NEWBORN SCREENING FOLLOW-UP

PANELS* AND SINGLE GENE TESTS

AMINO ACID DISORDERS

Argininemia
NEXTGEN SEQUENCING WITH CNV

11825	*ARG1*	**$890**

Argininosuccinate Lyase Deficiency
NEXTGEN SEQUENCING WITH CNV

9531	*ASL*	**$890**

Carbamoylphosphate Synthetase I Deficiency
NEXTGEN SEQUENCING WITH CNV

11829	*CPS1*	**$890**

Citrullinemia, Type I
NEXTGEN SEQUENCING WITH CNV

9535	*ASS1*	**$890**

Gyrate Atrophy of Choroid and Retina
NEXTGEN SEQUENCING WITH CNV

8339	*OAT*	**$890**

Homocystinuria
NEXTGEN SEQUENCING WITH CNV

10201	**5 GENES**	**$890**
CBS, MMADHC, MTHFR, MTR, MTRR		

Hypermethioninemia
NEXTGEN SEQUENCING WITH CNV

10133	**4 GENES**	**$890**
ADK, AHCY, GNMT, MAT1A		

Hyperphenylalaninemia
NEXTGEN SEQUENCING WITH CNV**

3403	**6 GENES**	**$640**
DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR		

Maple Syrup Urine Disease
NEXTGEN SEQUENCING WITH CNV**

3281	**3 GENES**	**$640**
BCKDHA, BCKDHB, DBT		

Ornithine Transcarbamylase Deficiency
NEXTGEN SEQUENCING WITH CNV

9723	*OTC*	**$930**

Phenylalanine Hydroxylase Deficiency
NEXTGEN SEQUENCING WITH CNV

9725	*PAH*	**$890**

SLC25A13-Related Disorders
NEXTGEN SEQUENCING WITH CNV

11857	*SLC25A13*	**$890**

** All genes in panels are available as single gene tests.*
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PANELS* AND SINGLE GENE TESTS

Tyrosinemia
NEXTGEN SEQUENCING WITH CNV

5011	**4 GENES**	**$890**
FAH, GSTZ1, HPD, TAT		

Urea Cycle Disorders
NEXTGEN SEQUENCING WITH CNV

10273	**9 GENES**	**$890**
ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15		

DISORDERS OF FATTY ACID OXIDATION

Disorders of Fatty Acid Oxidation
NEXTGEN SEQUENCING WITH CNV

10381	**27 GENES**	**$890**
ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, CPT1A, CPT2, DECR1, ECHS1, ETFA, ETFB, ETFDH, FLAD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, MLYCD, PPARG, SLC22A5, SLC25A20, TAZ		

Disorders of Folate Metabolism and Transport
NEXTGEN SEQUENCING WITH CNV

10121	**3 GENES**	**$890**
FOLR1, MTHFR, SLC46A1		

Glutaric Acidemia Type II
NEXTGEN SEQUENCING WITH CNV

10127	**3 GENES**	**$890**
ETFA, ETFB, ETFDH		

Medium Chain Acyl-CoA Dehydrogenase Deficiency
NEXTGEN SEQUENCING WITH CNV

9505	*ACADM*	**$890**

Mitochondrial Trifunctional Protein Deficiency and Long-Chain 3-Hydroxyacyl CoA Dehydrogenase Deficiency
NEXTGEN SEQUENCING WITH CNV

10635	**2 GENES**	**$890**
HADHA, HADHB		

Short Chain Acyl-CoA Dehydrogenase Deficiency
NEXTGEN SEQUENCING WITH CNV

8461	*ACADS*	**$890**

Smith-Lemli-Opitz Syndrome
NEXTGEN SEQUENCING WITH CNV

9573	*DHCR7*	**$890**

Systemic Primary Carnitine Deficiency
NEXTGEN SEQUENCING WITH CNV

9477	*SLC22A5*	**$890**

Very Long Chain Acyl-CoA Dehydrogenase Deficiency
NEXTGEN SEQUENCING WITH CNV

9507	*ACADVL*	**$ 890**

NEWBORN SCREENING FOLLOW-UP

PANELS* AND SINGLE GENE TESTS

ENDOCRINE DISORDERS

Carnitine Palmitoyltransferase 1A Deficiency
NEXTGEN SEQUENCING WITH CNV

9297	*CPT1A*	**$890**

Carnitine Palmitoyltransferase II Deficiency
NEXTGEN SEQUENCING WITH CNV

9563	*CPT2*	**$890**

Carnitine-Acylcarnitine Translocase Deficiency
NEXTGEN SEQUENCING WITH CNV

11859	*SLC25A20*	**$890**

Congenital Hypothyroidism and Thyroid Hormone Resistance
NEXTGEN SEQUENCING WITH CNV

