

Thyroid metastasis of lobular breast carcinoma[☆]

Metástasis tiroidea de carcinoma lobulillar de mama

The thyroid gland is an uncommon site for distant metastases from extra-cervical tumors. Malignant melanoma and kidney, lung, breast, and gastrointestinal carcinomas are the most common origins of these rare metastases. We report the case of a patient with rapidly progressive thyroid metastasis from a lobular breast carcinoma operated on three years previously.

An 83-year-old female patient underwent surgery in 2007 for a breast tumor by left radical mastectomy and axillary lymphadenectomy. The pathological report confirmed a grade 2, infiltrating lobular carcinoma 5 cm × 4.5 cm × 4.5 cm in size with no involvement of surgical margins and no tumor infiltration in resected lymph nodes (pT2N0). Estrogen receptor expression was 20% and progesterone expression was 5%. Treatment was therefore started with aromatase inhibitors. Subsequent oncological monitoring showed no tumor recurrence.

Three years after surgery, the patient experienced for one month a progressive dyspnea which worsened in a supine position, with no cough, expectoration, or fever. She reported a parallel neck enlargement with cervical tightness. Chest X-rays performed in the emergency room showed no evidence of cardiopulmonary disease, but a tracheal displacement suggesting goiter. Computed tomography (CT) of the neck was therefore performed. The CT scan confirmed the presence of a large multinodular goiter with severe tracheal stenosis (Fig. 1), after which the patient was referred to the endocrinology outpatient clinic.

At the clinic, the patient showed stridor with advanced respiratory difficulty. In a neck examination, a goiter of hard consistency, immobile on swallowing, and with no cervical adenopathies was palpated. The patient reported having had goiter since a young age, but it had never been studied. Thyroid function was normal, with a TSH level of 0.82 μ IU/mL (0.27–4.2) and a free T4 level of 1.28 ng/dL (0.93–1.71). Because of rapid goiter growth and symptomatic tracheal compression, urgent surgery was decided upon. During surgery, only left hemithyroidectomy plus isthmectomy could be performed because the contralateral lobe was found adhered to adjacent structures with gross tracheal infiltration. Dyspnea subsided after surgery. Pathological analysis of the specimen revealed thyroid metastatic infiltration by a lobular breast carcinoma (Fig. 2).

Two weeks after thyroidectomy, the patient attended the emergency room for anuria over the previous 24 h. A CT scan of the abdomen showed a big pelvic mass compressing both ureters with significant bilateral hydronephrosis. Nephrostomy was performed, and the patient was admitted to the oncology ward. In addition to pelvic mass, hepatic and bone metastases were found, with no evidence of local recurrence. The patient died one month after surgery.



Figure 1 CT of the neck showing tracheal stenosis.

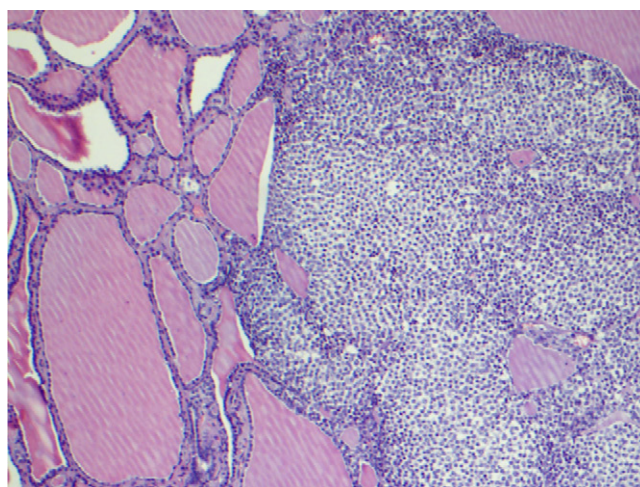


Figure 2 Thyroid follicles surrounded by tumor cells.

