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Discovery of somatic mutations in the progression of chronic myeloid leukemia by whole-exome sequencing

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ABSTRACT. We performed whole-exome sequencing in samples representing accelerated phase (AP) and blastic crisis (BC) in a subject with chronic myeloid leukemia (CML). A total of 12.74 Gb clean data were generated, achieving a mean depth coverage of 64.45 and 69.53 for AP and BC samples, respectively, of the target region. A total of 148 somatic variants were detected, including 76 insertions and deletions (indels), 64 single-nucleotide variations (SNV), and 8 structural variations (SV). On the basis of annotation and functional prediction analysis, we identified 3 SNVs and 6 SVs that showed a potential association with CML progression. Among the genes that harbor the identified variants, *GATA2* has previously been reported to play important roles in the progression from AP to BC in CML. Identification of these genes will allow us to gain a better understanding of the pathological mechanism of CML and represents a critical advance toward new molecular diagnostic tests for the development of potential therapies for CML.

Key words: Exome sequencing; Chronic myeloid leukemia; Accelerated phase; Blastic crisis

