**National Newborn Bloodspot Screening Programme**

**Notification of cystic fibrosis (CF) diagnosis for babies/children not identified through newborn bloodspot screening**

Newborn bloodspot screening (NBS) for cystic fibrosis (CF) was introduced in Ireland in July 2011. In order to understand how the programme is performing it is important to have a system of ongoing surveillance for cases of CF that went undetected by the CF NBS programme. Timely reporting of these cases results in a prompt and detailed review of the case by the Newborn Bloodspot Screening Programme Governance Group, and the case is included in the annual statistics of the CF NBS programme.

If you become aware of a case of CF recently diagnosed (by any means) in a child who was born in Ireland on, or after July 1st 2011, please complete this form and return to the National Newborn Bloodspot Screening Laboratory Temple Street, as a matter of urgency.

**Please send completed form to:**

National Newborn Bloodspot Screening Laboratory

Children’s Health Ireland at Temple Street.

Temple Street

Dublin 1

D01YC67

**Email:** [info.newbornscreening@cuh.ie](mailto:info.newbornscreening@cuh.ie)

**Please note:**

**Sections A and B to be completed by the Adult/Paediatric Cystic Fibrosis Centre reporting the case**

**Sections C and D to be completed by the National Newborn Bloodspot Screening Laboratory Children’s Health Ireland at Temple Street**

**Section A: Adult/Paediatric Cystic Fibrosis Centre**

|  |  |
| --- | --- |
| **Name of person completing this form:** |  |
| **Role on the team:** |  |
| **Today’s date:** |  |
| **Contact email address:** |  |
| **CF Consultant Paediatrician:** |  |
| **Specialist CF Nurse:** |  |
| **Paediatric CF Centre:** |  |

**Section B:**

1. **Details of child**

|  |  |
| --- | --- |
| **Full Name:** |  |
| **Date of Birth:** |  |
| **Hospital of Birth:** | If home birth please tick |
| **Gestational age at birth:** |  |
| **Maternal Surname:**  **(if different from child’s surname):** |  |
| **Ethnicity:** |  |
| **Was this child born in Ireland:** | Yes  No |
| **Current family address:**  **(Include eircode if known)** |  |
| **Address at birth (if different):**  **(Include eircode if known)** |  |
| **Date of referral to CF centre:** |  |
| **Date of first assessment at CF centre:** |  |
| **Date that CF diagnosis confirmed:** |  |

1. **Relevant clinical history – please tick all that apply from the following:**

|  |  |
| --- | --- |
| **Acute or persistent respiratory symptoms** |  |
| **Blood Transfusion** |  |
| **Electrolyte imbalance** |  |
| **Failure to thrive/malnutrition** |  |
| **Family history of CF** |  |
| **Genotype consistent with CF** |  |
| **Liver problems** |  |
| **Meconium ileus/other intestinal obstruction** |  |
| **Nasal polyps/sinus disease** |  |
| **Oedema** |  |
| **Prenatal screening (CVS, amnio)** |  |
| **Rectal prolapse** |  |
| **Steatorrhea/abnormal stools/ malabsorption** |  |
| **Unknown** |  |
| **Viral Illness** |  |
| **Other**  **(Specify)** |  |

1. **Investigations**

|  |  |  |
| --- | --- | --- |
| **Date of sweat test:** |  | |
| **Sweat chloride (result):** | mmol/L | |
| **Interpretation of sweat test result:** | Positive: |  |
|  | Equivocal  (30-60): |  |
|  | Negative: |  |
| **DNA analysis for CFTR mutations performed in the Dept of Clinical Genetics, Crumlin:** | Yes  No | |
| **Results of DNA analysis for CFTR performed in Dept of Clinical Genetics, Crumlin:** | Mutation 1:  Mutation 2: | |
| **DNA analysis for CFTR mutations performed in a laboratory other than the Dept of Clinical Genetics, Crumlin:** | Yes  No | |
| **Name of the laboratory and contact details:** |  | |
| **Did this testing involve Extended Gene Analysis (EGA) or a CF mutation panel, or other test** | EGA  CF mutation panel  Other (specify details): | |
| **Results of DNA analysis for CFTR mutations performed in the laboratory other than the Dept. of Clinical Genetics, Crumlin:** | Mutation 1:  Mutation 2: | |
| **Any other investigations (Nasal PD, jejunal Bx):** |  | |

