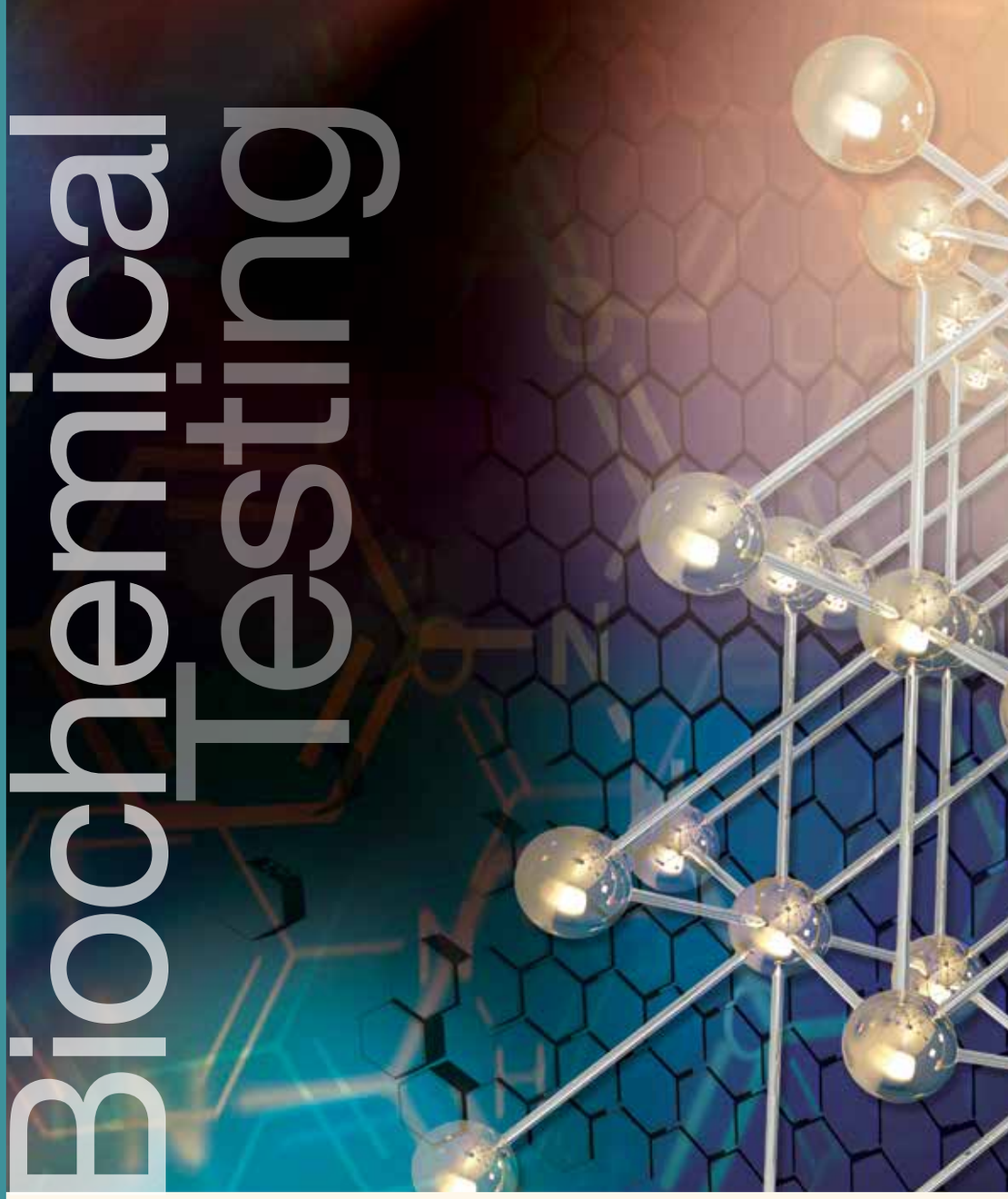
Baylor Miraca
Genetics Laboratories

Biochemical Testing

Biochemical Testing



The Biochemical Laboratory at Baylor Miraca Genetics Laboratories, performs specific testing for the purpose of diagnosing and monitoring patients with inborn errors of metabolism. The laboratory provides testing for the quantitative determination of a broad array of analytes. The lab offers a broad spectrum of quantitative analysis. In addition, over 30 enzyme assays are available for specific disorders. Professionals in human genetics provide reports with interpretations and telephone consultations are available with the director and medical geneticist.

Sample and Shipping Information

Specimen Type: 4901: Urine

Requirements: Send 2-4 cc of random urine.

Specimen Type: 4900: Plasma

Requirements: Draw blood in a EDTA (purple top) tube(s) and separate as soon as possible, freezing immediately. Send 1-2 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: 4902: Cerebrospinal fluid

Requirements: Send 1-2 cc of cerebrospinal fluid.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Forms or Sample Kits: Requisition and signed consent form must accompany specimen. Prior to any genetic testing, we recommend discussion of the risks and benefits of testing with a physician or genetic counselor. To receive our forms, additional information, or kits, please contact our laboratory: 1-800-411-GENE.

