

# MEDULLARY THYROID CANCER

## A comparative study between sporadic and familial cases in a reference care center in Spain

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### BACKGROUND

Hereditary Medullary Thyroid Cancer (MTC) accounts for 20-30% of cases, and it is related to RET germline mutations and multiple endocrine neoplasia type 2 (MEN2). Our aim was to analyze differences in diagnosis and outcome between sporadic and familial cases of MTC in a cohort of patients.

### MATERIALS & METHODS

Retrospective, unicentric cohort study that included all genotyped patients with MTC (n=48) diagnosed at Hospital Clínico San Carlos (Madrid) between 1984-2013; 42% were germline mutation carriers (45% moderate risk [category MOD], 45% high risk [category H], 10% highest risk [category HST]). Median follow-up was 61 (IQR 22-104) months. A comparative analysis was performed using the Student's t-test, the chi-squared test and the log-rank test.

### RESULTS

Mean age at diagnosis was 37.6 (SD 20.4) years in familial cases, and 62.5 (12.2) in sporadic cases (p<.001); local or distant metastases were present in 44% of familial cases and 61% of sporadic cases (p=NS). Most of the familial cases (55%) were asymptomatic and diagnosed after genetic screening.

Six months after total thyroidectomy, 24% of sporadic cases and no familial cases showed progression. During follow-up, 27% of sporadic cases and 6% of familial cases developed distant metastases (median time 30 [8-56] months).

Progression-free survival (PFS) and distant metastases-free survival (DMFS) were longer in familial cases (p=NS). Attributable mortality due to MTC was 23% in sporadic cases and 0% in familial cases; overall survival (OS) was longer in familial cases (p=.03).

