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An enigmatic hyperdiploid multiple myeloma with novel and complex cytogenetic abnormalities – A rare occurrence

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Abstract

Multiple myeloma (MM) is a cytogenetically heterogenous plasma cell malignancy. Based on the hallmark cytogenetic abnormalities, MM can be divided into hyperdiploid and non-hyperdiploid subtypes. The hyperdiploid subtype is characterized by trisomies of certain odd numbered chromosomes namely 3, 5, 7, 9, 11, 15, 19 and 21 whereas non-hyperdiploid subtype is characterized by translocations of the immunoglobulin heavy chain alleles at chromosome 14q32 with various partner chromosomes with the most important of which being t(4;14), t(6;14), t(11;14), t(14;16), and t(14;20). In general, hyperdiploid patients are considered a better prognostic group while non-hyperdiploid patients are considered a high risk group. Here we report one myeloma case presented with hyperdiploid karyotype along with other complex high risk abnormalities and also a novel abnormality which was not previously described. A 74 years old Malay lady presented with abnormal biochemical profile namely reversed albumin globulin ratio with very high globulin level, normochromic normocytic anaemia and acute kidney impairment. As part of MM diagnostic work up, bone marrow cytogenetic analysis was performed using conventional cytogenetics and fluorescent in situ hybridization (FISH). Conventional cytogenetic analysis showed hyperdiploid metaphases with 51-53 chromosome range and involving translocation (2;3)(q21;p21), trisomies 5,7,11,15,17,19 and 21. FISH analysis revealed presence of del(13)(q14.3), t(11;14)(q32;q13.3), t(4;14)(p16.3;q32.2) and del(17)(p13). The genetic profile of MM can acts as a determinant of patient survival and response to treatment. The reported patient's tumour cells exhibited a hyperdiploid chromosome count and translocation t(11;14) which are associated with good prognosis. However some of the tumour cells exhibited translocation t(4;14), deletions of chromosome 13q and 17p13. These abnormalities are associated with adverse prognosis. A novel abnormality translocation t(2;3) whose significance is unknown also was detected in the tumour cells. Hence this patient who is currently undergoing treatment (on dexamethasone, thalidomide, intravenous immunoglobulin and intravenous Zoledronic acid) is being closely follow up to determine whether the presence of these high risk cytogenetic abnormalities along with good prognosis abnormalities will confer her a more aggressive disease course. This case is presented because of the rare and simultaneous occurrence of these good prognostic, adverse prognostic abnormalities and also one novel abnormality in one patient.

Keywords: Multiple myeloma; hyperdiploid; non-hyperdiploid; complex cytogenetic abnormalities; interphase FISH

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