The Power of Knowing

BGI NOVA™ Metabolic Disease Genetic Test

About

BGI is proud to offer the BGI NOVA™ Metabolic Disease Genetic Test, a comprehensive screening test for inherited metabolic disorders (IMDs), which utilizes tandem mass technology to allow inexpensive screening for **48 different metabolic disorders**.

Many of these potentially fatal metabolic disorders are amenable to effective treatment upon timely diagnosis. Early detection and intervention can allow better clinical outcomes for the newborn, improving the quality of life for both them and their family.

Who is the BGI NOVA™ Metabolic Disease Genetic Test suitable for?

Any newborn suspected of a metabolic abnormality.

Sample Requirements

SAMPLE TYPE	REQUIREMENTS	SAMPLE SHIPMENT & STORAGE
Blood spot (heel)	Fed more than 6 times before blood draw	Shipped within one week under room temperature

Methodology

Tandem Mass Spectrometry & Next Generation Sequencing.

BGI provides newborn testing for inherited metabolic disorders based on tandem mass spectrometry coupled with liquid chromatography (LC-MS/MS). Testing can be performed from a dried blood spot sample, which can be safely taken from the newborn via a simple sampling procedure such as a heel prick.

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Conditions Screened

METABOLIC DISEASE CATEGORY	NO.	DISEASE NAME
Amino Acid Metabolism Disorders (21)	1	Maple Syrup Urine Disease
	2	Phenylketonuria
	3	Tetrahydrobiopterin Deficiency
	4	Tyrosinemia I
	5	Tyrosinemia II
	6	Tyrosinemia III
	7	Citrullinemia I
	8	Citrullinemia II
	9	Argininemia
	10	Ornithine Transcarbamylase Defeciency
	11	Hyperornithinemia with Gyralatrophy
	12	Carbamyl Phosphate Synthase Deficiency
	13	N-Acetyglutamate Synthase Deficiency
	14	Argininosuccinic Aciduria
	15	Homocysteinemia
	16	Hypermethioninemia Hyperornithinemia Homocitrullinuria Syndrome
	17	Hyperammonemia
	18	Hyperornithinemia
	19	Non Ketotic Hyperglycinemia
	20	Histidinemia
	21	Hypervlinemia
Organic Acid Metabolic Disorders (12)	22	Methyl Malonic Acidemia
	23	Propionic Acidemia
	24	2-Methylbutyryl CoA Dehydrogenase Deficiency
	25	Isovaleric Acidemia
	26	3-Methyl Crotonyl CoA Carboxylase Deficiency
	27	3-Methyl Glutaconic Aciduria
	28	2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency
	29	Multiple Carboxylase Deficiency
	30	3-Hydroxy-3-Methylglutayl-coenzyme A Lyase Deficiency
	31	Beta-Ketothiolase Deficiency
	32	Isobutyryl-CoA Dehydrogenase Deficiency
	33	Glutaric acidemia Type I
Fatty Acid Oxidation Metabolic Disorders (15)	34	Carnitine Palmitoyl Transferase Deficiency Type I
	35	Carnitine Palmitoyl Transferase Deficiency Type II
	36	Carnitine Uptake Defect
	37	Short Chain Acyl Coa Dehydrogenase Deficiency
	38	Medium Chain Acyl Coa Dehydrogenase Deficiency
	39	Very Long Chain Acyl Coa Dehydrogenase Deficiency
	40	Medium/Short Chain Hydroxy Acyl Coa Dehydrogenase Deficiency
	41	3 Hydroxy Long Chain Acyl Coa Dehydrogenase Deficiency
	42	Malonyl-CoA Decarboxylase Deficiency
	43	Ethylmalonic Encephalopathy
	44	Carnitine/Acylcarnitine Translocase Deficiency
	45	Trifunctional Protein Deficiency
	46	Glutaric Acidemia Type II
	47	Medium Chain 3- Ketoacyl CoA Thiolase Deficiency
	48	2,4- Dienyl-CoA Reductase Deficiency

Workflow

1. 

Conduct pre-test genetic counseling with patient and sign consent form

2. 

Take sample from patient and send it to BGI

3. 

Sequencing takes place at BGI laboratory

4. 

Receive test results 15 working days later

5. 

Conduct post-test genetic counseling with patient

Contact your local BGI representative for more information or email **info@bgi-international.com**. More information can also be found on our website. **www.bgi.com/global/**

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Testing services not currently available in the United States of America. Please contact a representative for regional availability.