

NIFTY® Mono

200+ Monogenic Diseases Screening

Characteristics of Dominant Monogenic Diseases

High Comprehensive Incidence

Dominant monogenic diseases account for more than 50% of monogenic diseases, and the comprehensive incidence of 200+ target diseases tested is more than 1/400.

Difficult to Discover before Delivery

74% of dominant monogenic diseases are caused by new mutations. Such cases are not inherited from parents, and the affected fetus does not necessarily have a phenotype in the uterus, or ultrasound abnormalities occur only in the middle and late pregnancy. Clinically, Such disease are lack of effective early screening methods, and are extremely susceptible to missed diagnosis before delivery.

Difficult to Treat after Delivery

The target disease detected is severe and may cause death, teratogenicity, and disability. Most of them lack effective treatment methods.

Non-invasive prenatal testing for multiple monogenic diseases can effectively prevent and control the initial onset of dominant monogenic diseases!

Product Introduction

NIFTY® Mono evaluates the risk of 202 dominant monogenic diseases associated with pathogenic or suspected pathogenic mutation sites in 155 target genes by collecting 10 mL of maternal peripheral blood, using dual unique molecular indexing (UMI) for library preparation, testing fetal free DNA fragments in maternal peripheral blood during pregnancy through target region capture and high-throughput sequencing technology, and combining with bioinformatics analysis technology. UMI analysis can significantly reduce the problem of false positives caused by PCR amplification deviations or the introduction of wrong bases during the library preparation process, and has very high specificity and sensitivity.

Product Name	NIFTY® Mono 200+ Monogenic Diseases Screening
Testing Range	Risk of 202 dominant monogenic diseases associated with pathogenic or suspected pathogenic mutation sites in 155 target genes
Library Preparation Method	Dual UMI + target region capture
Testing Method	High-throughput sequencing
Sequencing Depth	Average depth > 800 ×
Sensitivity*	>99%
Specificity*	>99%
TAT	22 working days

*Product performance data is R&D test data, for reference only

Testing Object

Applicable Gestational Age

10 weeks^{+0 day} - 24 weeks^{+6 days}

Applicable Population

All pregnant women with singleton pregnancy who wish to reduce the risk of childbirth in monogenic disease by non-invasive prenatal testing and who do not have a family history of monogenic disease and have a normal phenotype.

* For the specific testing population, please refer to the submission form and informed consent form for NIFTY® Mono.

References

[1] Yang, Y. et al. Molecular Findings Among Patients Referred for Clinical Whole-Exome Sequencing[J]. JAMA, 2014, 312(18):1870-1879.

[2] <https://www.ncbi.nlm.nih.gov/books/NBK1116/>.

[3] Toriello H V, Meck J M. Statement on guidance for genetic counseling in advanced paternal age[J]. Genetics in Medicine, 2008, 10(6):457-460.

[4] Kong A, Frigge M L, Masson G, et al. Rate of de novo mutations, father's age, and disease risk[J]. Nature, 2012, 488(7412):471-475.