NIPT

Down Sendromu ve
diğer trizomiler için
girişimsel olmayan
doğum öncesi tarama testi

OMEGA GENETİK

Nipt Test Nedir?

Gebeliğin erken döneminde , sık görülen kromozom anomalileri için bebeğinizin riskini ortaya koyan yüksek güvenilirlikte bir tarama testidir.

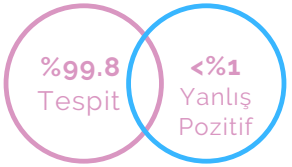
Neden Nipt Test Yaptırmalıyım?

- Bebeğin DNA'sına bağlı tarama testi olması nedeniyle %99.8 güvenilirlik oranına sahiptir.
- Bebeğe, amniyon sıvısına, göbek kordonuna, plasentaya direkt cerrahi müdahale yapılmaksızın sadece anne kolundan alınan kan ile çalışılır.
- Anne ve bebek için güvenlidir.
- Erken dönemde teşhis ve tanıya imkan verir.
- Gebeliğin 10. haftasından itibaren yaptırılabilir.
- Hızlı ve güvenilir sonuç verir.
- Hem otozomal hem de cinsiyet kromozomları için tek bir test ile tarama imkanı sunar.
- Tekil, ikiz ve tüp bebek gebelikler için uygundur.

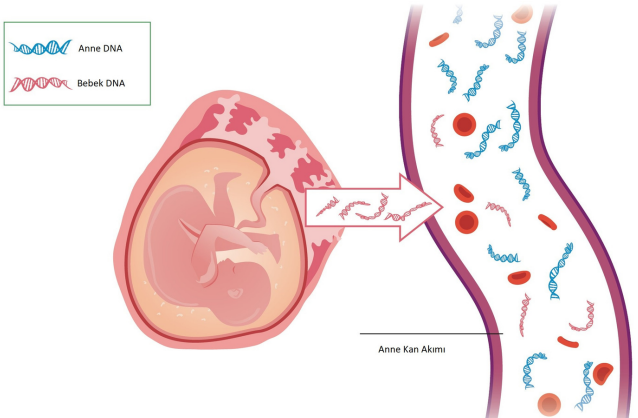
MomGuard™ Standard

Doğum Öncesi Tarama Testi

Down sendromu için Momguard testi ne kadar doğrudur?



Test	Standart
Trizomi 21 (Down Sendromu)	✓
Trizomi 18 (Edward Sendromu)	✓
Trizomi 13 (Patau Sendromu)	✓
Cinsiyet kromozomu anöploidi (Turner(mX), Kleinfelter (XXY) ve XXX Sendromu)	✓
Trizomi 1,2,3,4,5,6,7,8,9,10 11,12,14,15,16,17,19,20,22	✓



MomGuard™ Premium

Doğum Öncesi ve Sonrası Tarama Testi

Yenidoğan tarama testleri sayesinde saptanan yüksek risk, erken müdahaleye olanak sağlar.
Zamanında alınan önlemlerle henüz hastalık gelişmeden kontrol altına alınabilinir.

Test	Premium
MomGuard™ Standard	✓
Lizozomal Depo Hastalıkları (Gaucher,Fabry,Pompe,Hurler,Hunter,Sanfilippo Tip A,B,C,D ,Morquio Tip A,B,Maroteaux -Lamy, Sly Sendromu	✓
Glikojen Depo Hastalıkları Glikojenozis tip Ia,Ib,III,IV,,V,VI,VII	✓
Anormal Bakır Metabolizması (Wilson Hastalığı)	✓
Doğuştan İşitme Kaybı (Pendred,Usher Sendromu Tip1B,1D,Non-sendromik işitme kaybı,	✓

MomGuard™

Standard Test Report

Organization	
Patient	
DOB	80-12-06
Age / Gender	41 / F
Specimen	Whole blood

Registration No.	20220314-57002
Collection Date	2022-03-02
Receipt Date	2022-03-14
Analysis Date	2022-03-14
Report Date	2022-03-17

Patient Information

Number of Fetus	1
Ultrasound Gestational Age	10W 2D
Height / Weight	160cm / 59kg

Quality Control / Test Result

Cell Free DNA Quality	Sequencing Quality	Fetal Fraction	Standard Material Test Result
Pass	Pass	Pass(6.38%, Male)	Pass

INTERPRETATION Low Risk - The chance of the baby having a chromosomal abnormality is very low.

Result Details

Chromosome Aneuploidies

Items (Disease Type)	Result	Risk Score
Trisomy 21(Down Syndrome)	Low Risk	1/2145
Trisomy 18(Edward Syndrome)	Low Risk	1/3709
Trisomy 13(Patau Syndrome)	Low Risk	1/3476
Sex aneuploidy(mX, XXY, XXX)	Low Risk	1/2640

✳ *Test Method*
cfDNA is isolated from maternal blood and sequenced using Next Generation Sequencing (NGS) technology. Sequencing data is analyzed using in-house bioinformatics pipeline to identify fetal aneuploidy in the tested chromosomes. This test provides a result only when a sample meets the quality threshold.

