BC Newborn Screening Program Information Sheet

Positive Newborn Screen for Cystic Fibrosis: A Discussion Guide for Health Care Providers

This discussion guide is intended to help you counsel a family whose newborn has had a positive screen for Cystic Fibrosis (CF). A newborn "screens positive" for CF when his or her immunoreactive trypsinogen (IRT) level measures above the screening cutoff in a blood sample collected from the newborn's heel 24 hours after birth. This elevated IRT triggers the newborn screening lab to perform a DNA test that looks for the most common genetic changes (mutations) associated with CF. When a newborn is found to have one or more CF mutations, or the initial IRT level is very high, then further testing is recommended. Remember that "screen positive" means that the newborn MIGHT have CF but more tests are needed to determine if the newborn truly has CF.

You may choose to share this information sheet with the family, following your discussion, to help them understand their newborn's specific result. We have also included a more general information sheet for the family outlining the steps that will be taken to confirm or rule out CF. This newborn screening result will fall into one of the categories list below depending on the results of the initial IRT and DNA testing already completed.

Initial Newborn Result	**High IRT and <u>two</u> CF mutations**	**High IRT and <u>one</u> CF mutation**	**Very high IRT and <u>no</u> CF mutations**
Likelihood of CF	High	Moderate (less than 5% or 1/20)	Low (less than 1% or 1/100)
Next Step	Referral to BC Children's Hospital for Sweat Testing	Repeat blood spot card (IRT) at day 21 of life	Repeat blood spot card (IRT) at day 21 of life
Possible second IRT results	N/A	<u>Second IRT Elevated:</u> The newborn MAY have CF but a sweat test is required. This testing will be performed at BC Children's Hospital. <u>Second IRT normal:</u> CF is UNLIKELY. The newborn is most probably an unaffected carrier for CF. Although the risk of CF is very small (<1%), the family can opt to have sweat testing performed at BC Children's Hospital to definitively rule out CF.	<u>Second IRT Elevated:</u> The newborn MAY have CF but a sweat test is required. This testing will be performed at BC Children's Hospital. <u>Second IRT normal:</u> CF is HIGHLY UNLIKELY and no further testing is required.

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Initial Newborn Result	High IRT and <u>two</u> CF mutations	High IRT and <u>one</u> CF mutation	Very high IRT and <u>no</u> CF mutations
Possible Sweat testing results	<u>Abnormal:</u> This confirms the diagnosis of CF. The newborn will be referred to the CF Clinic at BC Children's Hospital. <u>Borderline:</u> The diagnosis of CF has been neither confirmed nor excluded. Further follow-up is required (repeat sweat testing and referral to the CF Clinic) <u>Normal:</u> This is an unexpected result. More testing will be required (repeat sweat testing and DNA testing) <u>Insufficient Quantity:</u> Not enough sweat was collected. The lab will arrange repeat testing.	<u>Abnormal:</u> This confirms the diagnosis of CF. The newborn will be referred to the CF Clinic at BC Children's Hospital. <u>Borderline:</u> The diagnosis of CF has been neither confirmed nor excluded. Further follow-up is required (repeat sweat testing and referral to the CF Clinic) <u>Normal:</u> The newborn DOES NOT have CF. Since one CF mutation was detected this newborn is a CF carrier. <u>Insufficient Quantity:</u> Not enough sweat was collected. The lab will arrange repeat testing.	<u>Abnormal:</u> This confirms the diagnosis of CF. The newborn will be referred to the CF Clinic at BC Children's Hospital. <u>Borderline:</u> The diagnosis of CF has been neither confirmed nor excluded. Further follow-up is required (repeat sweat testing and referral to the CF Clinic) <u>Normal:</u> The newborn DOES NOT have CF, not further testing required. <u>Insufficient Quantity:</u> Not enough sweat was collected. The lab will arrange repeat testing.
Genetic Counselling recommended	<u>Yes</u> We expect that both parents are CF carriers. Information about future reproductive risks and testing for other family members should be discussed.	<u>Yes</u> We expect that at least one parent is a CF carrier. Information about future reproductive risks and testing for other family members should be discussed.	<u>Possible</u> Genetic counseling recommended if CF is confirmed as we expect that both parents are carriers of a rare CF mutation.

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