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Hypocupremia associated cytopenia and myelopathy: A Scotland national retrospective review

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Copper is an essential trace element that is involved in a number of important enzymatic processes throughout the body. Recent single case reports and small studies have shown that deficiency of copper can cause reversible haematological changes and irreversible neurological injury. We chose to undertake a national study, looking at all cases of copper deficiency in Scotland over a 5-yr period using information from a national reference laboratory. From 16 identified patients, we determined that 86% had both haematological and neurological features of copper deficiency, while 18% had haematological features only at presentation. Twelve of the sixteen patients had high serum zinc concentrations (>18 lm/L) nine patients were using zinc-containing dental fixatives at time of diagnosis. 94% of patients had haematological features as an initial manifestation of copper deficiency, which included anaemia, thrombocytopenia and neutropenia. Patients who underwent later bone marrow testing had appearances in keeping with refractory cytopenia with multilineage dysplasia, refractory anaemia with excess of blasts, unclassified marrow dysplasia or probable myelodysplasia (MDS). 75% of patients had neurological symptoms or signs, including progressive walking difficulties and paraesthesia, or gait difficulties without sensory signs. Clinical examination was in keeping with spastic paraparesis (either with or without sensory neuropathy). Magnetic resonance imaging (MRI) showed multifocal T2 hyper intense foci in the subcortical white matter, and atrophy of the cerebrum and cerebellum was also seen on computerised tomography (CT). MRI of the spinal cord showed signal change in the dorsal columns in either the cervical or thoracic cord. 93% of cytopenias responded to copper replacement and addressing the original cause of the copper deficiency, but only 25% of patients had improvement in their neurological function, while 33% deteriorated and 42% remained unchanged. Our study demonstrates that copper deficiency is an under recognised cause of several types of cytopenia, which are reversible but can progress to significant neurological injury if left untreated. We illustrate the importance of identifying these patients early to prevent irreversible neurological injury.

Biography

Alemayehu A. Gebreyes has completed his MD at the age of 26 years from Addis Ababa University Ethiopia and has received MRCP UK after completing foundation year and core medical training in west Scotland. Currently, he is a Haematology trainee with special interest in Micronutrients and its role in haemopoiesis.

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