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Case Presentation
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Non-Syndromic Familial Papillary Thyroid Carcinoma: Early Onset and Multifocal Presentation in a mother and Daughter – A Case Report and Updated Literature Review

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Abstract

Introduction: Familial non-medullary thyroid carcinoma accounts for 3-7% of differentiated thyroid cancers. It is defined by the occurrence of papillary thyroid carcinoma in at least two first-degree relatives in the absence of a known hereditary syndrome.

Case Presentation: We report the case of a 56-year-old mother and her 35-year-old daughter, both diagnosed with papillary thyroid carcinoma discovered in EU-TIRADS V classified nodules. Histopathological examination revealed multifocal follicular-variant papillary microcarcinomas in the mother and an infiltrative papillary microcarcinoma in the daughter.

Conclusion: Non-syndromic familial papillary thyroid carcinoma may present with earlier onset and multifocal disease. Careful family history assessment and targeted screening of first-degree relatives may allow diagnosis at an early stage.

Keywords: Familial Papillary Carcinoma; Non-Medullary Thyroid Cancer; Multifocality; Family Screening

Introduction

Papillary Thyroid Carcinoma (PTC) accounts for more than 80% of differentiated thyroid cancers, with a continuously increasing global incidence [1].

Familial aggregation is observed in 3-7% of cases [2]. Familial Non-Medullary Thyroid Carcinoma (FNMTC) is defined by the occurrence of thyroid cancer derived from follicular cells in at least two first-degree relatives, in the absence of an identified hereditary syndrome [3].

Several genetic syndromes may be associated with differentiated thyroid carcinoma, including Cowden syndrome, familial adenomatous polyposis, Carney Complex, and Werner syndrome [3].

Non-syndromic FNMTC is suspected to follow an autosomal dominant inheritance pattern with incomplete penetrance [4]. Some studies suggest a more aggressive presentation compared to sporadic forms, with higher rates of multifocality and increased lymph node involvement [5, 6].

We report a mother–daughter case illustrating this entity.

Case Presentation

Case 1

A 56-year-old woman with no known family history at the time of diagnosis.

Neck ultrasound revealed an 18-mm hypoechoic nodule in the left thyroid lobe classified as EU-TIRADS V. Total thyroidectomy was performed.

Histopathological examination revealed:

- Three papillary microcarcinomas
- Follicular variant
- Stage: pT2NxMx

Postoperative course was uneventful.

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Case 2

Her 35-year-old daughter underwent screening following her mother's diagnosis.

Ultrasound revealed a 13.5-mm isthmic nodule classified as EU-TIRADS V.

Total thyroidectomy was performed.

Histology concluded:

- Infiltrative papillary microcarcinoma
- Stage: pT1aNxMx

Clinical outcome was favorable.

Discussion

Definition and Diagnostic Threshold

The diagnosis of FNMTC is based on the presence of at least two affected first-degree relatives [2].

However, true hereditary transmission is considered more likely when three or more family members are involved [7].

Genetic Basis

Unlike medullary thyroid carcinoma associated with RET mutations, no single causative gene has been identified in FNMTC.

Susceptibility loci have been proposed (2q21, 19p13), suggesting polygenic inheritance [4, 8].

Somatic mutations such as BRAF V600E, common in sporadic forms, are also found in familial cases without a clearly distinctive pattern [9].

Clinical Aggressiveness

Several meta-analyses have shown that familial PTC is associated with:

- Higher multifocality
- Increased bilaterality
- More frequent lymph node involvement
- Higher recurrence risk [5, 6]

However, no significant difference in disease-specific mortality has been demonstrated [10].

Therapeutic Implications

According to the 2015 American Thyroid Association guidelines [11], management of differentiated thyroid cancer is based on risk stratification.

Although no specific recommendations are dedicated to familial forms, some authors suggest a more extensive approach including total thyroidectomy and closer follow-up [5].

Family Screening

The benefit of systematic ultrasound screening remains debated.

Charkes estimated that when only two family members are affected, a significant proportion may represent coincidental aggregation [7].

Nevertheless, several recent reviews support targeted screening of first-degree relatives, particularly in cases of early onset [8].

In our case, screening allowed diagnosis at the microcarcinoma stage.

Conclusion

Non-syndromic familial papillary thyroid carcinoma remains a rare entity.

It may present with earlier onset and multifocal disease. Systematic family history assessment and individualized screening strategies could improve early diagnosis.

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