Infants with positive screening living within the boundary of Cardiff and Vale University Health Board

- Fax notification is received from New Born Screening Laboratory outlining patient details, IRT result and if one or two mutations are present. A plan is discussed for each new referral between consultant of the week and cystic fibrosis nurse specialist (CFNS).
- General Practitioner (GP) and health visitor (HV) are contacted by CFNS to discuss the result and ascertain the condition of the infant. If the infant is symptomatic it may be appropriate to arrange prompt review. If the family are not known to the HV it may be appropriate to contact the named midwife. Fax confirmation is sent to the GP and HV, including information re seeking advice if infant presents in the interim and guidelines for arranging the joint home visit (appendix 1 and 2)
- At this stage the GP and HV are requested not to discuss this information with the family. The HV is asked to arrange a joint home visit for the day prior to the sweat test, informing the parents the evening before or morning of the proposed visit to minimise the period of anxiety for the parents. An invitation is also extended to the HV to accompany the family on the day of the sweat test.
- Sweat tests are usually arranged for a Thursday via Heather Wheatley (biochemist) (extension 3560) at a time when both Consultant and Nurse Specialist are available. In the unlikely event of a Consultant not being available the SpR may be an appropriate alternative.

Joint home visit

- The screening result and the necessity for a sweat test are explained to the family. Genotype is not routinely discussed with families but the information is not with-held if requested
- Further information is provided according to the families needs.
- Medical history is taken and advice sought from Consultant of the week if there are any concerns regarding the infant.
- Written information and contact numbers are provided (appendix 3) including a sweat test information leaflet (appendix 4)

Day of the sweat test

The family and HV (if present) are met by one of the CFNS. Any questions are answered before introducing them to the biochemist. The results are usually available within 2 hours and a time arranged to meet with them. Results are always given face to face.

Sweat test results

Normal sweat test
(sweat chloride < 30 mmol/litre) and 1 CF mutation

- CFNS +/- consultant explain the sweat test is entirely normal, confirming the baby is a healthy unaffected carrier.
- Discuss referral to medical genetics. If parents agree referrals should be made to Dr Angus Clarke. Include parents' date of birth in the referral.
- Provide family with CF Trust 'Genetics' booklet.
- Inform Health Visitor of the result by telephone.
- Send Summary to GP and copy to parents Inform Dr Stuart J Moat or his deputy (Department of New Born Screening) of resul

Borderline/equivocal sweat test
(sweat chloride 30-60 mmol/litre) and 1 CF mutation

- CF Consultant +/- CFNS explain the result to the family
- Arrange to repeat sweat test
- Provide family with CF Trust 'Genetics' booklet
- Discuss referral to medical genetics. If parents agree referrals should be made to Dr Angus Clarke. Include parents date of birth in the referral
- Inform HV of result by telephone.
- Send summary to GP and copy to parents
- If two equivocal sweat tests consider sending DNA for further mutation analysis.
- Inform Dr Stuart J Moat or his deputy (Department of New Born Screening) of resul

Consistent with CF
(sweat chloride >60mmol/litre) and 1 or 2 CF mutations

- CF Consultant +/- CFNS explains the result to the family and counsel appropriately
- Repeat sweat test if only one mutation identified.
- Provide family with written information and contact numbers (appendix 5)
- Inform HV of result by telephone
- Send summary to GP
- Admit patient if symptomatic
- Commence treatment as appropriate (see management of newly diagnosed infant (appendix 6
- Inform Dr Stuart J Moat or his deputy (Department of New Born Screening) of result

Infants identified living outside Cardiff and Vale University Health Board

- Each local DGH should have a policy for disseminating to parents an abnormal CF newborn screening result.
- Fax notification is received from New Born Screening Laboratory outlining patient details, IRT result and if one or two mutations are present. A plan, including appropriate course of action and communication process is discussed for each new referral between UHW consultant of the week and CFNS.
- Availability of lead clinician in the relevant DGH is confirmed. If he/she is not available to meet with the family and perform the sweat test within a week of notification, alternative arrangements should be made to review the infant at the Children's Hospital for Wales, Cardiff (South Wales Patients). Those patients who reside in North Wales receive their shared-care with Liverpool and need to liaise with them as appropriate.
- Please confirm receipt of newborn screening report and report results of sweat test to the team at UHW when available (appendix 7)
- Dr Stuart J Moat or his deputy (Department of New Born Screening) also needs to be informed of result

Information for local clinics

Within Wales the majority of newly diagnosed infants with cystic fibrosis are identified as a result of newborn screening. This protocol has been written to ensure that information is disseminated between health professionals and parents of infants with an abnormal result in an efficient and effective manner.

Aims

- To diagnose the majority of patients with Cystic Fibrosis within four weeks of birth.
- To minimise the period of uncertainty for families. Ideally there should be less than 24 hours before the family being notified of an abnormal result and the sweat test being performed.
- Families should be be fully aware of, and understand the need for a sweat test

Objectives

- To achieve a co-ordinated approach between CF centre, local teams and members of Primary Care Team. This is usually through the General Practitioner and Health Visitor but may also include the midwife if appropriate.
- To minimise potential harm to families of infants identified as having an abnormal newborn screening result.
- To ensure that all affected families in Wales receive an equitable standard of care, irrespective of geographical location.

Process

- Each local DGH should have a policy for disseminating to parents an abnormal CF newborn screening result.
- If the sweat test is not able to be performed within a week of notification please seek advice re further management from either Dr Iolo Doull or Dr Julian Forton (South Wales patients)

Sweat test results

Normal sweat test

(sweat chloride < 30 mmol/litre) and 1 CF mutation

- Explain the sweat test is entirely normal, confirming the baby is a healthy unaffected carrier
- Offer referral offered to clinical genetics service for cascade screening.
- Provide family with CF Trust 'Genetics' booklet
- Inform Health Visitor of result
- Send letter sent to GP and parents and confirming child does not have CF.

Borderline/equivocal sweat test

(sweat chloride 30-60 mmol/litre) and 1 CF mutation

- Explain the result to the family
- Discuss with Dr Iolo Doull or Dr Julian Forton
- Repeat sweat test and ? DNA required for further mutation screening
- Arrange appropriate follow up appointment
- Offer referral offered to clinical genetics service for cascade screening
- Inform GP and Health Visitor of result

Consistent with CF

(sweat chloride >60mmol/litre) and 1 or 2 CF mutations

- Explain the result to the family and counsel appropriately
- See standards of care for infants in the first two years of life (appendix 5)
- Admit patient if symptomatic
- Provide with appropriate written information and contact numbers
- Inform Dr Iolo Doull or Dr Julian Forton and make arrangements for patient to meet the specialist CF team at the earliest opportunity, or at the following shared care clinic
- Inform GP and Health Visitor of result

Insufficient sweat

- Repeat sweat test as and when appropriate.
- Consider whether it is appropriate for it to be performed at Children's Hospital for Wales.

Appendices

appendix 1: Confirmation of abnormal screening test for cystic fibrosis (GP)
appendix 2:Confirmation of abnormal screening test for cystic fibrosis (HV)
appendix 3: Letter to family re abnormal screening test for Cystic Fibrosis
appendix 4
appendix 5: Standards of Care for Infants with Cystic Fibrosis in the First Two Years of Life: A Consensus Statement for Wales
appendix 6: Information for parents of a child newly diagnosed with Cystic Fibrosis
appendix 7: Suggested standard letter for GP and parents of child with negative sweat test
appendix 8: Letter to DGH consultant re abnormal screening result