Cystic fibrosis genetic testing questionnaire

Return completed form to: Duty Scientist, Molecular Genetics, Department of Clinical Genetics, Our Lady's Children's Hospital, Crumlin, Dublin 12, D12 N512 or fax (01) 4096971

Patient name: _____

Date of birth: _____

Ethnicity[1]: _____

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 partner[2]

Clinical symptoms for diagnostic requests:

Include sweat test result if available and whether sweat chloride or conductivity was measured[3]

_____

_____

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 Our Lady's Children's Hospital, Crumlin.