VERAgene® comprehensive NIPT

AFFIX
BARCODE LABEL – SIF
HERE

SAMPLE INFORMATION FORM

Please complete sections below in English.

MATERNAL SPECIMEN

FIRST NAME	LAST NAME
DATE OF BIRTH (DD/MM/YY)	ETHNICITY
ID	PHONE NUMBER
EMAIL	
ADDRESS	
CITY / POST CODE / COUNTRY	

PATERNAL SPECIMEN

FIRST NAME	LAST NAME
DATE OF BIRTH (DD/MM/YY)	ETHNICITY
ID	PHONE NUMBER
EMAIL	
ADDRESS	
CITY / POST CODE / COUNTRY	

REFERRAL INFORMATION

CLINIC NAME	CLINIC ID
REFERRING HEALTHCARE PROVIDER	
PHONE NUMBER	FAX
EMAIL	
ADDRESS	
CITY / POST CODE / COUNTRY	

CLINICAL AND TEST DETAILS

REQUESTED TEST
TICK ONLY ONE BOX BELOW

FOR SINGLETON PREGNANCIES

☐ TRISOMIES 13, 18, 21; PRESENCE OF Y; ANEUPLOIDIES X,Y; MICRODELETIONS; 100 SINGLE GENE DISEASES

FOR TWIN/VANISHED TWIN PREGNANCIES

☐ TRISOMIES 13, 18, 21; PRESENCE OF Y; MICRODELETIONS; 100 SINGLE GENE DISEASES

TEST INDICATIONS
TICK APPROPRIATE BOXES & ADD COMMENTS

☐ PATIENT/FAMILY HISTORY
☐ ABNORMAL ULTRASOUND
☐ ADVANCED MATERNAL AGE
☐ SERUM SCREEN RISK

T21 RISK SCORE:	1 IN
T18 RISK SCORE:	1 IN
T13 RISK SCORE:	1 IN

☐ CONSANGUINITY
☐ OTHER

CLINICAL INFORMATION
COMPLETE ALL SECTIONS BELOW

MATERNAL INFORMATION

GESTATIONAL AGE (WEEK + DAY)
WEIGHT (KG)
HEIGHT (CM)

TEST INFORMATION

REDRAW TEST:
☐ YES ☐ NO

COLLECTION DATE (DD/MM/YY): _____

NUMBER OF FETUSES

☐ 1 FETUS
☐ 1 FETUS – VANISHED TWIN
Collect 4 weeks after the vanishing event
☐ 2 FETUSES MONOCHORIONIC TWINS
☐ 2 FETUSES DICHORIONIC-TWINS

IVF INFORMATION

IVF PREGNANCY:
☐ YES ☐ NO

Only self-egg IVF pregnancies; not valid for donor egg or surrogates

HEALTHCARE PROVIDER COMMENTS

FOR LABORATORY USE ONLY
F-OPR-01/02.0-V8-EN

ORDER NUMBER	LAB ID NUMBER	KIT LOT NUMBER
COMMENTS	DATE & TIME OF RECEIPT (DD/MM/YY HH:MM)	RECEIVED BY

RESTRICTED

MEDICOVER GENETICS
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PATIENT CONSENT

By placing my signature signing below I hereby:

1. Confirm that I have read, or have had read to me, the Patient Informed Consent which is attached to this page and that I understand it.
2. Declare that I have had the opportunity to receive counseling from my referring healthcare provider on the VERAgene test and to discuss with the healthcare provider all aspects of the VERAgene test and this form including the benefits, risks and limitations of the VERAgene test, as well as the reasons for performing the test and availability of alternative testing options to my satisfaction.
3. Authorize my referring healthcare provider to collect the necessary biological samples (blood and buccal swab) and to submit this form and transport the samples to Medicover Genetics laboratories for the purposes of conducting the tests requested with this form.
4. Authorize Medicover Genetics to use any part of or the entirety of the biological samples (blood and buccal swab) for the purposes of conducting the tests requested with this form.
5. Authorize Medicover Genetics to communicate the results of the test to my referring healthcare provider.
6. Confirm that all the information on this form is true to the best of my knowledge.

Your test results and any unused biological material can help Medicover Genetics improve and further develop the quality, accuracy and effectiveness of the analysis and help us expand the scope of genetic testing. For this reason, Medicover Genetics would like to use your anonymized, de-identified (i.e. after removing all the personal information from which you can be identified) test results and unused biological material.

☐ For the above scope, I consent to the inclusion of my test results in Medicover Genetics' database, the coding, storing and using of biological material.