Thyroid metastases are uncommon in standard clinical practice, but not in post mortem studies, in which they are found with a frequency ranging from 0.5% in unselected autopsies to 24% in patients with tumor history.¹ In most autopsy series, the primary tumors most commonly causing thyroid metastases are breast and lung carcinomas. In clinical series, however, the most common cause is renal carcinoma.² The rarer cases reported include metastases from a choriocarcinoma.³ In a series of 43 patients with metastatic thyroid involvement, 33% of primary tumors were renal, followed by tumors in the breast (16%), lung (16%), esophagus (9%), and skin (5%).⁴ In another study analyzing 18,105 thyroidectomies and 29,708 cytologies of samples taken by fine needle aspiration, the prevalence rates of metastatic tumors were 0.13% and 0.07% respectively.⁵ Prevalence increased to 1.4% in a series of 1013 patients with thyroid cancers, 14 of which were metastatic.⁶ In all these series, the most prevalent breast cancer was ductal infiltrating carcinoma, while a lobular infiltrating carcinoma was found in the case reported here.⁴

Most patients are diagnosed by fine needle aspiration, although the surgical specimen is sometimes required for

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final diagnosis.⁶ Differential diagnosis between an anaplastic thyroid carcinoma and a secondary tumor is often difficult. The presence of cervical adenopathies, advanced patient age, and neoplastic history may suggest a metastatic tumor, but a positive thyroglobulin test will provide definite diagnosis of an anaplastic carcinoma.⁷ In the case reported, the patient's age, the rapid growth of previously known goiter, and stone-hard consistency suggested anaplastic carcinoma rather than metastasis. Thyroid metastases are usually diagnosed in advanced disease stages and are associated with a life expectancy of only a few months.⁵ Although treatment is palliative in nature, thyroid surgery may improve symptoms and the quality of life in these patients.

In conclusion, when faced with a patient with a history of tumor and a thyroid nodule, a cytological evaluation should be performed on a sample taken by fine needle aspiration to rule out a potential metastatic origin.⁷

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Patient with diabetes and impaired hearing[☆]

Paciente con diabetes e hipoacusia

We report the case of a 34-year-old female patient referred for assessment of diabetes mellitus (DM). Her personal history included bilateral hypoacusis, chronic left otitis media, asymptomatic antero-septal preexcitation with normal coronary arteries, and DM diagnosed at 24 years of age for mild hyperglycemia. Her family history included a maternal grandmother with DM and epilepsy, a mother with DM since 21 years of age and hypoacusis, a maternal uncle with DM and hypoacusis, and a diabetic brother diagnosed at 30 years of age, also with hypoacusis. The patient's mother had died from an episode of hypoglycemia, and her grandmother at 36 years of age during a seizure (Fig. 1).

The patient was initially treated with oral hypoglycemic drugs (metformin and sulfonylureas), and during the previous four years with insulin, with poor glycemic control in all cases. No acute or chronic metabolic complications were reported. Physical examination was normal, with a body mass index of 23 kg/m². The results of supplemental tests were as follows: glycosylated hemoglobin, 7.9%; C-peptide, 0.22 ng/mL (reference range, 1.00–4.00); and

negative anti-glutamic acid decarboxylase (GAD65), anti-pancreatic cell cytoplasm (ICA), anti-tyrosine phosphatase (IA-2), and anti-insulin antibodies. Because of her family history and DM data, a genetic study was performed of mitochondrial DNA (mtDNA) in blood (white blood cells). Using the direct sequencing method of the tRNA^{Leu} gene (transfer ribonucleic acid), the 3243A>G mutation was detected in mtDNA with heteroplasmy levels of approximately 31%. Using a PCR-RFLP method (polymerase chain reaction and restriction fragment length polymorphisms using microfluid detection in a 2100 Bioanalyzer [Los Angeles, USA]), heteroplasmy was 30%. The patient was diagnosed with maternally inherited diabetes and deafness (MIDD), treatment was started with Q10 coenzyme 150 mg/day, and insulin therapy was optimized. No muscle biopsy was performed to study the respiratory chain or for the detection of ragged red fibers because this is an invasive test that would not have provided additional data for diagnosis and treatment. Conductive hypoacusis with a mean threshold at 65 decibels (dB) in the right ear and mixed hypoacusis with a mean perception threshold at 65 dB in the left ear were confirmed as syndrome co-morbidities. She had an estimated 63% hearing impairment in her right ear, an estimated 95% hearing impairment in her left ear, and an estimated 65% impairment in binaural hearing. Eye fundus examination and echocardiogram were normal, as well as microalbuminuria.

It was inferred that her brother, mother, uncle, and grandmother carried the 3243A>G mutation. Her mother and grandmother had died from complications associated

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