

Request form for genetic studies of foetal remains or Products of Conception (POC). POC by NGS.

The fields marked with (*) are mandatory to carry out the test

PATIENT DETAILS	
* Name:	* Surname:
* Date of birth:	* Email address:
* Telephone:	* MRN:
PRESCRIBING DOCTOR DETAILS	BLOOD CLINIC DETAILS
* Prescribing doctor's name:	* Name of centre:
* Prescribing doctor's E-mail:	* Telephone:
* Name of centre:	* Telephone:
* Telephone:	* Email address:
TEST INDICATION	CLINICAL INFORMATION
<input type="checkbox"/> Advanced maternal age <input type="checkbox"/> Abnormal ultrasound <input type="checkbox"/> Family or personal history of aneuploidy or genetic disease ⁽¹⁾ ⁽¹⁾ Please specify <input type="checkbox"/> Previous miscarriages N° _____ <input type="checkbox"/> High-risk combined screening Specify value: _____ <input type="checkbox"/> Abnormal patient/partner Karyotype Specify: _____ <input type="checkbox"/> Other: _____ Language of the report: <input type="checkbox"/> Spanish <input type="checkbox"/> English	*Gestational age of foetal arrest (weeks): _____ + _____ *Date of gestational loss: _____ *Date of sample collection: _____ *Pregnancy: <input type="checkbox"/> Singleton <input type="checkbox"/> Twin <input type="checkbox"/> Multiple N° _____ Type of pregnancy: <input type="checkbox"/> Natural <input type="checkbox"/> Artificial Reproductive Treatment <input type="checkbox"/> HAI <input type="checkbox"/> PGT-A (PGS) <input type="checkbox"/> DAI <input type="checkbox"/> EMBRACE <input type="checkbox"/> FIV <input type="checkbox"/> PGT-SR (PGD) <input type="checkbox"/> ICSI <input type="checkbox"/> PGT-M (PGD)
Transferred embryos: <input type="checkbox"/> 1 <input type="checkbox"/> 2 <input type="checkbox"/> >2 Oocyte origin: <input type="checkbox"/> Own <input type="checkbox"/> Donated	
SAMPLE INFORMATION	
*Type of sample: <input type="checkbox"/> Guided biopsy <input type="checkbox"/> Conventional curettage <input type="checkbox"/> Spontaneous miscarriage <input type="checkbox"/> Other: _____ Sampling performed by (complete name): _____	
DIRECT ANALYSIS PERFORMED ON TISSUE (MEDICAL OR SURGICAL EVACUATION). Valid for singleton and multiple pregnancies.	
<input type="checkbox"/> POC by NGS <ul style="list-style-type: none"> Aneuploidy for all 24 chromosomes. Deletions and duplications greater than 10Mb. Triploidy and maternal cell contamination studied by complementary STRs analysis. 	
DOCTOR AUTHORISATION	
I certify that the information on this requisition form is correct to the best of my knowledge and that I have requested the above test based on my professional judgement of clinical indication. I have explained the limitations of this test and I have answered any question with medical judgement. I understand that Igenomix may need additional information and I agree to provide this information if necessary.	
*Doctor's signature: _____	*Date: _____
PATIENT CONSENT	
By signing this requisition form, I voluntarily request Igenomix to carry out the test indicated above. I have read and received a copy of the informed consent, included in the following pages. The risks, benefits and limitations of this test have been explained to me.	
*Patient's signature: _____	*Date: _____

Informed consent for analysis of foetal remains or products of conception (POC) screening for aneuploidy of all 24 chromosomes

DESCRIPTION, PURPOSE AND BENEFITS OF THE ANALYSIS

Chromosomal abnormalities can lead to foetuses with deformities, miscarriages or even neonatal deaths. Estimates of the frequency of chromosomal abnormalities in miscarriages range from 15% to 60%.

In the case of early termination of pregnancy or miscarriage, the diagnosis of a chromosomal abnormality may be important for planning future pregnancies.

Chromosomal tests on foetal remains, otherwise known as Products of Conception (POC) provide useful information and help patients and doctors to determine the causes of miscarriages, the risk of recurrent miscarriages and the subsequent risk of having children with chromosomal abnormalities.

POC test provides more information faster than traditional cytogenetic karyotyping. With the combination of NGS (Next Generation Sequencing) and STR (Short Tandem Repeat) techniques, the presence of extra or missing chromosomes is detected, as well as any partial chromosome losses or duplications, and the presence of maternal cell contamination (MCC) is either ruled out or detected.