Fig. 1. Age at diagnosis

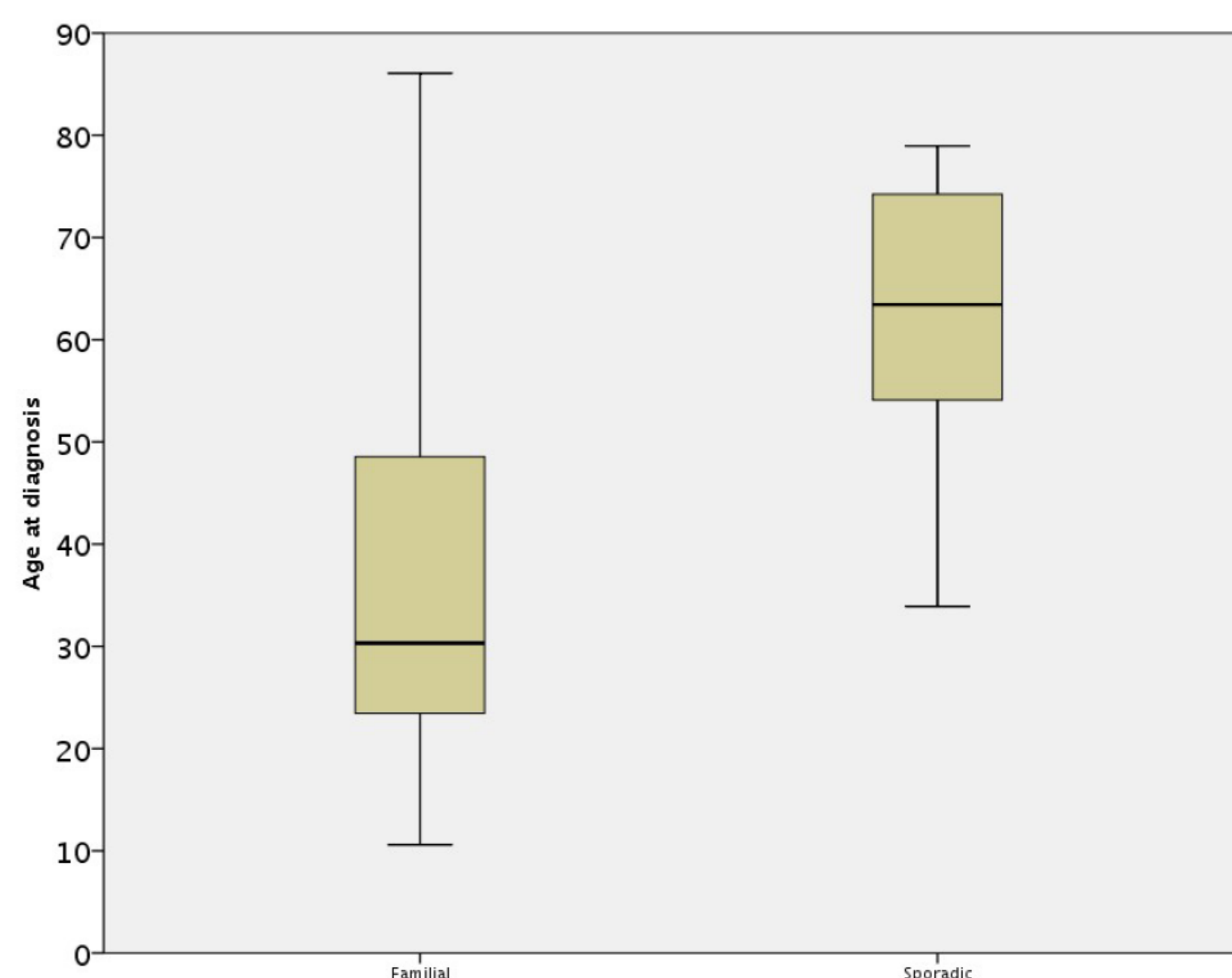


Fig. 2. Progression-free survival

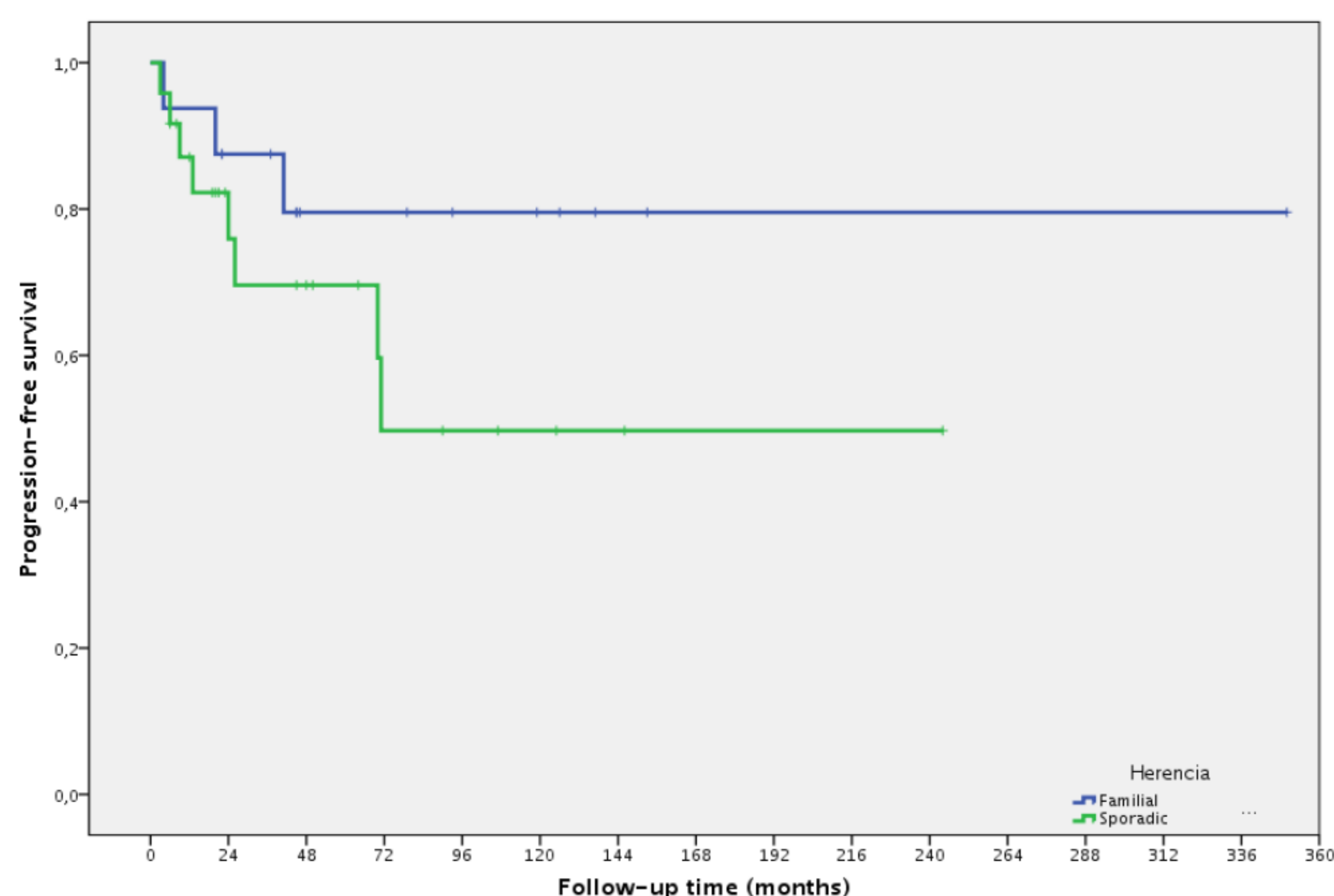
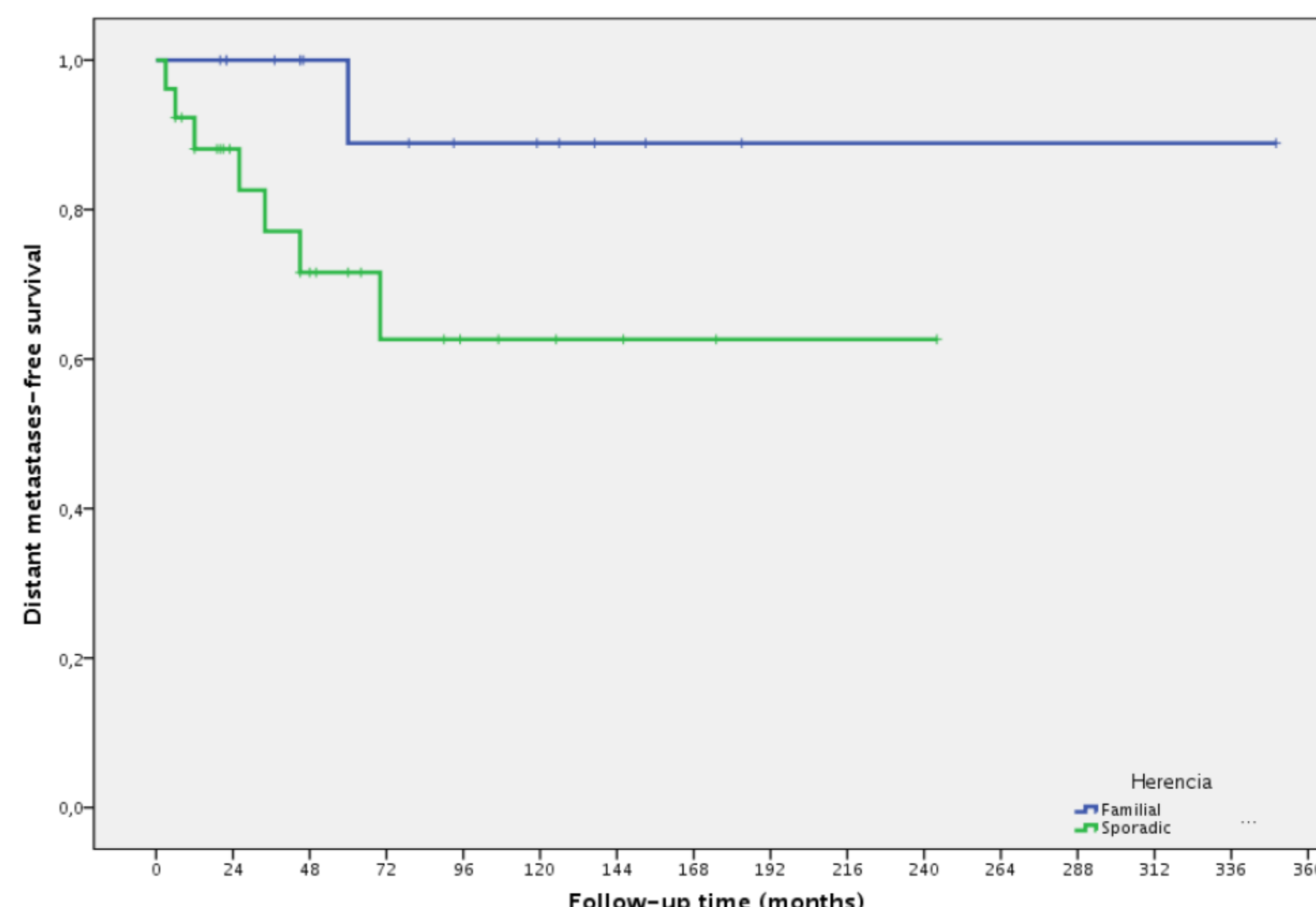


Fig. 3. Distant metastases-free survival



Age at diagnosis was significantly lower in familial cases of MTC. Genetic testing allowed for early diagnosis in asymptomatic mutation carriers, therefore familial cases had a better outcome and a longer survival.

