CASE REPORT

Cystic Fibrosis Compatible With a Full Term Army Engagement

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Abstract

A case of a specialist Senior Non-Commissioned Officer with Cystic Fibrosis (CF) is described. Partial expression of the CF trait is well known and sporadic cases are detected from time to time at recruitment, during recruit training and service. Respiratory symptoms may be mis-diagnosed as self-limiting asthma until a sweat chloride or other specific test for CF is performed.

Introduction

Cystic fibrosis (CF) is an inherited autosomal recessive disease caused by a mutation in the gene for the protein cystic fibrosis transmembrane conductance regulator (CFTR) leading to disordered regulation of the components of sweat, digestive juices and mucus. It is estimated that one in 25 people of European descent carry one gene for cystic fibrosis [1]. In 10-15% of CF patients with one severe and one mild CFTR mutation, or else two mild CFTR mutations, pancreatic exocrine sufficiency may permit normal growth and development. Whilst a known diagnosis of cystic fibrosis is a medical bar to military enlistment [1] genetic screening purely for pre-employment purposes is unlikely to be either ethically justifiable [2] or cost effective. As such, soldiers with mild CTFR mutations may present whilst in service.

Case Report

In 1989, a 17 year old Caucasian male with a childhood history of recurrent lower respiratory tract infections and sinusitis requiring washouts, was enlisted into the British Army. He successfully completed basic training but following a further respiratory tract infection was diagnosed with CF following a positive sweat chloride test, positive nasal Potential Difference (PD) and x-ray changes of early bronchiectasis (Figure 1). His CF genotyping at the time showed the Delta F508/N mutation. He nevertheless qualified as a Systems Engineering Technician, and for the first 10 years of his career was fully fit, after which he developed pancreatitis and was put on Creon tablets, subsequently undergoing laparoscopic cholecystectomy in 2000. He required six admissions for pancreatitis between 1999 and 2005. In Dec 2007 he suffered a further attack of pancreatitis. He smoked until November 2009.

Up until 2008, when not unwell he maintained fitness sufficient for his military duties, although his deployability outside UK since 1999 was very limited, reflected in his medical grading of P7 CPND (Geo) allowing deployments only to locations with unlimited access to first world medical care when the patient was stable and with the agreement of the local Senior Medical Officer. However following increased sputum production in mid-2008 he was unable to maintain military fitness, VO_2 max testing being associated with dyspnoea during the test and marked lassitude afterwards. He was investigated for apparent sleep apnoea, sleep studies revealing a slightly reduced mean saturation but his respiratory disturbance index was within normal limits. Medical boarding in July 2008 graded him non-deployable, as he was at that time unable to pass a Personal Fitness Test and undertook his own programme of PT.

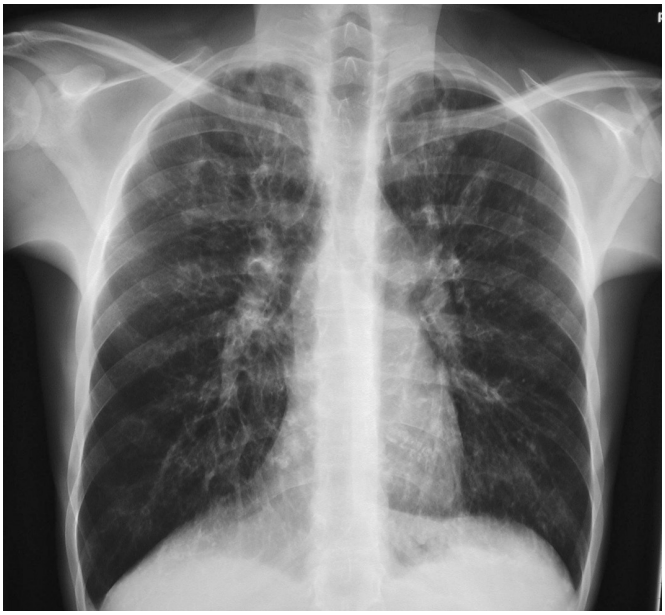

Figure 1. Chest X Ray of a case of Cystic Fibrosis showing accentuated hilar nodes and bronchial changes consistent with early atelectasis

Apart from an in-patient admission for pancreatitis in 2009, he continued to work in a non-deployable specialist capacity, which was valued by the Chain of Command as reflected by his senior appointment despite significant medical restrictions. His annual specialist review in 2010 noted that he continued to undertake physical training at least twice weekly and remained generally well, although noting some decrease in exercise tolerance over the past few years. *Staphylococcus aureus* was grown from his sputum for which he was successfully treated. He is expected to leave the British Army in 2011 on completion of a full 22 year engagement.

