CASE STUDY

BENHA UNIVERSITY

Empire Genomics Helps Characterize Rare Specific Gene Fusion in Acute Myeloid Leukemia

Background

Chromosomal rearrangements involving the KMT2A gene are common in acute myeloid leukemia. 135 different KMT2A rearrangements have been identified and 94 translocation partner genes characterized. Of these partner genes, 35 occur recurrently, and only 9 of them account for over 90% of cases. KMT2A-SEPT5 translocation is rare, and may be leukomogenically important and have an impact on disease course.

Objectives

In this study, a series of tests and analyses were performed using biotechnological devices, including flow cytometry, cytogenetic analysis, and dual-color FISH analysis. A rare case of translocation of KMT2A with SEPT5 in AML with t(11;22)(q23;q11.2) is characterized and presented along with a review of literature.

Approach

Following bone marrow biopsy, flow cytometry analysis, cytogenetic analysis, and dual-color fluorescent in situ hybridization were performed to characterize this case of a rare specific gene fusion in AML. To identify the genes involved in the translocation, a SEPT5 probe manufactured by Empire Genomics was used in conjunction with a KMT2A break-apart probe in FISH analysis of both interphase and metaphase cells.

Results

The procedures carried out as part of this case study helped to characterize this new case of de novo AML with KMT2A-SEPT5 fusion. There have only been nine cases reported with this same fusion, with varying prognoses. KMT2A-SEPT5 fusion may be important in leukemogenesis of AML with t(11;22)(q23;q11.2), but further studies are required to explore the role of septins in leukemogenesis, disease prognosis, and response to treatment.

Acute myeloid leukemia with KMT2A-SEPT5 translocaion: A case report and review of the literature

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Lead Organization

Benha University

Diseases

- Acute myeloid leukemia

Biomarkers Mentioned

- KMT2A
- SEPT5

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