




Test Code	Test Name	Genes Included
<b>Adrenal Disorders</b>		
<input type="checkbox"/> 816	<b>Primary Adrenal Insufficiency (Addison's disease)</b>	ABCD1, NROB1, AIRE
<input type="checkbox"/> 812	Autoimmune Polyglandular Syndrome (AIRE) DNA Sequencing Test	
<input type="checkbox"/> 815	ABCD1 DNA Sequencing Test (X-linked Adrenoleukodystrophy)	
<input type="checkbox"/> 814	NROB1/DAX1 DNA Sequencing Test (X-linked Adrenal Hypoplasia Congenita)	
<input type="checkbox"/> 881	Endocrine Hypertension (HSD11B2) DNA Sequencing Test (Apparent Mineralocorticoid Excess)	
<input type="checkbox"/> 855	PHEX DNA Sequencing Test (X-linked Hypophosphatemic Rickets)	
<input type="checkbox"/> 856	FGF23 DNA Sequencing Test (Autosomal Dominant Hypophosphatemic Rickets)	
<input type="checkbox"/> 879	<b>Congenital Adrenal Hyperplasia Evaluation</b> CYP21A2 sequencing and deletion, CYP11B1 sequencing	
<input type="checkbox"/> 880	CYP21A2 (CAH) DNA Sequencing and Deletion Test	
	Required: Indication for Study (check one or more below):	
<input type="checkbox"/>	Family history of CAH	
<input type="checkbox"/>	Virilization (ambiguous genitalia)	
<input type="checkbox"/>	Salt Wasting	
<input type="checkbox"/>	Parent/sibling of CAH patient	
<input type="checkbox"/>	17-hydroxyprogesterone (17-OHP) elevated concentration in serum	
<input type="checkbox"/>	Other _____	
<input type="checkbox"/> 875	CYP11B1 (CAH) DNA Sequencing Test	
<input type="checkbox"/> 874	Lipoid CAH (STAR) DNA Sequencing Test	
<input type="checkbox"/> 877	CYP17A1 DNA Sequencing Test	
<input type="checkbox"/> 878	HSD3B2 DNA Sequencing Test	
<input type="checkbox"/> 881	Endocrine Hypertension (HSD11B2) DNA Sequencing Test	
<b>Bone Diseases</b>		
<input type="checkbox"/> 860	<b>Osteogenesis Imperfecta Evaluation</b>	COL1A1, COL1A2
<input type="checkbox"/> 861	COL1A1 (OI) DNA Sequencing Test	
<input type="checkbox"/> 862	COL1A2 (OI) DNA Sequencing Test	
<input type="checkbox"/> 811	Osteoporosis-Pseudoglioma (LRP5) DNA Sequencing Test	
<input type="checkbox"/> 821	Idiopathic Osteoporosis (LRP5) DNA Sequencing Test	
<input type="checkbox"/> 857	<b>Hypophosphatemic Rickets Evaluation</b>	PHEX, FGF23
<input type="checkbox"/> 855	PHEX (Hypophosphatemic Rickets) DNA Sequencing Test	
<input type="checkbox"/> 856	FGF23 (Hypophosphatemic Rickets) DNA Sequencing Test	
<b>Congenital Hyperinsulinism</b>		
<input type="checkbox"/> 819	<b>Congenital Hyperinsulinism Evaluation</b> GLUD1, GCK, KCNJ11, ABCC8	
	Indication for Study (check one or more below):	
<input type="checkbox"/>	Diazoxide Responsive	
<input type="checkbox"/>	Diazoxide Non-Responsive	
<input type="checkbox"/>	Hypoglycemic	
<input type="checkbox"/>	Large for Gestational Age (LGA)	
<input type="checkbox"/>	Other (describe) _____	
<input type="checkbox"/> 822	GLUD1 (CH) DNA Sequencing Test	
<input type="checkbox"/> 823	GCK (CH) DNA Sequencing Test	
<input type="checkbox"/> 826	KCNJ11 (CH) DNA Sequencing Test	
<input type="checkbox"/> 827	ABCC8 (CH) DNA Sequencing Test	
<input type="checkbox"/> 042	<b>CH Parental Testing - To augment child/proband diagnosis</b> <b>For expedited diagnosis of proband, send parental testing samples as soon as possible and provide information below.</b>	
<input type="checkbox"/>	Mother <input type="checkbox"/> Father	
	Proband Name/Accession # _____	