Shipping Address:
Baylor Miraca Genetics Laboratories
2450 Holcombe Grand Blvd.-Recieving Dock, Houston, Texas 77021
1-800-411-GENE (4363) or 713-798-6555 • www.BMGL.com

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PYRIMIDINE METABOLISM DISORDERS AND 5-FLUOROURACIL TOXICITY SCREENING

Defects in the breakdown of pyrimidines are known to cause multiple autosomal reccessive inborn errors of metabolism, including dihydropyrimidine dehydrogenase (DPD) deficiency, dihydropyrimidinase (DHP) deficiency, and mitochondrial encephalopathy with gastro-intestinal symptoms (MNGIE). Key to the diagnosis of these disorders is the ability to detect urinary elevations of pyrimidine catabolic intermediates including uracil, thymine, dihydrouracil, dihydrothymine, and thymidine[1].

DPD and DHP deficiencies are disorders sharing similar clinical features that can range from asymptomatic to intellectual disability, microcephaly, epilepsy, and failure to thrive[1]. In addition, DPD deficiency has been linked to 50-75% of severe toxicities induced by the chemotherapeutic agent 5-fluorouracil (5-FU), leading some to recommend DPD testing prior to 5-FU administration[2]. DPD and DHP enzymes work sequentially in the breakdown of uracil/thymine to dihydrouracil/dihydrothymine and then to β-ureidopropionate/β-aminoisobutyrate. Thus, diagnosis of each disorder can be achieved by measuring the urinary excretion of uracil and thymine for DPD and dihydrouracil and dihydrothymine in the case of DHP[3].

MNGIE is a mitochondrial disorder resulting from defects in thymidine phophorylase. Patients with MNGIE typically present with cachexia within the first two decades of life; other clinical features include intestinal pseudo-obstruction, myopathy, and neuropathy[4]. Extreme elevation of thymidine in a patient's urine is diagnostic for this disorder[5].

Turn Around Time: 7 days

Test Code: Pyrimidine Panel 4215

CPT Codes: 82570X1, 82544X1

Related Tests
Plasma Thymidine 4330

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4901: Urine

Requirements: Send 2-4 cc of random urine.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

References

1. *Nyhan WL. Disorders of purine and pyrimidine metabolism. Molecular genetics and metabolism 86:25-33, 2005.*
2. *Ciccolini J, Gross E, Dahan L, Lacarelle B, Mercier C. Routine dihydropyrimidine dehydrogenase testing for anticipating 5-fluorouracil-related severe toxicities: hype or hope? Clinical colorectal cancer 9:224-8, 2010.*
3. *Jiang H, Jiang J, Hu P, Hu Y. Measurement of endogenous uracil and dihydrouracil in plasma and urine of normal subjects by liquid chromatography-tandem mass spectrometry. Journal of chromatography B, Analytical technologies in the biomedical and life sciences 769:169-76, 2002.*
4. *Garone C, Tadesse S, Hirano M. Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. Brain: a journal of neurology 134:3326-32, 2011.*
5. *Marti R, Lopez LC, Hirano M. Assessment of thymidine phosphorylase function: measurement of plasma thymidine (and deoxyuridine) and thymidine phosphorylase activity. Methods Mol Biol 837:121-33, 2012.*

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BIOCHEMICAL PANEL FOR AUTISM SPECTRUM DISORDERS

Autism Spectrum Disorders (ASDs) are an etiologically diverse group of neurodevelopmental disorders characterized by communication/social interaction difficulties and repetitive behaviors[1]. Inborn errors of metabolism affecting amino acid, creatine, and carnitine biosynthesis phenotypically overlap with or have been directly linked to ASD. Within the Austim Panel (4000) are eight tests designed to allow rapid screening for a wide variety of metabolic disorders that can lead to ASD or ASD-like symptoms.

Autistic-like features such as general cognitive impairment can result from organic acidurias and aminoacidopathies, e.g., phenylketonuria and ornithine transcarbamoylase defiency. More directly, within the branched chain amino acid metabolic pathway, mutations in branched-chain ketoacid dehydrogenase kinase (BCKDK) have been reported to cause autism[2]. To aid in the diagnosis of these disorders, we have included plasma amino acid, acylcarnitine and urine organic acid analysis in our autism panel.

Defects in creatine metabolism or transport can lead to the disruption of expressive and cognitive speech as well as cause intellectual disability, seizures, and movement disorders[3,4]. We at the Baylor Miraca Genetics Laboratories pioneered the tandem MS-based diagnosis of creatine metabolic disorders[5]. Included in the autism panel are two tests designed to detect the autosomal recessive arginine:glycine amidinotransferase (AGAT) and guanidinoacetate methylteansferase (GAMT) deficiencies, as well as X-linked creatine transporter defects.

In some instances of familial x-linked nondysmorphic autism, the causitive mutation resides in the carnitine biosynthetic enzyme trimethyllysine hydroxylase epsilon (TMLHE), thus implicating carnitine metabolism in autism[6]. In these particular cases of autism, diagnosis can be made via the quantification of urine or plasma levels of trimethyllysine and gamma butyrobetaine.

