Synthetic Control Materials

Disorder	Gene	Mutations	Notes	Source	Product	Cat. #
Cystic Fibrosis	CFTR	32 mutations including ACMG 25 panel	32 mutations in one vial	Molecular Controls	CF32 Control	CF32
Cystic Fibrosis	CFTR	delI507, 3199del6, D1152H, R1162X, 3659delC, 3849+10kbC>T, 3876delA	7 mutations in one vial	Molecular Controls	CF Reflex Control	CFRC
Cystic Fibrosis	CFTR	38 CFTR mutations including ACMG 25 panel (in blood-like matrix)	Mutations, 3 polymorphisms, and 5/7/9T variants	Maine Molecular Quality Controls, Inc.	INTROL CF	G106
Cystic Fibrosis	CFTR	28 mutations including ACMG 25 panel and 1 reflex variant	28 mutations and 1 reflex variant in 1 vial	AcroMetrix	OptiQual™ CF Mutation Controls	95-1402
Thrombosis	MTHFR	C677T, A1298C (in blood-like matrix)	contains: 1) wt MTHFR; 2) heterozygous 677C>T and 1298A>C genotypes; 3) homozygous 677C>T and 1298A>C genotypes	Molecular Controls	INTROL MTHFR	G105
Thrombosis	Factor II Factor V	G20210A (Factor II) G1691A (Factor V) (in blood-like matrix)	contains: 1) wt Factor II and Factor V; 2) heterozygous G20210A and G1691A genotypes; 3) homozygous G20210A and G1691A genotypes	Maine Molecular Quality Controls, Inc.	INTROL TRC Genotype Control	G104
Fragile X	FMR1	CGG repeat sizes: 20, 30, 41, 51, 60, 73, 93, 96, 118	1 CGG repeat size per control	National Institute of Standards and Technology	SRM2399	SRM2399
Bloom Syndrome, Canavan Disease, Familial Dysautonomia, Fanconi Anemia Type C, Gaucher Disease, Mucolipidosis Type IV, Niemann-Pick Disease, Tay Sachs Disease	BLM ASPA IKBKAP FANCC GBA MCOLN1 SMPD1 HEXA	32 mutations in 8 genes	32 mutations in 2 vials	Molecular Controls	AJP Control (for TM Bioscience Tag-It platform)	AJP32

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