

LETTERS TO THE EDITOR

A case of X linked agammaglobulinaemia complicated with systemic amyloidosis

EDITOR,—We recently cared for a 27 year old man who died of multiple organ failure caused by systemic amyloidosis. Immunoperoxidase staining of a rectal biopsy specimen showed AA-type amyloid fibrils. He had been diagnosed at the age of 14 months as having X linked agammaglobulinaemia (XLA), presenting with a history of recurrent pneumonia and skin infections, as well as undetectable B cells and very low immunoglobulin concentrations.

His serum IgG concentrations had been kept at about 4 g/l by monthly intramuscular, and subsequently intravenous, γ globulins. He had a reasonably good quality of life except for troublesome recurrent swelling and pain in his knees. Five years before his death he was diagnosed as having pulmonary tuberculosis and treated accordingly.

Amyloidosis is a rare complication of XLA. None of the 96 XLA patients reported by Lederman¹ had amyloidosis. Hermaszewski² presented two patients with renal amyloidosis out of 44 XLA cases. Renal amyloidosis and systemic amyloidosis were also described in two patients with hypogammaglobulinaemia and XLA.^{3,4} The type of amyloid fibrils in these patients was AA-type, as in our case. Patients with XLA cannot synthesise immunoglobulins and they do not have any plasma cells, therefore, the immunocyte derived amyloidosis consisting of AL proteins seems to be impossible. The pulmonary tuberculosis and rheumatoid arthritis-like process in our patient might have contributed to the development of amyloidosis.

In our clinic, among more than 100 patients with primary B cell deficiency followed up for 25 years, we have observed systemic amyloidosis in one patient with hyperimmunoglobulin M syndrome,⁵ and renal amyloidosis in a patient with mucocutaneous candidiasis. This suggests that there might be an additional factor, such as a genetic basis in the development of amyloidosis.

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Turner's syndrome, anorexia nervosa, and anabolic steroids

EDITOR,—We report two cases of anorexia nervosa in girls with Turner's syndrome from a group of 34 patients treated with a combination of oxandrolone and oestrogen.

Patient 1 started ethynyl oestradiol 1 μ g/day at age 9.8 years, increasing to 20 μ g/day over six years. At 10.8 years oxandrolone 1.25 mg/day was started, increasing to 2.5 mg/day after four years. Onset of anorexia nervosa occurred at age 16, before stopping oxandrolone. Weight loss was from 109% to 75% weight for height, and illness features were typical. Body dissatisfaction was not solely related to stature. Illness severity warranted a 10 week admission to a specialist unit. Patient 2 had a similar treatment protocol beginning at age 9 years. Anorexia nervosa began at 15.5 years with weight loss from 107% to 85% weight for height. Oxandrolone was discontinued after 5 kg weight loss. During her illness she also took an overdose of 50 paracetamol. Both patients are now well, with normal weight for height.

Oxandrolone has a long history of use for growth enhancement.¹ The anabolic effects include increased appetite and increase in lean body mass rather than fat mass. Obesity as a result of treatment is not usual.

The association of Turner's syndrome and anorexia nervosa has previously been attributed to chance.² However, Skuse and colleagues³ recently showed that females with Turner's syndrome whose X chromosome was maternally derived showed greater adjustment difficulties, poorer verbal and executive function, and poorer social skills, despite normal intelligence quotient. The authors link the dysfunction to disorders of social adjustment and cognition such as autism. More subtle deficits of social cognition may be a risk factor for anorexia nervosa, especially if associated with low self esteem, as is common in Turner's syndrome.⁴

The peak incidence of anorexia nervosa is during puberty. When puberty is artificially induced, good psychological support may prevent anorexia nervosa in girls (and boys) with low self esteem. Although oxandrolone is associated with disturbance of appetite and weight gain, we are unclear about the role of anabolic steroids in the association between anorexia nervosa and Turner's syndrome.

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The role of negative pressure ventilation

EDITOR,—Thomson's recent review of negative pressure ventilation¹ included results of our randomised controlled trial of 244 infants with neonatal respiratory failure.² She commented that there was "a small overall benefit of CNEP [continuous negative extrathoracic pressure] but with non-significant increases in mortality, pneumothorax rate, and cranial ultrasound abnormalities in the CNEP group". An important result not mentioned in her review was that the median duration of oxygen requirement was 33.6 days in the controls and 18.3 days in the infants treated with negative pressure (a difference of 15.3 days, 95% CI -0.2 to -30.4). We suggest, therefore, that negative pressure has an important role, particularly in the resolution phase of respiratory distress syndrome in preterm infants, and may reduce the risk of developing chronic lung disease. Newer methods for providing continuous nasal positive pressure support to preterm infants still fail to guarantee a transpulmonary pressure gradient, whereas negative pressure has provided this effectively, and quickly.³

We agree that, in school age children, the latest techniques for providing nasal mask ventilation may be preferable to negative pressure ventilation in the treatment of chronic respiratory failure. In addition, we have successfully used nasal mask positive pressure ventilation in three infants with congenital central hypoventilation syndrome.⁴

Our most frequent use of negative pressure ventilation is in the treatment of acute bronchiolitis. In infants with increasing oxygen requirements or recurrent apnoeic-hypoxaemic episodes, negative pressure ventilation may avoid the need for intubation and intensive care.⁵ The early use of negative pressure ventilation has resulted in a 0.4% intubation rate in our unit over the past four years. This compares favourably with published rates.

