

NEW TEST ANNOUNCEMENT REFERRAL

NOTIFICATION DATE: May 16, 2012 **EFFECTIVE DATE:** Immediately

ASHKENAZI JEWISH HEXA ONLY

Test ID: FFHEX

EXPLANATION: The following new orderable unit code, referred to Ambry Genetics, is available immediately for temporary referral during test down for Mayo test(s).

NOTE: This test will become obsolete when Mayo Test ID: TSD, Tay-Sachs Disease, Mutation Analysis, HEXA resumes.

REFERRAL LAB CODE: 1804HEXA

METHODOLOGY: DNA Sequencing

REFERENCE VALUES:

An interpretive report will be provided.

SPECIMEN REQUIREMENTS:

Draw blood in a lavender-top (EDTA) or yellow-top (ACD) tube(s) send 5 mL of EDTA or ACD whole blood refrigerate in plastic vial.

Note: Complete Ambry request form

SPECIMEN STABILITY INFORMATION:

Specimen Type	Temperature	Time
Blood	Refrigerate (preferred)	14 days
	Ambient	5 days

LIST FEE: \$ 200.00

CPT CODE: 83891 X 1; 83894 X 1; 83898 X 3; 83904 X 2; 83909 X 2; 83912 X 1

DAY(S) SET UP: Monday through Friday **ANALYTIC TIME:** 10 - 14 days

QUESTIONS: Contact Mary Erath, MML Laboratory Technologist Resource Coordinator Telephone: 800-533-1710

Ambry Genetics	

GENERAL TEST REQUISITION					► Ordering	Ordering Clinician Email			
Arrows "► " Mandatory for Processing					► Facility N	Name and Address	1	ID#	
Patient Information									
➤ DOB MM - DD - YEAR	► Last Name	► Firs	t Name	Middle	Initial				
► Gender □ F □ M	Gender □ F □ M ► Street Address, City, State, Zip			Additional	Additional Results Recipient				
► Ethnicity	-	•	•			Medical Pro	fessional Name:		
☐ African American ☐ Asian☐ Caucasian☐ Hispanic						Facility Nam	ne and Address		☐ Same As Above
☐ Jewish (Ashkenazi) ☐ Specify:	► Home Phone		Work/C	Cell					
Specimen			ous Test Histo	ory		Ph		Fx	
Collection Date: Previo		ously Detected Mutations:		Form Com	npleted by	► Phone			
Specimen ID: MR#:		16301	•						
MR# Specimen Type (See Requi		I allei	-	ed at Ambry? Yes		patient has bee	n supplied information rega	al or authorized person acknowing genetic testing and the	patient has given
, ,, ,	,	'	y previously teste :	d at Ambry? □ Yes Relation:		I confirm that th	nis is medically necessary fo	I and that the signed consent or the diagnosis or detection of	of a disease, illness,
☐ Blood ☐ Blood Spot	□ DNA		:			agement and tr	eatment decisions for this p		
☐ Cultured Amniocytes				Relation:		Weulcal Fibi	essional Signature [*]	Mandatory for Medicare/Medi Date:	icaid
		:	Relation:		* MD/DO, Clinical	inical Nurse Specialist, Nurse-Midwives, Nurse Practitioner, Physician Assistar		Assistant	
□ Other:		Does this patient give consent to the use of their sample for research? ☐ Yes ☐ No Consent is implied if a box is not marked							
► Indication for Testing (place Diagnostic	creening ☐ Researd ☐ Family	h History	Sweat C	ical Findings:					
Billing Information - Mand					form	ed nanerwork is o	n file no need to fill	out billing information	on again
AMBRY GENETICS provides a delayed until all of the billing re	selection of convenient	billing opt	ions. Please cho	ose an option below a				-	
⊠Bill Facility	☐ same as ordering	facility	☐ Bill Insura	nce Include card o	ору (both sides)	☐ Pre-Payment		
Facility Name		A completed Advance Beneficiary Notice of coverage (ABN) is required for Medicare patients. Ambry will pre-verify patient insurance coverage and if estimated patient out-of-pocket costs exceed \$350, patient is notified. Ambry Genetics will no longer perform Preverification for tests priced under \$200.		Payment Type ☐ Check ☐ Mastercard ☐ Discover ☐ Visa ☐ American Express					
Address, City, State, Zip PO Box 4100			Name of Insu	red		ation to patient? Self □ Parent	Card Number		Exp Date
Rochester, N	MN 55903-4	100	1			Spouse	Cardholder Name		Amount \$
Contact Person			insurance Co	mpany Name and	Addre	955	Signature X		Date
Contact Person Phone 800-447-6424			Insurance Ph	one				insurance benefits to be p	
			Member ID #		Grou	up #	medical information co	pration and authorize then ncerning my testing to my inancially responsible for	y insurer. I here-
			Authorization	#		Date]		
							X	Date	

