Delayed puberty—an occult systemic cause

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Case—A 17-year-old male presented with symptoms of absent secondary sexual characteristics, decreased appetite and infrequent non-specific abdominal pain. He had no headache, altered sense of smell, colour blindness or gynaecomastia. His other siblings had normal growth. He had a normal male body habitus, height below the 5th percentile, pre-pubertal body hair distribution with testes in the scrotum (12 ml bilaterally). Laboratory results revealed microcytic iron deficiency anaemia, hypogonadotrophic hypogonadism with 46XY on chromosome analysis.

Barium meal follow-through study (Figure 1) demonstrated multiple strictures in the small bowel, which was not resectable raising the suspicion of Crohn’s disease.

Figure 1. Barium meal follow through showing multiple strictures in the small bowel
Discussion—Delayed puberty can be a complication of underlying inflammatory bowel disease in young patients. Although systemic diseases are well recognised to cause hypogonadotropic hypogonadism, this is often less commonly perceived. Delay in the diagnosis and management can delay the onset of puberty indefinitely with potentially disastrous consequences on the pubertal growth spurt with a reduction in final adult height. Testosterone levels often return to normal after recovery from underlying disease. Clinicians should have a high index of suspicion to diagnose this well described, but less commonly perceived cause of delayed puberty.¹,²
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