1. **Genetic Follow On**

|  |  |
| --- | --- |
| **Referral made to genetic counselling:** | Yes  No |
| **Seen by genetic counsellor:** | Yes  No |
| **Parents tested for CF mutations:** | Yes  No |
| **Results of parental testing for CF mutations:** | Fathers genotype:  Mother genotype: |

**Section C – To Be Completed by the Newborn Bloodspot Screening Laboratory**

**Screening result**

|  |  |  |  |
| --- | --- | --- | --- |
| **Date of bloodspot sample(s):** |  | |  |
|  | |  |
|  | |  |
| 1. **IRT dried bloodspot result(s):**  * **Please include the IRT concentration** * **Please record the laboratory cut-off value in use at the time the sample was taken**   **IRT units ng/ml** | IRT concentrations <99th centile  Please record all IRT results | | IRT =  Cut off = |
| IRT concentrations ≥ 99th centile go to Q.2  Please record all IRT results | | IRT =  Cut off = |
| 1. **CF mutations detected?** | Yes  No | |  |
| If any mutation(s) detected please specify the mutation(s) | | Mutation 1:  Mutation 2: |
| No mutations detected with IRT concentration ≥99th centile | |  |
| **Infant referred to specialist CF centre for sweat test?** | Yes  No | | |
| **Name of specialist CF centre:** |  | | |
| **Date of Referral:** |  | | |
| **Date of successful Sweat Test:** |  | | |
| **Sweat chloride (result)** | mmol/L | | |
| **Interpretation of sweat test result:** | Positive |  | |
|  | Equivocal (30-60) |  | |
|  | Negative |  | |

**Section D:** **To Be Completed by Newborn Bloodspot Screening Laboratory after full review carried out into the undetected case**

**Reason**

|  |  |
| --- | --- |
|  | **Please tick** |
| **Not screened (not eligible)** | |
| Baby/child was not resident in Ireland for the first 6 weeks of life |  |
| Baby died prior to screening (but CFTR performed, no newborn bloodspot sample collected) |  |
| **Not screened (eligible)/diagnosis made before screening result** | |
| Baby/child identified as at risk (e.g. family history, antenatal anomaly) |  |
| Prenatal diagnosis |  |
| Baby diagnosed following meconium ileus  If Yes, record IRT concentration |  |
|  |
| Parents declined newborn bloodspot screening including CF  (If so, has opt out form been completed?) |  |
| Parents declined further investigations |  |
| **False negative screen** | |
| Baby/child who was screen reported as ‘Not suspected’ for CF  Baby /child who was screen reported as ‘carrier’ or at risk of CF following mutation analysis with one mutation detected and normal sweat test |  |
| Had any of the following occurred (please specify):   * Later screening of a moved-in or ‘older’ baby unsuitable for CF screening as older than 6 weeks at time of NBS sample * Delayed transit of sample (>14 days) and no repeat sample received * Blood transfusion < 72 hours and no valid repeat received * Blood transfusion < 72 hours and transfusion status not recorded on the newborn bloodspot screening card * Contaminated card with no repeat sample available/received * Meconium Ileus not recorded on screening card |  |
| **Screening programme deviation** | |
| No sample taken  Please specify why: |  |
| First or further samples taken after 6 weeks old and no sweat test performed |  |
| Sample did not arrive in laboratory/no record of sample |  |
| Repeat sample request not followed up by NNBSL or sample taker |  |
| Test failure/laboratory error (specify details)   1. NBS Lab: |  |
| 1. Genetic Lab: |  |
| Screen positive baby not referred by NNBSL |  |
| Non-attendance by parents/guardians for clinical appointments |  |