Contact and Organization Information ► Authorized Ordering Physician

▶ Ph 800-533-1710 ► FX 507-538-5340

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Patient Name

► Test Directory Requisition Form				
CANCER	CHROMOSOMAL MICROARRAY ANALYSIS (aCGH)			
Lynch Syndrome (HNPCC) - (EDTA Tube) □ 8518 HNPCC/Lynch Syndrome - MLH1, MSH2 & MSH6 gene sequence with MLH1, MSH2, MSH6 and EPCAM deletion/duplication (all concurrent)	□ 3002 Ambry CMA: 180K Oligo Array (1 EDTA + 1 Na Heparin) Note: This CMA has increased coverage on X chromosome			
□ 8500 HNPCC/Lynch Syndrome - Steps 1 and 2 (reflex option)	CHROMOSOME STUDIES			
Step 1 MLH1 & MSH2 gene sequence Step 2 MSH6 gene sequence and MLH1/MSH2/MSH6/EPCAM deletion/duplication 4646 PMS2 gene sequence and deletion/duplication	☐ 3660 High Resolution Chromosome Analysis/Karyotype (Na Heparin)			
Juvenile Polyposis Syndrome (JPS) - (EDTA Tube) 8004 BMPR1A and SMAD4 gene sequence and deletion/duplication (concurrent)	CEREBRAL CAVERNOUS MALFORMATIONS (CCM) - (EDTA Tube)			
☐ 8602 JPS - Steps 1 and 2 Step 1 BMPR1A & SMAD4 gene sequence Step 2 BMPR1A & SMAD4 deletion/duplication	□ 5370 CCM - All genes listed below for gene sequence and del/dup (concurrent) □ 5368 CCM - Steps 1 and 2 (reflex option) Step 1 KRIT1 gene sequence			
☐ 8600 BMPR1A and SMAD4 deletion/duplication ☐ 2820 BMPR1A gene sequence	Step 2 CCM2 and PDCD10 gene sequence with CCM2, KRIT1 and PDCD10 del/ dup			
☐ 1685 SMAD4 gene sequence Call JPS (single gene deletion/duplication) GENE	□ 5320 CCM2 gene sequence □ 5324 CCM2 deletion/duplication □ 5340 KRIT1 gene sequence □ 5344 KRIT1 deletion/duplication □ 5360 PDCD10 gene sequence □ 5364 PDCD10 deletion/duplication			
Familial Adenomatous Polyposis Syndrome (FAP) - (EDTA Tube) □ 8726 FAP - APC and MUTYH gene sequence and APC deletion/duplication (concurrent) □ 8722 FAP PLUS [APC gene sequence and deletion/duplication + MAP (see below)]	☐ 5366 CCM - All genes for del/dup			
Step 1 MUTYH associated Polyposis (MAP) Step 1 MUTYH specific mutation analysis of Y179C and G396D	DIAMOND BLACKFAN ANEMIA - (EDTA Tube)			
Step 2 If only one mutation detected, reflex to MUTYH gene sequence analysis	□ 8548 DBA—RPS19, RPL5, RPL11, RPL35A, RPS26, RPS10, RPS24, RPS17, RPS7			
☐ 4660 MUTYH gene sequence ☐ 3040 APC gene sequence and deletion/duplication	gene sequence (concurrent) 8540 DBA - Steps 1 through 3 8550 RPS19 gene sequence——Step 1			
Hereditary Diffuse Gastric Cancer - (EDTA Tube) ☐ 4726 CDH1 gene sequence and deletion/duplication	☐ 2460			
Other Genes and Syndromes - (EDTA Tube) □ 2866	□ 2588			
□ 2680 RET gene sequence (Multiple Endocrine Neoplasia Type 2) □ 4700 CDKN2A/p16 gene sequence (Malignant Melanoma) □ 2360 PALB2 gene sequence (Pancreatic Cancer) □ 2106 PTEN gene sequence and deletion/duplication	Additional Information			
□ 5426 RB1 gene sequence and deletion/duplication □ 2766 STK11 gene sequence and deletion/duplication (Peutz-Jeghers) □ 2606 VHL gene sequence and deletion/duplication (Von Hippel-Lindau Disease)				
Testing of Tumor Characteristics (Screening Tests for HNPCC) - (Na Heparin Tube) □ 8700(4) MMR Profile by IHC (MLH1, MSH2, MSH6, PMS2) □ 8702 MSI by PCR □ 7978 MLH1 Hypermethylation Analysis □ 7980 BRAF SMA V600E Analysis □ 7982 MLH1 Hypermethylation with BRAF (Analysis of V600E only) Analysis				
Paraganglioma-Pheochromocytoma Syndrome (PGL/PCC) - (EDTA Tube) □ 5418 PGL/PCC - SDHB, SDHC, SDHD, SDHAF2 gene sequence and deletion/duplication with TMEM127 gene sequence (concurrent) □ 5416 PGL/PCC - SDHB, SDHC, SDHD, SDHAF2 deletion/duplication □ 5380 SDHB gene sequence □ 5386 SDHC gene sequence □ 5392 SDHD gene sequence □ 5398 SDHAF2 gene sequence □ 5410 TMEM127 gene sequence □ 5410 Any single gene deletion/duplication GENE				
□ 2680 RET gene sequence (Multiple Endocrine Neoplasia Type 2) □ 2606 VHL gene sequence and deletion/duplication (Von Hippel-Lindau Disease)				