MATERNAL SIGNATURE (BIOLOGICAL MOTHER)	DATE

PATERNAL SIGNATURE (BIOLOGICAL FATHER)	DATE

HEALTHCARE PROVIDER ATTESTATION

I hereby certify and undertake that:

1. The patient has been informed that the test will only test for the disorder(s) requested on this form and has been duly and thoroughly counseled about the test and has received all the advice necessary to provide their informed consent, including the benefits, risks, and limitations of the VERAgene test.
2. I have answered all the patient's queries about the VERAgene test.
3. This form has been completed according to the wishes and instructions of the patients.
4. I have obtained the patient's informed consent and have attested their signature.

HEALTHCARE PROVIDER SIGNATURE	DATE

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PATIENT INFORMED CONSENT

VERAgene TEST: VERAgene is a Non-Invasive Prenatal Test (NIPT) which can be taken by pregnant women during or after the 10th week of pregnancy to screen for certain genetic conditions in the developing fetus before birth. VERAgene tests for the presence of an extra chromosome – a genetic condition called trisomy – in chromosomes 13, 18 and 21. VERAgene also offers testing for changes in the number of X and Y chromosomes (sex chromosome aneuploidies), and microdeletions (loss of a part of a chromosome). Additionally, VERAgene screens for fetal risk for 100 single gene diseases and can provide fetal sex information, if you opt to know. (Tables 1 & 3)

Table 1: Conditions Tested by VERAgene

	CONDITIONS	SIGNIFICANCE
Autosomal Chromosome Aneuploidies	Trisomy 13 - Patau syndrome	Life-threatening, high fetal mortality rate, reduced lifespan
	Trisomy 18 - Edwards syndrome	
	Trisomy 21 - Down syndrome	Mild to severe, with intellectual and physical disabilities, heart defects
Sex Chromosome Aneuploidies	Monosomy X - Turner syndrome	Fertility problems. Mild to severe learning difficulties & behavioral problems. Moderate to distinctive appearances
	Triple X syndrome, XXX	
	Klinefelter syndrome, XXY	
	Jacobs syndrome, XYY	
	XXYY syndrome	
Microdeletions	DiGeorge syndrome, 22q11.2 deletion	Several organs affected, mild to severe learning disabilities & behavioral problems. Distinctive appearances
	1p36 deletion	
	Smith-Magenis syndrome, 17p11.2 deletion	
	Wolf-Hirschhorn syndrome, 4p deletion	
Single gene diseases	Please see table 3 for complete list	Often severe with significant impact on quality of life

SAMPLE COLLECTION: VERAgene requires two tubes of blood (20ml) from the biological mother using standard phlebotomy practices and a buccal swab from the biological father. Samples from both biological parents are required for the test to be performed otherwise test results are not valid. Your healthcare provider will collect both samples and send them to Medicover Genetics laboratories for analysis. Additional sample may be needed if there is a shipping delay, breakage of the sample collection tubes, sample degradation or contamination, or if the sample has been collected or submitted incorrectly.

TESTING PROCESS: Genetic material (DNA) from the developing fetus's placenta is present in the pregnant woman's blood. With the help of specialized equipment and software, VERAgene uses an innovative, technology called 'Target Capture Enrichment Technology' to isolate the fetal DNA, and calculate whether there is an increased risk of the fetus having an aneuploidy or a microdeletion. Simultaneously, the maternal and paternal alleles (DNA) are analyzed for the 100 single gene diseases specified in table 3. If both biological parents are carriers of the same monogenic disease a 'high risk' result for the fetus is reported. A high risk result for single gene diseases indicates that the fetus has one in four chance (autosomal diseases) and one in two chance (X-linked diseases) of being affected. In a small number of cases the amount of DNA isolated from the maternal or the paternal sample is not sufficient for analysis and a redraw may be requested. Although rare, there is always a chance that a result will not be obtained due to lack of genetic material.

INTERPRETING NIPT RESULTS: Results are communicated to your healthcare provider in approximately 7 working days from sample receipt. Your healthcare provider is responsible to understand the specific uses and limitations of the test, communicate this information to you and answer any questions you may have. The healthcare provider is also responsible for counselling before and after the test, discussing possible next steps and clinical management including the provision of advice regarding the need for additional prenatal genetic testing. VERAgene NIPT is not a diagnostic but a screening test and results should always be considered in the context of other clinical criteria. Test results for risk of fetal aneuploidy, microdeletion or single gene disease are reported individually for each category and as a combined risk result. A negative result is reported as *VERY LOW RISK* for the specific condition and indicates that the possibility of the fetus having that condition is very low. A positive result is reported as *VERY HIGH RISK* for the specific condition and indicates that there is an increased possibility of the fetus having the specified condition. A high risk result for single gene diseases indicates that the fetus has one in four chance (autosomal diseases) and one in two chance (X-linked diseases) of being affected. A *VERY HIGH RISK* result in twin pregnancies indicates very high risk of at least one fetus having the specified condition. In twin pregnancies, detection of Y indicates the presence of at least one Y chromosome. As VERAgene is a screening test, a high risk result should always be confirmed by amniocentesis. Results and possible next steps should always be considered in the context of other clinical criteria and should be fully discussed with your healthcare provider.

