CASE REPORT

Diagnosis of cystic fibrosis in a patient of Egyptian background

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SUMMARY

Cystic fibrosis (CF) is the most common genetically inherited condition in European-derived populations. However, it is being increasingly recognised in other populations, including people of Asian, Black African and Caribbean descent. We present a case detailing the diagnosis of CF in a 12-year-old patient of Egyptian background who had been treated for difficult asthma. In doing so, we aim to highlight the importance of considering CF in all patients, regardless of ethnicity.

BACKGROUND

Cystic fibrosis (CF) is an autosomal recessive disease caused by a mutation in the CFTR (cystic fibrosis transmembrane conductance regulator) gene on chromosome 7. It affects 1 in 2500 babies born in the UK and is more common among people of European-derived populations.¹ Although previously presumed to be rare, CF is being increasingly recognised among non-Caucasian populations and is thought to be underdiagnosed in these patient groups.² Newborn screening for CF was introduced in the UK in 2007. However, screening will still miss a proportion of the population and vigilance must be maintained for those born prior to 2007 or born abroad who will not have been screened.

CASE PRESENTATION

A boy aged 12 years presented to the tuberculosis clinic (TB) with a several year history of a chronically productive cough with associated shortness of breath and wheeze. He also reported of lethargy, night sweats and weight loss. He had been screened for TB, with a negative Mantoux test, 5 years previously. Initially, the patient had been managed by his GP who had referred him on to secondary care with suspected asthma. At this point, he was started on a budesonide with formoterol preventer inhaler (Symbicort) and terbutaline sulfate turbohaler (Bricanyl) to variable effect. Owing to his ongoing symptoms, a chest X-ray was performed which revealed bilateral hilar lymphadenopathy, resulting in a referral to the TB clinic.

Born in the UK in 2002 at 38 weeks gestation, he was the fourth infant of non-consanguineous parents of Egyptian background. He had been fully immunised, including the BCG vaccination, and had had no known TB contact. The only positive family history was of atopy. His parents report he developed a cough soon after birth which persisted for most of his life. He was first acutely unwell with a productive cough and weight loss at the age of 5 years while on holiday in Egypt. On return to the

UK, he was hospitalised and improved following a course of intravenous antibiotics. It was 3 years after this episode that he was started on asthma treatment following the development of a chronic cough.

On initial examination at the TB clinic, his chest was clear on auscultation and he had several small submandibular lymph nodes. His height and weight were on the 9th centile for his age. Repeat TB investigations, including Mantoux, T-spot and sputum AFB, were negative. His sputum culture grew *Staphylococcus aureus* and he was treated for a lower respiratory tract infection with 2 weeks of co-moxiclav (amoxicillin with clavulanic acid).

At follow-up, he was still symptomatic with a chronically productive cough and poor appetite. Further questioning revealed abdominal pain and steatorrhoea. Examination at this point revealed finger clubbing (figures 1 and 2), prompting further investigations into an underlying chronic respiratory condition.

INVESTIGATIONS

High-resolution CT chest showed widespread bronchiectasis with evidence of mucus plugging

Figure 1 Clubbing of the thumbs.

Figure 2 Finger clubbing left hand.

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Figure 3 High-resolution CT chest showing bronchiectasis and mucus plugging.

(figure 3). Sweat test revealed a high chloride level of 81 mmol/L, with >60 mmol/L reflecting a likely diagnosis of CF. Subsequent genetic testing confirmed CF with a c.1040G>C p mutation and Exon 14b deletion present. Lung function tests at the time of diagnosis revealed an FVC (forced vital capacity) of 73% predicted and an FEV1 (forced expiratory volume in 1 s) of 54% predicted. There was also evidence of pancreatic insufficiency, as his faecal elastase level was <15 µg.

DIFFERENTIAL DIAGNOSIS

For a patient presenting with chronically productive cough, the differential diagnoses would include CF, asthma, tuberculosis, primary ciliary dyskinesia, immune deficiencies as well as gastro-oesophageal reflux disease and recurrent aspiration. Subtle signs such as finger clubbing and nasal polyps may help guide towards a diagnosis of CF.

TREATMENT

Following the above investigations, the patient was started on regular pancreatic enzyme replacement therapy, fat-soluble vitamins, nebulised DNase and prophylactic flucloxacillin. He was also seen in the multidisciplinary CF clinic, receiving dietetic and physiotherapy input. He had his first isolation of *Pseudomonas* several months after diagnosis which led to a 2-week admission for intravenous antipseudomonal antibiotics. Following this, he was started on regular nebulised colistimethate sodium.

OUTCOME AND FOLLOW-UP

After the initiation of treatment, the patient's long-standing symptoms, including cough and loss of appetite, improved. His weight and height increased to between the 9th and 25th centile. In addition, his lung function improved with an FVC of

89% predicted and FEV1 of 90% predicted following his admission for intravenous antipseudomonal antibiotics. He remains under regular review at the CF clinic.

DISCUSSION

In the last few decades, CF has been increasingly diagnosed in Latin America, the Middle East and in those from the Indian subcontinent.² There are little available data on the incidence of CF in Egypt. One of the only studies attempting to evaluate the number of patients with CF in the country was conducted in 1993 and involved over 18 000 newborns. It estimated the prevalent rate of CF in Egypt to be 1 in 2664.³ Supporting this, a study from neighbouring Jordan estimated the incidence of CF to be 1 in 2560.⁴ In a more recent study, 100 patients who were clinically suspected of having the disease were recruited for CF screening. CF was diagnosed in more than one-third of patients, suggesting that the condition may be going undetected due to a low index of suspicion.⁵

The growing recognition of CF in non-Caucasian populations, as supported by these studies, has implications for how CF is viewed among clinicians. The perception of CF as a condition solely attributed to people from European-derived populations must change to reflect our current understanding. Our case reported a patient with CF who was from an Egyptian background and had the classic signs and symptoms of CF. We wanted to highlight the importance of considering CF as a differential diagnosis in people regardless of their ethnic origin.

Learning points

- ▶ Look for clubbing in all patients with a chronic cough.
- ▶ Consider other diagnoses in patients diagnosed with asthma who are not responding to treatment.
- ▶ Be aware that although newborn screening exists since 2007, this is not 100% sensitive and may have missed older children and those not born in the UK.
- ▶ Consider a diagnosis of cystic fibrosis in patients presenting with appropriate symptoms regardless of ethnic origin.

Contributors Research and original writing conducted by RY. Revised by BR.

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REFERENCES

- 1 Davies JC, Alton EW, Bush A. Cystic fibrosis. *BMJ* 2007;335:1255–9.
- 2 World Health Organisation. *The Molecular Genetic Epidemiology of Cystic Fibrosis: Report of a Joint Meeting of WHO/ECFTN/ICF(M)/A/ECFS*. 19 June 2002. Genoa, Italy. http://www.who.int/genomics/publications/en/HGN_WB_04.02_report.pdf
- 3 Abdel-Salam E, Samuel S, Awad M, et al. Cystic fibrosis in Egyptian children: neonatal screening and high risk groups. *JAC* 1993;4:313–17.
- 4 Nazer HM. Early diagnosis of cystic fibrosis in Jordanian children. *J Trop Pediatr* 1992;38:113–15.
- 5 El-Falaki MM, Shahin WA, El-Basha NR, et al. Profile of cystic fibrosis in a single referral center in Egypt. *J Adv Res* 2014;5:563–8.

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