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Editorial

Hidden Depths

"For every mistake made from lack of knowledge, ten are made from lack of looking."

James Alexander Lindsay
Professor of Medicine
Queen's University of Belfast, 1899 -1921

The radiograph on this issue's front cover demonstrates a normal image of the abdomen. It is a favourite of mine and one that I use routinely in teaching undergraduate medical students. Take a good look. You may notice a curvilinear gas density projected within the pelvis. See it? This is what a tampon looks like (figure 1). It's a subtle finding, but once seen, the radiographic configuration is not forgotten. Once the tutorial group moves beyond the potentially salacious, 'Carry On Doctor' nature of the subject matter, I like to pose this question. A 24 year old lady is admitted to your hospital with a head injury. On the third day following admission, her clinical state rapidly deteriorates. Can you see anything on the radiograph that might be the cause? Clearly there might be many reasons for this deterioration, including an infection at the body piercing's skin site (also present on the radiograph) but if anyone had recognised the retained tampon then Toxic Shock Syndrome might enter the differential diagnosis. The point is: unless one looks, one never knows. Intimate examinations are, of course, problematic. Is there reluctance to perform one? Possibly. Is there an assumption that perhaps a nursing colleague has? Maybe.

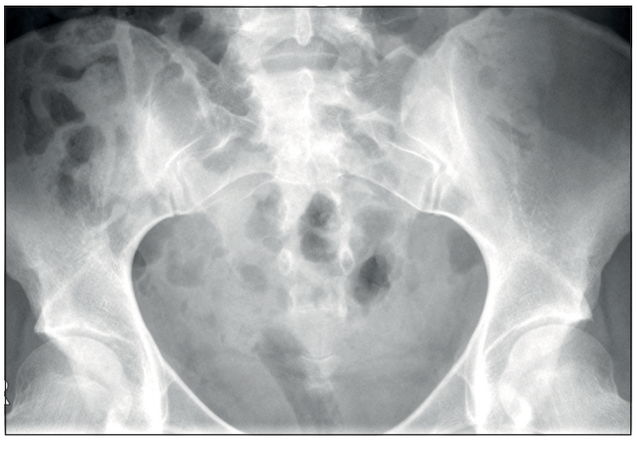


Fig 1. Vaginal tampon

And if it is so for adults, how much more charged is the atmosphere when children are concerned. Assumptions may be incorrect. For example, how distressing is it for a child or its parent if a necessarily intimate examination is required? Jarlath O'Donohoe's very interesting paper has some surprising results.

The Objective Structured Clinical Examination, (OSCE), is now ubiquitous in undergraduate medicine. More mature readers will recall the major case, and the attendant fretful preparation ("Hey, there's a 'Mitral Valve' in ward 8." "Now come on, she has a personality too." "What Schizophrenic as well?'). The examiners were often considered a lottery too. Some, it seemed from a youthful perspective, were possibly exhumed specifically for the express purpose of embarrassing and harassing with a series of incoherent pet questions. This appeared as inescapable an event as the certain knowledge that your friend would get the sweetest, most reasonable consultant who might ask how many legs the patient had, and ultimately ask to be remembered to her father. I'm sure it was never really like that, but the malady lingers on for some. More importantly, was that long case a good test? Was it valid or reliable? How was the passing standard reached? Did the examiners test for the minimally passing candidate or use regression analysis? Oh dear me. Sir Lancelot would be bristling with righteous indignation.

In his excellent review article on OSCEs, Gerry Gormley explains with commendable lucidity, the rationale for such an educational paradigm shift. One thing is certain: OSCEs don't happen by themselves. The examination is constructed with military precision, and requires a commensurate number of personnel. In this regard, Dr Gormley is a Field Marshall. His capacity for organisation is exceeded only by his calming influence on the snarling hordes -and that's just the examiners.

REVIEWERS

I thought it might be timely to salute those often-forgotten individuals: reviewers.

In an increasingly fractured professional existence, taking time to review papers can assume a relatively low priority. Two things are noteworthy. Firstly I have been heartened by the very high level of support from so many colleagues both near and very far who have risen immediately to that challenge. Perhaps more astonishingly, has been the care and scrutiny with which each paper has been evaluated.

Like blood donors, such reviewers display an altrusim and a belief in the necessity of this, the longer view and a sense of thoughtful enquiry. It is a an activity written, as it were, on water. Impossible to measure and I would wager, overlooked as important or even relevant when the scales are produced to gauge the cost of something, rather than its value.