Turn Around Time: 3 to 10 days

Test Codes: Autism Panel 4000

CPT Codes: 82017x1, 82379x3, 82540x1, 82543x4, 82570x1, 83150x1, 83789x6 ,83919x1, 83945x1

Related Tests

Plasma Amino Acid Analysis 4100

Plasma Acylcarnitine Profile 4300

Plasma Creatine/Guanidinoacetate Analysis 4130

Urine Creatine/Guanidinoacetate Analysis 4260

Urine Purine Analysis 4220

Urine Organic Acid Analysis 4200

Urine Carnitine Biosynthesis Panel 4135

Urine Pyrimidine Panel 4215

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4900: Plasma

Requirements: Draw blood in a EDTA (purple top) tube(s) and separate as soon as possible, freezing immediately. Send 1-2 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: 4901: Urine

Requirements: Send 2-4 cc of random urine.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

References

1. *Tchaconas A, Adesman A. Autism spectrum disorders: a pediatric overview and update. Curr Opin Pediatr 25:130-44, 2013.*
2. *Novarino G, El-Fishawy P, Kayserili H, et al. Mutations in BCKD-kinase lead to a potentially treatable form of autism with epilepsy. Science 338:394-7, 2012.*
3. *Schulze A. Creatine deficiency syndromes. Mol Cell Biochem 244:143-50, 2003.*
4. *Lion-Francois L, Cheillan D, Pitelet G, et al. High frequency of creatine deficiency syndromes in patients with unexplained mental retardation. Neurology 67:1713-4, 2006.*
5. *Sun Q, O'Brien WE. Diagnosis of creatine metabolism disorders by determining creatine and guanidinoacetate in plasma and urine. Methods Mol Biol 603:175-85, 2010.*
6. *Celestino-Soper PB, Violante S, Crawford EL, et al. A common X-linked inborn error of carnitine biosynthesis may be a risk factor for nondysmorphic autism. Proc Natl Acad Sci U S A 109:7974-81, 2012.*

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PYRIDOXINE-DEPENDENT EPILEPSY

Autosomal recessive pyridoxine-dependent epilepsy disorder (PDE; OMIM 266100) has a frequency of 1/400,000 – 1/700,000 and can present as clonic, generalized tonic-clonic and/or myoclonic seizures. Onset of seizures is typically in the first hours of life, but may present as late as three weeks of age. Accurate diagnosis is critical for patient management as seizures are often unresponsive to standard anticonvulsants and good seizure control is typically achievable with administration of pyridoxine hydrochloride. Until recently, definitive diagnosis of PDE was based on successful control of epilepsy with pyridoxine therapy and recurrence of seizures after pyridoxine withdrawal[1].

Recent research has demonstrated that in PDE, pipecolic acid (PA) and alpha-amino adipic semialdehyde (AASA) are markedly elevated in urine, plasma, and cerebrospinal fluid, and thus are diagnostic biomarkers of the disorder[2,3]. The neurotoxic affects of AASA are thought to result from the accumulation of its cyclic derivative L-Δ1- piperideine-6-carboxylate (P6C). In PDE, excess P6C condenses with pyridoxal-5'-phosphate (PLP) and inactivates this essential cofactor in neurotransmitter metabolism[4].

This panel determines both P6C and PA concentrations in either cerebral spinal fluid (test code 4812) or plasma (test code 4811) by tandem mass spectroscopy (under our assay conditions a majority of AASA is converted to P6C). Note that determination of PA alone is not specific for diagnosis of PDE because PA is also found elevated in peroxisomal disorders[5].

Turn Around Time: 6 days

Test Codes: PLASMA 4811, CSF 4812

CPT Codes: 82543x4

Related Tests
Pyridoxine-Dependent Seizures-ALDH7A1
Gene Sequence 6850

Alternative Disease Title: Pyridoxine Dependency With Seizures; AASA Dehydrogenase Deficiency; Pyridoxine-Dependent Seizures

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4900: Plasma

Requirements: Draw blood in a EDTA (purple top) tube(s) and separate as soon as possible, freezing immediately. Send 1-2 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: 4902: Cerebrospinal fluid

Requirements: Send 1-2 cc of cerebrospinal fluid.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

References

1. *Gospe M. Pyridoxine-dependent seizures: findings from recent studies pose new questions. Pediatr Neurol 26:181–185, 2002.*
2. *Plecko, B., Paul, K., Paschke, E., Stoeckler-Ipsiroglu, S., Struys, E., Jakobs, C., Hartmann, H., Luecke, T., di Capua, M., Korenke, C., Hikel, C., Reutershahn, E., Freilinger, M., Baumeister, F., Bosch, F., Erwa, W. Biochemical and molecular characterization of 18 patients with pyridoxine-dependent epilepsy and mutations of the antiquitin (ALDH7A1) gene. Hum. Mutat. 28: 19-26, 2007.*
3. *Sadilkova K, Gospe SM Jr, Hahn SH. Simultaneous determination of alpha-aminoadipic semialdehyde, piperideine-6-carboxylate and pipecolic acid by LC-MS/MS for pyridoxine-dependent seizures and folinic acid-responsive seizures. J Neurosci Methods, 2009.*
4. *Mills, P. B., Struys, E., Jakobs, C., Plecko, B., Baxter, P., Baumgartner, M., Willemsen, M. A. A. P., Omran, H., Tacke, U., Uhlenberg, B., Weschke, B., Clayton, P. T. Mutations in antiquitin in individuals with pyridoxine-dependent seizures. Nature Med. 12: 307-309, 2006.*
5. *Clayton, P. T. Pyridoxine-Dependent Epilepsy Due to □-Aminoadipic Semialdehyde Dehydrogenase (Antiquitin) Deficiency. The Online Metabolic & Molecular Bases of Inherited Disease.*

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CREATINE DEFICIENCY SYNDROMES

Creatine deficiency syndromes are characterized by intellectual disability, severe disturbance of expressive and cognitive speech, seizures (often poorly controlled), and movement disturbances[1,2]. A common hallmark is cerebral creatine depletion as detected by magnetic resonance spectroscopy[3].

