CASE STUDY

UNIVERSITY OF CALIFORNIA SANTA CRUZ

Empire Genomics Helps Characterize Rare Chromosomal Abnormality in AML

Background

Jumping translocations (JTs) are rare chromosomal abnormalities that involve a single donor chromosome and multiple recipient chromosomes. Only 21 JTs have previously been described in myeloid-lineage hematological disorders. JTs are generally associated with a poor prognosis. Although oligo-based array comparative genomic hybridization (oaCGH) has the advantage of precision, it has been underutilized in the study of JTs.

Objectives

In this study, a JT was characterized involving 3q13.31-qter and concomitant trisomy 8 in a previously healthy, 73 year-old patient with aggressive acute monocytic leukemia (AML), utilizing oaCGH analysis in conjunction with other advanced molecular cytogenetics, including conventional cytogenetic analysis (CCA), 24-color spectral karyotyping (SKY), fluorescence in situ hybridization (FISH), and quantification of telomere content and telomere lengths.

Approach

Many procedures were carried out in an attempt to characterize the JT acquired in the patient described. Combining the results of these analyses with a literature review of similar JTs and other related aberrations, inferences regarding clinical outcome can be drawn. A variety of biotechnological tools were utilized in this study, including the directly labeled locus specific BAC-probes manufactured by Empire Genomics, RP11-899P8, RP11-258A15, and RP11-661J1.

Results

Telomere FISH revealed a significantly reduced telomere content in the aberrant cells with JT compared with cytogenetically normal cells at diagnosis and in normal cells at complete remission. Additionally, locus-specific FISH with BAC-based probes from the 3q13.31-q13.32 region showed heterogeneity, a result made possible by Empire Genomics. Based on this study as well as a literature review, duplication of 3q13.31-qter may be a non-random, recurrent abnormality in rare, aggressive cases of AML. Further research is warranted, but it may be reasonable to classify patients with JTs as higher risk, even if other disease parameters indicate lower risk.

Characterization of an acquired jumping translocation involving 3q13.31-qter in a patient with de novo acute monocytic leukemia

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Lead Organization

University of California Santa Cruz

Diseases

- Acute monocytic leukemia (AML)

Biomarkers Mentioned

- TUSC7
- LSAMP

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