

Materials Developed & Characterized By Previous CDC Project

Disorder	Gene	Allele 1	Allele 2	Intron-8 Poly T	Additional Mutations (not characterized)	Repository	Cell line number	DNA number	DNA Sequencing	Characterization Methods** (# labs)
Nonsyndromic Deafness	Connexin 26 (CX26)	35delG	WT			Coriell	CD00001	CD00001	4 labs	ASAG (3), PCR/RD (2), PCR/hyb(1), Light (1)
Craniosynostosis/Muenke	FGFR3	C749G	WT			Coriell	CD00002	CD00002	6 labs	PCR/RGE (5)
Cystic Fibrosis	CFTR	1078delT	WT	7T/7T		Coriell	CD00003	CD00003		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	1898+1G>A	WT	7T/9T		Coriell	CD00004	CD00004		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	1898+1G>A	WT	7T/7T		Coriell	CD00005	CD00005		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	1898+1G>A	WT	7T/7T		Coriell	CD00006	CD00006		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	1898+1G>A	WT	7T/7T		Coriell	CD00007	CD00007		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	2184delA	WT	7T/9T		Coriell	CD00008	CD00008		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	394delTT	WT	7T/9T		Coriell	CD00009	CD00009		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	I148T	WT	9T/9T		Coriell	CD00010	CD00010		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	I148T	WT	7T/9T		Coriell	CD00011	CD00011		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	I148T	WT	7T/9T		Coriell	CD00012	CD00012		Inn (1), OLA (1), 87mut HA (1)
Cystic Fibrosis	CFTR	S1235R	WT	7T/7T		Coriell	CD00013	CD00013	6 labs	RFLP/Het/OLA (2), RFLP (1), Het/seq (1)
Cystic Fibrosis	CFTR	F508del	2184delA	7T/9T		Coriell	GM18799	NA18799		Inn (2), OLA (2), MALDI-TOF (1), Inv (1), ASAG (1)
Cystic Fibrosis	CFTR	F508del	1898+1G>A	7T/9T		Coriell	GM18800	NA18800		Inn (2), OLA (2), MALDI-TOF (1), Inv (1), ASAG (1)
Cystic Fibrosis	CFTR	3272-26A>G	?			Coriell	GM18801	NA18801		Inn (2), MALDI-TOF (1), ARMS (1)
Cystic Fibrosis	CFTR	Y122X	?	7T/7T		Coriell	GM18802	NA18802		Inv (1), MALDI-TOF (1), ARMS (1)
Cystic Fibrosis	CFTR	F508del	2183AA-G	7T/9T		Coriell	GM18803	NA18803		Inn (2), MALDI-TOF (1), ASAG (1), Inv (1)
Cystic Fibrosis	CFTR	F508del	2143delT	9T/9T		Coriell	GM18886	NA18886		Inn (2), MALDI-TOF (1)

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Cystic Fibrosis	CFTR	3120+G>A	621+1G>T	7T/9T	MTHFR C677T/WT	Coriell	GM07441	NA07441	4 labs	RFLP/Seq (1), Het (1), RFLP/Het/Seq (2), CHIP/Seq (1), OLA (1)
Cystic Fibrosis	CFTR	delF508	R117H	5T/9T	HFE H63D/H63D; MTHFR C677T/WT	Coriell	GM13591	NA13591	4 labs	RFLP/Seq (1), Het (1), RFLP/Het/Seq (2), CHIP/Seq (1), OLA (1)
Fragile X	FMR1	30 CGG repeats (male)				Coriell	GM07174	NA07174	1 lab	
Fragile X	FMR1	56 CGG repeats (male)				Coriell	CD00014	CD00014	1 lab	size & methylation (10), sized only (2) - (RD/PCR/ Southern)
Fragile X	FMR1	96 CGG repeats (male)				Coriell	GM06906	NA06906	1 lab	
Fragile X	FMR1	93 CGG repeats (male)				Coriell	GM06892	NA06892	1 lab	
Fragile X	FMR1	118 CGG repeats (male)				Coriell	GM06891	NA06891	1 lab	
Hemochromatosis	HFE	C282Y	WT			Coriell	CD00015	CD00015	2 labs	PCR/RGE (6), light (1), ASAG & PCR/RGE (1), Chip (1), PCR/ASH (2)
Hemochromatosis	HFE	C282Y	WT			Coriell	CD00016	CD00016	2 labs	PCR/RGE (7) light (1) PCR/ASH (2)
Hemochromatosis	HFE	H63D	H63D			Coriell	CD00017	CD00017	2 labs	light (1), ASAG & PCR/RGE (1), Chip (1), PCR/RGE (6), PCR/hyb (1), PCR/hyb & seq (1)
Hemochromatosis	HFE	H63D	WT			Coriell	CD00018	CD00018	2 labs	light (1), Chip (1), PCR/RGE (7), PCR/ASH (2)
Hemochromatosis	HFE	S65C	WT			Coriell	CD00019	CD00019	2 labs	PCR/RGE (4), light (1), ARMS & PCR/RGE (1), Chip (1), PCR/ASH (2)
Hemochromatosis	HFE	S65C	WT			Coriell	CD00020	CD00020	2 labs	light (1), PCR/RGE (7), PCR/ASH (2)