Patient Name

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DYSKERATOSIS CONGENITA (DC) - (EDTA Tube)	GENETICS - (EDTA Tube)		
□ 8161 DC - DKC1, TINF2, TERC, NHP2, NOP10, TERT gene sequence (concurrent) □ 8160 DC - Steps 1 through 3 □ 1960 DKC1 gene sequence □ 1980 TINF2 exon 6 sequence only □ 2120 TERC gene sequence □ 2060 NHP2 exon 4 sequence only □ 2080 NOP10 exon 2 sequence □ 2140 TERT gene sequence Step 3	□ 8640 AmbrySCREEN™ □ 1640 Alagille (JAG1 gene sequence and deletion/duplication) □ 1641 Alagille (JAG1 deletion/duplication) □ 8642 Amyotrohpic Lateral Sclerosis (SOD1, ANG, FIG4, FUS and TARDBP gene sequence) (concurrent) □ 8620 Amyotrohpic Lateral Sclerosis (SOD1 reflex to ANG, FIG4, FUS, TARDBP gene sequence) □ 8622 Amyotrohpic Lateral Sclerosis (SOD1 gene sequence) □ 1320 Aminoglycoside-Related Hearing Loss (MT-RNR1 gene sequence)		
□ 5160 WRAP53 gene sequence	☐ 5280 Andermann Syndrome (SLC12A6 gene sequence) ☐ 8520 Angelman Syndrome (SNRPN methylation reflex to UBEA3 gene sequence)		
EXOME SEQUENCING FOR CLINICAL DIAGNOSIS - (EDTA Tube)	☐ 2400 Angelman Syndrome (<i>UBE3A</i> gene sequence) ☐ 2420 Angelman-like Syndrome (<i>SLC9A</i> 6 gene sequence)		
TEST NOT ORDERABLE HERE. PLEASE OBTAIN THE EXOME TEST REQUISITION.	☐ 2440 Angelman/Prader-Willi Syndrome (SNRPN methylation) ☐ 1808 Ashkenazi Jewish Panel™ with all 16 conditions		
Additional Information	□ 1804 Ashkenazi Jewish FlexPanel™ as marked below □ Bloom (BLM) □ Glycogen Storage Disease 1a (GSD1a) □ Cystic Fibrosis (CFTR) □ Maple Syrup Urine Disease (BCKDHA/B) □ Maple Syrup Urine Disease Type 3 (DLD) □ Mucolipidosis Type IV (MLDV) □ Canavan (ASPA) □ Joubert Syndrome (TMEM216) □ Familial Dysautonomia (IKBKAP) □ Niemann-Pick A (SMPD1) □ Gaucher (GBA) □ Fanconi Anemia Type C (FANCC) □ Tay-Sachs (HEXA) □ Nemaline Myopathy (NEB) □ Usher Syndrome Type 1F (PCDH15) □ Usher Syndrome Type III (CLRN1) □ 4940 Aspartylglucosaminuria (AGA gene sequence) □ 1040 Beta Thalassemia Plus (HBB gene sequence with 619del check) □ 1226 Canavan (ASPA gene sequence and deletion/duplication) (concurrent) □ 1220 Canavan (ASPA gene sequence reflex deletion/duplication) □ 1370 Congenital Hyperinsulinism-Hyperammonemia (GLUD1 gene sequence) □ 1364 Congenital Hyperinsulinism (KCNJ11 gene sequence) □ 2380 CHARGE Syndrome (CHD7 gene sequence) □ 4960 Dihydropyrimidine Deyhyrogenase Deficiency (DPYD gene sequence) □ 720 Familial Mediterranean Fever (MEFV gene sequence) □ 1820 Gaucher Disease (GBA gene sequence) □ 1820 <t< td=""></t<>		
FAMILIAL HYPERCHOLESTEROLEMIA - (EDTA Tube)	☐ 1900 Glycogen Storage Disease Type Ib (SLC37A4 gene sequence)		
