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Simultaneous occurrence of thyroblastoma and papillary carcinoma

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Key words: *thyroblastoma; papillary thyroid carcinoma; DICER1 mutations*

A 35-year-old female presented to the Breast and Thyroid Surgery Department with a self-perceived mass on the right side of her neck. Upon neck evaluation, the ultrasound showed a dominant nodule measuring approximately 26 × 18 mm in the right lobe of the thyroid gland, and several abnormal lymph nodes were detected in the right cervical regions II–IV (Fig.1). The cervical computed tomography (CT) scan indicates a strong likelihood of thyroid malignancy. Subsequently, the patient underwent fine-needle aspiration (FNA) of the right lobe of the thyroid gland, which resulted in a suspicious malignancy, and poorly differentiated carcinoma could not be excluded. Afterwards, the patient underwent enlarged radical surgery for a tumour in the right lobe of the thyroid gland. Her other thyroid glands were normal and were saved. Meanwhile, all the excisional fresh specimens were sent to the pathology department for a rapid intraoperative diagnosis and a postoperative haematoxylin-eosin staining (H & E staining) diagnosis.

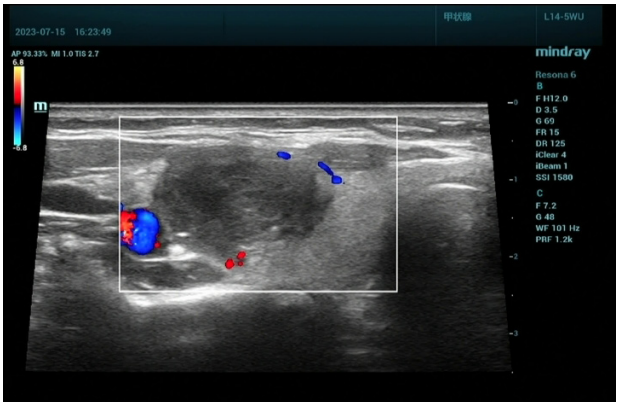

Figure 1. *Basal ultrasound (US) image showing irregular morphology of the nodule, with unclear borders, poorly demarcated anteriorly from the anterior periosteum, and uneven internal echogenicity, with strong echogenicity of a short rod-like pattern seen. The Doppler US suggests that punctate blood flow signals can be seen around the nodule*

On gross inspection, the nodule was greyish-white in colour on a cut section, measuring 2.5 × 2 × 1.5 cm. It was solid and hard, with regional cystic degeneration. Greyish-white and greyish-reddish material was observed inside the capsule. Upon microscopic examination, intraoperative frozen sections and postoperative H&E staining revealed hyperplasia of small round cells with deeply stained nuclei, visible cartilage, and choroid nodules (Fig. 2). Thyroblastoma (ThB) in combination with immunohistochemistry was considered. NGS gene testing suggested a missense mutation in exon 27 of the *DICER1* gene (p. E1813G) and a non-frameshift deletion mutation in exon 22 of the *DICER1* gene (p. S1046-L1058del).

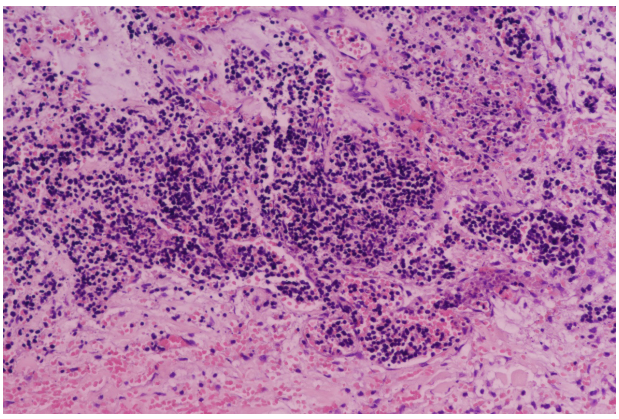

Figure 2. *The histology of thyroblastoma (ThB) in the 35-year-old female is shown. It invades the thyroid tissue, with a mixture of dense and sparse areas. Magnification 200×*

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Papillary thyroid carcinoma (PTC) with a diameter of approximately 1.8 mm was also microscopically identified. BRAF V600E mutation test: No mutation was detected, or it was below the lower limit of detection.

Meanwhile, the specimens of cervical lymph nodes, including central regional ones and region II, III, IV, V, and VI ones of the right neck, were sent to the pathology department for postoperative H & E staining diagnosis. The results indicated that 2 out of 17 lymph nodes on the right side, including areas 2A, 3, 4, and part of 5B, had tumour metastasis, consistent with components of thyroblastoma.

The patient recovered well and was regularly followed up after the operation. Three months post-operation, the patient remained asymptomatic.

Co-occurrence of ThB and PTC in the same patient is a rare phenomenon. To our knowledge, the simultaneous occurrence of 2 tumours of different origins in the thyroid gland is only 1% of thyroid malignancies, and among them, the collision of papillary thyroid carcinoma and medullary thyroid carcinoma is the most frequent [1]. Based on the small number of cases and complex histologic features, there is no clear histologic typing of thyroblastoma, and the case evidence that is currently available is insufficient to generalise and classify the histobiology of thyroblastoma [2]. Thyroblastomas are currently known to be associated with hotspot mutations in the *DICRE1* gene [3], and somatic cellular sources are common. Schultz et al. [4] defined *PTEN* hamartomatosis syndrome (PHTS), DICER1 syndrome, and hereditary smooth muscle neoplasia and renal cell carcinoma (HLRCC) syndrome as a class of multifactorial tumour predisposition syndromes with the potential for the simultaneous combination of several diseases. There is still a wealth of room for development regarding the screening and diagnosis of thyroblastoma.

The biological behaviour and aggressiveness of these cancers are completely different. Accurate diagnosis of this coexistence is very important because the treatment strategies and prognosis of these patients are very different. We herein report a case of thyroblastoma combined with papillary carcinoma. After surgical resection the patient was followed up without recurrence or metastasis. This case illustrates the importance of differential diagnosis and choice of surgical procedure to guide the treatment and diagnosis of this disease.

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Ethics statement
Approval has been obtained from the relevant local ethics committee. Participants have all provided informed consent to participate in the study.

Author contributions
Both authors contributed to the conception, design, and drafting of this submission in its final format.

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Conflict of interest
Both authors have no conflicts of interest relating to this article.

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