

Familial Mutation, Targeted Testing, Varies

Test ID: FMTT

Explanation:

On the effective date, Familial Mutation, Targeted Testing, Varies will update the test methodology and reporting names for several available genes, as noted below. Changes are indicated in bold and reflex removals are indicated by empty right-adjacent rows.

Please see the online setup file for the most up to date test build information.

<https://www.mayocliniclabs.com/test-notifications/index.html>

Current Methodology
Polymerase Chain Reaction (PCR) followed by DNA Sequencing Analysis, Gene Dosage Analysis by Array Comparative Genomic Hybridization (aCGH) , Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA), and/or Quantitative Polymerase Chain Reaction (qPCR)

New Methodology
Polymerase Chain Reaction (PCR) followed by DNA Sequencing Analysis, Gene Dosage Analysis by Multiplex Ligation-Dependent Probe Amplification (MLPA), and/or Quantitative Polymerase Chain Reaction (qPCR)

Current Reflex Tests	
Test ID	Reporting Name
MATCC	Maternal Cell Contamination, B
CULFB	Fibroblast Culture for Genetic Test
CULAF	Amniotic Fluid Culture/Genetic Test
_G001	Gene GRHPR
_G002	Gene PPOX
_G003	Gene CFTR_SEQ
_G004	Gene CFTR_MLPA
_G005	Gene MLH1
_G006	Gene MSH2

New Reflex Tests	
Test ID	Reporting Name
MATCC	Maternal Cell Contamination, B
CULFB	Fibroblast Culture for Genetic Test
CULAF	Amniotic Fluid Culture/Genetic Test
_G001	Gene GRHPR
_G002	Gene PPOX
_G003	Gene CFTR
_G005	Gene MLH1
_G006	Gene MSH2

Current Reflex Tests	
_G007	Gene MSH6
_G008	Gene MECP2_SEQ
_G009	Gene MLH3
_G010	Gene CHEK2
_G011	Gene IDUA
_G012	Gene AXIN2
_G013	Gene BMPR1A
_G014	Gene PTEN
_G015	Gene SMAD4
_G016	Gene STK11
_G017	Gene TP53
_G018	Gene IDS
_G019	Gene FLCN
_G020	Gene SPINK1
_G021	Gene PRSS1
_G022	Gene CTSC
_G025	Gene ABCD1
_G026	Gene CDH1
_G027	Gene NAGLU
_G028	Gene SGSH
_G029	Gene ARSB
_G030	Gene GNPTAB
_G031	Gene SEPT9
_G032	Gene ACADVL
_G033	Gene ACADM
_G034	Gene ACADS
_G035	Gene FECH
_G036	Gene MAPT
_G037	Gene PKHD1
_G038	Gene GRN
_G039	Gene FTCD
_G040	Gene CDKN1C

New Reflex Tests	
_G007	Gene MSH6
_G008	Gene MECP2
_G009	Gene MLH3
_G010	Gene CHEK2
_G011	Gene IDUA
_G012	Gene AXIN2
_G013	Gene BMPR1A
_G014	Gene PTEN
_G015	Gene SMAD4
_G016	Gene STK11
_G017	Gene TP53
_G018	Gene IDS
_G019	Gene FLCN
_G020	Gene SPINK1
_G021	Gene PRSS1
_G022	Gene CTSC
_G025	Gene ABCD1
_G026	Gene CDH1
_G027	Gene NAGLU
_G028	Gene SGSH
_G029	Gene ARSB
_G030	Gene GNPTAB
_G031	Gene SEPT9
_G032	Gene ACADVL
_G033	Gene ACADM
_G034	Gene ACADS
_G035	Gene FECH
_G036	Gene MAPT
_G037	Gene PKHD1
_G038	Gene GRN
_G039	Gene FTCD
_G040	Gene CDKN1C

