



Empire Genomics Probe Helps Researchers Report First Case of NUP98 Rearrangement in AML with t(11;19)(p15;p12) in the World

Background

Acute myelomonocytic leukemia is a group of diseases driven by various cytogenetic and molecular abnormalities. The present study is the first report of a case of AML harboring t(11;19)(p15;p12) in the world.

Objectives

In-depth evaluation of bone marrow samples was necessary to investigate this first reported case of AML harboring t(11;19)(p15;p12). This study aimed to use many methods of analysis to deduce findings regarding the diagnosis.

Approach

Analysis of bone marrow samples from this patient allowed for characterization of this rare case of AML, including a gene fusion between NUP98 and ZNF91. Bone marrow samples were sent out for whole-genome sequencing and RNA sequencing analysis. The WGS analysis revealed the presence of 16 in-frame gene fusions, including a fusion between NUP98 and ZNF91. FISH analyses were performed to clarify these findings.

Results

After the WGS analysis indicated a fusion between NUP98 and ZNF91, FISH analyses using the NUP98 break-apart FISH probe kit from Empire Genomics revealed the presence of a break-apart rearrangement chromosome 11p15 in 98.4% of cells. This first case of NUP98 rearrangement in AML with t(11;19)(p15;p12) suggests that more cases with similar karyotypes and genetic abnormalities should be evaluated.

NUP98 Rearrangement in Acute Myelomonocytic Leukemia With t(11;19)(p15;p12): The First Case Report Worldwide

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Lead Organization

Dong-A University College of Medicine

Diseases

- Acute Myelomonocytic Leukemia

Biomarkers Mentioned

- CD11c
- CD14
- CD33
- CD64
- CD71
- AML1-ETO
- PML-RARα
- CBFβ
- FLT3
- D835Y
- NUP98
- ZNF91
- HOXA9
- TP53