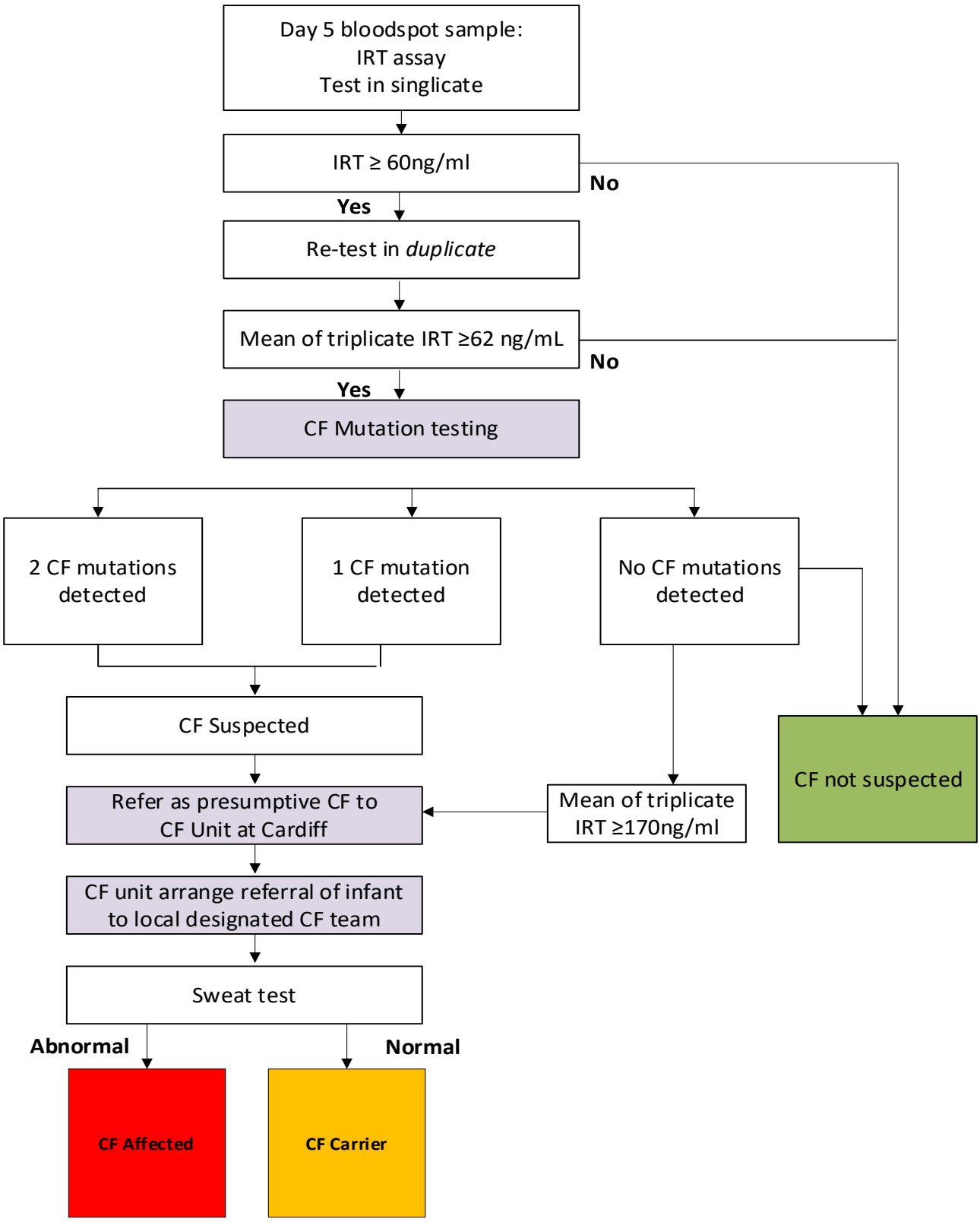
Sgrinio Smotyn Gwaed Newydd-anedig Cymru
Newborn Bloodspot Screening Wales