□ 8680 Familial Hypercholesterolemia Comprehensive Evaluation (LDLR and PCSK9 gene sequence and APOB partial gene sequence with LDLR deletion/duplication) □ 8582 Familial Hypercholesterolemia (LDLR and APOB partial gene sequence reflex to LDLR deletion/duplication) □ 2780 LDLR gene sequence □ 2784 LDLR deletion/duplication □ 2800 APOB partial gene sequence □ 2804 PCSK9 gene sequence	□ 2746 Hereditary Angioedema (SERPING1 gene sequence and deletion/duplication) □ 2708 Hirschsprung Disease (RET gene sequence) (concurrent) Hirschsprung Disease Steps 1 and 2 (RET) □ 2704 Step 1 only: exons 2,3,5,6,9,10,12,13,17 gene sequence □ 2706 Step 2 only: rest of gene sequence □ 1940 Hunter Syndrome (IDS gene sequence) □ 2160 Hurler Syndrome (IDUA gene sequence) □ 5020 Hyperoxaluria Type 2 (GRHPR gene sequence) □ 3200 Infantile Spasms (CDKL5 gene sequence) □ 4860 Lysosomal Free Sialic Acid-Storage (Salla) Diseases (SLC17A5 gene sequence) □ 8780 Marfan Syndrome NextGen Sequencing Panel		
GASTROENTEROLOGY - (EDTA Tube)	Marfan Syndrome NextGen Sequencing Panel Steps 1 and 2 Step 1 FBN1 gene sequence		
 □ 8022 Pancreatitis Plus (CFTR, PRSS1, SPINK1, CTRC gene sequence) □ 8020 Pancreatitis (CFTR, PRSS1, SPINK1 gene sequence) □ 8040 Pancreatitis Amplified (CFTR, PRSS1, SPINK1 with CFTR del/dup) □ 1100 PRSS1 gene sequence □ 1120 SPINK1 gene sequence □ 1660 CTRC gene sequence □ 1840 Wilson Disease (ATP7B gene sequence) □ 1440 Shwachman-Diamond Syndrome (SBDS gene sequence) Additional Information	Step 2 ACTA2, CBS, FBN2, MYH11, COL3A1, SLC2A10, SMAD3, TGFBR1, TGFBR2 gene sequence 4900 MCAD - Medium-chain acyl-CoA dehydrogenase (ACADM gene sequence) 5180 Mucolipidosis Type IV (MCOLN1 gene sequence) 1360 Neonatal Diabetes (KCNJ11 gene sequence) 1620 Neonatal Diabetes (INS gene sequence) 1860 Niemann-Pick Disease Types A & B (SMPD1 gene sequence) 18122 Primary Ciliary Dyskinesia NextGen Sequencing Panel 4840 Rhizomelic Chondrodysplasia Punctata Type 1 (PEX7 gene sequence) 1760 Phenylketonuria - PKU (PAH gene sequence) 1740 Pompe Disease (GAA gene sequence)		
Additional Information	□ 2180 Smith-Lemli-Opitz Syndrome (DHCR7 gene sequence) □ 1240 Tay-Sachs Plus (HEXA gene sequence) □ 5240 Tay-Sachs Enzyme Assay (HEXA Leukocytes) □ 1560 Transthyretin Amyloidosis (TTR gene sequence) □ 4920 VLCAD - Very long-chain acyl-CoA dehydrogenase (ACADVL gene sequence) □ 1700 Warfarin Sensitivity (CYP2C9 & VKORC1 SNP analysis) □ 5220 Y Chromosome Microdeletion Analysis Thrombophilia (5140) (1 EDTA Lavender Top) □ 5141 Factor II (Prothrombin G20210A) □ 5143 Factor V (Leiden) □ 5145 MTHFR (C677T and A1298C)		