Creatine deficiency syndromes include two autosomal recessive disorders, arginine:glycine amidinotransferase (AGAT) deficiency (OMIM 612718) and guanidinoacetate methyltransferase (GAMT) deficiency (OMIM 612736), as well as the X-linked creatine transporter deficiency (OMIM 300352).

In individuals with AGAT deficiency, both plasma and urine testing reveal extremely low guanidinoacetate (GAA) and creatine concentrations. In GAMT deficiency, high plasma GAA levels are characteristic. GAA is also mildly increased in urea cycle disorder patients with elevated or supplemented arginine levels. In individuals with X-linked creatine transporter deficiency, urine is required for diagnosis, as plasma levels of GAA and creatine are typically normal in this disorder. The characteristic finding in the urine of individuals with creatine transporter deficiency is an elevated creatine/creatinine ratio[4].

To diagnose all three creatine deficiency syndromes, the Creatine Panel determines GAA and creatine in both plasma and urine by UPLC-tandem mass spectroscopy[5].

Turn Around Time: 10 days

Test Codes: 4015 Creatine Panel or order individually Plasma 4130, Urine 4260

CPT Codes: 82540x1, 82570x2, 82543x4

Related DNA Tests

AGAT Deficiency: GATM Sequence 3455, Known Familial Mutation 3456, Prenatal Diagnosis 3457

GAMT Deficiency: GAMT Sequence 3145, Known Familial Mutation 3146, Prenatal Diagnosis 3147

Creatine Transporter Deficiency: SLC6A8 Sequence 3150, Known Familial Mutation 3151, Prenatal Diagnosis 3152

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4900: Plasma

Requirements: Draw blood in a EDTA (purple top) tube(s) and separate as soon as possible, freezing immediately. Send 1-2 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: 4901: Urine

Requirements: Send 2-4 cc of random urine.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

References:

1. *Schulze, A. Creatine deficiency syndromes. Molec. Cell Biochem. 244: 143-150, 2003.*
2. *Lion-Francois, L., Cheillan, D., Pitelet, G., Acquaviva-Bourdain, C., Bussy, G., Cotton, F., Guibaud, L., Gerard, D., Rivier, C., Vianey-Saban, C., Jakobs, C., Salomons, G. S., des Portes, V. High frequency of creatine deficiency syndromes in patients with unexplained mental retardation. Neurology 67: 1713-1714, 2006.*
3. *Stöckler-Ipsiroglu S, Salomons GS (2006) Creatine deficiency syndromes In: Fernandes J, Saudubray JM, van den Berghe G, eds. Inborn Metabolic Diseases. Springer Verlag; 2006:211-7*
4. *Sylvia Stöckler-Ipsiroglu, Carmen Stromberger, Chike B. Item, Adolf Mühl Stöckler-Ipsiroglu S, Battini R, de Grauw T, Schulze A. Disorders of creatine metabolism. In: Blau N, DuranM, Blaskovics ME, Gibson KM eds. Physician's Guide to the Laboratory Diagnosis of Metabolic Diseases. Springer Verlag; :467-80, 2003.*
5. *Sun, Q and O'brien, W. Diagnosis of creatine metabolism disorders by determining creatine and guanidinoacetate in plasma and urine. Methods Mol Biol. 603:175-85, 2010.*

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PURINE AND PYRIMIDINE METABOLISM DISORDERS

Purine Metabolism Disorders

Deficiencies in the enzymes involved in purine metabolism lead to nonspecific, mostly neurologic, symptoms, e.g., intellectual disability, seizures, muscular hypotonia, or urinary tract calculi. Several inborn errors of metabolism result in the accumulation of elevated quantities of various purine bases[1] and purine nucleosides in the urine or cerebrospinal fluid. These include a wide range of disorders from primary immune deficiency, such as adenosine deaminase deficiency and purine nucleotide deficiency, to disorders leading to severe intellectual disability, like Lesch-Nyhan disease, adenylosuccinase deficiency and molybdenum cofactor deficiency.

Adenosine deaminase deficiency and purine nucleoside phosphorylase deficiency are autosomal recessive disorders that are the basis of severe combined immunodeficiency or SCID. Symptoms of SCID include pneumonia, chronic diarrhea, growth retardation and neurological problems such as developmental delay and motor function disorders.

Lesch-Nyhan disease (LNS) is an X-linked recessive disorder caused by the deficiency of enzyme hypoxanthine-guanine phosphori-bosyltransferase (HGPRT). Three characteristic symptoms of LNS are hyperuricemia, neurological defects and cognitive and behavioral abnormalities, such as self-mutilation [2].

Pyrimidine Metabolism Disorders

Defects in the breakdown of pyrimidines are known to cause multiple autosomal recessive inborn errors of metabolism, including dihydropyrimidine dehydrogenase (DPD) deficiency, dihydropyrimidinase (DHP) deficiency, and mitochondrial encephalopathy with gastro-intestinal symptoms (MNGIE).

