Germline mutations in multiple endocrine neoplasia; a summary of Thai case reports

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Multiple endocrine neoplasia (MEN) is an important medical disorder characterized by several endocrine gland abnormalities. There are various types of MEN and some types have parathyroid gland involvement. The MEN1 and MEN2A are the two main types of MEN that parathyroid gland abnormalities are observable (1). The underlying genetic factors of MEN is an interesting issue. There are few data on this aspect. In Thailand, a tropical country in Southeast Asia, the MEN is sporadically reported and the examination of underlying genetic mutations in cases with MEN is little mentioned. In this specific short study, the authors summarize and review the data reported in publications from Thailand on MEN1 (Wermer’s syndrome) and MEN2A (Sipple syndrome) (1). According to the literature search in international databases (PubMed and SCOPUS) and local Thai referencing databases (Thai Index Medicus and TCI), there are only four reports on genetic mutations seen in Thai patients with MEN1 and MEN2A. According to the collected data, there are 4 reports on 2 MEN1 and 9 MEN2A cases (2-5). Focusing on germline mutations, the heterozygous for IVS6 + 1G to A ss seen in one case and the deletion mutation at exon 10 (1793delG) is seen in other case. Focusing on MEN2A cases, missed sense mutation mutations are identified at codon 634 exon 11 p.Cys634Tyr, p.Cys634Arg and p.Cys634Trp in 5, 2 and one cases and the left other case has heterozygous for 1900T>C (C634R). Here, it can show that various underlying genetic mutations contributing to MEN1 and MEN2A exist in Thailand.

Authors’ contribution
SY and VW wrote the manuscript equally.

Implication for health policy/practice/research/medical education
Various underlying genetic mutations contributing to MEN1 and MEN2A are exist in Thailand.

Keywords: Parathyroid glands, Germline mutations, Multiple endocrine neoplasia

Conflicts of interest
The authors declare no conflicting interest.

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References

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