

PHAEOCHROMOCYTOMAS AND PARAGANGLIOMAS

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

BACKGROUND

Hereditary Phaeochromocytoma (PCC) and Paraganglioma (PGL) accounts for 30-35% of cases, and it is related to germline mutations in 12 genes with two phenotypes: pseudohypoxic and MAP-kinase. Our aim was to analyze differences in diagnosis and outcome between sporadic and familial cases of PCC/PGL in a cohort of patients.

MATERIALS & METHODS

Retrospective, unicentric cohort study that included all genotyped patients (n=36, 27 with PCC and 9 with PGL) diagnosed at Hospital Clínico San Carlos (Madrid) between 1984-2013; 33% were germline mutation carriers (25% pseudohypoxic [PH] phenotype, 75% MAP-kinase [MAPK] phenotype). Median follow-up was 98 (IQR 56-141) months. A comparative analysis was performed using the Mann-Whitney U test, the chi-squared test and the log-rank test.

RESULTS

Median age at diagnosis was 35.3 (21.9-45.1) years in familial cases, and 64.4 (48.2-73.0) in sporadic cases (p=.02). Most of the sporadic cases were diagnosed incidentally (61% vs 17%, p=.01). Multifocality was more frequent in familial cases (50% vs 8%, p=.01).

Recurrent disease after surgery was present in 40% of familial cases and in no sporadic cases (p=.006, median time 73 [30-85] months); it was more frequent in the PH group (67%) than in the MAPK group (17%). Progression-free survival (PFS) was longer in sporadic cases (p=.009).

