BGI NOVA™ Metabolic Disease Testing

Introduction

BGI is proud to offer the BGI NOVA™ Metabolic Disease Testing, 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 Testing suitable for?

- Any newborn suspected of a metabolic abnormality.

Sample Requirements

Sample Type	Requirements	Sample Shipment & Storage
Dried 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.

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 Deficiency
	11	Hyperornithinemia with Gyral Atrophy
	12	Carbamoyl Phosphate Synthetase Deficiency
	13	N-Acetyglutamate Synthase Deficiency
	14	Argininosuccinic Aciduria
	15	Homocysteinemia
	16	Hypermethioninemia Hyperornithinemia Homocitrullinuria Syndrome
	17	Hyperammonemia
	18	Hyperornithinemia
	19	Nonketotic Hyperglycinemia
	20	Histidinemia
	21	Hypervalinemia
Organic Acid Metabolic Disorders (12)	22	Methylmalonic Acidemia
	23	Propionic Acidemia
	24	2-Methylbutyryl-CoA Dehydrogenase Deficiency
	25	Isovaleric Acidemia
	26	3-Methylcrotonyl-CoA Carboxylase Deficiency
	27	3-Methylglutaconic Aciduria
	28	2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency
	29	Multiple Carboxylase Deficiency
	30	3-Hydroxy-3-Methylglutacyl-CoA Lyase Deficiency
	31	Beta-Ketothiolase Deficiency
	32	Isobutyryl-CoA Dehydrogenase Deficiency
	33	Glutaric Acidemia Type I
Fatty Acid Oxidation Metabolic Disorders (15)	34	Carnitine Palmitoyltransferase Deficiency Type I
	35	Carnitine Palmitoyltransferase 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 Hydroxyacyl-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- Dienoyl-CoA Reductase Deficiency

Workflow

1. Conduct pre-test genetic counseling with patient and sign a consent form
2. Take a sample from the patient and send it to BGI
3. Sequencing takes place at BGI laboratory
4. Receive test results 5 working days later
5. Conduct post-test genetic counseling with the patient

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Published October 2022.

Unless otherwise informed, all sequencers and sequencing reagents are not available in Germany, USA, Spain, UK, Hong Kong, Sweden, Belgium or Italy.
Certain sequencing services are not available in USA and Hong Kong. Please contact a representative for regional availability. The company reserves the right of final interpretation.

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