1989	**26 GENES**	**$890**

DUOX2, DUOXA2, FOXE1, GLIS3, GNAS, HESX1, IGSF1, IYD, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC5A5, TG, THRA, THRB, TPO, TRH, TRHR, TSHB, TSHR, UBR1

HEARING LOSS

Deafness, Autosomal Dominant 3A (DFNA3A) and Deafness, Autosomal Recessive 1A (DFNB1A)
NEXTGEN SEQUENCING WITH CNV**

7677	*GJB2*	**$640**

Hereditary Hearing Loss and Deafness
NEXTGEN SEQUENCING WITH CNV

5063	**203 GENES**	**$1,290**

A2ML1, ABHD12, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, AP1B1, ARSG, ASIC5, ATP1A3, ATP2B2, ATP6V1B1, BCS1L, BDP1, BSND, CABP2, CACNA1D, CATSPER2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLDN9, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL1A1, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, CRYL1, CRYM, DCDC2, DIABLO, DIAPH1, DIAPH3, DMXL2, DNAJC3, DSPP, DTNA, EDN3, EDNRB, ELMOD3, EPS8, EPS8L2, ERAL1, ESPN, ESRRB, EYA1, EYA4, FAM136A, FDXR, FGF3, FGFR3, FITM2, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPRASP2, GPSM2, GRAP, GREB1L, GRHL2, GRXCR1, GRXCR2, GSDME, HARS, HARS2, HGF, HOMER2, HOXA2, HSD17B4, IFNLR1, ILDR1, KARS1, KCNE1, KCNJ10, KCNQ1, KCNQ4, KITLG, LARS2, LHFPL5, LMX1A, LOXHD1, LRTOMT, MAFB, MARVELD2, MCM2, MEPE, MET, MIR96, MITF, MPZL2, MRPS2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NLRP3, NOG, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PDE1C, PDZD7, PEX1, PEX26, PEX6, PJVK, PLS1, PLS3, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRKCB, PRPS1, PTPRQ, RAI1, RDX, REEP6, REST, RIPOR2, ROR1, S1PR2, SEMA3E, SERPINB6, SIX1, SIX2, SIX5, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC44A4, SLC52A2, SLC52A3, SLC9A1, SLITRK6, SMPX, SNAI2, SOX10, SPNS2, STRC, SYNE4, TBC1D24, TBX1, TCOF1, TECTA, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMT10C, TRRAP, TSHZ1, TSPEAR, TUBB4B, TWNK, USH1C, USH1G, USH2A, WBP2, WFS1, WHRN, XYLT2

Pendred Syndrome and Nonsyndromic Hearing Loss Associated with Enlarged Vestibular Aqueduct
NEXTGEN SEQUENCING WITH CNV**

