

## SPECIMEN REQUIREMENT CHANGE- INFORMED CONSENT

**NOTIFICATION DATE:** June 15, 2010 **EFFECTIVE DATE:** July 1, 2010

## **NEW YORK STATE INFORMED CONSENT REQUIREMENTS**

**EXPLANATION:** In order to assist in your compliance with New York Statutes relating to informed consent for genetic testing, Mayo Medical Laboratories (MML) will be implementing the following on July 1, 2010:

- 1. It is the expectation of MML that the ordering provider obtains informed consent from the patient for genetics testing.
- 2. Please document on the request form, batch sheet or electronic order that a copy of the informed consent is on file. (Note: Do not send the consent document to MML)
- 3. If MML does not receive notification that a copy of the informed consent is on file, MML will contact you to confirm.
- 4. All orders will go to the laboratory to start the processing of the test; testing will not be delayed. However, results will not be released until MML has confirmation that informed consent is on file.
- 5. Upon contact, MML will document the name of the individual at the client site who provides the verbal confirmation.

For your reference, attached is a list of MML orderable tests in which informed consent may be required according to New York State statutes. Please review with your laboratory staff.

QUESTIONS: Contact your Mayo Medical Laboratories' Regional Manager or MML Technologist Support Telephone: 800-533-1710

Unit	
Code	Test Name
86208	15q Deletion, Type I and Type II Characterization, Prader-Willi/Angelman Syndromes, FISH
89365	15q Duplication, FISH
88541	1p36.3 Del, FISH
81129	22q11.2 Del, VCF, DiGeorge, FISH
81648	5,10-Methylenetetrahydrofolate Reductase, Mutation, Blood
88898	aCGH, Whole Genome, Constitutional
83643	AGXT Mutation Analysis (G170R)
8815	Alpha-Fucosidase, Fibroblasts
8814	Alpha-Fucosidase, Leukocytes
8785	Alpha-Galactosidase, Leukocytes
8784	Alpha-Galactosidase, Serum
9499	Alpha-Globin Gene Analysis
8780	Alpha-L-Iduronidase, Fibroblasts
8773	Alpha-Mannosidase, Fibroblasts
8772	Alpha-Mannosidase, Leukocytes
8782	Alpha-N-Acetylglucosaminidase, Serum
80334	Amniotic Fluid Cluture for Genetic Test
83674	Amyloidosis, Transthyretin-Associated Familial, Reflex, Blood
8783	A-N-Acetylglucosaminidase, Fibroblasts
81081	Aneuploidy Detection, POC, FISH
89097	Apolipoprotein B-100 Molecular Analysis, R3500Q and R3500W
80905	Apolipoprotein E Genotyping, Blood
88912	ARPKD Known Mutation
88911	ARPKD Mutation Screen
8778	Arylsulfatase A, Fibroblasts
8779	Arylsulfatase A, Leukocytes
8777	Arylsulfatase A, Urine
8151	Arylsulfatase B, Fibroblasts
86329	Ashkenazi Jewish Panel with CF
88887	Ashkenazi Jewish Panel without CF
83278	Beckwith-Wiedemann Syndrome
8787	Beta Glucosidase, Fibroblasts
8788	Beta Glucosidase, Leukocytes
8008	Beta-Galactosidase, Fibroblasts
8486	Beta-Galactosidase, Leukocytes
8006	Beta-Glucuronidase, Fibroblasts
88205	Biotinidase, Serum

85316	Bloom Syndrome, Molecular Analysis
89012	BTD Gene, Full Gene Analysis
89013	BTD Gene, Known Mutation
89742	BTK Full Gene Panel, Blood
89307	BTK Full Gene Sequence
89306	BTK Gene, Known Mutation
89740	BTK Known Mutation Panel, Blood
88829	C4 Acylcarnitine
88831	C5-DC Acylcarnitine
88830	C5-OH Acylcarntitine
81780	Canavan Disease Mutation
89891	Carbohydrate Deficient Transferrin - CDG
83704	CASR Known Mutation
83703	CASR Mutation Screen
83817	CASR Mutation Screen, Biochem and Genetic
88876	CFTR Gene, Full Gene Analysis
88880	CFTR Gene, Known Mutation
80602	Chromosome Anomalies, Misc, FISH
8426	Chromosomes, Amniotic Fluid
80257	Chromosomes, Chorionic Villus Sample
8696	Chromosomes, Congenital, Blood
81332	Chromosomes, MMC Stress, Blood
8887	Chromosomes, POC, Autopsy, Stillbirth
926	Chromosomes, Sister Chromatid Exchange
8425	Chromosomes, Skin Biopsy
88536	Cri-du-chat, 5p Del, FISH
89081	CYP21A2 Full Gene Analysis
89082	CYP21A2 Known Mutation
81148	Cystathionine B-Synthase, Fibroblasts
9497	Cystic Fibrosis Mutation Panel
80226	DNA Analysis B
81801	DRPLA Gene Analysis
88264	Fabry Disease Full Gene Analysis
88266	Fabry Disease Known Mutation
88264	Fabry Disease, Full Gene Analysis
81419	Factor V Leiden (R506Q)
83705	Familial Amyloidosis Known Mutation
83667	Familial Amyloidosis, DNA Sequence
85319	Familial Dysautonomia, Mutation
85318	Fanconi Anemia, Molecular Analysis
83001	FAP Known Mutation

