



Genotype phenotype correlation in RET positive Medullary Thyroid Cancer: A single institution experience

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Introduction

- Demographics, presentation and prognosis differ between MTC subtypes based on RET mutation
- Screening is important for early diagnosis and prophylactic thyroidectomy

Objective

- To analyze mutation profile in RET positive MTCs for differences in demographics, presentation and outcomes

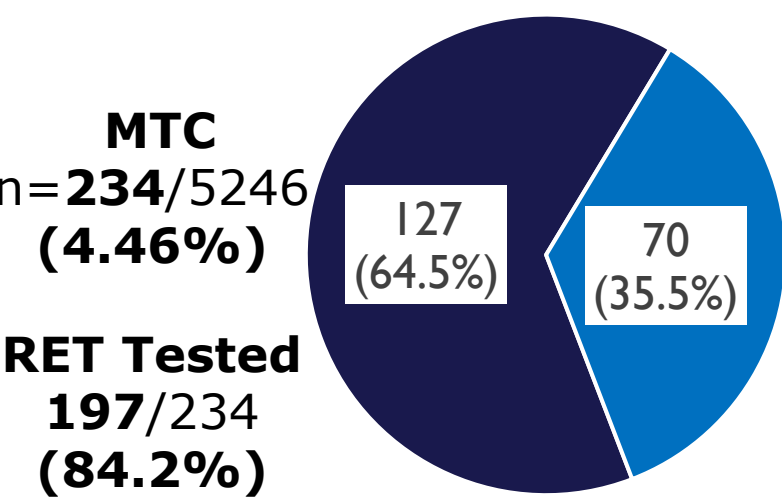
Conclusion

- 35.5% Hereditary MTC - 634 - most common mutation (65.7%)
- Occurrence of pheochromocytoma and HPT in 634 consistent with literature
- 804 mutation had higher basal calcitonin and nodal recurrence

Methodology

- Retrospective study- single institution
- January 2002 - December 2022

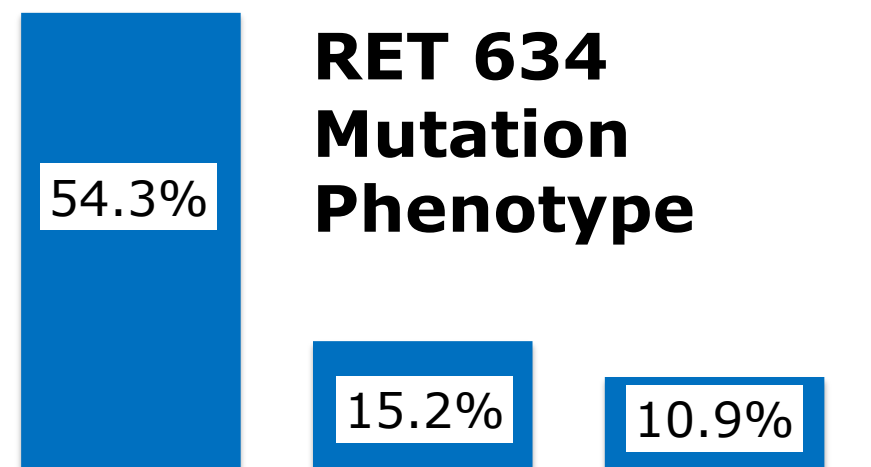
Results



Demographics	634 n=46 (65.7%)	804 n=12 (17.1%)	618 n=4 (5.7%)	918 n=4 (5.7%)	790 n=2 (2.9%)	620 n=2 (2.9%)
Median age (yrs)(range)	27 (9-68)	43 (22-64)	37 (18-39)	16.5 (8-37)	43 (29-57)	37 (33-41)
Gender(M:F)	20:26	6:6	2:2	2:2	0:2	2:0

Presentation

- Goitre with LN most common presentation
- 21.7% (10/46) 643 mutation and 25% (1/4) 918 mutation presented with pheochromocytoma
- 33/64 individuals of 37 index families screened tested positive for RET mutation
- 5 underwent prophylactic thyroidectomy (634-2,804-2,618-1)



- HPT seen only in 634 mutation

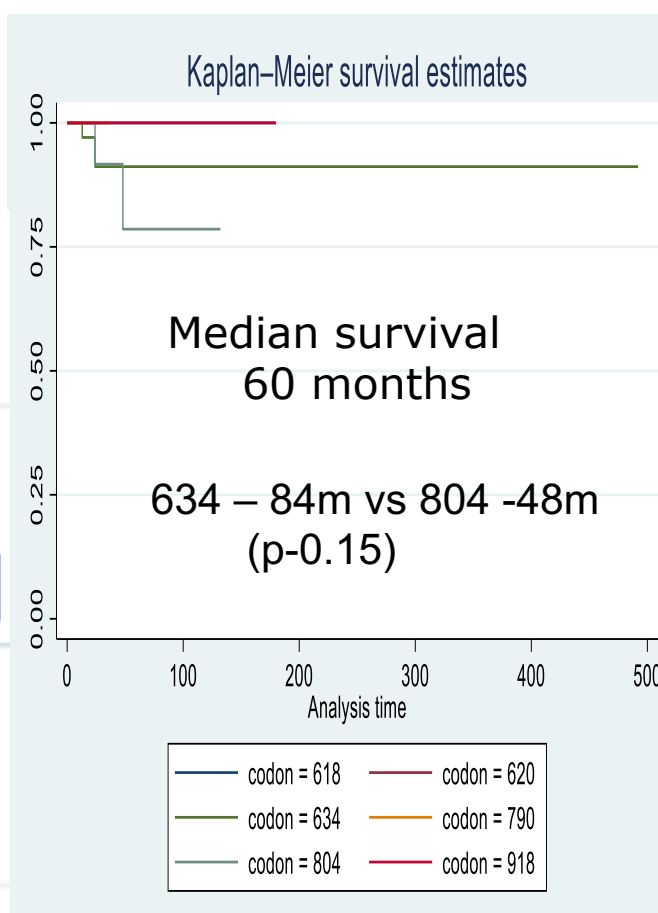
Pheochromocytoma

	Incidence	Synchronous	MTC before Pheo	Pheo before MTC
634	54.3% n=25/46	68% (mean age 35.7yrs)	28% (mean duration 15.6 yrs)	4% (duration 10 yrs)
918	25% n=1/4	100% (37 yrs)	-	-

Outcomes	634	804	618	620	790	918
Nodal mets at presentation	50% (23)	66.6% (8)	50% (2)	50% (1)	50% (1)	75% (3)
Metastasis at presentation	17.4% (8)	8.3% (1)	25% (1)	-	-	-
Nodal recurrence	17.4% (8)	25% (3)	25% (1)	50% (1)	p=0.021 634 vs 804	25% (1)
Metastasis on follow up	8.6% (4)	8.3% (1)	-	50% (1)	-	-

	634	804	618	620	790	918
Basal median Calcitonin (pg/ml)	1969	2000	10645	9874	3604	10110
						p<0.05
Biochemical cure(< 50pg/ml)	34.8%	33.3%	50%	0	50%	0
						p=0.52

Overall survival



Disease free survival