4861	*SLC26A4*	**$640**

PANELS* AND SINGLE GENE TESTS

HEMOGLOBIN DISORDERS

Alpha Thalassemia Deletion / Duplication and Constant Spring
MLPA**

6070	**2 GENES**	**$540**
HBA1, HBA2		

Alpha Thalassemia
SANGER SEQUENCING AND MLPA**

6090	**2 GENES**	**$1,470**
HBA1, HBA2		

Beta-Thalassemia and Hemoglobinopathy
NEXTGEN SEQUENCING WITH CNV**

7693	*HBB*	**$640**

LYSOSOMAL STORAGE DISORDERS

Fabry Disease
NEXTGEN SEQUENCING WITH CNV

7681	*GLA*	**$640**

Gaucher Disease
SANGER SEQUENCING

479	*GBA*	**$990**

Glycogen Storage Disease Type II (Pompe Disease)
NEXTGEN SEQUENCING WITH CNV

9605	*GAA*	**$890**

Krabbe Disease
TARGETED DELETION

632	*GALC Exons 11-17 (502T/Del)*	**$250**

Krabbe Disease
NEXTGEN SEQUENCING WITH CNV

7883	*GALC*	**$640**

Lysosomal Storage Disorders
NEXTGEN SEQUENCING WITH CNV

13065	**146 GENES**	**$1,280**

ABCC8, ABCD1, ABHD5, ACOX1, ACY1, ADAMTS10, ADAMTSL2, ADSL, AGA, ALDH7A1, AMACR, AMT, ANTXR2, AP3B1, ARG1, ARSA, ARSB, ASAH1, ASPA, ATG5, ATP13A2, ATP6AP1, BLOC1S3, BLOC1S6, BTD, CLCN5, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, DHCR7, DNAJC5, DNM1L, DPYD, DTNBP1, DYM, ETFA, ETFB, ETFDH, FAR1, FBN1, FH, FIG4, FOLR1, FUCA1, GAA, GALC, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GRN, GUSB, HEXA, HEXB, HGSNAT, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HSD17B4, HYAL1, IDS, IDUA, KCTD7, KMT2D, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, LTBP2, LYST, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MLPH, MOCS1, MOCS2, MYO5A, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, NPR2, OCRL, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PHYH, PNPLA2, PPT1, PSAP, QDPR, RAB27A, RAI1, SCARB2, SCP2, SGSH, SLC17A5, SLC25A15, SLC46A1, SLC9A6, SMPD1, SUMF1, SUOX, TCF4, TPP1, TRIM37, VPS33A

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NEWBORN SCREENING FOLLOW-UP

PANELS* AND SINGLE GENE TESTS

Mucopolysaccharidosis Type I
NEXTGEN SEQUENCING WITH CNV

7713	*IDUA*	**$640**

Mucopolysaccharidosis Type III
NEXTGEN SEQUENCING WITH CNV

3419	**4 GENES**	**$640**

GNS, HGSNAT, NAGLU, SGSH

Niemann-Pick Disease Type C
NEXTGEN SEQUENCING WITH CNV

3425	**2 GENES**	**$640**

NPC1, NPC2

Niemann-Pick Disease Types A and B
NEXTGEN SEQUENCING WITH CNV

7833	*SMPD1*	**$640**

ORGANIC ACID DISORDERS

Methylmalonic Aciduria and Homocystinuria
NEXTGEN SEQUENCING WITH CNV

10245	**10 GENES**	**$960**

ABCD4, CD320, HCFC1, LMBRD1, MMACHC, MMADHC, PRDX1, TCN2, THAP11, ZNF143

ß-Ketothiolase Deficiency
NEXTGEN SEQUENCING WITH CNV

9499	*ACAT1*	**$890**

3-Hydroxy-3-MethylGlutaryl-CoA Lyase Deficiency
NEXTGEN SEQUENCING WITH CNV

9653	*HMGCL*	**$890**

3-Methylcrotonyl-CoA Carboxylase Deficiency
NEXTGEN SEQUENCING WITH CNV

10069	**2 GENES**	**$890**

MCCC1, MCCC2

3-Methylglutaconic Aciduria Type I
NEXTGEN SEQUENCING WITH CNV

11827	*AUH*	**$890**

Disorders Related to Metabolism of Cobalamin, Folate and Homocysteine
NEXTGEN SEQUENCING WITH CNV

10397	**39 GENES**	**$960**

ABCD4, ACSF3, ADK, AHCY, ALDH6A1, AMN, CBS, CD320, CUBN, DHFR, FOLR1, FTCD, GIF, GNMT, HCFC1, HIBCH, LMBRD1, MAT1A, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MTHFD1, MTHFR, MTR, MTRR, PCCA, PCCB, PRDX1, SLC46A1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143

Ethylmalonic Encephalopathy
NEXTGEN SEQUENCING WITH CNV

7637	*ETHE1*	**$640**

PANELS* AND SINGLE GENE TESTS

Glutaric Acidemia Type I
NEXTGEN SEQUENCING WITH CNV

9617	*GCDH*	**$890**

Holocarboxylase Synthetase Deficiency
NEXTGEN SEQUENCING WITH CNV

9651	*HLCS*	**$890**

Isovaleric Acidemia
NEXTGEN SEQUENCING WITH CNV

9667	*IVD*	**$890**

Malonyl-CoA Decarboxylase Deficiency
NEXTGEN SEQUENCING WITH CNV

8567	*MLYCD*	**$890**

Methylmalonic Acidemia
NEXTGEN SEQUENCING WITH CNV

10295	**11 GENES**	**$930**

ACSF3, ALDH6A1, CD320, MCEE, MLYCD, MMAA, MMAB, MMADHC, MMUT, SUCLA2, SUCLG1

Metabolic Hypoglycemia
NEXTGEN SEQUENCING WITH CNV

10365	**38 GENES**	**$960**

ACADM, ACADVL, ACAT1, ACSF3, AGL, ALDOB, CA5A, DGUOK, ETFA, ETFB, ETFDH, FBP1, G6PC, GALT, GK, GYS2, HADH, HMGCL, HMGCS2, MLYCD, MPV17, NNT, OXCT1, PC, PCK1, PCK2, PGM1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, SLC16A1, SLC22A5, SLC25A20, SLC2A2, SLC37A4, TANGO2