Patient Name

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HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT) - (EDTA Tube)	PULMONOLOGY - (EDTA Tube)			
□ 8662 HHT ACVRL1, ENG and SMAD4 gene sequence with ACRL1 and ENG deletion/duplication (concurrent) □ 1680 HHT ACVRL1 & ENG gene sequence and deletion/duplication □ 8660 HHT Steps 1 through 3 □ 1683 Step 1 ACVRL1 & ENG gene sequence □ 1681 Step 2 ACVRL1 & ENG deletion/duplication □ 1684 Step 3 SMAD4 gene sequence Call HHT Single Gene Deletion/Duplication GENE	Cystic Fibrosis - (EDTA Tube) □ 1002 508 FIRST™ (deltaF508 reflex to CFTR Amplified) □ 1012 508 ONLY™ (deltaF508 mutation only) □ 1007 CFTR Amplified (CFTR gene sequence and deletion/duplication) (concurrent) □ Report PolyT / TG Status □ 1006 CFTR Amplified (CFTR gene sequence reflex deletion/duplication) □ Report PolyT / TG Status □ 1000 CFTR gene sequence □ 1004 CFTR deletion/duplication			
NEUROLOGY / INTELLECTUAL DISABILITY	☐ 1010 CFTR TG repeat analysis (Poly T Variant & TG Repeat)			
8630	Primary Ciliary Dyskinesia +/- CFTR - (EDTA Tube) 8122			
Maternal Cell Contamination - (EDTA Tube)	RETT SYNDROME - (EDTA Tube)			
□ 1260 MCC for amniotic fluid culture or cvs (run concurrently with requested test) □ 1262 MCC Reference for maternal blood sample (No Charge)	□ 2028 Rett Syndrome - CDKL5 and MECP2 gene sequence with MECP2 del/dup (concurrent) 8200 Rett Syndrome - Steps 1-3 (reflex to next step when negative) 2020 Step 1 MECP2 gene sequence 2022 Step 2 MECP2 deletion/duplication			
NOONAN/LEOPARD SYNDROME - (EDTA Tube)	 □ 2040 Step 3 CDKL5 gene sequence □ 2026 MECP2 gene sequence reflex deletion/duplication 			
□ 8402 Noonan Syndrome - PTP11, SOS1, KRAS gene sequence and RAF1 partial (concurrent) □ 8400 Noonan Syndrome - Steps 1 and 2 (reflex to next step when negative) □ 2280 PTPN11 — Step 1 □ 2300 SOS1 — Step 2 □ 2320 RAF1 — Step 2 □ 2340 KRAS — KRAS — KRAS — Step 2 □ 8460 LEOPARD Syndrome (PTPN11 and partial RAF1 gene sequence) The following will be requested when ordering known mutation analysis for a mutation identified	SPECIFIC MUTATION / GENE ANALYSIS / DEL/DUP ANALYSIS - (EDTA Tube) Gene Sequence Analysis (GSA) Single Site-Mutation Analysis (SMA) Single Site-Del/Dup Analysis Gene Name: Mutation(s): Gene Name: Mutation(s):			
in an outside laboratory: 1) Proband report (mandatory) and 2) Positive Control (recommended). ACMG guidelines, CAP, and CLIA regulatory provisions recommend use of a positive control to provide evidence of amplification when interrogating a specific sequence alteration. It is recommended that individuals for a known genotype for the locus tested be included as a positive control to ensure assay performance.	□ Positive Control Not Available □ Positive Control Sent / To Be Sent Reporting Options □ Report Amino Acid changing polymorphisms (silent polymorphisms available on request)			