PROCEDURE, RISKS AND LIMITATIONS

In order to process the sample, it will be necessary that the test request document is correctly completed. Otherwise, the analysis may be stopped until the required information is provided to the laboratory.

POC by NGS

The genetic analysis is performed directly on the evacuated foetal. The sample must be collected (either by the patient if it is a spontaneous abortion or by curettage by the gynaecologist if the abortion is missed or incomplete) and sent using the sample collection kit provided by Igenomix. To rule out or detect contamination with maternal cells, it will be necessary to obtain a biological sample from the patient (blood in EDTA tube, preferably using the one sent in the kit), which will be sent together with the POC sample. The risks derived from these procedures should be explained by the professionals in charge of performing them.

Once in the laboratory, whenever possible, foetal versus maternal tissue is identified in the POC sample received. The sample is cleaned, dissected, DNA is extracted and analysed using 24-chromosome bulk sequencing (NGS). The STR protocol is used to detect or rule out maternal cell contamination as well as some of the polyploidy.

Before performing the genetic study, you should consider the implication of the possible results. There are three possible results:

- a. Positive results: one or more alterations are detected that could be the cause of the gestational loss.
- b. Negative results: No genetic alterations are detected within the resolution of the platform used. A negative result does not imply the absence of pathology of genetic cause, since it depends on the extent of the study requested according to the diagnostic suspicion and the limitations of the technique used.
- c. Non-informative results: Exceptionally, contamination of the sample, poor quality or low quantity of the sample may result in no results being obtained.

Due to the complexity of genetic testing and the important implications of the test results, the results obtained must be interpreted together with other clinical data, within the general context of a medical consultation to be conducted by healthcare professionals. Reports of the results will be kept strictly confidential. The test result will be available within 12 working days following receipt of the sample. A small percentage of samples may experience a variable delay due to unforeseen causes. Should this occur, the relevant clinical manager will be informed of the delay and Igenomix will not be responsible, under any circumstances, for any delay beyond the aforementioned period.

The main limitations of the POC test consist of:

1. POC test detects the most frequent genetic conditions associated with gestational losses but does not detect all genetic conditions and/or all possible chromosomal abnormalities. The following are the conditions that may not be detected:
 - Balanced chromosomal rearrangements.
 - POC test does not test for specific genes and cannot detect conditions caused by individual gene mutations, such as sickle cell disease, cystic fibrosis, or Tay-Sachs disease.
 - Complete trisomies of acrocentric chromosomes will not be distinguishable from trisomies caused by a Robertsonian translocation (affecting chromosomes 13, 14, 15, 21, or 22) or an isochromosome in one of the parents.
 - Detection of uniparental disomy (a condition in which both copies of a chromosome come from the same parent).
 - Other causes of miscarriage not yet identified.
2. Risk of misdiagnosis due to incorrect sample identification, inaccurate parentage information, mosaicism or other unidentified genetic abnormalities. Diagnostic errors due to test failure account for <1%.
3. There is the possibility of unpredictable and uncontrollable problems with transportation, such as those related to weather and air transport, or other circumstances beyond control, which would not allow timely results. There is also the possibility that the sample received at the laboratory is unacceptable for analysis, and therefore, results cannot be obtained from the sample provided.
4. POC by NGS detects most polyploidies, such as triploidy 69,XXY, 69,XXX, and tetraploidy 92,XXX and 92,XXYY. However, it cannot detect some tetraploidies, such as 92,XXXX or 92,XXYY.
5. Mosaicism (presence of more than one distinct cell line in the sample). POC by NGS does not detect low levels of mosaicism, less than 30%.
6. POC by NGS test cannot detect gains or losses of chromosomal material below 10Mb
7. There is a possibility that the result may not be conclusive because of the probability of contamination with maternal cells.

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DATA PRIVACY, STORAGE AND RESEARCH USE OF THE SAMPLES

Your privacy is a priority for the Igenomix Group. Your identity and all data referring to your personal information will be confidential and only Igenomix personnel will be permitted access to this information, along with the relevant authorities when required by the laws of the applicable jurisdiction.

