GeneMind

GeneMind Expanded Carrier Screening(ESC) Solution

Explore Life's Mysteries for Better Healthcare

Highlight:

Screen for a broad range of inherited disorders

A 216-gene panel targeting the entire coding region enables the analysis of 191 genetic diseases, along with the detection of the MTHFR gene 677 locus related to folic acid metabolism

Consolidate stand-alone assays to improve lab efficiency

NGS enables simultaneous screening of hundreds of genes in one experiment, providing information on a broader range of targets with a faster turn-around time than traditional techniques such as PCR or Sanger sequencing.

A complete end-to-end solution from a single vendor

GeneMind could provide the “extraction kits -library preparation kits -flow cell -sequencing kits -analysis software” streamlined workflow and help to ensure obtaining reliable results

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Background:

Carrier screening is a type of genetic sequencing that can tell you whether you carry a gene for certain genetic disorders. When it is done before or during pregnancy, it allows you to find out your chances of having a child with a genetic disorder. Currently, there are over 9,000 single-gene genetic diseases, mostly fatal, causing malformations or disabilities. Only 5% of them have effective treatments, and these treatments are often prohibitively expensive for ordinary families. Nowadays, an increasing number of experts suggest that carrier screening for single-gene genetic diseases is of great importance and necessary before people get marriage. NGS enables rapid carrier screening research across the broadest range of disorders, with a scalable, cost-effective solution. ESC is a genetic screening method for would-be and soon-to-be parents that can detect whether the parents unknowingly carry genetic conditions that may pass to their children.

Research shows that, on average, each person carries 2.8 pathogenic genes for recessive genetic diseases, and 84% of people carry at least one pathogenic mutation gene. Carriers generally do not exhibit symptoms, but the greatest risk of recessive inheritance arises if both partners carry the same pathogenic gene, potentially resulting in an affected child.

GeneMind ESC solution is based on the latest Carrier Screening Guidelines from the American College of Medical Genetics and Genomics (ACMG). It screens for over 200 single gene diseases, including autosomal recessive genetic diseases and X-linked recessive genetic diseases. It helps couples of childbearing age screen for carrier status of pathogenic variations in their own single-gene recessive genetic diseases, allowing them to know the potential reproductive risks, effectively prevent birth defects and avoid the tragedy of having an affected child. The program also includes screening for the MTHFR 677 locus related to folate metabolism, providing comprehensive, accurate, and efficient one-stop genetic testing services for couples preparing for pregnancy.

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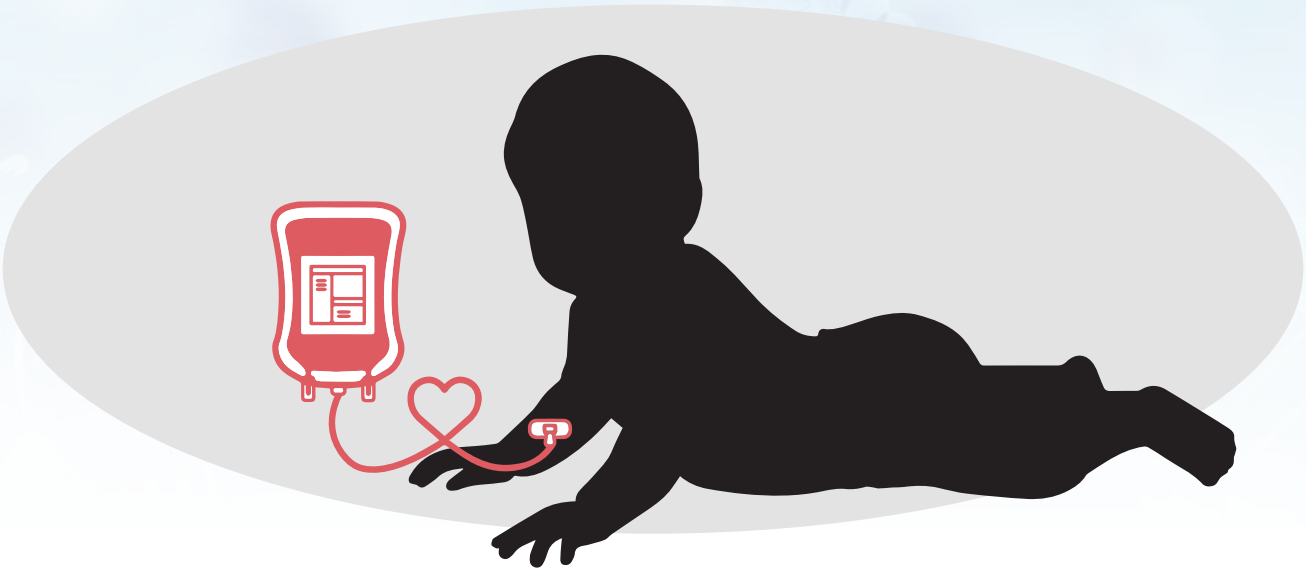
Product Introduction:

✧ GeneMind expanded carrier screening panel covering 191 genetic diseases within 216 genes

