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Analysis of mutation occurrence in patients with acute myeloid leukaemia using next-generation sequencing

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The aim of the study was to determine the frequency of genetic changes among patients of the Holy Cross Cancer Centre in Kielce diagnosed with bone marrow cancer.

A group of 75 patients hospitalized in 2019–2021 was subjected to the study. Detection of genetic changes was performed using high-throughput next-generation sequencing, NGS, using Ion Torrent technology. The panel used detects changes associated with the development of bone marrow cancer (40 genes, 29 fusions).

The study group consisted of 75 patients diagnosed with various types of cancer. The largest group were patients with acute myeloid leukaemia, AML (n = 47); the next groups were patients with myelodysplastic syndromes, MDS (n = 14), and other myeloproliferative neoplasms (n = 14). Among all patients, 62 people had at least one mutation (83%). In the group of patients with AML, at least one mutation was observed in 46 people (98%). Among patients from the AML group, changes in 27 genes and 5 types of fusion were detected. The most frequently observed changes in the group of patients with AML were internal tandem duplication in the FLT3 gene (26%) and mutations in the nucleophosmin 1 gene, NMP1 (17%). In the group of patients with MDS, six (43%) had at least one lesion. Of the 11 types of change observed in patients with MDS, nine have been reported in patients with AML.

The results of the research revealed high genetic heterogeneity among the studied AML patients. The presence of changes in the same genes in patients from the MDS and AML groups may indicate the possibility of transformation of MDS into AML.

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