



About

BGI is proud to offer the BGI NOVA™ Newborn Genetic Test, a comprehensive screening test that determines a baby's risk for **87 inherited disorders**, as well as providing personalized genetic information on the likely response of **32 pediatric drugs**. Utilizing Next Generation Sequencing technology and with access to BGI's industry leading genetics bioinformatics software, the BGI NOVA™ Newborn Genetic Test offers the most comprehensive and accurate newborn screening test on the market with a positive predictive value (PPV) of >99%.

The Power of Knowing

BGI NOVA™ Newborn Genetic Test

Who should consider the BGI NOVA™ Newborn Genetic Test?

- Parents who want a comprehensive genetic screen for their baby
- Parents who would like to learn their baby's drug-related genetic status
- Babies who have missed out on regular screening
- Babies from parents with a family history of inherited disorders or from a population identified as at higher risk for genetic disease

Testing is not suitable for:

- Definitive diagnosis of a disorder
- Newborns with numerical or structural changes of the chromosome, copy number variations and/or germ cell mosaicism
- Newborns who have received blood transfusions, organ transplants or stem cell therapy

Highlights

- Unique target sequencing
- Library with trace DNA: can obtain >100ng DNA from samples and build a library based on just 50ng DNA (1ug=1000ng is the minimal requirement for standard library)
- A comprehensive database comprising 12,000 mutations of all listed genes is used to generate report automatically.

Conditions Screened

CONDITION CATEGORY	87 INHERITED DISORDERS
Amino Acid Metabolism Disorders	20
Fatty Acid Oxidation Disorders	16
Organic Acid Disorders	13
Carbohydrate Disorders	3
Creatine Metabolism Disorders	1
Lysosomal Storage Disease	10
Copper Metabolism Disorder	2
Primary Immunological Deficiency	14
Blood Lipid Disorders	3
Miscellaneous Genetic Conditions	5

DRUG CATEGORY	32 PAEDIATRIC PHARMACOGENOMICS
Neurology Drugs	11
Anti-infection Agents	10
Cancer Drugs	2
CRheumatism Drugs	1
Cardiovascular Drugs	3
Gastroenterology Drugs	3
Others	2

Sample Requirements

SAMPLE TYPE	SAMPLE SHIPMENT & STORAGE
Saliva (oral swab)	Shipped within one week under room temperature
Blood spot (cord blood or peripheral blood)	Shipped within one week under room temperature

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/

Workflow

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Conduct pre-test genetic counseling with patient and sign consent form
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Take sample from patient and send it to BGI
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Sequencing takes place at BGI laboratory
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Receive test results 15 working days later
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Conduct post-test genetic counseling with patient