

GeneMind Expanded Carrier Screening (ESC) Solution

Explore Life's Mysteries for Better Healthcare

Highlight:

- Screen for a broad range of monogenic disorders

A 215-gene panel enables the analysis of 191 genetic diseases, along with the detection of the *MTHFR* gene 677 locus—a critical variant in folic acid metabolism that plays a key role in pregnancy health and the prevention of neural tube defects.

- Consolidate stand-alone assays to improve lab efficiency

NGS enables simultaneously screening of hundreds of genes in one run, providing information on a broader range of targets with a shorter turnaround time than traditional techniques such as PCR or Sanger sequencing.

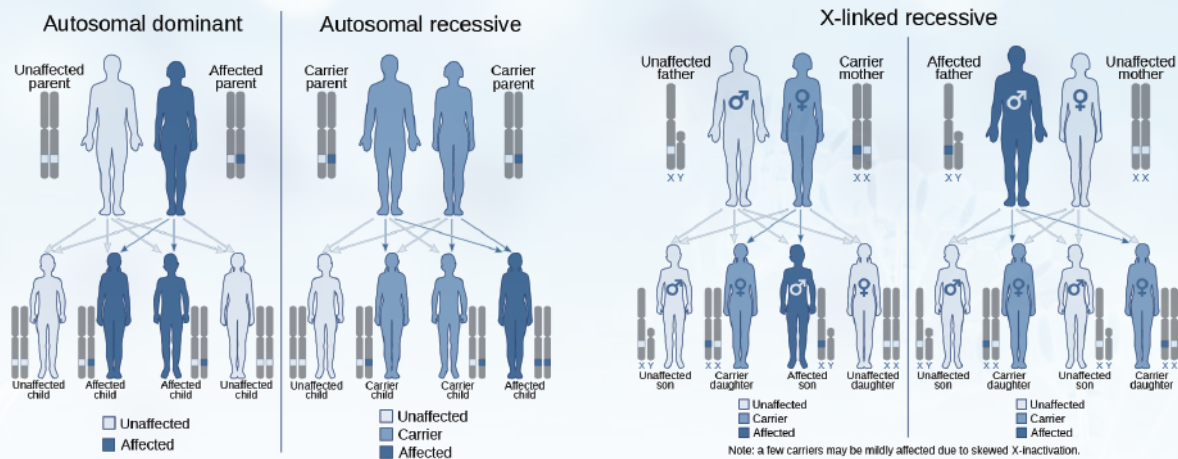
- Complete end-to-end solution from a single provider

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



Background

There are over 6,000 types of monogenic disorders ^[1], Around 1 in 50 people are affected by a known monogenic disorder, while around 1 in 263 are affected by a chromosomal disorder ^[2]. However, only about 5% of these conditions have effective drug treatments available ^[3].



[https://en.wikipedia.org/wiki/Dominance_\(genetics\)](https://en.wikipedia.org/wiki/Dominance_(genetics))

https://en.wikipedia.org/wiki/X-linked_recessive_inheritance

GeneMind Expanded Carrier Screening (ECS) solution is developed in accordance with the latest Carrier Screening Guidelines from the American College of Medical Genetics and Genomics. It screens for 191 monogenic disorders, including autosomal recessive and X-linked conditions, helping couples of reproductive age assess their carrier status and understand potential reproductive risks.

Targeted customer

Carrier screening is recommended for couples planning pregnancy or in the first trimester, 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

Sample Requirements

Sample Type	Requirements	Storage condition	transport
Blood	≥ 2 mL	2-8 °C for one week -20°C for one year	Cold-chain transportation
Buccal Swab	3-4	Room temperature storage	Room temperature transport

