Title	Cystic Fibrosis Screen Positive, Inconclusive diagnosis CFSPID
Version	1.0

1. Introduction

An unintended consequence of CF newborn screening is the identification of infants with an inconclusive diagnosis. This is called CF Screen Positive, Inconclusive diagnosis (CFSPID). The majority of these infants remain healthy and they should not be treated as though they have CF. However, a small number may convert to a diagnosis of CF and those that do not, may develop a CF Transmembrane Conductance Regulator Related Disorder (CFTR-RD) later in life. The most common CFTR-RD are congenital bilateral absence of the vas deferens and pancreatitis.

2. CFSPID Definition:

An asymptomatic infant with a positive NBS result for CF and either:

1. A sweat chloride value <30 mmol/L and two CFTR variants at least one of which has unclear phenotypic consequences OR
2. An intermediate sweat chloride value (30–59 mmol/L) and one or zero CF causing variants

3. Follow-up for CFSPID

This does not need to be done in the CF clinic. After the initial assessment, children should be reviewed in clinic at 6 months of age and then annually until the age of 6 years. At 6 years of age, the child should undergo a more extensive evaluation and may be considered for discharge if all if normal.

4. Investigations

- A sweat test should be performed as part of the initial assessment and then at 6 months, 2 years and 6 years of age.
- A faecal elastase should be performed as part of the initial assessment and then at 12 months and 6 years of age.
- Unless clinically indicated, a chest x-ray does not need to be performed until 6 years of age.
- Spirometry and LCI (if available) should be performed aged 6 years.
- Respiratory cultures do not need to be performed unless the child is symptomatic (persistent productive cough).

5. Management

There is no evidence to support the routine use of CF therapies for the treatment of CFSPID infants/children.