

## عنوان مقاله:

Calcium-sensing receptor SE haplotype in patients with primary hyperparathyroidism; expected magnitude of lowered serum calcium level

## محل انتشار:

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## خلاصه مقاله:

In primary hyperparathyroidism (PHT), the problem of calcium metabolism is an important problem (۱). The underlying molecular mechanism is very interesting. The calcium-sensing receptor (CSR) is widely mentioned for its effect on blood calcium control (۲). The genetic polymorphism of CSR is related to the different phenotypic manifestation of blood calcium level in patients with PHT (۳). The CSR AQ haplotype in patients with PHT is found to relate to the lower blood calcium level (۴,۵). In this short report, the authors assess the molecular change due to the mutated type SE haplotype and discussed on the magnitude of lowered serum calcium level. This work is a quantum chemistry analysis using mathematical modeling study using the same technique as previously published by Joob and Wiwanitkit (۶). The basic calculation for the molecular mass in the wild and mutated types of CSR polymorphisms was done. The focused studied polymorphisms are A۹۸۶S is ۱۶ and Q۱۰۱۱E. First, the molecular masses of the wild types (۹۸۶A and ۱۰۱۱Q) were performed. After that, the mutation S and E are assigned to ۹۸۶A and ۱۰۱۱Q to form ۹۸۶S and ۱۰۱۱E mutated types. Then the similar calculation for molecular masses of the mutated types (۹۸۶S and ۱۰۱۱E) was performed. The change of molecular mass after mutation assignment is calculated. Comparison of magnitude of change was then done. According to the analysis, the molecular change due to the mutated type of AQ haplotype is shown in Table ۱. The estimated the magnitude of change in mutated types of A۹۸۶S is higher than Q۱۰۱۱E. Calcium is an important bio-mineral. The alteration of blood calcium is a common problem seen in PHT. Risk of CSR genotypes are widely mentioned in the patients with PHT. It was suggested that some genetic variants may predispose to calcium stone formation or hypocalcemia. Among patients with PHT, the cases with AQ haplotypes (۹۸۶A and ۱۰۱۱Q) are reported for clinical association with lower serum calcium levels and hypercalciuria comparing to the SQ haplotypes (۳,۴). Based on this work, the lowering blood calcium due to the effect of genetic polymorphism in CSR should be higher in A۹۸۶S polymorphism comparing to Q۱۰۱۱E. In the patient with ۹۸۶S, higher blood calcium level could be expected

## کلمات کلیدی:

Primary hyperparathyroidism, Parathyroid gland, Calcium, Calcium-sensing receptor, Calcium stone

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