

Cystic Fibrosis Genetic Testing Questionnaire

Return completed form to: Duty Scientist, Molecular Genetics, Department of Clinical Genetics, Children's Health Ireland at Crumlin, Cooley Road, Dublin D12 N512 or fax to (01) 4096971

Patient name: _____

Date of birth: _____

Ethnicity¹: _____

Is this patient/this patient's partner currently pregnant? yes / no

GP/Consultant name and address: _____

Indications for testing:

- Suspected diagnosis of symptomatic patient
- Patient has definite clinical diagnosis of CF
- Carrier testing as a prerequisite for assisted reproduction
- Carrier testing due to family history of CF in this patient or their partner²

Clinical symptoms for diagnostic requests:

Include sweat test result if available and whether sweat chloride or conductivity was measured³

Family History:

- No known family history of CF
- Family history of CF present (*complete all fields*)
 - Name of family member _____
 - Date of birth of family member (*required*) _____
 - Are they affected with CF or a carrier? _____
 - Genetic test results (*if available*)⁴ _____
 - Relationship to patient _____
- Partner has family history of CF / is a confirmed CF carrier / has a diagnosis of CF
 - Partner's name _____
 - Partner's date of birth _____
 - Partner's family history _____
 - Has partner been tested for CF? (*provide genetic test results if available*)⁴ _____

1. Our testing panel detects approximately 93.5% of the CF mutations found in the Irish population. Coverage may be reduced or unknown for other populations.

2. The recommended minimum age for carrier testing is 16 years in accordance with internationally recognised guidelines.

3. Sweat chloride reference values in patients >6 months of age: <40mmol/L – negative; 40 – 60mmol/L – borderline/equivocal; >60mmol/L – consistent with a diagnosis of CF. Sweat conductivity alone is not an adequate test for diagnosis of CF.

4. Include copy of genetic test report if individual was not tested in Department of Clinical Genetics, Children's Health Ireland at Crumlin.