

## **Case series: Tuberculosis (TB): Where Are We Heading?**

**DR M Ranjn, Dr V Cheruvalli, Northern Lincolnshire and Goole NHS Trust**

### Introduction

CF is a genetic condition. People are born with CF and cannot catch it later in life.

To have cystic fibrosis, an individual must carry two gene mutations. The new-born screening looks for the most common mutations. Hence, it's possible for someone with a rare genetic mutation to be missed, until later in life. Cystic fibrosis can vary widely in its severity and symptoms, and can mimic other lung diseases such as asthma, making diagnosis challenging

### Case report

A 10yrs old child attended emergency department as she collapsed in the school following a coughing bout. The history revealed recurrent productive cough for nearly two years with weight loss. She was managed at primary care as recurrent infective exacerbation of asthma.

She had received 6 courses of antibiotics and her sputum showed repeated heavy growth of Haemophilus influenza in the months preceding admission.

Assessment revealed poor growth, finger clubbing and coarse crepitation on chest. The CXR showed extensive abnormal density of the lung fields. CT chest showed moderately severe diffuse cylindrical bronchiectasis involving both lung fields. Her sweat test showed chloride of 40mmol/l (sweat collected 20ul) initially and later chloride of 56mmol/l (sweat collected 45ul). Genetic testing showed single mutation.

Hence, she was referred to tertiary CF team. After discussing with the family, she is being treated as CF based on her clinical course, sweat test & genetic mutation. They have planned to repeat genetic test for further mutation and repeat the sweat test.

### Conclusion:

This case illustrates

1. The importance of revisiting the diagnosis when the presentation is unusual
2. New-born screening failures do occur
3. Single gene mutation does not imply CF, mutation analysis usually done for common mutations.
4. As CF has lifelong implications, diagnosis must be considered carefully