DPD and DHP deficiencies are disorders sharing similar clinical features that can range from asymptomatic to intellectual disability, microcephaly, epilepsy, and failure to thrive[2]. In addition, DPD deficiency has been linked to 50-75% of severe toxicities induced by the chemotherapeutic agent 5-fluorouracil (5-FU), leading some to recommend DPD testing prior to 5-FU administration[3]. DPD and DHP enzymes work sequentially in the breakdown of uracil/thymine to dihydrouracil/dihydrothymine and then to β-ureidopropionate/ β-aminoisobutyrate[4].

MNGIE is a mitochondrial disorder resulting from defects in thymidine phosphorylase. Patients with MNGIE typically present with cachexia within the first two decades of life; other clinical features include intestinal pseudo-obstruction, myopathy, and neuropathy[5,6].

Disease	Metabolites
Adenosine Deaminase Deficiency	Deoxyadenosine, Adenosine[1]
Purine Nucleoside Phosphorylase Deficiency	Inosine, Deoxyinosine, Guanosine, Deoxyguanosine
Molybdenum Cofactor Disease & Xanthinuria	Xanthine, Hypoxanthine
Adenylosuccinase Deficiency	Succinyladenosine
Lesch-Nyhan Disease	Adenine, Hypoxanthine, Sulfocysteine
APRT Deficiency	Adenine, 8-hydroxyadenine, 2,8-dihydroxyadenine
PRPP Synthetase Deficiency	Adenine, Hypoxanthine
Dihydropyrimidine Dehydrogenase (DPD) deficiency	Uracil and Thymine
Dihydropyrimidinase (DHP) Deficiency	Dihydrouracil, Dihydrothymine, Uracil and Thymine
MNGIE	Thymidine

Purine and Pyrimidine Metabolism Disorders

Turn around time: 7 days

Test code: Purine Panel 4220
Pyrimidine Panel 4215
Purine & Pyrimidine Panel 4010

Related tests: Plasma Thymidine 4330
Urine Sulfocysteine 4225

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4901: Urine

Requirements: Send 2-4 cc of random urine.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

References:

1. *Ito, T, van Kuilenburg, ABP, Bootsma, AH, Haasnoot, AJ, van Cruchten, A, Wada, Y, van Gennip, AH. Rapid screening of high-risk patients for disorders of purine and pyrimidine metabolism using HPLC-electrospray tandem mass spectrometry of liquid urine or urine-soaked filter paper strips.Clinical Chemistry. 46:445-452, 2000.*
2. *Nyhan WL. Disorders of purine and pyrimidine metabolism. Molecular Genetics and Metabolism. 86:25-33, 2005.*
3. *Ciccolini J, Gross E, Dahan L, Lacarelle B, Mercier C. Routine dihydropyrimidine dehydrogenase testing for anticipating 5-fluorouracil-related severe toxicities: hype or hope? Clinical Colorectal Cancer. 9:224-8, 2010.*
4. *Jiang H, Jiang J, Hu P, Hu Y. Measurement of endogenous uracil and dihydrouracil in plasma and urine of normal subjects by liquid chromatography-tandem mass spectrometry. Journal of Chromatography B, Analytical Technologies in the Biomedical and Life Sciences. 769:169-76, 2002.*
5. *Garone C, Tadesse S, Hirano M. Clinical and genetic spectrum of mitochondrial neurogastrointestinal encephalomyopathy. Brain: A Journal of Neurology. 134:3326-32, 2011.*
6. *Marti R, Lopez LC, Hirano M. Assessment of thymidine phosphorylase function: Measurement of plasma thymidine (and deoxyuridine) and thymidine phosphorylase activity. Methods Molecular Biology. 837:121-33, 2012.*

GLOBAL METABOLOMIC ASSISTED PATHWAY SCREEN (GLOBAL MAPS)SM

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Metabolomic profiling is a newly-developed, large scale, semi-quantitative screening test that looks at perturbations in both individual analytes and pathways related to biochemical abnormalities, including but not limited to amino acid, organic acid, lipid, and nucleotide metabolism. This is a small molecule screen for compounds 50-1500 Da, intended as a screening tool for individuals who have an undifferentiated phenotype or as supportive evidence in individuals with equivocal mutations in genes related to metabolic processes. Any abnormalities detected on the Metabolomic Profile should be confirmed by diagnostic biochemical or molecular diagnostic testing. This test must be ordered by a physician and must be accompanied by detailed clinical information for the most informative interpretation.

Patient should NOT be on TPN, special diet, dietary supplements, or drug therapies for most informative results.

This test is not intended to screen for lysosomal storage disorders, congenital disorders of glycosylation, mucopolysaccharidoses, mucolipidoses, or other similar large molecule disorders.

Indications for Testing:

This screening test may be considered for individuals with

- Non-syndromic intellectual disability
- Seizures
- Failure to thrive
- Hypoglycemia
- Recurrent vomiting
- Autism spectrum disorder
- Speech/language delay
- Equivocal molecular test results in a gene known to be involved in small molecule metabolism

Use of this test in combination with exome analysis may be most beneficial in more difficult cases or in cases where a small molecule inborn error of metabolism is suspected.