Below are listed the different purposes for which your personal data will be processed either by the Clinic or the healthcare provider, or by Igenomix:

Processing of your personal data for the purpose of carrying out the genetic test. For this purpose, Igenomix UK ("Igenomix") is processing your personal data exclusively on behalf of your clinician/healthcare provider. Your clinician/healthcare provider is the data controller and Igenomix is the data processor. This entails that for this purpose, Igenomix will only process your personal data following the instructions and directions of your clinician/health care provider. Hence, the purpose and means for the processing of your personal data to carry out the test is mainly determined by your clinician/healthcare provider, who will have provided you with all relevant privacy information regarding the processing of your personal data in line with the applicable regulations.

Therefore, if you have any queries regarding the processing of your personal data for this purpose, you may contact the data controller (your clinician/healthcare provider) at the address it has provided you in its own privacy policy. If you need to contact Igenomix, you can reach out to our Data Protection Officer through the following e-mail address privacy@igenomix.com or with postal mail to the address Igenomix UK, 40 Occam Road, Guildford GU2 7YG, UK.

The sample will be analysed by Igenomix or an associated group selected by Igenomix on an international level. Igenomix reserves the right to carry out part or all of the analyses included in the test through third party laboratories certified with recognised international quality standards, and with the same level of data protection security measures. Any results obtained in this way will be inspected by Igenomix and this circumstance will be indicated in the final report issued.

Use of Pseudonymized Information for Continuous Improvement, Internal Training and Knowledge Development (Not Biomedical Research)
In accordance with applicable international standards (ISO 15189, CAP, CLIA) and national legislation, we are required to carry out ongoing validations, quality controls, internal audits, and staff training. These activities are essential to ensure the accuracy, reliability, and safety of the services provided by Igenomix. As part of this, test results generated from Igenomix's marketed services may be used in a key-coded (pseudonymized) form, meaning they cannot be directly linked to your identity, and access to identifying information is strictly restricted. The data may be used internally for: (1) Continuous Improvement; Optimise protocols, techniques and tools, ensuring increasingly effective and safe care. (2) Team Training; To help our staff maintain a high level of competence using real but de-identified case data (3) Internal Knowledge Development; To deepen the understanding of genetic and reproductive processes, to support learning and refinement of our laboratory systems and practices. (4) Regulatory Post-Marketing Surveillance – to conduct retrospective statistical analyses required by the EU In-Vitro Diagnostic Regulation (IVDR) to confirm the ongoing safety and performance of our tests. Findings may be disclosed to competent authorities and to the scientific community only in aggregated or anonymised form (e.g. conference presentations or peer-reviewed publications).

Note: These activities are not considered biomedical or scientific research. The use of your data is strictly for internal operational purposes, such as quality assurance, training, and process improvement, and does not involve the creation of new generalizable scientific knowledge, publication of research results, or use of your sample/data in external studies.

These internal uses in no way compromise the confidentiality or integrity of your personal information. Your identity cannot be accessed by the staff conducting these activities, and all processing is subject to strict safeguards.

Sample storage

Samples collected as part of Igenomix marketed services will be stored temporarily in accordance with applicable laws and standards, including ISO 15189, which ensures quality and integrity. The sample will be kept under controlled conditions (e.g., proper temperature and security) to maintain its suitability for testing.

Samples are retained only for as long as necessary for the intended purpose after which it will be securely destroyed or anonymized in a manner that protects your privacy.

Exceptions to this process shall only be made where longer retention is legally required or explicitly authorized by the individual, with their informed consent.

HAVING READ AND UNDERSTOOD THE ABOVE, I AM INFORMED OF:

The indication, procedure, probability of success, risks and complications of the proposed treatment, as well as the economic cost of such test(s). The willingness of the health personnel to expand on any aspect of the information that has not been sufficiently clarified.

I understand the explanations provided to me in clear and simple language, and the attending physician has allowed me to make all the observations and has clarified all the doubts I have raised, also stating that I can freely renounce this consent at any time.

I state that I am satisfied with the information received and that I freely give my consent for the foetal remains collected to be sent to the Igenomix facilities in order for the corresponding POC test to be performed.

I also agree that the results of the diagnosis(s) may be communicated to my physician so that he/she may advise me appropriately in the planning of future pregnancies.

1 For patients residing outside the U.S.: Customers domiciled outside the United States in certain jurisdictions may have the option to request that their personal information be removed at any time from our active databases, subject to the applicable laws and regulations of that jurisdiction. Although we may remove your personal information from our active databases, some or all of your personal information will remain archived in backup copies to comply with legal, regulatory and other requirements. Information that has already been encrypted and/or anonymized may not be retrievable or traceable for destruction, deletion or modification. If you choose to have your personal information removed from our active databases, please contact us at privacy@igenomix.com.