Highly elevated ferritin levels are associated with haemophagocytic lymphohistiocytosis – are we missing treatable diagnoses? A retrospective service evaluation of diagnosis in patients with ferritin >10,000 μg/L

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Background
Haemophagocytic lymphohistiocytosis (HLH) is a hyperinflammatory syndrome which can complicate sepsis, malignancy or autoimmune disease and may lead rapidly to critical illness and death. Early treatment reduces mortality but diagnosis requires a high index of suspicion and correct interpretation of laboratory results. Highly elevated ferritin levels (HEF) >10,000 μg/L are highly specific for HLH and should prompt consideration/exclusion of hyperinflammation. Diagnostic guidelines for HLH were published in 2004 requiring the presence of ≥5/8 criteria.

Aim
To assess recognition of HLH in a paediatric population with HEF.

Methods
This retrospective study was conducted at 8 UK centres. Biochemistry databases identified patients ≥16 years with serum ferritin >10,000 μg/L between 01.04.2014 and 31.03.2017. A standardised proforma was used to collect data. Cases were assessed against the 2004 HLH criteria. Due to limited access to some laboratory tests, modified criteria using a threshold of ≥4/5 (excluding tissue haemophagocytosis, decreased natural killer cell function, increased soluble interleukin-2 receptor) were also applied.

Results
Data were collected for 51 patients (63% female; mean age at surgery 14.7 years). Diagnoses included adolescent (62.8%), juvenile (7.8%) and infantile (5.9%) idiopathic scoliosis, congenital (7.8%) and syndromic scoliosis (2%) and Scheuermann’s kyphosis (13.7%). Mean (±SD) thoracic curvature was 61.3±15.2° and mean kyphosis in those with Scheuermann’s was 92.4±7.3°. No correlation was elicited between HEF1 or FVC (%predicted) and VO2peak (%predicted). Greater thoracic curves were associated with lower HEF (%predicted) and VO2peak (%predicted). A standardised proforma was used to collect data.

Conclusion
Contrary to previously published data, those with lower thoracic curves and therefore poorer lung function show improved exercise capacity and later onset AT, likely as a consequence of improved physical conditioning. These patients also report better quality of life, which may be due to maintenance of normal exercise levels. These findings suggest that physical adaptation occurs in scoliosis with impaired lung function, and that this may be protective to patients’ mental health.