# Life & Times The stable rower:

# identifying a rare disease

# Life & Times Books

After reading the May 2018 BJGP, I feel that I have experienced a recent case that amalgamates many of the themes of that issue. My case highlights the importance of appropriate safety netting,1 rare diseases,2 unintentional weight loss,<sup>3</sup> and reassurance.<sup>4</sup>

#### PATIENT PRESENTING WITH COUGH

A student attended my morning clinic with a chest infection, which, considering I work in a GP practice associated with a university, is so much my bread and butter that I may as well have put the sultanas on it and baked the pudding myself.

My patient reported a 2-week history of cough with no sputum production and no shortness of breath. He didn't feel unwell. There were bilateral bronchial crackles and no raised lymph nodes. ICE suggested he wanted antibiotics. Whilst I'm here doc, can I weigh myself? I'm eating big meals all the time but I'm still not gaining weight. He was 1.90 m with BMI of 22.8. He had joined the rowing society and been told to gain 10% of his body weight. Subsequently he had started eating roughly double the recommended daily intake of food. He had seen several different GPs for five similar coughs since starting at university 6 months ago and on two occasions had been prescribed antibiotics. He had a stable sexual partner and did not inject drugs. Each time he was told to return if his condition got worse, but, as his condition either got better or remained the same, he did not return. He believed he had Fresher's Flu. Prior to university, he had never seen a doctor for anything other than his immunisations.

It occurred to me how often he was attending for the same complaint despite looking well and not striking me as health anxious. I organised for an infective and immunological blood screen to be taken that day, and started him on oral amoxicillin. After 2 days his first blood test returned including normal tests for FBC, HIV, and CRP. On day 3, the immunoglobulin test returned: IgA, IgM, and IgG were all negligible. I picked up the phone and referred the patient immediately to immunology for immunoglobulin deficiency. The next day he attended an outpatient appointment and was diagnosed with X-linked agammaglobulinaemia.

Initially the patient was started on prophylactic azithromycin for 2 months while the education and arrangements regarding maintenance solely on regular immunoglobulin infusions were made.

Presenting with symptoms this late is incredibly rare as patients typically present with recurrent illness from 6 months old at termination of breast feeding.<sup>5</sup>

#### **REASSURANCE**

He came back to see me, 6 weeks after I first met him. I was struck by how well he looked; he literally bounced across the room as he walked: 'Doc, check out my weight'. He beamed at his 5 kg gain in 6 weeks. How can I help you today?' I asked. 'Oh nothing, I just wanted to thank you for listening to me.' We talked briefly about his condition but the immunology team had educated him fully on the condition, and then off he bounded to his new life of vitality. The ray of zebra sunshine that made my entire week worthwhile.

His case highlights several learning points: safety netting should include reassessment if the patient does not follow an improving health trajectory, not just a deteriorating one; the role of a GP includes referring to secondary care when something rare is possible for specialist assessment; weight change below what is intended is a marker for potential cancer, metabolic and haematological conditions; and reassurance for the patient came not from identifying his rare illness (5/1 000 000 births) or identifying that with treatment his risk of serious pathology was low,5 but that his X-linked agammaglobulinaemia was explained fully to him.

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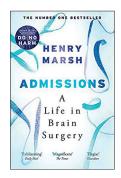
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## Admissions: A Life in Brain Surgery **Henry Marsh**

Weidenfeld & Nicolson, 2018, PB, 288pp, £8.99, 978-1474603874



#### RETIREMENT CONUNDRUMS

I was in Waterstones perusing the books and spotted the paperback Admissions: A Life in Brain Surgery in a carefully laidout pile. Despite a reluctance to pick up medical autobiographies I decided to read the opening paragraph. This was an error on my part because, after reading the first sentence, I was instantly hooked! Admissions is a wonderful title as it carries a double entendre: the obvious being patient stories but a description too of Marsh's own personal foibles. Marsh worked as a consultant neurosurgeon in London hospitals for most of his career before becoming a celebrity doctor by writing his first book, the bestseller Do No Harm: Stories of Life, Death and Brain Surgery, and by featuring in award-winning documentary films Your Life in Their Hands and The English Surgeon. Here in this memoir he explores his medical life, which occurs both here in the NHS and in the Ukraine and Nepal. He is definitely a man who works with his hands, not only in surgery but also with a huge collection of professional handyman tools. The book follows his retirement from the NHS and the purchase of a dilapidated lock keeper's cottage. He decides to divide his retirement time by renovating this cottage in England and by continuing to work abroad.

Marsh is an obsessional perfectionist who does not suffer fools gladly and resigns from the NHS in a fit of anger over inflexible intensive care guidelines, but not before squeezing the nose of a hapless intensive care nurse. It is the personal admissions that lift the book from simply being a collection of