While this test may also enable the identification of new biomarkers associated with specific genetic conditions, as well as biomarkers associated with medications, disease status, and diet-related treatment, we will not report xenobiotics, plant biproducts, or microbial findings

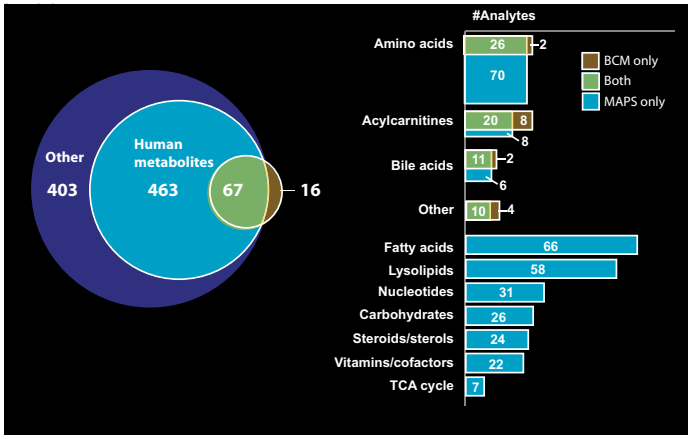

Figure 1. *MAPS analysis is a comprehensive screen, replacing many single tests. The average findings from a single MAPS analysis of plasma are compared to the findings from all plasma tests offered by the Baylor Miraca Genetics Laboratories clinical biochemical genetics laboratory (BCM).*

Technical Information - Methodology: The Global MAPS test will identify known metabolites in plasma, or cerebrospinal fluid. The test will analyze hundreds of compounds simultaneously using a combination of LC-MS/MS and GC-MS technologies (>900 compounds are regularly detected in human plasma).

Significant abnormalities in metabolic pathways and/or in selected single analytes may be identified. In these situations, we will recommend targeted follow-up testing (molecular and/or biochemical) for confirmation.

Test Type: Analyte Analysis

References Values or Ranges: Metabolites and biomarkers that fall out of the normal range will be reported relative to the general population. Individual concentrations will not be reported.

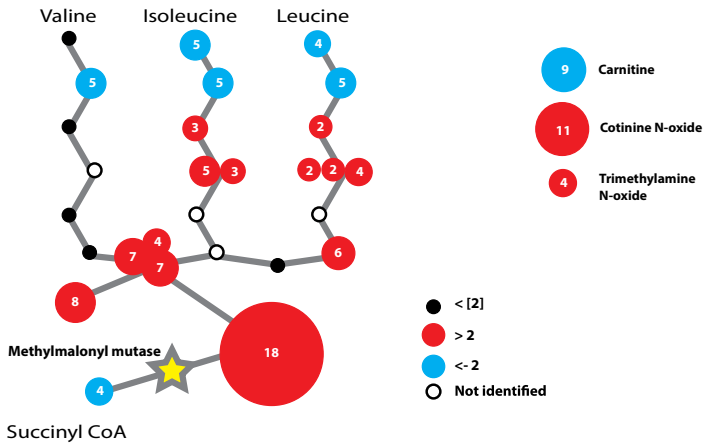

Figure 2. *Global MAPS data for a patient receiving treatment for methylmalonyl mutase deficiency. Each node represents a step in the branched chain amino acid degradation pathway.*

SAMPLE & SHIPPING INFORMATION

Specimen Type: 4900: Plasma

Requirements: Draw blood in a EDTA (purple top) tube(s) and separate as soon as possible, freezing immediately. Send 1-2 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: 4902: Cerebrospinal fluid

Requirements: Send 1-2 cc of cerebrospinal fluid.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Turn Around Time: 21 days

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

BILLING INFORMATION

List Price: For insurance or institutional prices, please call 713-798-3566.