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Discussion

The diagnosis of cystic fibrosis is usually made within the first six months of life, but recently, the diagnosis of cystic fibrosis in adults has been reported with increasing frequency since the first adult report in 1946 [3]. This has led some researchers to believe that adults presenting with cystic fibrosis later in life constitutes a unique population of patients with different characteristics from those patients diagnosed earlier in life. Rodman et al [4] studied 55 patients who had been diagnosed with cystic fibrosis and divided them into an early diagnosis group (range, 0.1-15 years; n = 28), and a later diagnosis group (range 24-72.8 years, n = 7). Patients with a later diagnosis of cystic fibrosis had a significantly lower prevalence of pancreatic insufficiency and cystic fibrosis related diabetes; they also had better lung function. McCloskey et al [5] also studied patients diagnosed with cystic fibrosis from one centre in Northern Ireland and divided them into an early diagnosis or late diagnosis (after the age of 10). McCloskey, also found that later diagnosis was associated with pancreatic sufficiency, but had a weaker association with pulmonary function. McCloskey also looked at the genetics of the population who were diagnosed later in life and found that late diagnosis was significantly related to carriage of the R117H mutation.

Hodson et al [6], identified 366 patients who had survived 40 years and longer from four centres with large cystic fibrosis clinics (London, Minneapolis, Toronto, and Verona). In all the centres males survived longer than females, FEV_1 and BMI appeared to stabilise after 40 years of age. FEV_1 was on average 12% higher in patients who were pancreatic sufficient ($p > 0.0001$).

Inés et al [7] also studied adult cystic fibrosis patients treated at one centre and divided them into an early diagnosis (younger than 14 years, n = 39), and later diagnosis (over 14 years at diagnosis, n=50). Delta F508 was the most common genetic mutation present in 50 patients (56.2%). Twenty one of these patients were diagnosed early in life and 29 were diagnosed later. In the early diagnosis group, the Delta F508 mutation was associated with an exocrine pancreatic insufficiency in 26/29 patients with the mutation, where as in the late diagnosis group it was associated with pancreatic insufficiency in only 4/21 patients with the mutation (19%). Overall, pancreatic insufficiency was present in 33/39 patients (84.6%), and in 8/50 patients in the late diagnosis group (16%). More patients in the early diagnosis group were malnourished (10/39) compared with the late diagnosis group (2/50).

In the past CF has been viewed as an invariably fatal disease by, at best, early adulthood; however recent surveys have shown this not to be so. The variable genetic expression necessarily means that individuals with the CF trait can, as in this case, perform normally in the early years of their career, presenting with a typical symptomatology later. Colleagues in military medical services of other NATO nations report isolated similar cases [8] but for every one of these there are several more who have failed recruit training for lack of aerobic exercise capacity and subsequently are found to have CF, usually by sweat chloride test. We are aware of at least two such cases enlisted into the British Army during the past decade who were thus discharged.

A sweat chloride test although not especially expensive in laboratory costs (£200) nevertheless involves a significant investment in time by both investigator and patient to complete the test, and given the low numbers of CF positive personnel amongst those successfully enlisting for basic training we do not recommend sweat chloride estimations as a screening test for all recruits. Equally, pre-employment genetic testing is inappropriate as not only does it fail the Wilson and Jungner Screening criteria (9), but also it is ethically unjustifiable. At present the test may be undertaken on grounds of clinical indication by any physician, usually a specialist respiratory or general physician, caring for the military patient.

Conclusion

We recommend that an unexplained series of respiratory infections and/or failure at physical testing such as annual fitness tests should alert the military doctor to the possibility of CF, given the widespread distribution of the gene in the UK population.

References

1. Joint Services Publication. The influence of Particular Conditions on PULHHEEMS assessment for Entry. MOD. London 2006.346. Ch3 Leaflet 2
2. Faculty of Occupational Medicine. Guidance on Ethics for Occupational Physicians. FOM. London. 2006.
3. Hellerstein H. Cystic fibrosis of the pancreas in an adult. Ohio Med Journal 1946;42:616-7
4. Rodman D, Polis J, Heltshe S et al. Late diagnosis defines a unique population of long-term survivors of cystic fibrosis. Am J Resp Crit Care Med 2005;171:621-6
5. McCloskey M, Redmond A, Hill A, Elborn J. Clinical Features Associated with Delayed Diagnosis of Cystic Fibrosis. Respiration 2000;67:402-7
6. Hodson M, Simmonds N, Warwick W et al. International/ multicentre report in patients with cystic fibrosis (CF) over the age of 40 years. Cyst Fibros 2008; 10:1016:1-6
7. Modolell I, Alvarez A, Guarner L, .De Grazia J, Malagelada J. Gastrointestinal, Liver and Pancreatic Involvement in Adult Patients with Cystic Fibrosis. J Neuroend Tum Panc Dis Sci 2001;22(4):395-9
8. Lee R. Cystic Fibrosis in US Naval Personnel: Personal Communication. 2009
9. Wilson J, Jungner G. Principles and Practice of Screening for a Disease. World Health Organisation. Geneva.1968