Organic Aciduria
NEXTGEN SEQUENCING WITH CNV

10391	**29 GENES**	**$930**

ACAT1, ACSF3, ALDH6A1, BCKDHA, BCKDHB, CD320, D2HGDH, DBT, DLD, ETFA, ETFB, ETFDH, GCDH, HLCS, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, PCCA, PCCB, SLC25A1

Propionic Acidemia
NEXTGEN SEQUENCING WITH CNV

3291	**2 GENES**	**$640**

PCCA, PCCB

OTHER DISORDERS

Biotinidase Deficiency
NEXTGEN SEQUENCING WITH CNV

9551	*BTD*	**$890**

Comprehensive Congenital Heart Disease
NEXTGEN SEQUENCING WITH CNV

13008	**398 GENES**	**$1,490**

See our website for a complete list of genes.

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NEWBORN SCREENING FOLLOW-UP

PANELS* AND SINGLE GENE TESTS

Cystic Fibrosis and CF-Related Disorders
NEXTGEN SEQUENCING WITH CNV**

3035	*CFTR*	**$640**

Epimerase Deficiency Galactosemia
NEXTGEN SEQUENCING WITH CNV

8335	*GALE*	**$890**

Galactosemia Panel
NEXTGEN SEQUENCING WITH CNV

5013	**4 GENES**	**$890**

GALE, GALK1, GALM, GALT

Galactosemia Type I (Classic and Variant Galactosemia)
NEXTGEN SEQUENCING WITH CNV

9613	*GALT*	**$890**

Glycine Encephalopathy
NEXTGEN SEQUENCING WITH CNV

10129	**3 GENES**	**$890**

AMT, GCSH, GLDC

Inborn Errors of Immunity / Primary Immunodeficiency
NEXTGEN SEQUENCING WITH CNV

13999	**486 GENES**	**$1,490**

See our website for a complete list of genes.

Neonatal Crisis (NICU)
PGXOME SEQUENCING PANEL WITH CNV DETECTION

7383	**1,837 GENES**	Patient Only NICU panel of patient only	**$1,890**
10065	**1,837 GENES**	Family - Duo NICU panel of patient + 1 additional family member	**$2,290**
10066	**1,837 GENES**	Family - Trio NICU panel of patient + 2 additional family members	**$2,490**

Sequencing of Additional Family Member	**$390**
Report for Additional Family Member	**$490**
Reflex to whole exome	**$390**

See our website for a complete list of genes and a list of disorders related to this panel.

Peroxisomal Disorders
NEXTGEN SEQUENCING WITH CNV

10369	**27 GENES**	**$1,030**

ABCD1, ABCD3, ACOX1, AGPS, AGXT, AMACR, CAT, DNM1L, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2, TRIM37

PANELS* AND SINGLE GENE TESTS

Primary Ciliary Dyskinesia (PCD) / Immotile Cilia Syndrome and Cystic Fibrosis
NEXTGEN SEQUENCING WITH CNV

10415	**47 GENES**	**$930**

AK7, ARMC4, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CFAP298, CFAP300, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAL1, DRC1, FOXJ1, GAS2L2, GAS8, INVS, LRRC56, LRRC6, MCIDAS, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, SPEF2, TTC12, TTC25, ZMYND10

Pyruvate Dehydrogenase Complex Deficiency
NEXTGEN SEQUENCING WITH CNV

5035	**6 GENES**	**$890**

DLAT, DLD, PDHA1, PDHB, PDHX, PDP1

Severe Combined Immunodeficiency (SCID)
NEXTGEN SEQUENCING WITH CNV

5209	**17 GENES**	**$890**

ADA, AK2, CD247, CD3D, CD3E, CORO1A, DCLRE1C, IL2RG, IL7R, JAK3, LAT, LIG4, NHEJ1, PRKDC, PTPRC, RAG1, RAG2

Spinal Muscular Atrophy (SMA)
MLPA**

6064	**2 GENES**	**$540**

SMN1, SMN2

X-Linked Adrenoleukodystrophy
NEXTGEN SEQUENCING WITH CNV**

7557	*ABCD1*	**$640**

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PREVENTION GENETICS

3800 South Business Park Avenue • Marshfield, Wisconsin 54449
Phone: (715) 387-0484 • **www.PreventionGenetics.com**
Email: support@preventiongenetics.com