✳ *Test Purpose & Limitation*
This test is a screening test for T21, T18, T13 and sex chromosome aneuploidy under the consent of the mother.
No analysis for sex chromosome aneuploidy is to be performed in case of twin.
This test is highly accurate, but not diagnostic. Therefore, if a confirmatory test is required according to the test result or the clinical situation of the mother, an amniocentesis or CVS should be performed. Very rarely, fetal sex information can be wrong for reasons such as vanishing twin.

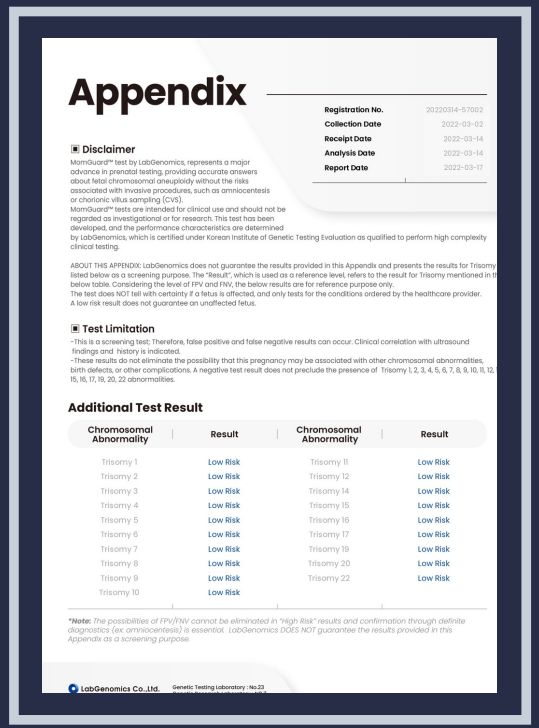
MomGuard Performance Table

	Sensitivity	Specificity
Chromosome 21	98.65%	99.94%
Chromosome 18	100%	99.98%
Chromosome 13	100%	99.97%

- Overall PPV = 0.8932, NPV=0.9999
- Overall False Positive (FP) % = 0.02
- Overall False Negative (FN) % = 0.009

LabGenomics Co.,Ltd. Genetic Testing Laboratory : No.23 Genetic Research Laboratory : NO.7

Medical Doctor : SY Kim M.D., Ph.D.
Analysis officer : KR Kim Ph.D.
Laboratory officer : MS Kim Ph.D.



Appendix

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▣ Disclaimer

MomGuard™ test by LabGenomics, represents a major advance in prenatal testing, providing accurate answers about fetal chromosomal aneuploidy without the risks associated with invasive procedures, such as amniocentesis or chorionic villus sampling (CVS).
MomGuard™ tests are intended for clinical use and should not be regarded as investigational or for research. This test has been developed, and the performance characteristics are determined by LabGenomics, which is certified under Korean Institute of Genetic Testing Evaluation as qualified to perform high complexity clinical testing.

ABOUT THIS APPENDIX: LabGenomics does not guarantee the results provided in this Appendix and presents the results for Trisomy listed below as a screening purpose. The "Result", which is used as a reference level, refers to the result for Trisomy mentioned in t below table. Considering the level of FPV and FNV, the below results are for reference purpose only.
The test does NOT tell with certainty if a fetus is affected, and only tests for the conditions ordered by the healthcare provider. A low risk result does not guarantee an unaffected fetus.

▣ Test Limitation

-This is a screening test; Therefore, false positive and false negative results can occur. Clinical correlation with ultrasound findings and history is indicated.
-These results do not eliminate the possibility that this pregnancy may be associated with other chromosomal abnormalities, birth defects, or other complications. A negative test result does not preclude the presence of Trisomy 1, 2, 3, 4, 5, 6, 7, 8, 9, 10, 11, 12, 15, 16, 17, 19, 20, 22 abnormalities.

Additional Test Result

Chromosomal Abnormality	Result	Chromosomal Abnormality	Result
Trisomy 1	Low Risk	Trisomy 11	Low Risk
Trisomy 2	Low Risk	Trisomy 12	Low Risk
Trisomy 3	Low Risk	Trisomy 14	Low Risk
Trisomy 4	Low Risk	Trisomy 15	Low Risk
Trisomy 5	Low Risk	Trisomy 16	Low Risk
Trisomy 6	Low Risk	Trisomy 17	Low Risk
Trisomy 7	Low Risk	Trisomy 19	Low Risk
Trisomy 8	Low Risk	Trisomy 20	Low Risk
Trisomy 9	Low Risk	Trisomy 22	Low Risk
Trisomy 10	Low Risk		

**Note: The possibilities of FPV/FNV cannot be eliminated in "High Risk" results and confirmation through definite diagnostics (ex. amniocentesis) is essential. LabGenomics DOES NOT guarantee the results provided in this Appendix as a screening purpose.*

LabGenomics Co.,Ltd. Genetic Testing Laboratory : No.23

Türkiye'nin
Her Bölgesinde
Uzman Sağlık
Personelimiz İle
Hizmetinizdeyiz.

MomGuard™
Non Invasive Prenatal Test

LabGenomics

illumina®

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