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

[1] <https://www.omim.org/statistics/entry>

[2] Mayo Clinic Proceedings. 76 (8): 777–783. doi:10.4065/76.8.777

[3] <https://esperare.org/en/addressing-rare-diseases>

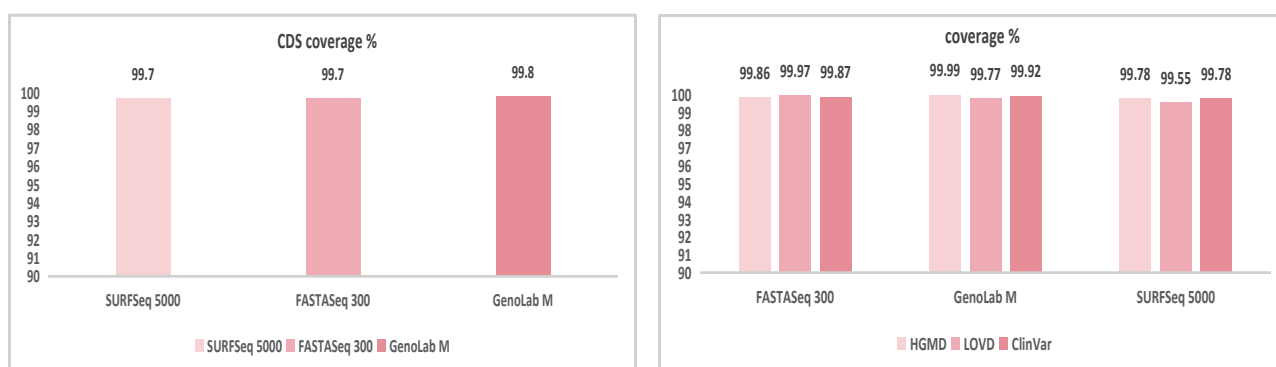
Testing Workflow



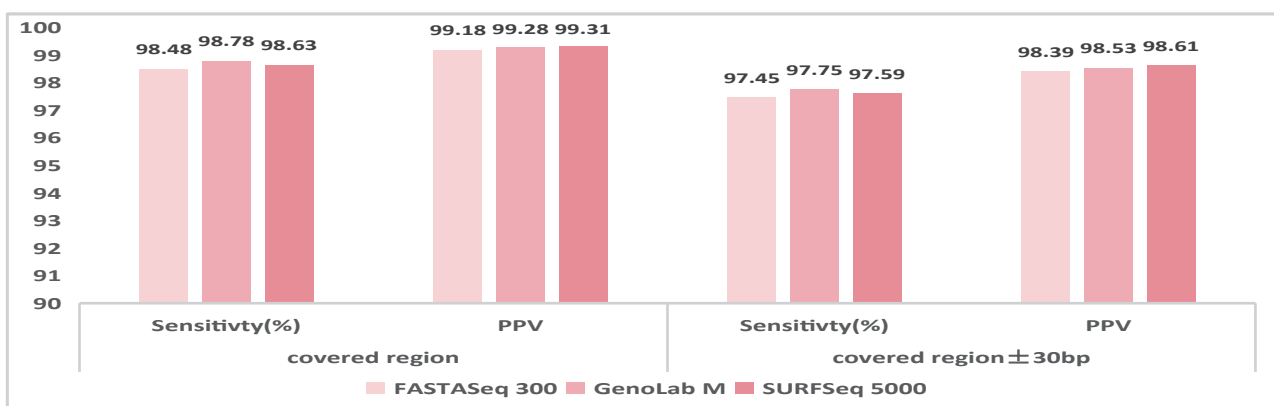
Parameter	Carrier Screen			
Sample type	gDNA/Tissue/blood/ buccal swabs			
Method	Capture sequencing			
Panel Size	Region size: 2152.76 Kb, probe coverage size: 2690.81 kb			
Sequencing Platform	FASTASeq™ S	FASTASeq™ 300	GenoLab™ M	SURFSeq™ 5000
Number of samples per run (1 Positive Control +1 Negative Control included)	1 FCL: 4 1 FCM: 8	1 FCM: 25 1 FCH: 56 1 FCP: 100	1 FCM: 50 1 FCH: 100	1 FCM: 100 1 FCH: 400
Read length	PE150			
Required data/ sample	≥1.2 Gb raw data			
Turnaround time ¹	3-10 days			
Report generation	Local analysis and report system			

1. Please note that the turn-around-time shown in the table is validated in GeneMind's lab, just for reference. It may be different due to the local situation, such as the sample volumes, lab equipment and the proficiency of operator.

Performance Data



➤ The CDS regions of NM transcripts from 215 genes achieve 99% coverage.



➤ Mutation detection results from the GM12878 cell line demonstrate excellent sensitivity and positive predictive value across the three platforms, with high consistency.

191 genetic diseases and 215 genes

System	Targeted Diseases	Number of Targeted Genes
Metabolic Systems	(76) Phenylketonuria (PKU); Tyrosinemia Type1; Maple Syrup Urine Disease (MSUD); Methylmalonic Acidemia; Glutaric Acidemia Type1; Glycogen Storage Disease; Galactosemia; Mucopolysaccharidosis; Niemann-Pick	91
Endocrine Systems	(6) 46,XY Sex Reversal, Type 2; Hyperinsulinemic Hypoglycemia, Familial; Combined Pituitary Hormone Deficiency 2; Adrenal Hypoplasia, Congenital, with Sex Reversal, etc.	5
Neurological and Neuromuscular Disorders	(35) Duchenne Muscular Dystrophy; Wilson Disease; Joubert Syndrome; X-linked Centronuclear Myopathy, etc.	38
Respiratory, Visual, and Auditory Systems	(13) Deafness; Retinitis Pigmentosa; Cystic Fibrosis, etc.	13
Hematologic and Immunologic Disorders	(12) Thalassemia; Hemophilia; Fanconi Anemia, etc.	19
Dermatologic Disorders	(10) Oculocutaneous Albinism; Epidermolysis Bullosa; Xeroderma Pigmentosum; Ichthyosis, etc.	17
Skeletal Disorders	(9) Cerebrooculofacioskeletal Syndrome (COFS); Short-Rib Thoracic Dysplasia 3 with or without Polydactyly; Epiphyseal Dysplasia; Achondroplasia, etc.	6
Digestive and Urinary Systems	(5) Progressive Familial Intrahepatic Cholestasis Type 2; Polycystic Kidney Disease, etc.	6
Multisystem Disorders	(25) Cerebro-oculo-facio-skeletal Syndrome; Meckel Syndrome; Kyphoscoliotic Ehlers-Danlos syndrome type 1, etc.	20
Sum	191	215

✖ Disclaimer

This document is based on company data of October, 2025, and is provided for reference only.

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