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HEREDITARY VERSUS SPORADIC MEDULLARY THYROID CARCINOMA: A SINGLE TERTIARY CENTRE COHORT STUDY

Qin Zhi Lee[1], Raja Nurazni Raja Azwan[1], Chin Voon Tong[1], Zanariah Hussein[1]
[1]*Endocrinology Unit, Department of Medicine, Hospital Putrajaya*

INTRODUCTION
Medullary thyroid carcinoma (MTC) comprises sporadic and hereditary forms, the latter commonly associated with multiple endocrine neoplasia type 2 (MEN2) which is identifiable through genetic screening of germline RET proto-oncogene. We compared the clinicopathological features and outcomes of hereditary and sporadic MTC in our single centre.

METHODOLOGY
We conducted a retrospective audit of patients with MTC from 2000 to 2026. Patients were classified as either hereditary or sporadic based on genetic testing and/or family history of MTC/MEN2A syndrome. Variables analyzed included age at diagnosis, mode of presentation, pre-operative calcitonin, tumor size, lymph node (LN) involvement, post-operative biochemical cure, repeat surgery and presence of structural residual/recurrent disease.

RESULTS
A total of 57 patients were included (18 hereditary [31.6%], 39 presumed sporadic [68.4%]) with a median follow-up of 7.5 years (IQR 1.5–12.4). Hereditary MTC was diagnosed at a significantly younger age than sporadic MTC (33.0 vs. 44.7 years, $p = 0.010$) where 33% of the cases were diagnosed via screening detection whereas sporadic MTC more commonly presented with symptomatic neck swelling (84.6% vs. 50.0%, $p = 0.008$). There were no significant differences in median pre-operative calcitonin (513.5 vs. 1148.0 pg/mL, $p = 0.101$), tumor size (24.5 vs. 22.5 mm, $p = 0.301$), or LN involvement (41.7% vs. 61.5%, $p = 0.307$) between hereditary and sporadic MTC. Long-term outcomes were also comparable, with no differences in biochemical status, need for repeat surgery or residual/recurrent structural disease.

CONCLUSION
Hereditary MTC presents earlier and is more frequently detected due to screening, whereas sporadic MTC often presents symptomatically. Long-term outcomes are not primarily determined by hereditary status alone. Early access to RET mutation testing with a view to initiating prophylactic treatments rather than post detection surgery may improve disease burden.