ELIGIBILITY CRITERIA:

1. VERAgene is available for singleton pregnancies and twin pregnancies, including in-vitro fertilization (IVF) pregnancies of at least 10 weeks of pregnancy where the biological parents' gametes are used.
2. Twin pregnancies in which loss of one fetus (vanished twin) occurred are eligible for testing on or after the 10th week of pregnancy and 4 weeks after the vanishing event.
3. Twin and vanished twin pregnancies are not eligible for X and Y aneuploidy detection.
4. The VERAgene test cannot be performed on pregnancies achieved with egg/sperm donation or surrogacy.
5. Patients with malignancy or history of malignancy, patients with bone marrow or organ transplant or recent transfusions are not eligible for the test.

Consult with your healthcare provider to determine if VERAgene is appropriate for you. Please see table below for eligibility.

Table 2: Eligibility for VERAgene NIPT

	Trisomies 13, 18, 21	Aneuploidies X, Y	Microdeletions	Presence of Y	Single Gene
Singleton	✓	✓	✓	✓	✓
Twin / Vanishing Twin	✓		✓	✓	✓
IVF Pregnancy (Self Egg Used)					
Singleton	✓	✓	✓	✓	✓
Twin / Vanishing Twin	✓		✓	✓	✓

DISCLOSURE: VERAgene only tests and reports on the tests selected on the information form. The VERAgene non-invasive prenatal test is not intended and is not validated for the detection of mosaicism, triploidy, partial trisomy or translocations. The test will not identify all deletions associated with each microdeletion syndrome. This test has been validated on full region deletions and may be unable to detect smaller deletions. VERAgene investigates a number of pathogenic/likely pathogenic mutations associated with moderate or severe phenotype but not all of them. Therefore, a negative result or low risk result reduces but does not eliminate the possibility of the fetus to be affected or to carry the mutation. Although this test is highly accurate there is still a possibility of a false positive and false negative results. This is due to technical and/or biological limitations, including but not limited to confined placental mosaicism (CPM) or other types of mosaicism, maternal constitutional or somatic chromosomal abnormalities, residual cfDNA from a vanished twin or other rare molecular events. The test does not report on the parental carrier status for the monogenic diseases tested.

QUALITY IMPROVEMENT: Please choose the relevant option on the consent form to grant us permission to anonymously use your remaining sample to improve the quality, accuracy and effectiveness of VERAgene.

Please make sure you read and understand the information on this document before signing it, and complete all relevant information accurately as incorrect information can lead to inaccurate test results. Please discuss any questions you may have with your healthcare provider. For additional information please visit our website at ***www.medicover-genetics.com****.*

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Table 3: Single Gene Diseases Screened by VERAgene

