Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Normal	Cystic Fibrosis	Normal			
Normal	Cystic Fibrosis	Normal	Immunoreactive Trypsinogen CFTR Mutation	Normal 0 Mutations Detected	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Normal. None of the CFTR variants in the DSHS panel were detected. However, the presence of other variants not included in the panel cannot be ruled out.
Abnormal	Cystic Fibrosis	Indeterminate	Immunoreactive Trypsinogen	Elevated	Repeat the newborn screen within 72 hours. Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level on the first specimen. The second screening specimen is required to determine if result is significant.
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen	Elevated	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. Elevated IRT level is consistent with the previous newborn screening result. None of the CFTR variants in the DSHS panel were detected in the previous specimen.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen	Elevated	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. This result is consistent with the previous newborn screening specimen results for the same baby. CF cannot be ruled out.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen	Elevated	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. This result is consistent with the previous newborn screening specimen results for the same baby. The previous specimen results are indicative of CF.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	Immunoreactive Trypsinogen CFTR Mutation	IRT Elevated CFTR Mutation Panel may be 0, 1, or 2 Mutations Detected	Revised Screening Result for Cystic Fibrosis (CF). Additional testing using a CFTR Mutation Panel has been performed. *(Note: Result notes vary depending on the results applied for CFTR Mutation Panel)* [The specimen was originally reported as Indeterminate for CF. The original screening result note read "Repeat the newborn screen within 72 hours. Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level on the first specimen. The second screening specimen is required to determine if result is significant."]
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen CFTR Mutation	Elevated 0 Mutations Detected	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected. However, there is a minimal risk for Cystic Fibrosis due to variants not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Very Elevated 0 Mutations Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Very Elevated. None of the CFTR variants in the DSHS panel were detected. Although there is a minimal risk for CF in the absence of detected variants, a very elevated IRT result may be indicative of CF due to variants not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1078delT (c.948delT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1717-1G>A (c.1585-1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1898+1G>A (c.1766+1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2183AA>G (c.2051_2052delAAinsG), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2184delA (c.2052delA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2789+5G>A (c.2657+5G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3120+1G>A (c.2988+1G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3659delC (c.3528delC), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3849+10kbC>T (c.3717+12191C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3849+4A>G (c.3717+4A>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3876delA (c.3744delA), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3905insT (c.3773_3774insT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 394delTT (c.262_263delTT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 621+1G>T (c.489+1G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 711+1G>T (c.579+1G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, A455E (c.1364C>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, D1152H (c.3454G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, DF508 (c.1521_1523delCTT), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, DI507 (c.1519_1521delATC), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant which is not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, E60X (c.178G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G542X (c.1624G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G551D (c.1652G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G85E (c.254G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, N1303K (c.3909C>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Q493X (c.1477C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1162X (c.3484C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H with IVS8-9T/9T (c.[350G>A;1210-12[9]/1210-12[9]]), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R334W (c.1000C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R347H (c.1040G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R347P (c.1040G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R553X (c.1657C>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R560T (c.1679G>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549N (c.1646G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549R A>C (c.1645A>C), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S549R T>G (c.1647T>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, V520F (c.1558G>T), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, W1282X (c.3846G>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y1092X C>A (c.3276C>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y1092X C>G (c.3276C>G), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 1 Mutation Detected	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Y122X (c.366T>A), in the CFTR gene was identified. CF cannot be ruled out due to a possibility of a second variant not included in the panel.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. An apparently homozygous CF-causing variant, DF508 (c.1521_1523delCTT), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G542X (c.1624G>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and G551D (c.1652G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and N1303K (c.3909C>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and W1282X (c.3846G>A), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R553X (c.1657C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and 621+1G>T (c.489+1G>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-5T/7T (c.[350G>A;1210-12[5]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Analyte(s)	Analyte Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated 2 Mutations Detected	Probable Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. Two potential CF-causing variants, DF508 (c.1521_1523delCTT) and R117H with IVS8-9T/9T (c.[350G>A;1210-12[9]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Inconclusive	Immunoreactive Trypsinogen	Elevated	Immunoreactive Trypsinogen (IRT) Elevated. Many unaffected infants have an elevated IRT level. Additional testing for a panel of mutations in the CFTR gene is in progress to determine if result is significant. Final report with CFTR Mutation panel results to follow.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen	Very Elevated	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Very Elevated. Although there is a minimal risk for CF in the absence of detected variants, a very elevated IRT result may be indicative of CF. Additional testing for a panel of mutations in the CFTR gene is in progress. Final report with CFTR Mutation panel results to follow.
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 1 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 2 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated or Normal 3 Mutations Detected	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated Abnormal	free text
Abnormal	Cystic Fibrosis	Abnormal	Immunoreactive Trypsinogen CFTR Mutation	Elevated Unsatisfactory	free text
Normal	Cystic Fibrosis	Revised Result			free text
Abnormal	Cystic Fibrosis	Revised Result			free text
Unsatisfactory	Cystic Fibrosis	Revised Result			free text