Fig. 1. Age at diagnosis

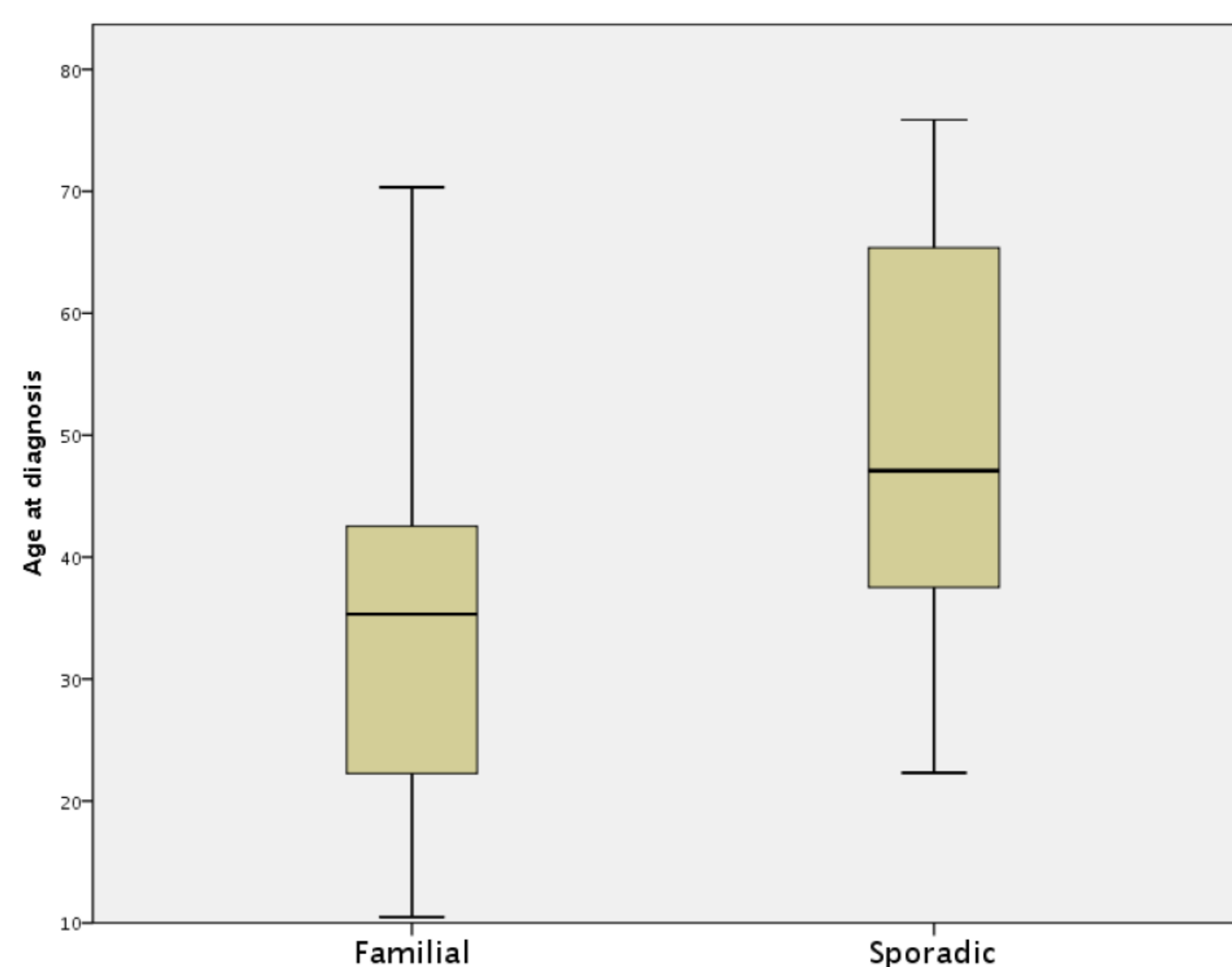


Fig. 2. Genotypes of familial cases

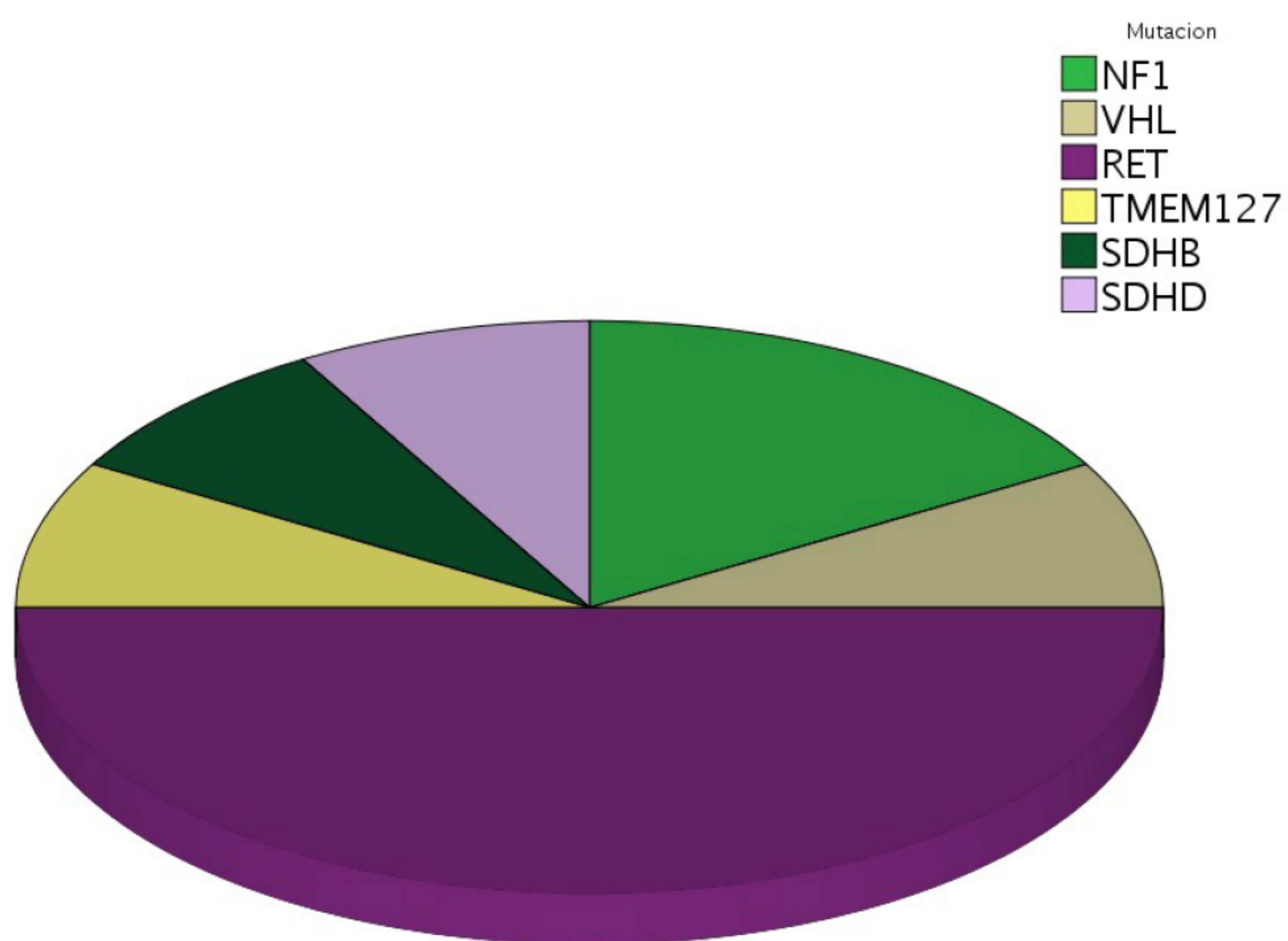
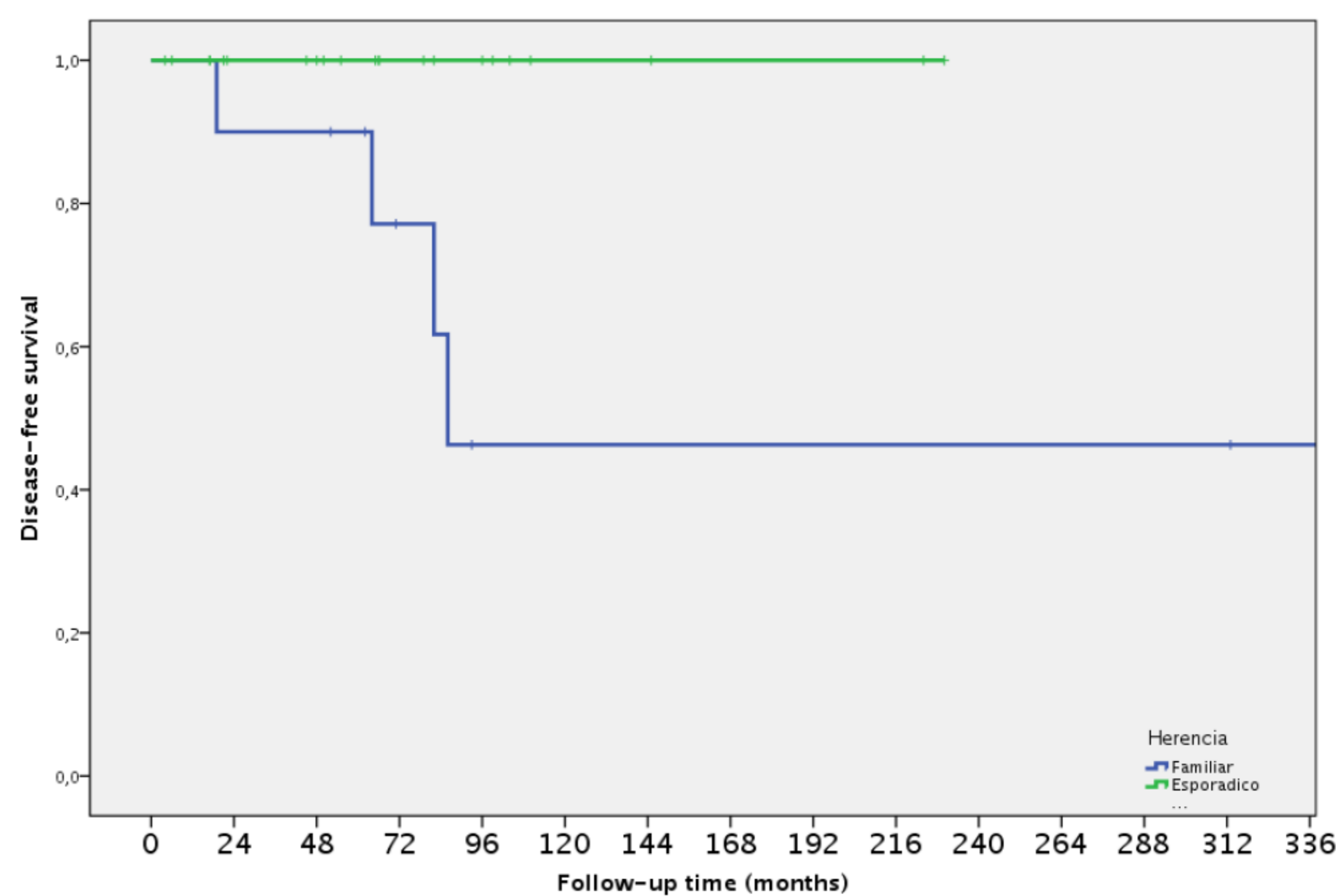


Fig. 3. Long term relapse comparison.



Age at diagnosis was significantly lower in familial cases of PCC/PGL. Malignant behaviour and multifocality were associated with familial cases. Genetic testing allowed for early diagnosis in asymptomatic mutation carriers, although sporadic cases had significantly longer PFS.

