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Multiple Endocrine Neoplasia type 1 in Poland: a two center experience.

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Abstract

INTRODUCTION: Multiple Endocrine Neoplasia type 1 (MEN1) has been causing problems for clinicians since it was described in 1954 by Wermer. Not only its rarity, but also variable clinical manifestations and no genotype-phenotype correlation make it hard to establish evidence-based guidelines for management of this syndrome. Nationwide registers and population-based researches are the best means to improve knowledge about this rare disease. By now, there was no example of such research in the Polish population of MEN1 patients.

MATERIALS AND METHODS: We performed a retrospective analysis of clinical and genetic data of patients diagnosed with MEN1 syndrome and followed-up in two polish referral centers in years 1994-2018.

RESULTS: We analysed 79 patients out of which majority were women. Mean age of population was 43 years, mean age at MEN1 diagnosis was 37.95 years and a mean interval from initial symptoms to MEN1 diagnosis was 6.93 years. Primary hyperparathyroidism (PHP), gastroenteropancreatic neuroendocrine tumor (GEP-NET) and pituitary adenoma (PA) developed in 90%, 52% and 47% of patients respectively. The dominance of insulinoma with low prevalence of gastrinoma is the most vivid difference, when compared to previously described populations. Moreover, we found a 3.5 times higher risk of developing a pituitary tumor in patients with a frameshift mutation with STOP codon of MEN1 gene.

CONCLUSIONS: Polish population of patients with MEN1 is different than previously described European and Asian populations primarily in prevalence of functional NETs. Frameshift mutation with STOP codon of MEN1 gene increase significantly risk of PA. Further studies, with a larger

cohort of patients, are needed to fully describe the Polish population and improve diagnosis and management of the syndrome.

KEYWORDS: MEN1; Multiple Endocrine Neoplasia type 1; genotype-phenotype correlation.; polish population

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