Current Reflex Tests	
_G041	Gene CPOX
_G042	Gene ATP7B
_G043	Gene GAA
_G044	Gene HMBS
_G045	Gene GALT
_G046	Gene GLA
_G047	Gene BTD
_G048	Gene HEXA
_G049	Gene AGXT
_G050	Gene APC
_G051	Gene MLYCD
_G052	Gene MMACHC
_G053	Gene GBA
_G054	Gene SMPD1
_G055	Gene CPT2
_G056	Gene TTR
_G057	Gene UBE3A
_G058	Gene GALC
_G059	Gene GSN
_G060	Gene LYZ
_G061	Gene FGA
_G062	Gene APOA1
_G063	Gene APOA2
_G064	Gene MMADHC
_G065	Gene SLC25A20
_G066	Gene ARSA
_G067	Gene NPC1/2_SEQ and NPC1/2_MLPA
_G068	Gene PMS2_LR and PMS2_SEQ
_G069	Gene PMS2_MLPA
_G070	Gene RAI1
_G071	Gene MUTYH

New Reflex Tests	
_G041	Gene CPOX
_G042	Gene ATP7B
_G043	Gene GAA
_G044	Gene HMBS
_G045	Gene GALT
_G046	Gene GLA
_G047	Gene BTD
_G048	Gene HEXA
_G049	Gene AGXT
_G050	Gene APC
_G051	Gene MLYCD
_G052	Gene MMACHC
_G053	Gene GBA
_G054	Gene SMPD1
_G055	Gene CPT2
_G056	Gene TTR
_G057	Gene UBE3A
_G058	Gene GALC
_G059	Gene GSN
_G060	Gene LYZ
_G061	Gene FGA
_G062	Gene APOA1
_G063	Gene APOA2
_G064	Gene MMADHC
_G065	Gene SLC25A20
_G066	Gene ARSA
_G067	Gene NPC1/2_SEQ and NPC1/2_MLPA
_G068	Gene PMS2
_G070	Gene RAI1
_G071	Gene MUTYH

Current Reflex Tests	
_G072	Gene HGSNAT
_G073	Gene GNS and GRHPR_MLPA
_G074	Gene PSAP
_G075	Single-gene Large Del/Dup
_G076	Gene MECP2_MLPA
_G077	Gene RET
_G078	Gene SUMF1
_G079	Gene CASR_Seq
_G080	Gene VHL_SEQ
_G081	VHL_MLPA
_G082	Gene SHDP_MLPA
_G083	Gene SDHB, SDHC, and SDHD_MLPA
_G084	Gene SDHB, SDHC, SDHD_Seq
_G085	Gene BRCA1
_G086	Gene BRCA2
_G087	Gene DMD_MLPA
_G088	Gene PMP22_MLPA
_G089	Gene MPZ_MLPA
_G102	Gene SERPINA1
_G112	Gene SDHAF2
_G113	Gene TMEM127
_G114	Gene MAX
_G115	Gene SMN1
_G125	Gene PMP22_SEQ
_G126	Gene G6PD_SEQ
_G127	Gene GJB2_SEQ
_G128	Gene HBA1/HBA2_SEQ
_G129	Gene HBB_SEQ
_G130	Known Familial Variant,Other
G168	Gene CSTB
G169	Gene CACNA1A

New Reflex Tests	
_G072	Gene HGSNAT
_G073	Gene GNS and GRHPR_MLPA
_G074	Gene PSAP
_G077	Gene RET
_G078	Gene SUMF1
_G079	Gene CASR_Seq
_G080	Gene VHL_SEQ
_G084	Gene SDHB, SDHC, SDHD_Seq
_G085	Gene BRCA1
_G086	Gene BRCA2
_G087	Gene DMD_MLPA
_G089	Gene MPZ_MLPA
_G102	Gene SERPINA1
_G112	Gene SDHAF2
_G113	Gene TMEM127
_G114	Gene MAX
_G115	Gene SMN1
_G125	Gene PMP22
_G126	Gene G6PD
_G127	Gene GJB2
_G128	Gene HBA1/HBA2
_G129	Gene HBB
_G130	Known Familial Variant,Other
G168	Gene CSTB
G169	Gene CACNA1A

Current Reflex Tests	
CRYOB	Cryopreserve for Biochem Studies
FIBR	Fibroblast Culture

New Reflex Tests	
CRYOB	Cryopreserve for Biochem Studies
FIBR	Fibroblast Culture

Questions

Contact Michelle Rath, Laboratory Technologist Resource Coordinator at 800-533-1710.