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Diagnostic difficulties in myelodysplasia: Role of morphology and molecular techniques in age related clonality, cytopenia and life-style choices

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Back ground: MDS are heterogenous clonal disorders ranging from single to multi-lineage cytopenia and dysplasia. The link between haematopoietic aging, clonality and bone marrow failure syndromes remains unclear. There are diagnostic difficulties. To this end we present a case of myeloneuropathy and myelodysplasia.

Methods: 47-year old care worker referred for worsening myalgia and weakness in lower limbs. She was anemic with low BMI and short stature. Neurological examination revealed loss of pin-prick/vibration up to both knees and mild tandem gait. Her Complete blood count (CBC) showed HB:82g/L,WCC:1.5x109/L,Plt:219x109/L,ANC:0.6x109/L, no blasts. All other blood and radiology investigations normal. Neutropenia got worse (WCC:0.8x109/L). A bone marrow test and molecular myeloid makers were performed. Bone marrow Fish for trisomy 8. Monosomy7,5qwerenormal.Furtherbiochemicaltestsincluding trace elements for lead, mercury, copper, zinc was performed. Lumber puncture and CSF examination was unremarkable.

Results: Trace element results show normal levels of Selenium, lead and mercury. Serum iron and Vit D was low. Serum Copper low <1.0 (NR=11-22umol/L). 24-hour urinary copper was low at 0.06umol/L (NR: 0.1-1), high ceruloplasmin, raised serum zinc 27.1umol/L (NR:11-22) . Patients diet and Upper GI endoscopy normal. Patient was commenced on Iv copper and Iron, no growth factors or blood products were used. Further investigations revealed that patient has been using dentures for 16-years and some of the denture cream/glue contain zinc which may have caused zinc overload and neurological symptoms.

Conclusions: In patients with unexplained neutropenia and anemia, trace elements and copper level should be checked and denture questions should be asked in patients with MDS/cytopenia.

Speaker Biography

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