Reviewer’s report

**Title:** Familial hypocalciuric hypercalcemia: A novel Mutation in the Calcium Sensing Receptor gene in an Irish Pedigree

**Version:** 1  **Date:** 27 October 2009

**Reviewer:** Yong-Kwei Tsau

Which of the following best describes what type of case report this is?: Findings that shed new light on the possible pathogenesis of a disease or an adverse effect

Has the case been reported coherently?: Yes

Is the case report authentic?: Yes

Is the case report ethical?: Yes

Is there any missing information that you think must be added before publication?: No

Is this case worth reporting?: Yes

Is the case report persuasive?: Yes

Does the case report have explanatory value?: Yes

Does the case report have diagnostic value?: Yes

Will the case report make a difference to clinical practice?: Yes

Is the anonymity of the patient protected?: Yes

Comments to authors:

In this case report, clinical diagnosis of familial hypocalciuric hypercalcemia is established hypercalcemia with relative hypocalciuria (FeCa < 1%) and an (autosomal) dominant family history. The authors demonstrate an association between the presence of a novel heterozygous CASR gene (A213E) point mutation and affected family members. Although not a direct causative relationship for abnormal calcium sensing receptor function, molecular diagnosis of this disease may indeed avoid unnecessary treatment, such as parathyroidectomy.