3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	Glycine Encephalopathy (GLDC-related)	Neuronal Ceroid Lipofuscinosis (CLN8-related)
3-Methylcrotonyl-CoA Carboxylase Deficiency 1	Glycogen Storage Disease, Type 1A	Neuronal Ceroid Lipofuscinosis (MFSD8-related)
3-Methylcrotonyl-CoA Carboxylase Deficiency 2	Glycogen Storage Disease, Type 1B	Neuronal Ceroid Lipofuscinosis (TPP1-related)
Abetalipoproteinemia	Glycogen Storage Disease, Type 3	Nijmegen Breakage Syndrome
Acyl-CoA Oxidase I Deficiency	Glycogen Storage Disease, Type 7	Omenn Syndrome (RAG2-related)
Aicardi-Goutières Syndrome	GRACILE Syndrome	Ornithine Aminotransferase Deficiency
Alport Syndrome, X-Linked	Hereditary Fructose Intolerance	Ornithine Translocase Deficiency [Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH) Syndrome]
Alstrom Syndrome	Homocystinuria, Type cblE	Pendred Syndrome
Andermann Syndrome	Hydrolethalus Syndrome	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX1-related)
Aromatase Deficiency	Inclusion Body Myopathy, Type 2	Peroxisome Biogenesis Disorders Zellweger Syndrome Spectrum (PEX2-related)
Arthrogryposis Mental Retardation Seizures	Isovaleric Acidemia	Phenylalaline Hydroxylase Deficiency (Phenylketonurea)
Asparagine Synthetase Deficiency	Joubert Syndrome, Type 2	Pontocerebellar Hypoplasia, Type 1A
Aspartylglycosaminuria	Junctional Epidermolysis Bullosa, Herlitz Type	Pontocerebellar Hypoplasia, Type 2D
Autosomal Recessive Polycystic Kidney Disease	Lamellar Ichthyosis, Type 1	Pontocerebellar Hypoplasia, Type 2E
Bardet-Biedl Syndrome (BBS1-related)	Leber Congenital Amaurosis (LCA5-related)	Primary Ciliary Dyskinesia (DNAH5-related)
Bardet Biedl Syndrome (BBS12-related)	Leigh Syndrome, French-Canadian Type	Primary Ciliary Dyskinesia (DNAI1-related)
Beta Thalassemia	Leukoencephalopathy with Vanishing White Matter	Primary Hyperoxaluria, Type 3
Biotinidase Deficiency	Leydig Cell Hypoplasia [Luteinizing Hormone Resistance]	Pycnodysostosis
Canavan Disease	Limb Girdle Muscular Dystrophy, Type 2E	Pyruvate Dehydrogenase Deficiency (PDHB-Related)
Carpenter Syndrome	Lipoamide Dehydrogenase Deficiency [Maple Syrup Urine Disease, Type 3]	Retinal Dystrophy (RLBP1-related) [Bothnia Retinal Dystrophy]
Choreacanthocytosis	Lipoprotein Lipase Deficiency	Retinitis Pigmentosa 25 (EYS-related)
Choroideremia, X-Linked	Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	Retinitis Pigmentosa 59 (DHDDS-related)
Citrin Deficiency	Lysinuric Protein Intolerance	Sanfilippo Syndrome, Type D [Mucopolysaccharidosis IIID]
Combined Oxidative Phosphorylation Deficiency 3	Maple Syrup Urine Disease, Type 1B	Severe Combined Immunodeficiency, Type Athabaskan
Congenital Disorder of Glycosylation, Type 1A (PMM2-related)	Methylmalonic Acidemia (MMAA-related)	Severe Combined Immunodeficiency, X-Linked
Congenital Neutropenia (HAX1-related)	Methylmalonic Aciduria, Type Mut(0)	Sickle-Cell Disease
Crigler Najjar Syndrome, Type I	Methylmalonic Aciduria and Homocystinuria, Type cblC	Sjögren-Larsson Syndrome
Cystic Fibrosis *	Methylmalonic Aciduria and Homocystinuria, Type cblD	Steroid-Resistant Nephrotic Syndrome
Factor XI Deficiency	Mucopolysaccharidosis, Type II [Hunter Syndrome], X-Linked	Stuve-Wiedemann Syndrome
Familial Dysautonomia	Mucopolysaccharidosis, Type IIIC [Sanfilippo C]	Tay-Sachs Disease
Fanconi Anemia, Type C	Multiple Sulfatase Deficiency	Usher Syndrome, Type 1F
Fanconi Anemia, Type G	Myotubular Myopathy, X-Linked	Usher Syndrome, Type 3
Gaucher Disease	Navajo Neurohepatopathy [MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome]	Wolman Disease
Glutaric Acidemia, Type 2A		

* The VERAgene 100 panel tests for mutations that cause the classic Cystic Fibrosis phenotype.

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PATIENT PRIVACY SUMMARY

This privacy notice provides a summary of how Medicover Genetics Limited collects and processes your personal data with this form. It is important that you read this privacy notice together with our full privacy policy which contains more detailed information about our data processing. A copy is available online at www.medicover-genetics.com.

1. Important information and who we are

Medicover Genetics is responsible for processing the personal data collected on this form.

We have appointed a data protection officer (DPO). If you have any questions about this privacy notice or our data protection practices, please contact the DPO.

CONTACT DETAILS

Full name of legal entity: Medicover Genetics Limited (HE 418406)

Email address: dpo.cy@medicover.com

Postal address: 31 Neas Engomis Street, 2409 Engomi, Nicosia, Cyprus

Telephone number: + (357) 22266888

2. The data we collect about you

We collect, use, store and transfer personal data about you as follows:

- Identity Data.
- Contact Data.
- Sensitive data (ethnicity, medical/clinical data).

3. How we use your personal data

We will only use your personal data for the purpose for which we collected it. This includes the following:

- To register you as a new customer.
- To conduct the selected test and to process and deliver your results.
- To manage your relationship with us and to provide customer support, where applicable.
- To contact you or your referring healthcare provider on your results.
- To invoice the referring healthcare provider.

4. How we share your personal data

We share your personal data with your referring healthcare provider, so we can communicate the results of your test to them. Medicover Genetics stores personal information on its database which is hosted by cloud service providers.

5. International transfers

We do not transfer, store or process your personal data outside the European Economic Area unless you or your referring healthcare provider are located outside the EEA.

6. Your legal rights

Under certain circumstances, you have rights under data protection laws in relation to your personal data including the right to receive a copy of the personal data we hold about you, the right to erasure ('right to be forgotten'), the right to restriction of processing and the right to make a complaint at any time to the Office of the Commissioner for Personal Data Protection.

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