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Letter to Editor

Error in Reporting a Case of NF-κB2 Deficiency

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I would like to address an important concern regarding the case report titled "[*NF-κB2* Mutation in a Patient with Lymphopenia and Extreme Cold Sensitivity (a case report)]" published by Immunology and Genetics Journal in 2019 with Doi: 10.22034/igj.2019.212824.1029. In this article, the authors reported a case with a chief complaint of cold intolerance or cold sensitivity in all seasons, associated with a nonsense mutation (c.1831C > T) in the *NF-KB2* gene (1)The variant is reported incorrectly, as the correct variant is a frameshift variant (c.457delA) located at chromosome position Chr10:104127118. This variant has not been previously reported and is considered novel. Based on the American College of Medical Genetics and Genomics (ACMG) guidelines, it is likely pathogenic.

As reported, the patients had no significant infection, and cold intolerance in all seasons was the main complaint. The laboratory data at the diagnosis showed mild lymphopenia, decreased B and T cell counts, and normal immunoglobulin levels. However, the patient's subsequent tests did not observe lymphopenia and decreased T-cell counts. To further investigate, we measured the number of B and T cell subsets in the patient and T cell function. We found only a mild decrease in the frequency of total B cells (3.3%); while the number of T cells and T cell function were normal (data not shown). We also measured the protein expression of p52 in the patient and found no abnormal expression (data not shown), highlighting that the variant likely had no significant effect on protein expression.

Patients with *NFKB2* mutations are at higher risk of viral infections, pituitary gland involvement, and ectodermal dysplasia (2), and they present a CVID-like phenotype (3). Laboratory data from the patient show a weak correlation with the disease, and this patient does not exhibit a typical CVID phenotype. However, we cannot exclude the possibility of NF-κB2 deficiency, and the case should be evaluated using a functional assay to determine whether it is NF-κB2 deficient, as the mutation is likely pathogenic.

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