Mutation in Rh48: Assessment for possible mutation prone point

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Abstract

Rh antigen is a widely studied but not already known antigen. RH48 (JAL) is a low-incidence Rh antigen of unknown molecular background and is proposed for association with weakened expression of RhCE antigens. Here, the author performed a bioinformatics approach to study the probability of JAL mutation. It can be shown that JAL is a highly unstable molecule that is prone for mutation.

Keywords JAL · Mutation · Probability

Introduction

Rh blood group is an important blood group system in transfusion medicine. Rh antigen is a widely studied but not already known antigen. Rh48 (JAL) is a low-incidence Rh antigen of unknown molecular background and is proposed for association with weakened expression of RhCE antigens [1–2]. The recent studies showed some mutations within the JAL antigen [1–2] and this is believed to be the cause of low expression. The mutation within R48 is of interest since the exact genetic is not well clarified. Here, the author performed a bioinformatics approach to study the probability of JAL mutation.

Materials and methods

The standard protocol for assessment the mutation prone point within the JAL molecule was done. The protocol has already published in previous reference works and acceptable for its reliability [3–7]. In brief, the sequence of the studied molecule, JAL, was primarily searched via standard database, PubMed then further determination of mutation prone point within its molecule was done by computational tool namely GlobPlot. This process is standard technique and used.

Results

The derived sequence of studied molecule, human JAL, (CAD88169), poses 50 residues and 37 residues are prone to mutation (Fig. 1). Calculating for probability, there can be up to 2237 mutations of JAL molecule.

iqLATMSAMS vlisagavlg kvnlaqlvrm vlvevtalgt lrmvISNIFN

Fig. 1 Showing mutation resisted residues within JAL. (in capital letters).

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Discussion

JAL is a focused interest in transfusion medicine. The solution to the finding on low antigenic expression due to JAL is believed to be due to underlying genetic defect [1–2]. The author hereby used the standard published bioinformatics technique and identified mutation prone positions within R48. Further calculation on the number of probable mutations of the JAL was also done based on the fact that each mutation prone point can be freely changed, changing to any of other 22 amino acids. It can be seen that thousands of mutations can be expected.

It can be shown that JAL is a highly unstable molecule that is prone for mutation. However, since the Rh antigen has its nature of low prevalence, the prevalence of JAL should be low. In addition, it might be noted that although there are numbers of possible mutations those mutations might be non-sense. At least, the derived results from this study can be used as the first step information for further work to realize the clinical importance of possible mutated JAL.

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