Cystic Fibrosis (CF) Clinical Referral Guidelines for Newborn Bloodspot Screening Wales

October 2020 Version 1

CF newborn screening protocol



Stage of Process	Guidelines
Defining a CF suspected screening result:	If a sample from a baby is found to have an immunoreactive trypsinogen (IRT) concentration equal to or greater than 60ng/ml, a repeat test should be performed in duplicate on the original bloodspot card. If the mean of triplicate IRT is greater than 62ng/ml the sample is sent to for DNA analysis. If no CF gene variants are detected and the IRT is <170ng/ml, **CF is not suspected**. If there are one or two CF gene variations detected then **CF is suspected**. If there are no CF gene variants detected and the IRT is greater or equal to 170ng/ml then **CF is suspected**.
Referral of babies with a positive ('CF suspected') screening result:	Wales Newborn Screening Laboratory notifies the lead consultant for CF AND the CF team at the Children's Hospital for Wales (CHfW) on the day of the result. The clinical referral must be by email using a standard template letter. The CF team at CHfW will contact the local CF team, ensuring that there is local expertise available.

First family contact	The local team cystic fibrosis nurse specialist (CFNS) or consultant will contact the baby's GP and Health Visitor (HV) to discuss the result and ascertain the condition of the baby. At this stage they are requested not to discuss the newborn screening result with the family. If the baby is symptomatic it may be appropriate to arrange a prompt review.

	A joint visit of the CFNS and the health visitor is planned the day prior to the sweat test to allow information to be shared with the family by a professional experienced in CF care and preferably accompanied by a professional who knows the family. The HV is asked to arrange a home visit for the day prior to the sweat test. On the morning of the planned visit the family should be contacted again to inform them that a nurse from the hospital will be accompanying them to discuss newborn screening result. If the family asks for further information they can be informed it is the CF screening result. In circumstances when a CFNS is unavailable to attend the visit, the HV can liaise with the CFNS and respiratory consultants for further information and support prior to the visit. The CF Trust recommend that this information is given by someone who has comprehensive knowledge of CF (Cystic Fibrosis Trust, (2011) Standards for the clinical care of children and adults with cystic fibrosis in the UK .2nd edition). In some areas of Wales specialist training and support is provided to HVs to enable them to undertake this visit confidently. At this visit, the screening result and the necessity for a sweat test are explained to the family. Genotype is not routinely discussed with the families but the information is not withheld if asked for. Written information and contact numbers are provided.

First family face to face review with lead consultant for CF **(Sweat test**)	This review should occur within **28 days of birth**. The family is met by the CFNS prior to the sweat test and any further questions are answered. The sweat test is undertaken and the results are available the same day. The results will be given face to face. Sweat chloride concentration less than 30mmol/L with no or 1 CF variant – **Unaffected carrier**. If 1 variant

	identified, offer referral to genetics service to discuss cascade screening. Equivocal sweat chloride concentration (30-59mmol/L) with no or 1 CF variant – arrange to repeat sweat test in a timely manner. Sweat chloride concentration greater than or equal to 60mmol/L - **Consistent with CF** Insufficient sweat – repeat sweat test within a week. (RCPCH endorsed guidance Guidelines for the Performance of the Sweat Test for the Investigation of Cystic Fibrosis in the UK v.2)

Upon confirmation of diagnosis	Obtain a full medical and family history. Collect a stool sample for faecal elastase. Arrange extended CFTR variant testing (if only one CF gene variant identified). Commence pancreatic enzyme replacement therapy (creon micro). Consider commencing fat soluble vitamin supplements. Obtain cough swab for microscopy, culture and sensitivity. Document weight and length. Introduce to physiotherapist and dietitian within 7 days. Provide written information and contact telephone numbers. Discuss sweat testing siblings. Discuss entry of data onto the CF registry. Offer referral to genetics service to discuss cascade screening.