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AVAILABLE BIOCHEMICAL TESTS	ANALYTE ANALYSIS		ENZYME ASSAY		DNA ANALYSIS
Disease Name	Test Code	Specimen Type	Test Code	Specimen Type	Test Code Analysis
3-Methylcrotonyl-CoA Carboxylase Deficiency	4200	Urine			3635, 3638, 3639, 3640, 3643, 3644
3-Methylglutaconic Aciduria Type I	4200	Urine			3913, 3913, 3910
5-Oxoprolinuria	4200	Urine			1300
Acid Sphingomyelinase Deficiency | Niemann-Pick Disease Type A, B			4607, 4608	SFC/WBC	6942, 6944, 6047 | MA
Adenine Phosphoribosyltransferase Deficiency	4220	Urine			2825 | APRT SEQ
Adenosine Deaminase Deficiency	4220	Urine	4509	RBC	5010 | ADA SEQ
Adenylosuccinase Deficiency | ADSL	4220	Urine			3695 | ADSL SEQ
Alkaptonuria	4200	Urine			1300
Arginase Deficiency | Argininemia	4100	Plasma	4536	RBC	3425 | ARG1 SEQ
Arginine: Glycine Amidinotransferase (AGAT) Deficiency	4260, 4130	Urine /Plasma			3455 | GATM SEQ
Argininosuccinic Aciduria | Argininosuccinate Lyase Deficiency	4100	Plasma	4524	RBC	6360 | ASL SEQ
Arylsulfatase A Deficiency | Metachromatic Leukodystrophy			4537, 4538	WBC	6380 | ARSA SEQ
Aspartylglycosaminuria | Aspartylglucosaminuria	4300	Plasma	4514	SFC	2205 | AGA SEQ
Biotinidase Deficiency	4200, 4350	Urine	4555	Serum	3495 | BTD SEQ
Canavan Disease	4200	Urine			6942, 6944, 6070 | MA
Carbamoylphosphate Synthetase Deficiency | CPSI	4100	Plasma	4561	Liver	2085, 3345 | CPS1 SEQ
Carnitine Deficiency, Systemic | OCTN2	4300	Plasma			3360 | SLC22A5 SEQ
Carnitine Palmitoyltransferase I Deficiency | CPTI	4300	Plasma			3365 | CPT1A SEQ
Carnitine Palmitoyltransferase II Deficiency | CPTII	4300	Plasma			3160 | CPT2 SEQ
Carnitine-Acylcarnitine Translocase Deficiency	4300	Plasma			3435 | SLC25A20 SEQ
Cobalamin Disorders	4150, 4140	Plasma			2120 PANEL
Transcobalamin II Deficiency | TCN2	4150, 4140	Plasma			2120, 3695
Transcobalamin Binding Protein Deficiency | CD320	4150, 4140	Plasma			3965
Cobalamin a | MMAA	4150, 4140	Plasma			2120, 3575
Cobalamin b | MMAB	4150, 4140	Plasma			2120, 3580
Cobalamin c | MMACHC	4150, 4140	Plasma			2120, 3440
Cobalamin d | MMADHC	4150, 4140	Plasma			2120, 3885
Cobalamin e | MTRR	4150, 4140	Plasma			2120, 2565
Cobalamin f | LMBRD1 | Lysosomal B12 Transporter	4150, 4140	Plasma			2120, 2560
Cobalamin g | MTR | Methionine Synthase	4150, 4140	Plasma			2124
Cobalamin j | ABCD4	4150, 4140	Plasma			5134
Methylmalonyl-CoA Mutase Deficiency | MUT	4150, 4140	Plasma			2120, 3585
Citrin Deficiency	4100	Plasma			3155 | SLC25A13 SEQ
Citrullinemia Type I	4100	Plasma	4545	SFC	6180 | ASS1 SEQ
Coenzyme Q10 Deficiency	4800	Muscle			3850, 3415, 3775, 3405, 3410 | CABC1, COQ2, COQ9, PDSS1, PDSS2 SEQ
Creatine Transporter Deficiency | CRTR	4260	Urine			3150 | SLC6A8 SEQ
Cystinosis	4627	WBC			1300

AVAILABLE BIOCHEMICAL TESTS	ANALYTE ANALYSIS		ENZYME ASSAY		DNA ANALYSIS
Disease Name	Test Code	Specimen Type	Test Code	Specimen Type	Test Code Analysis
Dihydropyrimidinase (DHP) deficiency	4215	Urine			1300
Dihydropyrimidine Dehydrogenase (DPD) deficiency	4215	Urine			1300
Fabry Disease			4517	WBC	6063 | GLA SEQ
Fatty Acid Oxidation Disorders | FODs	4300	Plasma			2095 | 2300 | 2000
Fumarate Hydratase Deficiency | Fumaric Aciduria	4200	Urine			3740 | FH SEQ
Gaucher Disease			4554	WBC	6942, 6944, 6033 | MA
Glutaric Aciduria Type I	4200	Urine			3685
Glutaric Aciduria Type II | Multiple Acyl-CoA Dehydrogenase Deficiency	4300	Plasma			2349
Glycerol Kinase Deficiency | GK	4200	Urine			8466
Glycine Encephalopathy *	4100, 4160*	Plasma/CSF*			5034
GM1 Gangliosidoses			4548, 4549	SFC/WBC	1300
Guanidinoacetate Methyltransferase (GAMT) Deficiency	4260, 4130	Urine /Plasma			3145 | GAMT SEQ
Hawkinsinuria	4200, 4100	Urine/Plasma			2075
Hexosaminidase A Deficiency | Tay-Sachs Disease | Sandhoff Disease			4569	Serum	6942, 6944, 6066 | MA
HMGCoA Lyase Deficiency	4300, 4200	Urine /Plasma			5064 | 5060
Holocarboxylase Synthetase Deficiency	4350, 4200, 4300	Urine/Plasma			3540 | HLCS SEQ
Homocystinuria caused by Cystathionine Beta-Synthase Deficiency	4140, 4100	Plasma			3970
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome | HHH	4100, 4240	Urine/Plasma			3235 | SLC25A15 SEQ
Hyperprolinemia Type II	4100, 4240	Urine /Plasma			5135 | 5139
Hypophosphatasia	4240	Urine			2250 | ALPL SEQ
Ichthyosis X-linked | Steroid Sulfatase Deficiency			4614, 4615	SFC/WBC	8485 | FISH
Isovaleric Acidemia	4200	Urine			3680 | IVD SEQ
Krabbe Disease			4565, 4566	SFC/WBC	6415 | GALC SEQ
Lesch Nyhan Syndrome	4220	Urine	4572, 4573	SFC/RBC	6240 | HPRT1 SEQ
Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency | LCHAD	4300	Plasma			3120, 3122 | HADHA SEQ, MA
Long Chain Acyl-CoA Dehydrogenase Deficiency | LCAD	4300	Plasma			3385 | ACADL SEQ
Lowe Syndrome			4585	SFC	6039 | OCRL SEQ
Lysinuric Protein Intolerance	4100, 4240	Urine /Plasma			1300
Malonyl-CoA Decarboxylase Deficiency	4200	Urine			1300
Maple Syrup Urine Disease	4100	Plasma			3460, 3865 | DLD, DBT SEQ
Medium Chain Acyl-CoA Dehydrogenase Deficiency | MCAD	4300, 4350	Urine /Plasma			3115, 3117 | ACADM SEQ, MA
Methylmalonic Acidemia | MMA	4150, 4300	Urine/Plasma			3575, 3580, 3585, 3885 | MMAA, MMAB, MUT, MMADHC SEQ
Methylmalonic Acidemia and Homocystinuria, cblC type	4150, 4140	Plasma			3440 | MMACHC SEQ
Mitochondrial Neurogastrointestinal Encephalopathy Disease | MNGIE	4330, 4215	Urine/Plasma			3060 | TYMP SEQ
Molybdenum Cofactor Deficiency	4220, 4225	Urine			MOCSI-3595 SEQ | MOCS2-3615 SEQ
Mucolipidosis I | Sialidosis			4603	SFC	1300

