Early Medullary thyroid Carcinoma (MTC) in an infant with Multiple endocrine neoplasia type 2B (MEN2B, RETS Mutation codon 891)

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INTRODUCTION

An age-related progression from C-cell hyperplasia to medullary thyroid carcinoma (MTC) is associated with various germ-line mutations in the rearranged during transfection (RET) proto-oncogene that could be used to identify the optimal time for prophylactic surgery.

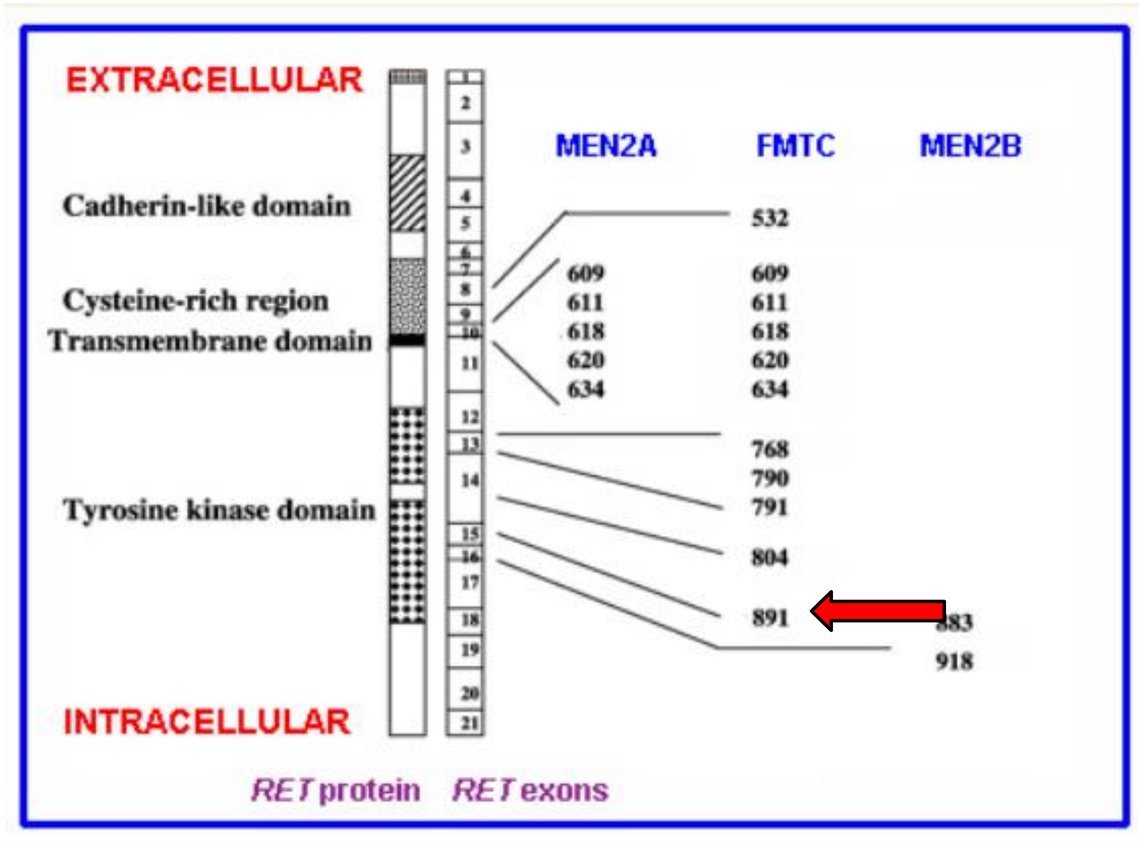
In 207 patients from 145 families there was a significant age-related progression from C-cell hyperplasia to MTC. Thus, early diagnosis and prevention are particularly crucial.

Case Study

We describe a 24 months Qatari male who was diagnosed with medullary thyroid cancer. At age 22 months he was screened for RET proto-oncogene because his mother found had MEN2B proven germline RET mutation.

Physical examination showed stable vital signs, coarse facial features, enlarged lips and thickened gum, disproportionate short stature with decrease U/L ratio and height SDSD = -3.69. There was no goitre or cervical lymphadenopathy. No skin pigmentation. No palpable abdominal mass.

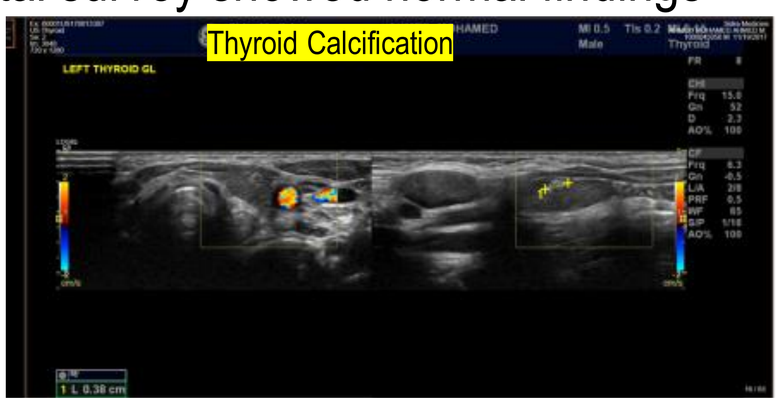
His genetic screening showed a germline RETS mutation in codon 891. Mother was anxious to minimize the future risk and therefore wish to undertake thyroidectomy as early as possible .Therefore, screening for MTC at age of 24 months was elected. Unfortunately the screening showed positive findings for MTC.



Thyroid U/S revealed a small foci of globular dystrophic calcification in the left lobe concerning for an early medullary cell carcinoma. Abdominal U/S was normal.

Labs showed very high serum level of basal Calcitonin but normal CEA, PTH and Free T4 and TSH levels.

Skeletal survey showed normal findings



Management

The patient has been considered for early total thyroidectomy.

Discussion

Patients with MEN2B and RETS mutation in codon 891mutation have moderate gene penetrance and the recommended age to begin annual screening for MTC is 5 years and the suggested timing of prophylactic thyroidectomy is childhood or young adulthood.

This infant, with this mutation, had evident MTC at the age of 2 years that raises concern about late screening for these patients.

Conclusions and recommendations

We report for the first time the occurrence of MTC at an early age (2 years) in an asymptomatic toddler with MEN2B, with RETS mutation in codon 891.

Genetic and biochemical screening is mandatory for appropriate individual timing of elective total thyroidectomy before age of 5 years.

Delayed screening may miss cases of MTC in these children. Therefore timing of total thyroidectomy could be individualized based on codon-specific prognosis.