Test Code	Test Name	Genes Included
<b>Diabetes</b>		
<input type="checkbox"/> 8800	<b>Monogenic Diabetes (MODY) 4-Gene Evaluation</b>	GCK, HNF1A, HNF1B, HNF4A
<input type="checkbox"/> 8801	<b>Monogenic Diabetes (MODY) 3-Gene Evaluation</b>	GCK, HNF1A, HNF1B
<input type="checkbox"/> 8802	<b>Monogenic Diabetes (MODY) 2-Gene Evaluation</b>	GCK, HNF1A
<input type="checkbox"/> 885	<b>Monogenic Diabetes (MODY) 5-Gene Evaluation</b>	HNF1A, GCK, HNF4A, HNF1B, IPF1
<input type="checkbox"/> 802	HNF4A (MODY1) DNA Sequencing and Deletion Test	
<input type="checkbox"/> 803	GCK (MODY2) DNA Sequencing and Deletion Test	
<input type="checkbox"/> 804	HNF1A (MODY3) DNA Sequencing and Deletion Test	
<input type="checkbox"/> 834	IPF1 (MODY4) DNA Sequencing Test	
<input type="checkbox"/> 805	HNF1B (MODY5) DNA Sequencing and Deletion Test	
<input type="checkbox"/> 837	CEL (MODY8) DNA Sequencing Test	
<input type="checkbox"/> 882	<b>Neonatal Diabetes Mellitus Evaluation</b>	IPF1, GCK, KCNJ11, INS, ABCC8
<input type="checkbox"/> 841	IPF1 (NDM) DNA Sequencing Test	
<input type="checkbox"/> 842	GCK (NDM) DNA Sequencing Test	
<input type="checkbox"/> 843	KCNJ11 (NDM) DNA Sequencing Test	
<input type="checkbox"/> 853	INS (NDM) DNA Sequencing Test	
<input type="checkbox"/> 876	ABCC8 (NDM) DNA Sequencing Test	
<b>Nephrogenic Diabetes</b>		
<input type="checkbox"/> 854	<b>Nephrogenic Diabetes Insipidus Evaluation</b>	AVPR2, AQP2
<input type="checkbox"/> 851	Nephrogenic Diabetes Insipidus (AVPR2) DNA Sequencing Test	
<input type="checkbox"/> 852	Nephrogenic Diabetes Insipidus (AQP2) DNA Sequencing Test	
<b>Familial Cancer Syndromes</b>		
<input type="checkbox"/> 818	MEN1 (MEN1) DNA Sequencing Test	
<input type="checkbox"/> 889	<b>Pheochromocytoma Evaluation</b>	RET, VHL, SDHB
<input type="checkbox"/> 813	MEN2 (RET) DNA Sequencing Test	
<input type="checkbox"/> 858	von Hippel-Lindau Syndrome (VHL) DNA Sequencing Test	
<input type="checkbox"/> 888	SDHB DNA Sequencing Test	
<b>Familial Hypocalciuric Hypercalcemia</b>		
<input type="checkbox"/> 829	Familial Hypocalciuric Hypercalcemia (CASR) DNA Sequencing Test	
<b>Familial Testing - Targeted Analysis</b>		
<input type="checkbox"/> 800	<b>Familial DNA Sequence Evaluation</b> This test detects previously identified sequence variants in at-risk family members. This test is available for HNF4A, GCK, TCF1, IPF1, TCF2, COL1A1, COL1A2, MEN1, and RET mutations Proband Accession # _____ Relationship _____	
<b>Lipid Disorders</b>		
<input type="checkbox"/> 895	<b>Hypercholesterolemia Evaluation</b>	LDLR, APOB
<input type="checkbox"/> 894	LDLR (Hypercholesterolemia) DNA Sequencing Test	
<input type="checkbox"/> 893	APOB Mutation Analysis	
<b>Obesity</b>		
<input type="checkbox"/> 884	<b>Early Onset Obesity Panel</b>	LEPR, MC4R
<input type="checkbox"/> 883	Early Onset Obesity (LEPR) DNA Sequencing Test	
<input type="checkbox"/> 640	Early Onset Obesity (MC4R) DNA Sequencing Test	
<input type="checkbox"/> 887	<b>Bardet-Biedl Syndrome Evaluation</b>	BBS1, BBS2, BBS10
<input type="checkbox"/> 871	BBS1 (BBS) DNA Sequencing Test	
<input type="checkbox"/> 872	BBS2 (BBS) DNA Sequencing Test	
<input type="checkbox"/> 886	BBS10 (BBS) DNA Sequencing Test	

**Note:** Test requisitions become outdated. For the most accurate and up-to-date test offering, please visit [AthenaDiagnostics.com](http://AthenaDiagnostics.com).