Finally, Thomson recommends monitoring during negative pressure ventilation with pulse oximetry and end tidal carbon dioxide. We find transcutaneous carbon dioxide monitoring more practical as this avoids the need for expired air sampling, and displaying and interpreting a capnogram.

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WESTMINSTER BRIEFING

The following items are from *Children & Parliament*, winter–spring 1998. *Children & Parliament* is an abstracting service based on *Hansard* and produced by the National Children's Bureau. It covers all parliamentary business affecting children and is available on subscription via the internet. The *Children & Parliament* web site provides direct links to full text *Hansard*, government department sites, the sites of the Office for National Statistics, Ofsted, and other relevant organisations. For further details contact Lisa Payne, Editor, *Children & Parliament*, National Children's Bureau, 8 Wakley Street, London EC1V 7QE, UK (tel: +44 (0) 171 843 6000; fax: +44 (0) 278 9512). (The *Hansard* reference is given in parentheses.)

- Speakers in both Houses backed the idea of a national radio channel dedicated to children. It was suggested that such a channel could potentially improve children's language and verbal skills and encourage listening and concentration. Although it is unwilling to intervene directly the government seems to support the idea. (12 Jan 98, 13 Jan 98, Col 118–124,194.)
- Government consultation on the subject of parental discipline and corporal punishment is likely to involve the Home Office, the Lord Chancellor's Department, the Department for Education and Employment, the Department of Health, and the Scottish, Welsh, and Northern Ireland Offices in the spring of 1999. (12 Jan 98, Col 118.)
- The next survey of children's dental health will be in 2003. (12 Jan 98, Col 148.)
- A government white paper proposes the setting up of a Food Standards Agency to provide for public protection against food related health hazards. (14 Jan 98, Col 351–366, 1079–1094.)
- The government aims to give teachers more time to spend on teaching basic literacy and numeracy while allowing them more flexibility as regards history, geography, art, design and technology, music, and physical education. But science, information technology, and religious education will maintain their curriculum positions. (13 Jan 98, Col 174–175.)
- Lottery funding for sports in schools so far includes over £48 million (143 awards) to state secondary schools and colleges, over £8 million (8 awards) to state primary schools, nearly £3 million (6 awards) to private secondary schools and colleges, and nearly £39 million (30 awards) to the higher and further education sectors. (15 Jan 98, Col 265–266.)
- According to Unicef some 32% of children under five in Iraq are chronically malnourished, mainly as a result of international sanctions. (21 Jan 98, Col 985–990.)
- The number of inpatient episodes for children under 16 with asthma fell from 64 500 in 1991–92 to 54 300 in 1995–96. (27 Jan 98, Col 23–24.)
- Total births in England, Wales, and Scotland in 1996 numbered some 710 000. The name of the father was not registered in about 8% of cases. (26 Jan 98, Col 104.)
- The government has set up a task group to consider what research is necessary to develop effective programmes to reduce the rate of unwanted pregnancies, especially among teenagers. (26 Jan 98, Col 73–74.)
- There are about 320 schools for children with emotional and behavioural difficulties in England. Fifteen such schools have closed since May 1997. (13 Feb 98, Col 253–254.)
- There is to be a free vote in the House of Commons on the abolition of corporal punishment in independent schools. (26 Feb 98, Col 360.)
- By the end of January 1998 a total of 1239 people with haemophilia had received compensation for HIV infection from blood products. The payments began in 1988 and have now reached £90 million. (6 Mar 98, Col 824.)
- The Transport Research Laboratory estimates that about 450 deaths and serious injuries each year would be prevented if the UK were to switch to European Central Time; reducing the driving limit for alcohol from 80 to 50 mg/100 ml would save another 300. (9 Mar 98, Col 16–17.)
- It has been estimated that some 37% of primary school children and 14% in secondary schools are bullied. In 10% and 4% the bullying occurs at least weekly. (16 Mar 98, Col 483–484.)

Correction

Two errors occurred in the medical education article "Current means of obtaining a MD degree in the UK" by Joseph Raine (*Arch Dis Child* 1998;78:174–7). Both in the abstract and results section the term "submitting" should have read "starting" (The time from starting the MD to confirmation that it had been obtained was 21–102 months (median 54).) Also in the results section it should have read "Six per cent of candidates had daily [not regular] meetings with their supervisors ..." These errors are regretted.