

Li-Fraumeni Syndrome and Breast Cancer

Sung Hun Kim, MD, Bong Joo Kang, MD, Jeongmin Lee, MD, Ga Eun Park, MD

Department of Radiology, Seoul St. Mary's Hospital, College of Medicine, The Catholic University of Korea, Seoul, Korea

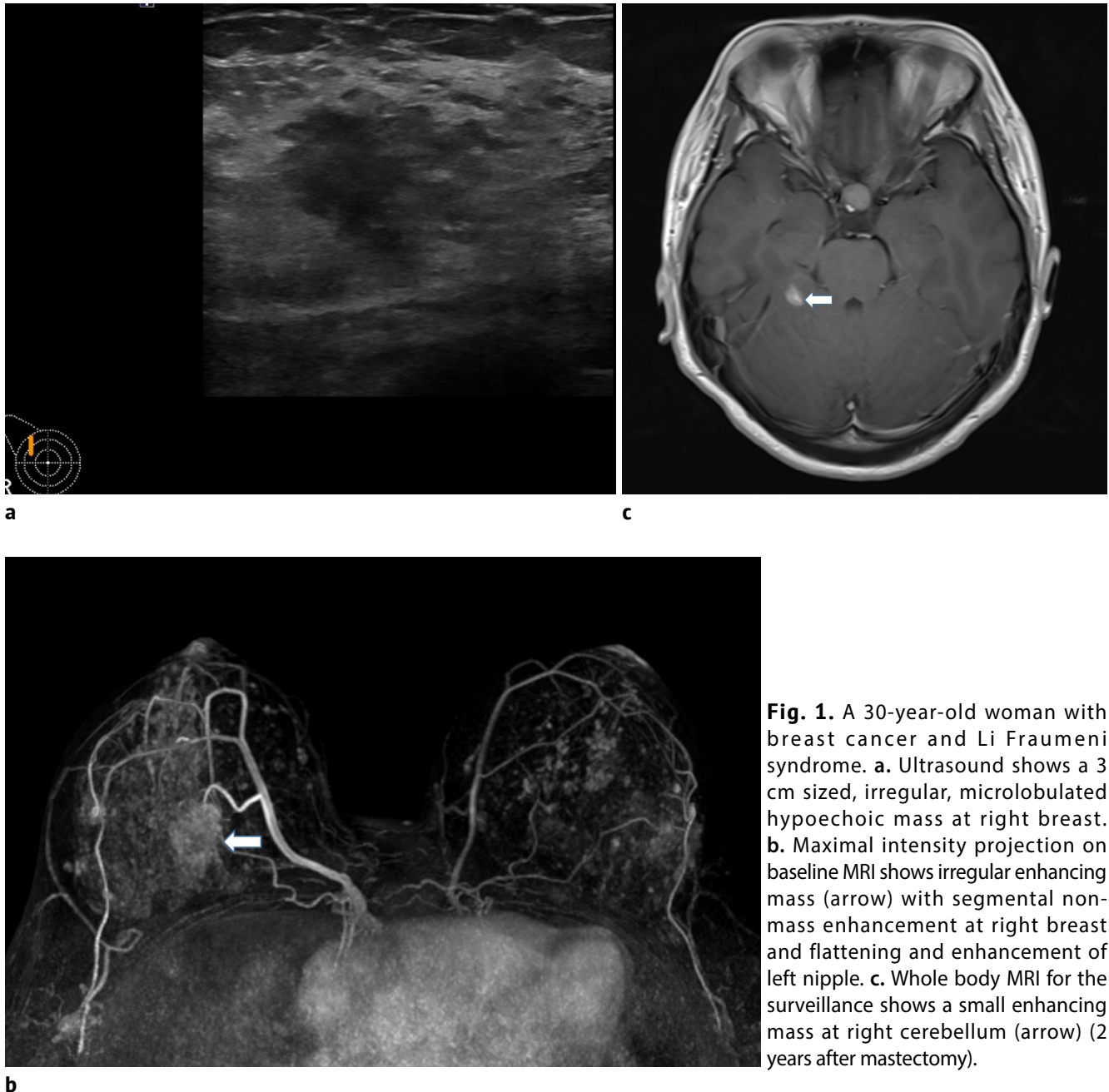


Fig. 1. A 30-year-old woman with breast cancer and Li Fraumeni syndrome. **a.** Ultrasound shows a 3 cm sized, irregular, microlobulated hypoechoic mass at right breast. **b.** Maximal intensity projection on baseline MRI shows irregular enhancing mass (arrow) with segmental non-mass enhancement at right breast and flattening and enhancement of left nipple. **c.** Whole body MRI for the surveillance shows a small enhancing mass at right cerebellum (arrow) (2 years after mastectomy).

Corresponding Author: Sung Hun Kim, M.D.

Department of Radiology, Seoul St. Mary's Hospital, College of Medicine, The Catholic University of Korea, Banpo-daero 222, Seocho-gu, Seoul, 06591, Korea

Tel. +82-2-2258-1455 Fax. +82-2-2258-1457

E-mail: rad-ksh@catholic.ac.kr

A 30-year-old woman presented with a palpable lump on her right breast. US guided core needle biopsy of right mass confirmed the Invasive ductal carcinoma (ER/PR/HER2 -/-/+, Ki-67, 61%) and punch biopsy of left nipple confirmed the Paget disease. US and MRI are presented as Figure 1 (a, b). she had a family history of osteosarcoma and skin cancer in her older brother and lung cancer in her father. Genetic counseling and test was done. TP53 mutation was positive and BRCT mutation was negative. Li Fraumeni syndrome was confirmed.

She completed 6 cycles of Docetaxel/Carboplatin/Trastuzumab/Pertuzumab (TCHP). Bilateral mastectomy was done and surgical pathology showed no remaining cancer in right breast and ductal carcinoma in situ in left breast.

After 2 years, brain metastasis was developed (Figure 1c).

Li-Fraumeni syndrome is a cancer predisposition syndrome caused by a germline mutation of the TP53 gene on chromosome 17p13. It has an autosomal dominant pattern of inheritance with high penetrance. These patients have a very high lifetime

cumulative risk of developing multiple malignancies and have a strong family history of early-onset malignancies. TP53 gene mutations cause the loss of function of p53, leading to downstream events permissive for development of various malignancies throughout life. The Li Fraumeni syndrome component tumors include soft tissue sarcomas, osteosarcoma, premenopausal breast cancer, brain tumors, and adrenal cortical carcinomas (1). The surveillance should include every year clinical examination, WBMRI, breast MRI in females from 20 until 65 years and brain MRI until 50 years (2).

References

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