LEUCEMIAS

Myeloproliferative neoplasms with t(8;22)(p11.2;q11.2)/BCR-FGFR1: a meta-analysis of 20 cases shows cytogenetic progression with B-lymphoid blast phase.

Montenegro-Garreaud X, Miranda RN, Reynolds A, Tang G, Wang SA, Yabe M, Wang W, Fang L, Bueso-Ramos CE, Lin P, Medeiros LJ, Lu X.

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Abstract

Rearrangements of FGFR1 result in the 8p11 myeloproliferative syndrome, a group of rare diseases that features a myeloproliferative neoplasm (MPN) that commonly progresses to lymphoblastic leukemia/lymphoma or acute myeloid leukemia. The most common partner of FGFR1 is ZMYM2, and patients with the ZMYM2-FGFR1 fusion often present with MPN and T-lymphoblastic lymphoma. There are 14 other partners that can fuse with FGFR1, and of interest is the BCR-FGFR1 fusion that results from t(8;22)(p11.2;q11.2). Patients with t(8;22) often show leukocytosis and present with an MPN resembling chronic myeloid leukemia or very rarely, with B-lymphoblastic leukemia (B-ALL). In this study, we analyzed the clinicopathological, cytogenetic, and molecular features of 2 new patients with the t(8;22)(p11.2;q11.2)/BCR-FGFR1 who presented with B-ALL. An underlying MPN became apparent when a morphologic remission of B-ALL was achieved after chemotherapy. We subsequently reviewed the literature and identified 18 additional cases reported with B-ALL in a background MPN or with the MPN as a chronic phase. Our data suggest that the t(8;22)(p11.2;q11.2)/BCR-FGFR1 may arise from a myeloid/B progenitor cell. It is important to recognize that neoplasms carrying the t(8;22)/BCR-FGFR1, although rare, can commonly with B lymphoblastic leukemia at the initial diagnosis, which could distract one from recognizing a possible underlying 8p11 myeloproliferative syndrome.

Mutations in the BCR-ABL1 gene in a peruvian patient with acute lymphoblastic leukemia resistant to therapy.

Ortiz CA, Alvarez YP, Dongo-Pflucker KL, Valdivia E, Mendoza Fernández J, Dávila S, Mora-Alférez P.

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Abstract

CONTEXT: The fusion gene BCR-ABL1 is present in at least the fourth part of B-cell acute lymphoblastic leukemia adult cases. Patients with this fusion gene are candidates to tyrosine kinase inhibitors treatment, and the response to this therapy can be measure by quantification of BCR-ABL1 transcripts. Some patients relapse because the presence of mutations in the tyrosine kinase domain of BCR-ABL1.

CASE REPORT: This is a report of a patient with BCR-ABL1 who initially achieved molecular response with imatinib therapy, relapsing after fifteen months. The treatment was changed to dasatinib, but the patient doesn't achieve molecular response. Retrospectively, we analyzed the tyrosine kinase domain of BCR-ABL1 and we found three mutations (E459K, E255K and V299L).

CONCLUSIONS: We conclude that gain of mutations during treatment with TKIs has strong impact in the progress of disease, being relevant the detection of BCR-ABL1 mutations in relapsed patients or in case of BCR-ABL1 persistence.