Biochemical Tests

AVAILABLE BIOCHEMICAL TESTS	ANALYTE ANALYSIS		ENZYME ASSAY		DNA ANALYSIS
Disease Name	Test Code	Specimen Type	Test Code	Specimen Type	Test Code Analysis
Mucolipidosis II | I-cell Disease			4569	Serum	6925
Mucopolysaccharidosis Type I | Hurler Disease | MPS I			4575, 4576	SFC/WBC	6385 | IDUA SEQ
Multiple Acyl-CoA Dehydrogenase Deficiency	4300, 4200	Urine /Plasma			3840, 3855, 3860 | ETFDH, ETFA, ETFB SEQ
Ornithine Aminotransferase Deficiency | Gyrate Atrophy	4100	Plasma			5280
Ornithine Transcarbamylase Deficiency | OTC	4210, 4100	Urine /Plasma	4582	Liver	3140 | OTC SEQ
Orotic Aciduria	4210	Urine			1300
Phenylalanine Hydroxylase Deficiency | Phenylketonuria | PKU	4100, 4120	Plasma/Blood Spot			3135 | PAH SEQ
Propionic Acidemia	4200, 4300, 4350	Urine/Plasma			3765, 3770 | PCCA, PCCB SEQ
Purine Nucleoside Phosphorylase Deficiency | PNP	4220	Urine	4592, 4593, 4594	SFC/RBC/ WBC	5025 | PNP SEQ
Pyridoxine-Dependent Seizures	4844, 4812	Plasma/CSF			6950 | ALDH7A1 SEQ
Pyruvate Dehydrogenase Deficiency | PDHA1	4200	Urine			3165 | PDHA1 SEQ
Ribose 5-Phosphate Isomerase Deficiency	4340	Urine			
Sandhoff Disease | Tay-Sachs Disease			4569/4620	Serum/WBC	HEXA SEQ 6925
Succinic Semialdehyde Dehydrogenase Deficiency | SSADH	4200	Urine			
Sulfocysteinuria	4225	Urine			
Transaldolase Deficiency	4340	Urine			
Trifunctional Protein Deficiency | TFP	4300	Plasma			3120, 3630 | HADHA, HADHB SEQ
Tyrosinemia Types I, II, and III	4250	Urine			3445 | FAH SEQ
Very Long Chain Acly-CoA Dehydrogenase Deficiency | VLCAD	4300	Plasma			3355 | ACADVL SEQ
Vitamin D Deficiency	4360	Plasma			6565
Wolman Disease | Lysosomal Acid Lipase Deficiency | Cholesterol Ester Storage Disease			4502, 4503, 4504	SFC/WBC/ Liver	6430 | LIPA SEQ
Xanthinuria, Type I	4220	Urine			5025
** Must send both plasma & CSF samples for clinical diagnosis.*	*RBC=Red Blood Cell WBC=White Blood Cell; SFC=Skin Fibroblast Culture CSF=Cerebrospinal Fluid*				*SEQ=Sequencing MA=Mutation Analysis*

SAMPLE & SHIPPING INFORMATION

Specimen Type: Plasma
Requirements: Draw blood in heparin (green top) tube(s) and separate as soon as possible, freezing immediately. Send 2-3 cc of plasma. Store the specimen frozen at -20C. Specimen may be stored frozen for up to 7 days.

Specimen Type: Serum
Requirements: Draw blood in a No Additive (red-top) or Serum Gel (red/gray-top) tube(s) and separate as soon as possible. Send 1-2 cc of serum.

Specimen Type: Urine
Requirements: Send 2-4 cc of random urine.

Specimen Type: Cerebrospinal fluid
Requirements: Send 1-2 cc of cerebrospinal fluid.

Shipping Conditions: Ship frozen on 3-5 lbs dry ice in an insulated container by overnight courier.

Shipping Address: 2450 Holcombe, Grand Blvd. - Receiving Dock, Houston, Texas 77021-2024

Baylor Miraca
Genetics Laboratories

2450 Holcombe Boulevard, Suite O-100, Houston, Texas 77021 • 1-800-411-GENE (4363) or 713-798-6555 • www.BMGL.com

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