CASE 1.

Case Background

A 34 year old female presented to hospital with a 4 week history of lethargy and shortness of breath.

A full blood count was performed and showed a white cell count of $78x10^9$/L with 78% circulating blasts. A bone marrow biopsy was performed and showed a markedly hypercellular aspirate and 92% blasts with monoblastic morphology.

Baseline molecular testing revealed mutated *NPM1* (NPMA). No other somatic or germline mutations were detected on routine molecular testing. Cytogenetics showed a normal karyotype. Immunophenotyping confirmed blasts to have a myeloid phenotype with aberrant CD56 expression and uniform CD34 negativity.

The patient was diagnosed with AML with mutated *NPM1* and stratified as 'favorable' genetic risk as per the ELN2022 genetic risk classification. She underwent intensive induction chemotherapy and was not planned for a CR1 allograft at diagnosis.

Bone marrow biopsy following cycle 1 induction revealed 4% blasts, CR by ELN AML response criteria. However, there was a small persistent population of blasts with aberrant immunophenotype by standard immunophenotyping in the bone marrow sample. Post-C2 induction a bone marrow biopsy was performed showing continued morphologic CR (1% blasts). Post-C2 aberrant blasts were not detected on standard immunophenotyping.