82582	FAP Mutation Screen
81927	Fatty Acid Oxidation Probe Assay, Fibroblast Culture
89308	FBN1 Full Gene Sequence
89311	FBN1 Gene, Known Mutation
83375	FH/ADH Reflex Panel
9569	Fragile X Syndrome, Molecular Analysis
8333	Gal-1-P Uridyltransferase, RBC
8628	Galactokinase, Blood
80341	Galactose-1-Phosphate Uridyltransferase Biochemical Phenotype, RBC
84366	Galactosemia Gene Analysis
84367	Galactosemia Known Mutation
8816	Galactosylcer B-Galactosid, WBC
8297	Galactosylcer B-Galactosidase, Fibroblasts
88877	GALT Gene, Full Gene Analysis
81235	Gaucher Disease, Molecular Analysis
81508	Hemochromatosis HFE Gene Analysis, Blood
80297	Hemophilia A Molecular Analysis for Inversion
88691	Hered Pancreatitis, Known Mutation
83019	Hered Pancreatitis, Mutation Screen
89278	HEXA Gene, Full Gene Analysis
89283	HEXA Gene, Known Mutation
8776	Hexosaminidase A & Total, Fibroblasts
8775	Hexosaminidase A & Total, WBC
80350	Hexosaminidase A (MUGS), Serum
82943	Hexosaminidase A and Total, Leukocytes/Molecular Reflex
8774	Hexosaminidase A and Total, Serum
80945	Iduronate Sulfatase, Fibroblasts
83644	IVD Mutation Analysis (A282V)
88540	Kallmann, Xp22.3, FISH
81183	LDLR Gene, Known Mutation
89073	LDLR Large Deletion/Duplication
81013	LDLR, Full Gene Sequence
89406	Lysosomal Disorders Screen, Blood Spot
87925	MAPT Known Mutation
87924	MAPT Screening Sequence Analysis
88636	Maternal Cell Contamination
83934	MCAD, Known Mutation
89284	MECP2 Gene, Full Gene Analysis
89285	MECP2 Gene, Known Mutation
81082	MEN2 (2A,2B,FMTC) Known Mutation
80573	MEN2 (2A,2B,FMTC) Mutation Screen

81921	Methylmalonic Acid, Amniotic Fluid
88533	Miller-Dieker, 17p13.3 Del, FISH
83002	MLH1 Known Mutation
83015	MLH1 Mutation Screen
83191	MLH1/MSH2 Mutation Screen
89830	MLYCD Gene, Full Gene Analysis
89831	MLYCD Gene, Known Mutation
89436	MMACHC Gene, Full Gene Analysis
89135	MMACHC Gene, Known Mutation
83082	MSH2 Known Mutation
83016	MSH2 Mutation Screen
83706	MSH6 Known Mutation
83723	MSH6 Mutation Screen
85321	Mucolipidosis IV, Mutation Analysis
84304	MYH Gene Analysis
80946	N-Acetylgalactosamine-6-Sulfate Sulfatase, Fibroblasts
89314	Neonatal Marfan Syndrome, FBN1 Gene
81409	Newborn Aneuploidy Detection, FISH
85322	Niemann-Pick Disease, Types A and B
9313	Niemann-Pick Type C, Fibroblasts
83118	NPC Known Mutation
89015	NPC Mutation Screen
8380	Phenylalanine and Tyrosine, Plasma
89898	Pompe Disease, Full Gene Sequencing
89897	Pompe Disease, Known Mutation
81153	Prader-Willi/Angelman Syndrome Molecular Analysis
81424	Prenatal Aneuploidy Detection, FISH
89188	Progranulin Gene Full Gene Analysis
89187	Progranulin Gene, Known Mutation
83003	Protein S Heerlen (S460P)
81742	Prothrombin G20210A
83899	Pyruvate Dehydrogenase Comp, Fibroblasts
83947	SCAD Known Mutation
83939	SCAD Mutation Screen
86374	Sex Chromosome Mosaicism, FISH
81595	Smith-Lemli-Opitz Scrn, Plasma
88534	Smith-Magenis, 17p11.2 Del/Dup, FISH
82971	Specimen Source Identification
8481	Sphingomyelinase, Fibroblasts
81176	Spinobulbar Muscular Atrophy, Kennedy's Disease
88537	SRY, Yp11.3, FISH

88539	STS, Xp22.3, FISH
82583	Subtelomeric Region Anomalies, FISH
84388	TACI Full Gene Sequencing
89122	TACI Gene, Known Mutation
82588	Tay-Sachs, Mutation Analysis
89461	TGFBR2, Full Gene Sequence
89462	TGFBR2, Known Mutation
80291	Thiopurine Methyltransferase, RBC
89396	UGT1A1 Gene, Known Mutation
89611	UGT1A1 Sequence, Hyperbilirubinemia
82970	Uniparental Disomy
8599	Uroporphyrinogen Decarboxylase, RBC
80288	Uroporphyrinogen III Co-Synthase, RBC
89211	VHL Deletion Detection
89083	VHL Full Gene Analysis
89084	VHL Known Mutation
81662	von Willebrand Disease 2N (Subtype Normandy), Blood
81163	Williams Syn, 7q11.23 Del, FISH
83698	Wilson Disease Known Mutation
83697	Wilson Disease Mutation Screen
88535	Wolf-Hirshhorn, 4p Del, FISH
8668	X, Y, Aneuploidy, Buccal Smear, FISH
88538	XIST, Xp22.3, FISH
82992	Y Microdeletion
81252	Zygosity Testing (Multiple Births)