A case of young girl with high risk RET mutation successfully diagnosed as medullary thyroid carcinoma in very early stage.

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Introduction

- Medullary thyroid carcinoma (MTC) in Multiple Endocrine Neoplasia type 2 (MEN2) shows near complete penetration.
- Presentation and prognosis of MTC highly depend on RET proto-oncogene mutation.

American Thyroid Association (ATA) guidelines recommend prophylactic surgery for the patients of MEN2 with high risk RET mutations.

Case Presentation

age 5 month-old
- The patient’s mother was diagnosed as MEN2A.
- Gene analysis ↓
- RET codon C634G mutation was detected (figure1)

Annual calcium infusion test to monitor calcitonin level and neck ultrasonography

8 year-old
- Result of the test became positive (table1) but ultrasonography finding was negative
- Stage I MTC (T1, N0, M0) was confirmed (figure2)

Table1 Stimulated calcitonin level by calcium infusion test

<table>
<thead>
<tr>
<th>age</th>
<th>serum calcitonin level</th>
<th>reference value</th>
</tr>
</thead>
<tbody>
<tr>
<td>0'</td>
<td>11.3</td>
<td>6.40 pg/mL (ECLIA)</td>
</tr>
<tr>
<td>2'</td>
<td>333</td>
<td></td>
</tr>
<tr>
<td>5'</td>
<td>248</td>
<td></td>
</tr>
</tbody>
</table>

- Both basal and stimulated calcitonin levels elevated at 8 year-old.
- Elevation of basal calcitonin level was mild.

Discussion

Phenotype of MTC varies even among family members with the same genetic defect.

Prophylactic thyroidectomy has some debatable problems.

- Higher risk of surgical complications at younger age
- Hormone replacement therapy for longer time
- Public health insurance coverage

Instead of prophylactic treatment...

- Detecting early stage MTC could be another major follow-up method.
- The timing of thyroidectomy should be decided for each individual case.

How?

- Serum calcitonin level is a sensitive marker for detecting MTC.
- Deciding right time for thyroidectomy by serum calcitonin could relieve above mentioned problems.

Conclusions

We recommend that annual calcium infusion test should be performed for RET gene mutation carriers to detect MTC in early stage and perform thyroidectomy without delay.

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Reference