Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Normal	Cystic Fibrosis	Normal	
Normal	Cystic Fibrosis	Normal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Result may be Abnormal or Inconclusive	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	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	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	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	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	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 406-1G>A (c.274-1G>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 935delA (c.803delA), 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	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1677delTA (c.1545_1546delTA), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 1898+5G->T (c.1766+5G>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2055del9>A (c.1923_1931del9insA), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2143delT (c.2012delT), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 2307insA (c.2175_2176insA), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3791delC (c.3659delC), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, 3199del6 (c.3067_3072delATAGTG), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, A559T (c.1675G>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, CFTRdele2,3 (c.54-5940_273+10250del21kb), 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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G178R (c.532G>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	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, G330X (c.988G>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, K710X (c.2128A>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, L206W (c.617T>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, M1101K (c.3302T>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, Q890X (c.2668C>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R75X (c.223C>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R117H (c.350G>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	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/5T (c.[350G>A;1210-12[5]/1210-12[5]]), 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	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/9T (c.[350G>A;1210-12[5]/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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1066C (c.3196C>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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, R1158X (c.3472C>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S1196X (c.3587C>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, S1255X (c.3764C>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	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Elevated. One variant, W1089X (c.3266G>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	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	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	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	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.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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	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	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.
Abnormal	Cystic Fibrosis	Abnormal	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	Abnormal	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, 935delA (c.803delA), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, 1078delT (c.948delT), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, G551D (c.1652G>A), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, Q493X (c.1477C>T), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, R117H with IVS8-7T/7T (c.[350G>A;1210-12[7]/1210-12[7]]), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, 3876delA (c.3744delA), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, L206W (c.617T>G), in the CFTR gene was identified.
Abnormal	Cystic Fibrosis	Abnormal	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, D1152H (c.3454G>C), in the CFTR gene was identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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 1717-1G>A (c.1585-1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 2184delA (c.2052delA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 2307insA (c.2175_2176insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 2789+5G>A (c.2657+5G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 3120+1G>A (c.2988+1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 3876delA (c.3744delA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 D1152H (c.3454G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 DI507 (c.1519_1521delATC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 G85E (c.254G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 Q493X (c.1477C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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 R1162X (c.3484C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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/9T (c.[350G>A;1210-12[5]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 S1255X (ex. 20) (c.3764C>A), in the CFTR gene were identified. A variant of uncertain significance, S1255X (ex. 19) (I1203V, c.3607A>G), was also identified. While S1255X (ex. 19) and S1255X (ex. 20) are two separate mutations, they are generally considered a haplotype with the two mutations occurring together (Cutting G, et al. 1992. Am. J. Hum. Genet. 50:1185-1194).
Abnormal	Cystic Fibrosis	Abnormal	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 S549N (c.1646G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 3791delC (c.3659delC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 G330X (c.988G>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 2055del9>A (c.1923_1931del9insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 1898+1 G>A (c.1766+1 G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 3659delC (c.3528delC), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 3849+10kbC>T (c.3717+12191C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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 3905insT (c.3773_3774insT), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 406-1G>A (c.274-1G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 A455E (c.1364C>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 CFTRdele2,3 (c.54-5940_273+10250del21kb), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 R1158X (c.3472C>T), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 R347H (c.1040G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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 R347P (c.1040G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, DI507 (c.1519_1521delATC) and D1152H (c.3454G>C), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, G542X (c.1624G>T) and 2055del9>A (c.1923_1931del9insA), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, G542X (c.1624G>T) and L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, L206W (c.617T>G) and Q493X (c.1477C>T), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Abnormal	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, L206W (c.617T>G) and S549N (c.1646G>A), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, 2789+5G>A (c.2657+5G>A) 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	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, D1152H (c.3454G>C) 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	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, R347H (c.1040G>A) 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	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, R560T (c.1679G>C) 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	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, 3199del6 (c.3067_3072delATAGTG) and R117H with IVS8-7T/9T (c.[350G>A;1210-12[7]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, 2307insA (c.2175_2176insA) 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	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, G542X (c.1624G>T) and R117H with IVS8-7T/9T (c.[350G>A; 1210-12[7]/1210-12[9]]), in the CFTR gene were identified.
Abnormal	Cystic Fibrosis	Abnormal	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, G551D (c.1652G>A) 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	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, D1152H (c.3454G>C) and L206W (c.617T>G), in the CFTR gene were identified.
Abnormal	Galactosemia	Abnormal	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/5T (c.[350G>A;1210-12[5]/1210-12[5]]), in the CFTR gene were identified.

Table 9: Cystic Fibrosis

Overall Result	Disorder	Screening Result	Screening Result Note
Abnormal	Cystic Fibrosis	Normal	Possible Cystic Fibrosis (CF). Recommend referral for confirmatory sweat testing and consider genetic counseling within 7 days. Immunoreactive Trypsinogen (IRT) Normal. 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	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected except a “No Call” result for both the wild type and variant R75X loci. Possible reason for a “No Call” result includes a variant or polymorphism within or around the primer annealing sites. There is a minimal risk for Cystic Fibrosis due to a variant not included in the panel. Clinical evaluation not necessary unless symptomatic.
Abnormal	Cystic Fibrosis	Inconclusive	No further evaluation necessary unless clinically indicated. Immunoreactive Trypsinogen (IRT) Elevated. None of the CFTR variants in the DSHS panel were detected except a “No Call” result for both the wild type and variant R1162X loci. Possible reason for a “No Call” result includes a variant or polymorphism within or around the primer annealing sites. There is a minimal risk for Cystic Fibrosis due to a variant not included in the panel. Clinical evaluation not necessary unless symptomatic.
Abnormal	Cystic Fibrosis	Inconclusive	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	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	free text
Normal	Cystic Fibrosis	Revised Result	free text
Abnormal	Cystic Fibrosis	Revised Result	free text
Unsatisfactory	Cystic Fibrosis	Revised Result	free text