Name:	Individual One	**Name:**	Individual Two
Birth date:	01/01/1990	Birth date:	02/02/1992
CHN/ID:	000000	CHN/ID:	123456
Sex:	Female	Sex:	Male
Sample type:	Blood	Sample type:	Blood
Sample arrival date:	25/03/2025	Sample arrival date:	25/03/2025

Report date: 27/03/2025

PGT-M study for *HBB*

Indication

PGT-M (Preimplantation Genetic Testing for Monogenic Disorders) study from an embryo biopsy obtained during an IVF cycle. This study is indicated after obtaining a conclusive result from an informativity study performed in family members.

Results

After the analysis of 375 polymorphisms, 54 informative polymorphisms were employed to determine the embryos status successfully (Table 1).

Table 1. Results obtained from PGT-M study with PGD-SEQ kit.

Embryo ID	Embryo Status
EMB1	Carrier from Mother
EMB2	Carrier from Father
EMB3	Affected
EMB4	Non-carrier

Sample Name	State	Sex
EMB4-HBB case	Non Carrier	unknown
EMB1-HBB case	Carrier From Mother	unknown
EMB2-HBB case	Carrier From Father	unknown
EMB3-HBB case	Affected	unknown

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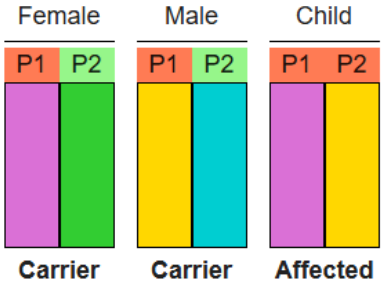
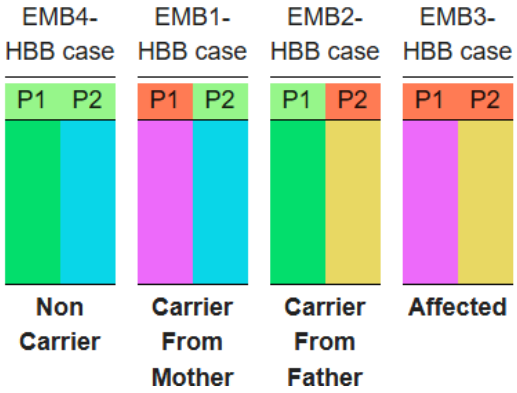
Healthy allele | Carrier allele | Likely recombinant

Family Members



Embryos



Methodology

The test is performed from a DNA sample isolated from an embryo biopsy. The DNA was amplified and after that the PGT-M study was performed using the PG-Seq™ Core Panel kit (Revvity) and the Illumina or Element Biosciences sequencing platforms by Next Generation Sequencing.

Previously to the PGT-M study, the polymorphisms surrounding the mutation(s) present in the family line are analyzed to obtain linkage information between these polymorphisms and the mutation of interest (informativity study). This information is subsequently used to determine the possible carrier, non-carrier or affected status of each embryo in an indirect manner. A direct study can be also performed additionally depending on the nature of the alteration and only if it is possible.

Name:	Individual One	**Name:**	Individual Two
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The interpretation provided in this report serves as guidance; the timing of embryo transfer is at the discretion of the clinic, including advice on the need for genetic counseling or diagnostic tests. Any test results should be interpreted in the context of all available clinical findings. The analysis of the results, including the determination of each embryo status, and report generation have been carried out using Journey Genomics software. Accuracy of the results is based on the assumption that samples received were correctly identified, family relationships are true, and the clinical diagnosis of relatives is correct.

PGT-M study limitations

- A prior informativity study has been conducted for this test, which has been classified as SUITABLE. Every PGT-M study comprises two parallel studies: a direct study (which may be optional, depending on the variant to be analyzed) and an indirect study, which is indispensable and is initiated in the informativity study.
- This test is designed exclusively to detect the variants indicated by the clinic or patients. No other variants or diseases not previously indicated are analyzed.
- The embryonic status (defined as carrier, affected, or non-carrier) refers exclusively to the disease described in the Indication section, always based on the information provided by the clinic or patients regarding the presence or absence of the alteration(s) in each of the family members participating in the study.
- This test represents only a small part of the embryo and may not necessarily represent the content of the entire embryo.
- The PGT-M study is subject to artifacts that may lead to a misclassification of the embryo's status. These artifacts mainly include mosaicism and allele drop-out; the presence of maternal or external contamination cannot be completely ruled out. The entire PGT-M study is designed to minimize the possibility of diagnostic error due to these factors, and their presence can be detected at various points in the process. These artifacts are defined as follows:
    - Mosaicism: A mosaic embryo is one that has two or more cell lines with different genetic compositions. In the case of a specific disease, a mosaic embryo would have genetic material from both alleles inherited from the affected parent.
    - Allele drop-out: During the laboratory process, since it is not possible to specifically guide the chemical reactions in each sample, one allele of the embryo may be preferentially represented over the other. In this way, only one of the alleles present in the sample would be shown, while the other would remain hidden. If this occurs with the allele carrying the alteration, the embryo could appear non-carrier while actually being a carrier. This is the main reason why a direct study alone is not recommended in PGT-M techniques, and an additional indirect study is performed.
- The sensitivity and specificity of the technique are high. The error rate has been minimized through the application of various control points throughout the process. However, according to scientific publications, there is an approximate 7% probability of error in the global PGT-M analysis. Therefore, since no technique is infallible, it is recommended to confirm the results in case of pregnancy.
- The percentages regarding the inheritance or transmission of traits or diseases are purely theoretical and involve numerous processes beyond our control. Hence, the final number of carrier, healthy, or affected embryos may not match expectations, considering these inheritance rules.

Name:	Individual One	**Name:**	Individual Two
Birth date:	01/01/1990	Birth date:	02/02/1992
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