Commentary: Parents' view

Patient's parents with the support of S Ellis

Lister Hospital,
Stevenage, Herts
SG1 4AB
S Ellis
consultant
rheumatologist
Spencer.ellis@nhs.net

When doctors asked us to participate we agreed without hesitation, knowing that it would bring new knowledge to the doctors and patients. When our daughter was first admitted to hospital, we thought she had an infection that could be simply and quickly cured. However, as time went on and all the tests returned negative, it became clear that there was something more sinister and it became increasingly frustrating that doctors could not come up with a definite diagnosis despite numerous investigations.¹

As she developed many rare complications associated with this illness, we naturally lost confidence in our attending doctors. Although the diagnosis of

adult onset Still's disease was made, we still sought a second medical opinion because of our daughter's deterioration.

Despite her complications, we were reassured and kept regularly informed of her development and progress. Now, we are working together with the doctors for further improvement.

Competing interests: None declared.

1 Sivakumar R, Pavulari S, Ellis S. Fever of unknown origin: case outcome. *BMJ* 2006; doi: 10.1136/bmj.38950.395868.68.

doi 10.1136/bmj.38975.459688.68

Commentary: Few clues to diagnosis

Paul Emery, Jane Freeston

Academic Section
of Musculoskeletal
Disease, Chapel
Allerton Hospital,
Leeds LS7 4SA
Paul Emery
head
Jane Freeston
specialist registrar in
rheumatology

Correspondence to:
P Emery p.emery@leeds.ac.uk

Adult onset Still's disease is a difficult clinical diagnosis. There are few, if any, diagnostic features, with the possible exception of a very high ferritin concentration. Sivakumar and colleagues describe a not atypical case history of a patient presenting with fever.¹ Not unnaturally, the patient was assumed to have an infection. This is understandable and correct, as missing infection could clearly have had serious consequences. For every patient with adult onset Still's disease there are a significant number of patients with severe infection, and it is crucial that infection is the first diagnosis to be excluded.

The persistence of the fever after treatment was the first clue that the patient may have something other

than infection. Development of a rash was perhaps the first concrete clue to her diagnosis. Her raised ferritin concentration was highly suggestive, although very high concentrations are more specific. The problem with the Yamaguchi diagnostic criteria for diagnosis is that they are quite non-specific. The presence of spiking fever and rash and the deterioration in cardiac function should perhaps have triggered a trial of immunosuppression with steroids. Her doctors were correct to meet the request for a second opinion; these cases require as much expertise and experience as is available.

Ferritin concentrations

Serum ferritin concentrations above a threshold of 1000 µg/l (five times the upper limit of normal) have been used to suggest adult onset Still's disease in most studies.² However, similar concentrations can be produced by infection, liver disease, haemophagocytic syndrome, and malignancy. A French study of the validity of hyperferritinaemia as a diagnostic tool reported that a ferritin concentration five times the upper limit of normal had a specificity of 41% and a sensitivity of 80%.³ Others have suggested that concentrations above 3000 µg/l are diagnostic, but only after infection has been excluded.⁴

Another approach has been to look at the fraction of glycated ferritin, which in healthy subjects constitutes 50-80% of ferritin. In inflammatory diseases, however, saturation of glycosylation mechanisms causes the glycated fraction to drop to 20-50%, and in Still's disease it is often less than 20%. Fautrel and colleagues showed that when this diagnostic criterion was applied in conjunction with a fivefold rise in ferritin, the specificity of this combination rose to 93% but with a fall in sensitivity to 43%.³

Bone damage in adult onset Still's disease

WELLCOME PHOTO LIBRARY