So to all of them, I, on behalf of the editorial board, say thank you. Please continue to send me your good papers.

Barry Kelly

Gaucher Disease may be underdiagnosed in the UK and Northern Ireland

Gaucher is currently underdiagnosed in the UK

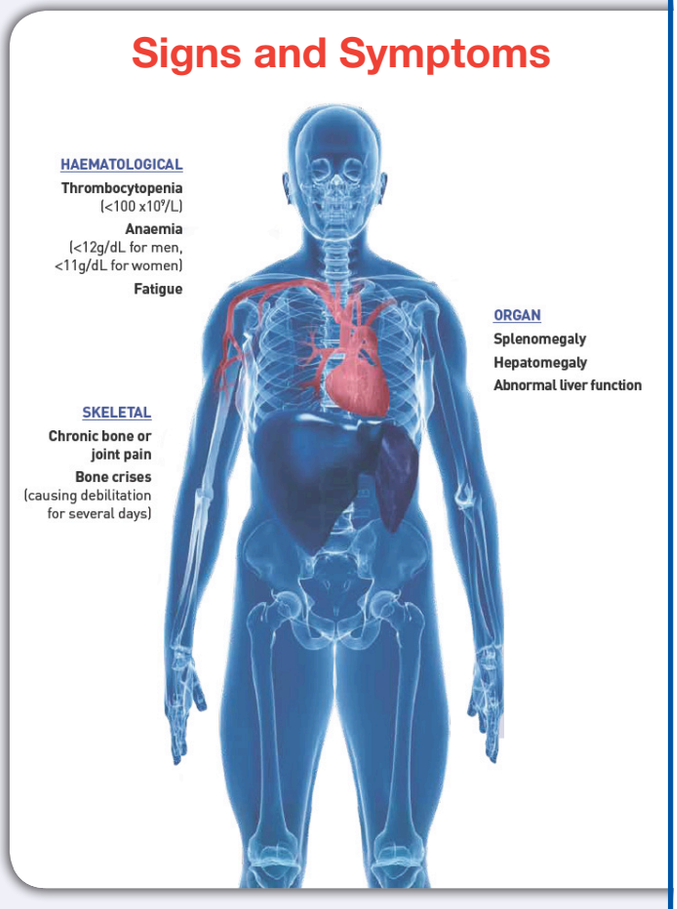
Gaucher disease is a rare inherited lysosomal storage disorder caused by the deficiency of the glucocerebrosidase enzyme, which results in the accumulation of glucocerebroside within the lysosomes of macrophages.

The prevalence of type Gaucher disease is estimated at 1 in 50,000-100,000 in the general population (and 1 in 850 in the Ashkenazi Jewish population).[1,2] However, there are only around 300 patients in the UK and only 3 cases in Northern Ireland with known Gaucher disease (approximately 1 in 200,000 of the population).[2]

This may partly be because Gaucher disease is a heterogenous multi-systemic disorder with variable symptoms and progression. The presenting symptoms can also be similar to other conditions, which may delay diagnosis and access to appropriate management.[1]

Misdiagnoses include leukaemia, immune thrombocytopenia purpura, autoimmune disease, hepatic cirrhosis, idiopathic avascular necrosis, viral disease, idiopathic splenomegaly, and anaemia of chronic disease.[1]

When haematological malignancies have been ruled out, Gaucher disease should be considered[1]



WHEN SHOULD YOU TEST FOR GAUCHER

All patients requiring splenectomy with no diagnosis

Any patient with any of the following: anaemia, thrombocytopaenia, bone pain, Splenomegaly, Hepatomegaly, Monoclonal gammopathy of undetermined significance

HOW TO TEST:

Blood should be sent for glucocerebrosidase assay in the first instance which requires 5 ml in EDTA. The biochemical marker chitotriosidase is usually markedly elevated in this condition as well. It is a good idea to speak to your local biochemistry laboratory before sending the sample in. DNA testing may be carried out to identify the mutation in a particular case but this is not the first line investigation.

For further information or advice about Gaucher disease please contact the Department of Medical Genetics at Belfast City Hospital or the Gaucher disease association www.gaucher.org.uk

[1] Mistry PK et al. Am J Hematol 2011; 86(1): 110–5.
[2] Connock M et al. The clinical effectiveness and cost-effectiveness of enzyme replacement therapy for Gaucher's disease: a systematic review. Health Technology Assessment 2006; 10(24): 1–156.