

A 19-year-old woman presented with a lump in her right breast. She had macrocephaly and milimetric papules on the tongue giving a cobble stone appearance. The breast ultrasonography (US) showed more than twenty circumscribed, oval masses in each breast. An US-guided core biopsy was performed for the palpable masses which were confirmed as juvenile fibroadenomas. After the biopsy, a breast US was done as follow-up every 6 or 12 months for 3 years. During the surveillance period, core biopsies were performed six times, followed by US-guided vacuum-assisted core needle excisions or surgical excisions of lesions with increased sizes. Pathologic results were fibroadenoma, tubular adenoma or atypical ductal hyperplasia (ADH) involving fibroadenoma. At the age of 19 years, the patient underwent a neck US for a palpable mass in the left neck. Several indeterminate nodules were revealed in both thyroid glands. A follow-up US revealed that some of those nodules increased in size. The patient underwent a total thyroidectomy and the pathologic result revealed an invasive follicular carcinoma in the left thyroid and follicular adenoma in the right thyroid (Fig.1A). At the age of 21 years, she visited the hospital due to a swelling of her left cheek. Head and neck CT scan revealed a vascular mass in the left parotid gland extended into the left forehead. An external carotid angiography confirmed the diagnosis of an AVM with feeder vessel arising from the left internal maxillary artery (Fig.1B). A partial embolization decreased the blood flow through the AVM, alleviating the patient's symptom. She was referred to genetic counseling under the suggestion of a Cowden syndrome and the PTEN DNA sequencing test of her blood sample revealed a frameshift mutation, c.301dupA (p.I101NfsX6). At the age of 22 years, the patient presented with a mass with increased size and increased vascularity in the left breast. An US-guided vacuum-assisted core needle excision revealed a ductal carcinoma in situ (DCIS) of non-comedo type and a low grade involving fibroadenoma (Fig.1C). The contrast-enhanced breast MRI for the preoperative evaluation demonstrated multiple, well-circumscribed, enhancing masses in both breasts (Fig.1D). All masses showed high or intermediate signal intensity on T2-weighted images (Fig.1E). Several masses showed an early rapid enhancement with washout kinetic pattern on time-signal intensity curve evaluation, which tends to be associated with malignancy (Fig.1F). A bilateral prophylactic mastectomy with immediate breast reconstruction was performed in view of multiple bilateral breast masses with suspicious kinetic features on breast MRI and a high risk for breast cancer of Cowden syndrome. The surgical histopathology revealed ADH involving tubular adenoma without residual carcinoma in the left breast and multiple tubular adenomas, fibroadenomas and intraductal papillomas in both breasts. The patient had no family history of breast cancer or Cowden syndrome.