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SHORT COMMUNICATION

WILEY

Next-generation sequencing in two cases of *de novo* acute basophilic leukaemia

Takuya Shimizu[1] | Tadakazu Kondo[1] | Yasuhito Nannya[2] | Mizuki Watanabe[1] | Toshio Kitawaki[1] | Takero Shindo[1] | Masakatsu Hishizawa[3] | Kouhei Yamashita[1] | Seishi Ogawa[2] | Akifumi Takaori-Kondo[1]

[1]Department of Hematology and Oncology, Graduate School of Medicine, Kyoto University, Kyoto, Japan

[2]Department of Pathology and Tumor Biology, Graduate School of Medicine, Kyoto University, Kyoto, Japan

[3]Department of Hematology, Kyoto-Katsura Hospital, Kyoto, Japan

Correspondence
Tadakazu Kondo, Department of Hematology and Oncology, Graduate School of Medicine, Kyoto University, 54-Shogoin-kawaramachi, Sakyo-ku, Kyoto 606-8507, Japan.
Email: tadakazu@kuhp.kyoto-u.ac.jp

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Abstract

Acute basophilic leukaemia (ABL) is a rare subtype of acute myeloid leukaemia (AML); therefore, few data are available about its biology. Herein, we analysed two ABL patients using flow cytometry and next-generation sequencing (NGS). Two cell populations were detected by flow cytometry in both patients. In Case no. 1, blasts ($CD34^+$, $CD203c^-$, $CD117^+$, $CD123dim^+$) and basophils ($CD34^-$, $CD203c^+$, $CD117^{\pm}$, $CD123^+$) were identified, both of which were found by NGS to harbour the 17p deletion and have loss of heterozygosity of *TP53*. In Case no. 2, blasts ($CD33^+$, $CD34^+$, $CD123^-$) and basophils ($CD33^+$, $CD34^+$, $CD123^+$) were identified. NGS detected *NPM1* mutations in either blasts or basophils, and *TET2* in both. These data suggest an overlap of the mutational landscape of ABL and AML, including *TP53* and *TET2* mutations. Moreover, additional mutations or epigenetic factors may contribute for the differentiation into basophilic blasts.

KEYWORDS

acute basophilic leukaemia, gemtuzumab ozogamicin, next-generation sequencing

1 | INTRODUCTION

Acute basophilic leukaemia (ABL) is a rare subtype of acute myeloid leukaemia (AML) recognized by the 2016 World Health Organization (WHO) classification. The proposed diagnostic criteria for ABL are as follows: (a) myeloblasts + metachromatic blasts (>20%) and basophils (>40%) of nucleated bone marrow or peripheral blood cells; and (b) persistent hyperbasophilia.[1] However, the molecular cytogenetic data available on ABL are scarce. Indeed, it remains unclear whether ABL shares a similar mutational profile with AML. Moreover, there is no established treatment strategy for ABL, highlighting the need for a better understanding of this malignancy.

Here, we diagnosed two patients with ABL based on morphology and cytology. In addition, bone marrow samples were analysed using next-generation sequencing (NGS). This report will add new information about the mutational landscape of this rare entity.

2 | CASE DESCRIPTION

2.1 | Case no. 1

The patient was a 59-year-old Japanese man with a history of cardiac arrhythmia. One month before presentation to our institution, he