Meta-analysis of children with Multiple Endocrine Neoplasia (MEN) Type 2A from 1995-2014: Impact of RET mutation screening on age at thyroidectomy and frequency of metastatic disease

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The authors have nothing to disclose

Take home message
Age at total thyroidectomy and metastatic medullary thyroid cancer rate decreased overall and in high risk mutation carriers. However, still 70% of high risk mutation carriers are thyroidectomised beyond recommended age of 5 years.

Background

Medullary thyroid cancer (MTC) in MEN 2A is caused by mutations in RET. Guidelines (2001/2009/2015) recommend prophylactic total thyroidectomy (TT) based on mutation specific risk levels (ATA 2015: high/moderate), Figure 1.

The aim of this study was to analyse changes of age at TT, frequency of metastatic MTC (MMTC), and frequency of TT according to guidelines since introduction of RET testing in 1995.

Methods

Patients in publications from 1995-2014 aged 0-20 years with individual information on age at TT, histology, and RET-mutation, if available, were included. Patients were grouped according to publication year: groups A=1995-1999, B=2000-2004, C=2005-2009, and D=2010-2014. Median age at TT, rate of MMTC, and rate of TT according guidelines were compared in the four groups.

Results

In 110 publications 604 patients were identified.

Figure 1: Median age at TT, frequency of MMTC and frequency of TT according to guidelines since introduction of RET testing in 1995.

Figure 2: The median age at TT did not differ in the four groups (11, 11, 10, 8 years).

Figure 3: The median age at TT did not differ between groups (9, 11, 12, 10 years).

Figure 4: The median age at TT decreased significantly in the four groups from 13, 11, 9 to 7 years (P<0.01).

Figure 5: Significant decrease of MMTC in group A vs. D (27% vs. 8%, P<0.01).

Figure 6: Children thyroidectomised before 5 years remained low (15%, 22%, 20%, 30%) despite genetic testing.