

ABSTRACT

The present disclosure relates to a genetic variation identification method, genetic variation identification apparatuses, and a storage medium. The method includes: obtaining at least one gene sequencing read corresponding to a genetic variation candidate site (11); obtaining base arrangement features of the genetic variation candidate site (12); determining non-base arrangement features of the genetic variation candidate site based on non-base arrangement information of the at least one gene sequencing read within a preset site interval, where the non-base arrangement features remain unchanged after a base arrangement order changes (13); and identifying genetic variation of the genetic variation candidate site based on the base arrangement features and the non-base arrangement features of the genetic variation candidate site (14). In the method, in consideration of the feature that non-base arrangement features are not constrained by a base arrangement order, pseudo-genetic variation caused by germ-line genetic variation and interferences such as noise and errors can be better screened out, so that genetic variation is better identified and accuracy of genetic variation identification is improved.