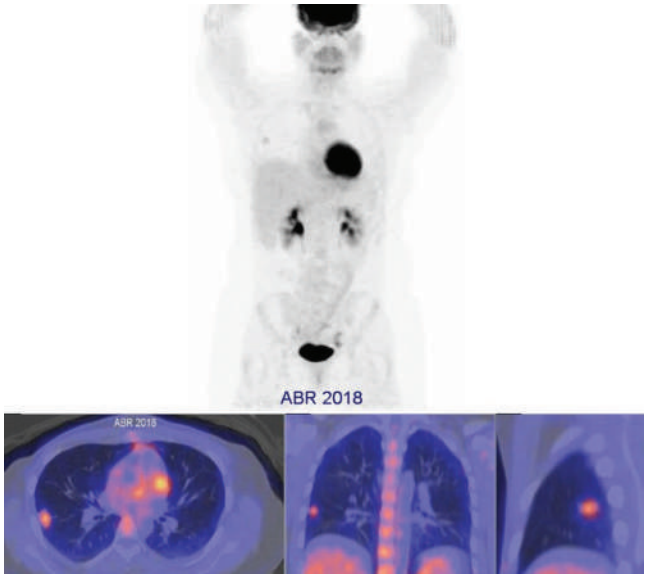

Figure 1. A 26-year-old female diagnosed with bilateral breast carcinoma (invasive ductal carcinoma), hormonal-receptor-negative, and Her-2-Neu-positive. The genetic panel for breast cancer gene 1 (BRCA1) and BRCA2 mutations was negative, with positive heterozygous germline mutations on TP53, considering Li-Fraumeni syndrome. Post-surgical staging with 2-[18F]-fluorodeoxyglucose (FDG) positron emission tomography/computed tomography (PET/CT) reported a hypermetabolic pulmonary module in the superior segment of The right lower lobe.

Li-Fraumeni syndrome is a rare disorder caused by abnormalities of the tumor suppressor protein *P53* gene (TP53) (1), and an autosomal dominant inheritance that affects approximately 400 families worldwide (1,2).

The importance of P53 as an anticancer filter lies in the activation of DNA repair proteins and induction of apoptosis (1,2). When altered, it represents a greater risk of developing neoplasms (90% in females and 73% in males) (1). The most frequent cases in children are adrenocortical carcinoma, osteosarcomas, and rhabdomyosarcomas (1), while those in adults less than 46 years old are sarcomas, brain tumors, breast cancer, leukemia, and bronchoalveolar carcinoma (2,3).

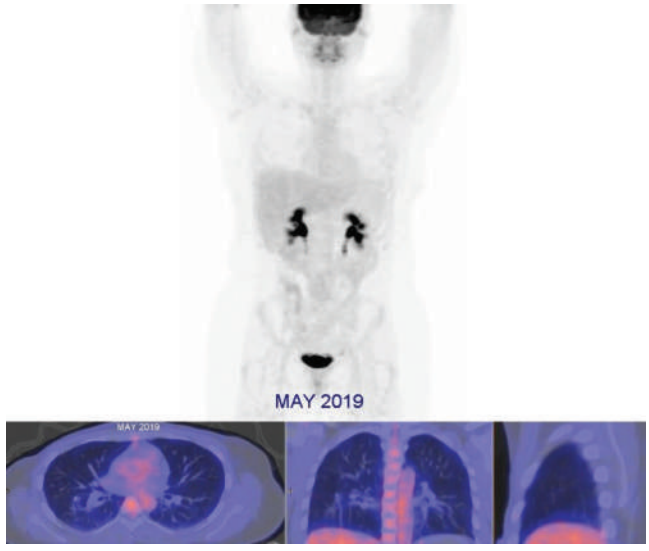

Figure 2. A lobectomy was accomplished with a histopathology report of pulmonary adenocarcinoma with lepidic pattern. A year later, oncological follow-up was indicated with 2-[18F]-FDG PET/CT without evidence of abnormalities.

The diagnostic images play a role in the follow-up. It is recommended to have an ultrasound of the abdomen and pelvis every 3 months in children and annually in adults (2) or an annual total body magnetic resonance (1,2) and annual images of the breasts for females aged 20 years old and above. Ionizing radiation is not recommended, given the hypothetical risk of developing another neoplasm (2).

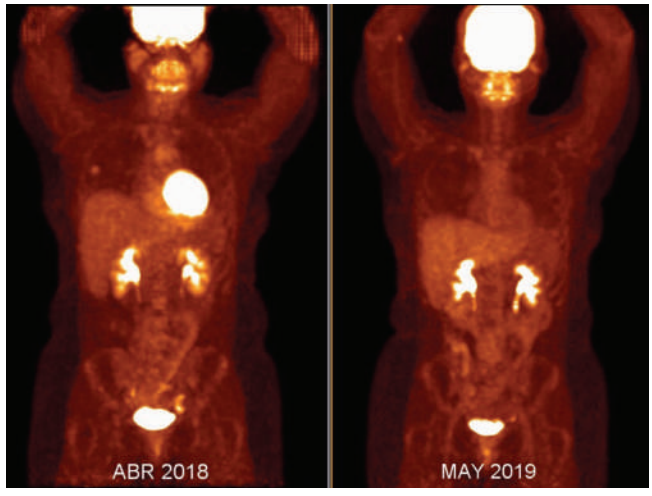

Figure 3. Comparison of 2-[18F]-FDG PET/CT used for cancer follow-up

According to the cohort of 30 patients from Nogueira et al. (3) in Brazil, although the detection of tumor lesions with 2-[18F]-FDG PET/CT in this syndrome can be as low as 20%, this type of diagnostic test helps confirm the presence of lesions, as is demonstrated in our case, and should be considered as a determining factor for the choice of this technique with respect to other modalities because a report of a metabolically lesion requires immediate histopathological confirmation.

Similarly, the replacement of other diagnostic imaging methods and tranquility of the patient and medical staff in charge of a suspected associated neoplasm contribute to a 2-[18F]-FDG PET/CT-negative result during the follow-up.

Ethics

Informed Consent: Informed consent was obtained from patient.

Peer-review: Externally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: M.A.H., I.F.V.G., L.L.V., C.M.A., Concept: M.A.H., I.F.V.G., Design: M.A.H., I.F.V.G., Data Collection or Processing: M.A.H., I.F.V.G., L.L.V., C.M.A., Analysis or Interpretation: M.A.H., I.F.V.G., L.L.V., C.M.A., Literature Search: M.A.H., Writing: M.A.H., I.F.V.G., L.L.V., C.M.A.

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