Test Code	Test Name	Genes Included
<b>Reproductive Disorders</b>		
<input type="checkbox"/> 817	Male Precocious Puberty (LHCGR) DNA Sequencing Test	
<input type="checkbox"/> 679	<b>Complete Kallmann/IHH Evaluation</b>	CHD7, KAL1, PROK2, PROKR2, FGFR3, FGFR1, GnRHR, GnRH1, KISS1R, TACR3
<input type="checkbox"/> 667	<b>Normosmic Kallmann/IHH Evaluation</b>	PROK2, PROKR2, FGFR1, GnRHR, GnRH1, TACR3, KISS1R
<input type="checkbox"/> 462	<b>Anosmic Kallmann/IHH Evaluation</b>	KAL1, PROK2, PROKR2, FGFR3, FGFR1, GnRHR, KISS1R
<input type="checkbox"/> 173	KAL1 DNA Sequencing Test	
<input type="checkbox"/> 175	PROK2 DNA Sequencing Test	
<input type="checkbox"/> 180	PROKR2 DNA Sequencing Test	
<input type="checkbox"/> 195	FGFR3 DNA Sequencing Test	
<input type="checkbox"/> 196	FGFR1 DNA Sequencing Test	
<input type="checkbox"/> 279	GnRHR DNA Sequencing Test	
<input type="checkbox"/> 343	GnRH1 DNA Sequencing Test	
<input type="checkbox"/> 358	TACR3 DNA Sequencing Test	
<input type="checkbox"/> 364	KISS1R DNA Sequencing Test	
<input type="checkbox"/> 461	CHD7 DNA Sequencing Test	

Test Code	Test Name	Genes Included
<b>Short Stature</b>		
<input type="checkbox"/> 865	<b>Combined Pituitary Hormone Deficiency Evaluation</b>	PROPI, POU1F1
<input type="checkbox"/> 863	PROPI (CPHD) DNA Sequencing Test	
<input type="checkbox"/> 864	POU1F1 (CPHD) DNA Sequencing Test	
<input type="checkbox"/> 848	<b>Growth Hormone Deficiency (GHD) Evaluation</b>	GH1 and GHRHR Seq.; SHOX Seq. and Del.
<input type="checkbox"/> 866	GH1 (GHD) DNA Sequencing Test	
<input type="checkbox"/> 868	GHRHR (GHD) DNA Sequencing Test	
<input type="checkbox"/> 847	SHOX (GHD) DNA Sequencing and Deletion Test	
<input type="checkbox"/> 867	GHR (SS) DNA Sequencing Test	
<b>Noonan Syndrome</b>		
<input type="checkbox"/> 846	Noonan Syndrome (PTPN11) DNA Sequencing Test	
<input type="checkbox"/> 658	<b>KRAS/RAF1/SOS1 DNA Sequencing Evaluation</b>	SOS1, RAF1, KRAS
<input type="checkbox"/> 662	SOS1 DNA Sequencing Test	
<input type="checkbox"/> 663	RAF1 DNA Sequencing Test	
<input type="checkbox"/> 664	KRAS DNA Sequencing Test	

**Specimen Requirements & Shipping Information (applies to all tests)**

- Specimen Type:** Whole blood, 8 mL in yellow or lavender top (pediatric minimum volume: 2 mL)
- Stability:** Hemolysis may compromise DNA recovery and integrity after 48 hrs. It is recommended to ship samples immediately after draw. Samples can be stored for short periods only. Send specimen overnight at room temperature.
- Shipping:** Send specimen overnight at room temperature. If you have any questions on sample requirements or shipping, contact our client service department at 00-1-508-756-2886, extension 2.

**NOTE: Specimen tube(s) must be labeled with two of the following forms of identification: name, date of birth, last four digits of SS#, patient ID no. These same two forms of ID must also be indicated on the test requisition.**

Athena Diagnostics Client Service Representatives are available from 8:30am to 6:30pm Eastern Time (U.S.).

International Customers please call  
**00-1-508-756-2886**

or Fax 00-1-774-843-3721



200 Forest Street, 2nd Floor  
 Marlborough, MA 01752 • [AthenaDiagnostics.com](http://AthenaDiagnostics.com)

To: Whom it may concern  
From:  
CC:  
Date:  
Re: International Blood Sample

Please be advised that:

The items contained in this shipment under Air bill number \_\_\_\_\_ are samples of whole human blood from a patient in plastic specimen collector tubes that will be used for Laboratory testing only.

The specimen collection tubes in the shipment are of "Human material containing no animal material "or" Non-human primate material or other non-primate animal material.

We hereby declare that the human blood samples from a patient contained in this shipment under Air bill number \_\_\_\_\_ "are not of tissue culture origin or any imported material that is a human vaccine in final dosage form.

We also so state that the samples contained in this shipment under Air bill number \_\_\_\_\_, are non-cultured, non-recombinant, non-infectious, containing no animal content and/or bovine serum albumin.

We also declare that the human blood samples from a patient contained in this package under Air bill number \_\_\_\_\_, have not had any previous testing.

The sample is being sent to Athena Diagnostics, Inc., the only clinical reference laboratory dedicated exclusively to testing for certain neurological disorders, their mission extends to providing diagnostic results that encompass interpretation, counseling and educational resources for healthcare professional and the patients for whom they care.

If the above advisories do not meet with the protocol standards, please advise. The telephone number is \_\_\_\_\_.

Sincerely,

\_\_\_\_\_