

Adult E-Poster

vating mutations in the MEN 1 tumor suppressor gene. Germline mutations in MEN 1 show high penetrance and account for approximately 70–80% of diagnosed MEN 1 cases. Several polymorphisms have also been identified within the MEN 1 gene region, with at least 12 benign variants reported in the general population. While these variants are typically considered non-pathogenic, the c.1621A>G variant has been reported in some studies as potentially contributing to a low-penetrance MEN 1 phenotype in certain carriers.

CASE

We report a case of a 56-year-old female who presented with a 3-month history of painless jaundice and anorexia. She had no personal or family history of malignancy or endocrine disorders. Investigations revealed cholestatic jaundice (bilirubin 89 $\mu\text{mol/L}$), and hypercalcemia (2.88 mmol/L). Imaging showed a solitary 1.7 cm enhancing pancreatic head lesion. Biochemical workup indicated primary hyperparathyroidism (intact-PTH 22.9 pmol/L [normal range; 1.96 – 8.49], calcium/creatinine clearance ratio 0.04). She underwent Whipple's procedure, and histopathology confirmed a 2.1 cm grade 1 pancreatic neuroendocrine tumor (T2N0). Gallium-68 PET/CT showed no distant disease but identified a right lower thyroid lobe focus, suggestive of a parathyroid adenoma. Pituitary MRI was unremarkable.

She met the 2 hallmark features for MEN 1; primary hyperparathyroidism and a pancreatic neuroendocrine tumor, although her presentation occurred later than is typical for MEN 1 cases. The whole exome sequencing showed no pathogenic MEN 1 mutation but detected a c.1621A>G variant that is classified as non-deleterious polymorphism. Interestingly, pathogenic variants in TP53 and BRCA1 were identified without phenotypic expression to date.

CONCLUSION

This case raises questions about the possible pathogenic role of the MEN 1 c.1621A>G variant, especially considering previous reports linking it to low-penetrance MEN 1. Its coexistence with mutations in TP53 and BRCA1 further suggests potential gene-gene interactions or modifier effects, warranting further investigation.

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METASTATIC POORLY DIFFERENTIATED THYROID CANCER: A CASE REPORT

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INTRODUCTION/BACKGROUND

Poorly differentiated thyroid carcinoma (PDTC) is an aggressive subtype of thyroid cancer, representing 0.23%–2.6% of cases.¹ Due to its rarity, the role of thyroglobulin (Tg) monitoring and the effectiveness of radioactive iodine (RAI) ablation have not been clearly defined. Elevated Tg levels in PDTC are associated with higher recurrence suggesting prognostic significance. While RAI avidity is variable, 25% of PDTC cases maintain the ability to uptake iodine. In such cases, RAI ablation significantly improves survival after thyroidectomy.

CASE

A 51-year-old female presented to a private hospital with a one-year history of neck swelling in April 2023. Initial blood investigations, including thyroid function tests were normal and she was advised that no surgical intervention was necessary. There was progressive enlargement of the neck, and by January 2024, she developed airway compression. CT scan showed a large multinodular goiter, an ill-defined hypodense mass in the left thyroid lobe and pulmonary nodules measuring 0.5–2 cm suggesting metastases. She underwent total thyroidectomy and histopathology confirmed PDTC with lymphovascular spread (pT3aNx, high risk).

She was referred to Endocrinology post-thyroidectomy and was started on TSH suppression therapy and given RAI ablation (150 mCi) in April 2024. Baseline stimulated Tg was >500 ng/mL with negative anti-Tg antibodies.

CONCLUSION

This is the first case of PDTC in our center. A multidisciplinary team was important in management. Our case highlights the prognostic role of Tg, the need for more evidence on the efficacy of each treatment modality and the importance of a standardized treatment algorithms for PDTC.