System	Targeted Diseases	No. of Targeted Genes
Metabolic and endocrine systems	(82) Phenylketonuria, tyrosinemia type I, maple diabetes mellitus, methylmalonic acidemia,glutaracidemia type I, Glycogen accumulation disease, galactosemia, mucopolysaccharidosis,Niemann-Pick disease, etc	96
Nervous system	(35) Progressive pseudohypertrophic muscular dystrophy, hepatolenticular degeneration, Joubert syndrome, etc	37
Breathing, sight, hearing	(13) Deafness, retinitis pigmentosa, cystic fibrosis, etc	13
Blood and immunity	(11) Thalassemia, hemophilia, Fanconi anemia, etc	13
Skin	(10) Oculocutaneous albinism, epidermolysis bullosa, xeroderma pigmentosum, ichthyosis, etc	17
Skeleton	(9) Craniofacial skeletal syndrome, short rib thoracic dysplasia type 3 with or without polydactyly and epiphyseal dysplasia Good, achondroplasia, etc	6
Digestive and urinary systems	(5) Progressive familial intrahepatic cholestasis type 2, polycystic kidney, etcepiphyseal dysplasia Good, achondroplasia, etc	6
Multi-system	(25) Brain-eye-facial-skeletal syndrome, Meckel syndrome, and kyphotic Ehlers-Danlos synthesis Type 1, etcGood, achondroplasia, etc	20
Sum	**191**	**216**

✧ GeneMind expanded carrier screening panel covering 191 genetic diseases within 216 genes

The MTHF gene, which encodes methylene tetrahydrofolate reductase (MTHFR enzyme), is a key enzyme in the folate-methionine metabolic pathway.The mutation of MTHFR gene could cause the enzyme changes in folate activity and folate metabolism disorders, leading to a variety of diseases, among which neonatal defects and hyperhomocysteinemia caused by stroke is the most serious.



The latest study showed that the mutation at site 677 is the key factor leading to the decrease of MTHFR enzyme activity and thermal stability. So MTHFR gene 677 locus detection can be useful to screening the high risk population of folic acid deficiency, set personalized folic acid supplementation program, reduce birth defects and avoid the side effects of folic acid overdose.

Parameter		Carrier Screen
Sample Volume		2 mL of a single tube of blood/ 3ml mouth swabs
Library preparation		PCR
Method		Capture NGS
No. of samples per run(1 PC +1 NC included)	GenoLab M*	1 FCM :37 2 FCM/1 FCH :75 1 FCM+1 FCH: 112 2 FCH: 150
	FASTASeq 300*	1 FCH : 37 1 FCM : 15
Read length		PE150
Average raw data/ sample		⩾2G
TAT		GenoLab M*: 6-10 d FASTASeq 300*: 3-7 d
Report generation		Local analysis and report system

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Clinical Cases:

Background: Hepatolenticular degeneration (OM # 277900) is an autosomal recessive genetic disease. The mutation of ATP7B gene on chromosome 13 leads to the disturbance of copper ion transport and excretion in the body, and copper accumulates in the liver, nervous system, cornea, kidney and other organs, showing a series of clinical manifestations.

Tester information: Couples(female, 28 years old; male:30 years old), both denial of a history of major illness

Testing method: Expanded carrier screening is performed by hybridization capture targeted enrichment and sequencing

Sex	Gene Name	Hereditary Mode	RNA	Variation information	zygotic type	Pathogenicity
Female	ATP7B	AR	NM_001042351	c.2333G > T:p.R778L	Heterozygosis	Yes
Male	ATP7B	AR	NM_001042351	c.2975C > T:p.P992L	Heterozygosis	Yes

Suggestion: The tested couples are carriers of ATP7B gene locus, the disease is autosomal recessive, and the probability of the offspring developing HLS disease is about 25%, it is recommended to carry out related genetic counseling, and if the offspring are born, it is recommended to carry out prenatal diagnosis or preimplantation diagnosis.

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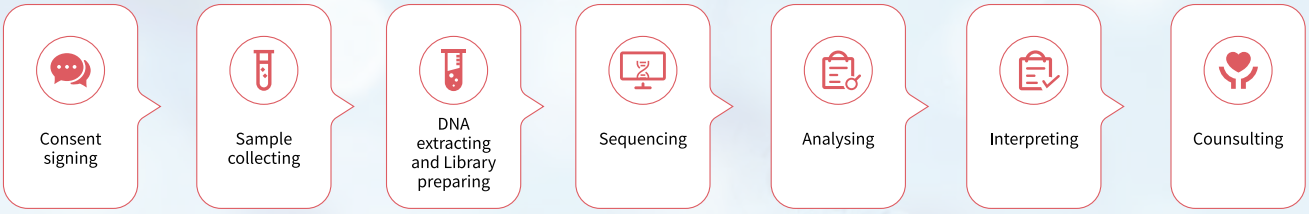
Testing Workflow:



Targeted customer

Carrier screening is recommended for any expectant or early trimester couples (before 16 weeks of pregnancy), including:

- ☑ All couples of childbearing age with normal phenotype and no family history of genetic disease
- ☑ All couples who want to have a healthy baby through assisted reproductive technology
- ☑ Couples who have consanguineous relations

Screening model:

- ☑ Simultaneous screening：The couple are screened at the same time
- ☑ Sequential screening：One spouse gets screened first and the other spouse should be screened based on the former screening results

Sample Requirements:

Sample Type	Requirements	Storage condition	transport
Blood	⩾3 mL	2-8 °C for one week -20°C for one year	Cold-chain transportation
Swab	6	Room temperature stroage	Room temperature transport

*Unless otherwise informed, GeneMind sequencing platform and related sequencing reagents are not available in the USA, Canada, Australia, Japan, Singapore, Western Europe, Southern Europe and Nordic countries yet.

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