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Identification and functional characterization of a cytoplasmic nucleophosmin leukaemic mutant generated by a novel exon-11 NPM1 mutation

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Nucleophosmin (NPM1) mutations occur in 50–60% of adult acute myeloid leukemia (AML) with normal karyotype.1, 2 About 40 NPM1 mutations2 have been so far identified, all clustering in exon-12. In spite of molecular heterogeneity, all mutations cause common changes at the C terminus of NPM mutants, i.e. loss of tryptophans 288 and 290 (or 290 alone) and creation of a new nuclear export signal (NES) motif.2 As a consequence, NPM mutants aberrantly accumulates in the cytoplasm of leukaemic cells;3, 4 hence, the term NPMc+ (cytoplasmic-positive) AML.1, 2 Here, we report on the identification and functional characterization of a cytoplasmic nucleophosmin mutant generated by a novel exon-11 NPM1 mutation in a patient with AML.