

Title	Late Diagnosis
Version	1.0

1	Background
2	Clinical Presentation
	Clinical Severity for Late Diagnosis of CF
3	Confirming the Diagnosis of Cystic Fibrosis
4	Other investigations
5	Psychosocial Impact of Late Diagnosis
6	Helpful Resources

1. Background

- Although the majority of patients with cystic fibrosis are now diagnosed by newborn screening, late diagnoses still occur. This can occur at any time from infancy to old age
- Patients more likely to diagnosed later in life include:
 - Those who did not undergo CF newborn screening
 - Those with CFTR mutations associated with a milder phenotype
 - Those with non-Caucasian ethnicity

2. Clinical Presentation

- There are a wide range of potential presentations for individuals with a late diagnosis of CF. These include:
 - Recurrent lower respiratory tract infections
 - Chronic wet cough
 - Confirmed bronchiectasis
 - Failure to thrive
 - Recurrent rectal prolapses
 - Sinus disease / nasal polyps
 - Recurrent pancreatitis
 - Infertility

3. Clinical Severity for Late Diagnosis of CF

There are few data on the clinical outcomes of individuals diagnosed with CF in later life. The Canadian CF Registry (Reference 1) does provide reassuring data, with those who received a diagnosis as adults tending to have a milder disease than the general CF population, with

- a median forced expiratory volume in 1 second of 81%,
- lower prevalence of pancreatic insufficiency (15%),
- less CF-related diabetes (3%), and
- a lower culture positivity with *Pseudomonas aeruginosa* (30.4%).

The CFTR mutations in this Registry group differ in prevalence from those described for the CF population as a whole, with only 4.7% of patients being F508del homozygous and 38.1% being F508 heterozygous as opposed to >45% F508del homozygous and >40% F508del heterozygous.

4. Confirming the diagnosis of CF

- A sweat test required to confirm the diagnosis of CF
- This is supported by identification of two CFTR mutations

5. Other investigations

- Other investigations may be undertaken around the time of the CF diagnosis to assess the severity of the lung disease, the presence of CF complications and guide treatment options. These include:
 - Chest x-ray
 - High resolution chest CT scan
 - Flexible bronchoscopy
 - Sputum culture / cough swab
 - Abdominal USS

- Faecal elastase
- Glucose tolerance test
- Blood tests as per annual review

6. Psychological impact of late diagnosis

- A late diagnosis of CF can have a huge psychological impact on the individual and those close to them. This can potentially adversely affect adherence with treatment. Early input from CF Psychologist is a vital to support the individual through this difficult time.

7. Helpful Resources

- The CF Trust has a helpful publication on the late diagnosis of CF, available on its website: [Late diagnosis factsheet Sep 2020.pdf \(cysticfibrosis.org.uk\)](https://www.cysticfibrosis.org.uk/late-diagnosis-factsheet-sep-2020.pdf)

8. References

1. Desai S, Wong H, Sykes J, Stephenson AL, Singer J, Quon BS. Clinical characteristics and predictors of reduced survival for adult-diagnosed cystic fibrosis: analysis of the Canadian CF Registry. *Ann Am Thorac Soc* 2018;15:1177–1185.