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Hemochromatosis	HFE	H63D	S65C			Coriell	CD00021	CD00021	2 labs	light (1), PCR/RGE (7), PCR/ASH (2)
Huntington Disease	HD	31 CAG repeats	18 CAG repeats			Coriell	CD00022	CD00022		PCR/PAGE 1- (6), PCR/PAGE 2- (3), PCR/CE 1 - (2)
MTHFR Thermolabile Poly	MTHFR	C677T (A222V)	C677T (A222V)			Coriell	CD00023	CD00023		ASAG (1), PCR/RGE (4), PCR/RD (2), PCR/ASH (3)
MTHFR Thermolabile Poly	MTHFR	C677T (A222V)	C677T (A222V)			Coriell	CD00024	CD00024		ASAG (1), PCR/RGE (4), PCR/RD (2), PCR/ASH (3)
MTHFR Thermolabile Poly	MTHFR	C677T (A222V)	C677T (A222V)			Coriell	CD00025	CD00025		PCR/RGE (2), Invader (1)
MTHFR Thermolabile Polymorphism	MTHFR	C677T	WT		Prothrombin G20210A/WT; Factor V Leiden R506Q/WT; HFE S65C/WT	Coriell	GM16028	NA16028		ASAG (1), PCR/RGE (6), PCR/ASH (3)
Alpha-Thalassemia	HBA1	Type 1 SEA	WT			Coriell	CD00026	CD00026		g-Southern (3), g-Southern & PCR/GE (3), PCR/CE (1), Q-PCR (1), PCR (1), g-Southern & PCR & PCR/GE (1)
Alpha-Thalassemia	HBA1	Type 1 SEA	WT			Coriell	CD00027	CD00027		g-Southern & PCR & PCR/GE (1), PCR/GE & g-southern (1)
Thrombophilia; Factor V mutation	Factor V Leiden	R506Q	WT		HFE C282Y/H63D	Coriell	GM14641	NA14641		ASAG (2), PCR/RGE (4), Invader (2), PCR/ASH (3)
Thrombophilia; Prothrombin mutation	Prothrombin mutation	G20210A	G20210A		HFE H63D/WT; MTHFR C677T/WT	Coriell	GM16000			PCR/RD (5), Invader (2), PCR/RGE (2), PCR/ASH (2)
Sickle Cell/Hemoglobin C Disease/ Hemoglobin SC	HBB	HbS	HbC			Coriell	GM16266		3 labs	PCR/Het (1), PCR/RGE (2), PCR/ASH (4), PCR/RD (1)

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****METHODS LISTED ON FOLLOWING PAGE**

Methods**

ASAG = Allele-specific amplification assay with gel electrophoresis such as ARMS

PCR/RD = PCR with mismatched primer introducing allele-specific restriction enzyme site and gel electrophoresis

PCR/hyb = PCR + allele-specific hybridization

PCR/Het = PCR and heteroduplex analysis

Light = lightcycler

PCR/RGE = PCR + restriction endonuclease digestion and gel electrophoresis

Inn = Innogenetics reverse slot blot

OLA = Oligonucleotide Ligation Assay

RFLP = Restriction Fragment Length Polymorphism

Het = heteroduplex analysis

87 mut HA = 87 mutation Heteroduplex Assay

RFLP/Seq = Restriction Fragment Length Polymorphism + Sequencing

RFLP/Het/Seq = Restriction Fragment Length Polymorphism + Heteroduplex analysis + Sequencing

RFLP/Het/OLA - Restriction Fragment Length Polymorphism + Heteroduplex analysis + Oligonucleotide Ligation Assay

Het/Seq = Heteroduplex analysis + Sequencing

Chip/Seq = Motorola Life Sciences chip technology eSensor + Sequencing

RD/PCR/Southern = Restriction digest, PCR, Southern blot

PCR/PAGE 1= PCR and Polyacrylamide gel electrophoresis, one primer set

PCR/PAGE 2= PCR and Polyacrylamide gel electrophoresis, two primer sets

PCR/CE 1 = PCR, capillary electrophoresis, one primer set

Chip = Motorola Life Sciences chip technology eSensor

PCR/ASH = PCR and allele-specific hybridization

g-southern = genomic southern blot

quant PCR= quantitative PCR