Prevalence of germline mutations in pancreatic neuroendocrine tumors

ENETS EUROPEAN NEUROENDOCRINE TUMOR SOCIETY

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INTRODUCTION

Approximately 10% of pancreatic neuroendocrine tumors (PanNETs) develop due to inherited syndromes. However, generally genetic counseling and testing is not performed routinely and little date accumulated on the prevalence of PanNETs associated with the presence of germline mutations.

METHODS

Two groups of patients were formed.

- Group A – patients with asymptomatic PanNETs T1 under observation without surgical or systemic treatment (n=50).
- Group B – patients with locally advanced or metastatic PanNETs (n=16).

The patients underwent genetic testing using the new generation whole-genome sequencing method (NGS EVOGEN-GENOME panel).

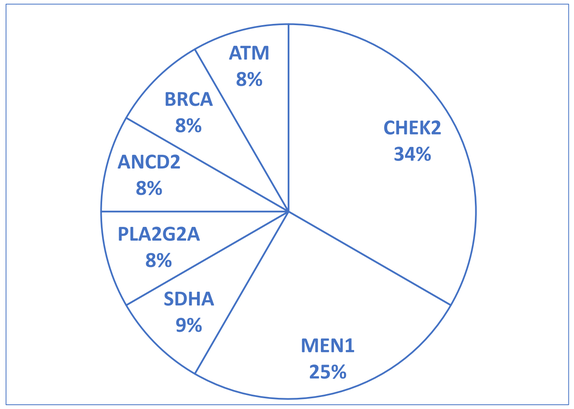
CONCLUSION

Genetically determined pancreatic NETs are more common than it is believed. Dominance of MEN1 among germline mutations was not observed in our work. Among small non-functioning tumors, a mutation in CHEK2 gene occurred in a third of cases, and in MEN1 - in a quarter. At the same time this mutation was not detected in the group of locally advanced and metastatic tumors. **It is possible the mutation in CHEK2 gene is a previously unaccounted factor in the development of pancreatic NETs or the favorable course of the disease**, what requires further research.

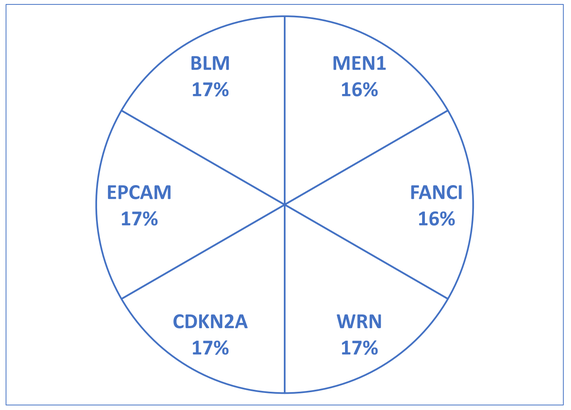
RESULTS

- In group A a germinal mutation was identified in 12 patients (24.0%), what exceeds the literature data.
- A mutation in the CHEK2 gene was found in 4 patients (8%), MEN1 - in 3 patients (6%).
- Also 5 mutations were observed: SDHA, PLA2G2A, ANCD2, BRCA, ATM.
- In group B germinal mutations were found in 6 patients (37.5%) in the FANCI, WRN, CDKN2A, EPCAM, BLM, and MEN1 genes.

MUTATIONS IN GROUP A



MUTATIONS IN GROUP B



FUTURE DIRECTION FOR RESEARCH

Further genetic tests are required to determine correlation between PanNET genesis, germinal mutations and clinical prognosis.