# DONOR*ASSAY*™

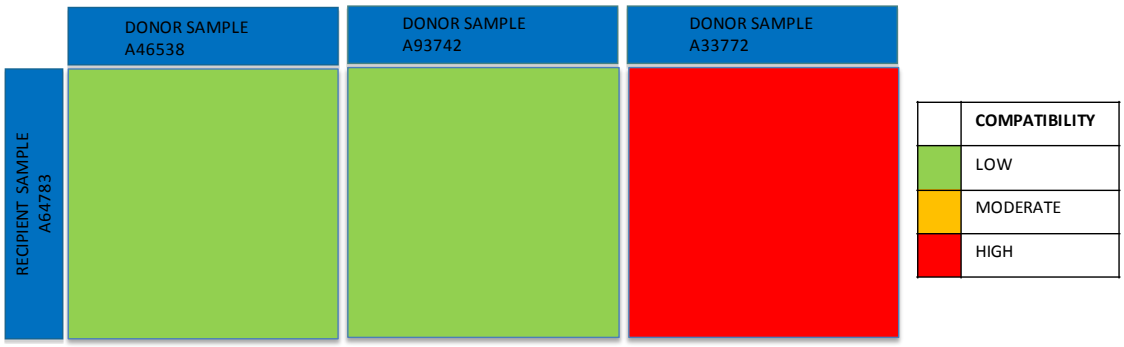
## SUMMARY

Every fertility clinic takes the health of their donors and recipients and their offspring very seriously. Both donors and recipients undertake rigorous family history enquiries and a range of tests to minimise the risk of certain diseases being passed to or inherited by any child created by artificial insemination.

But despite this, there is still a possibility that the combination of recipient and donor DNA can create a risk of the child being born suffering from an inheritable disease, if, for example, the donor has developed a new mutation in his sperm production pathways, or if both donor and recipient carry in their DNA the same single copy of a genetic mutation that, when paired, can cause a specific disease.

**DONOR***ASSAY* enables clinics offering artificial insemination by donor to undertake a comprehensive comparison of donor genetic profiles against those of intended recipients, enabling them to identify potential matches where there is a risk of producing a child with or carrying any one of 700 inherited diseases and reports it clearly as a high, medium, or low compatibility between donor and recipient (Figure 1).

*Figure 1: Example DONOR / RECIPIENT compatibility report.*




## THE ASSAY

The test searches for over 300 genetic markers in the DNA of the proposed recipient and compares them with the same set of genetic markers in the DNA of each prospective donor.

Each genetic marker is known to relate to a specific disease; the test determines whether a child conceived by artificial insemination using each donor would carry any risk of either having or carrying any one of 700 inherited diseases and if so gives a measure of the risk.

1. Samples of cheek cells, which contain DNA, are collected from the recipient by gently rubbing the inside of the mouth using up to 5 non-invasive, cotton-tipped swabs (buccal swabs).
2. A sample of semen, containing sperm, is provided for DNA analysis by each prospective donor.
3. DNA is extracted from both recipient buccal cells and donor semen samples and is analysed using next generation sequencing and interpreted using bioinformatics analysis.

## RESULTS AND REPORTING

The results of the analysis and interpretation are reported as the compatibility between recipient and each donor.
However, if preferred, and where country legal regulations allow, it can be reported as a confidential analysis of each party.

## ASSAY PATHWAY

The assay is based on the analysis of the sequence of over 300 genes and genetic markers that have been shown to be relevant to over 700 diseases, which are categorised as being dominant, recessive or mitochondrial.

The DNA sequences are compared using HEALI's decision algorithm:

I. DNA sequences are searched for specific forms of each genetic marker;

II. Marker forms relevant to diseases found in the recipient are compared with those of each donor;

III. Where matches are found, they are categorised for the disease type and inheritance profile.

IV. The matches at risk of being inherited as diseases are reported in terms of the compatibility of donors with recipient.

## CONFIDENTIALITY

The assay has been designed to be completely confidential. HEALI cannot identify the identity of either prospective donors or recipient and fertility clinics are not informed of the genetic markers of either donors or recipient.

## VALIDATION AND DEVELOPMENT

**DONOR***ASSAY* analyses genes listed in the NIH Genetic Testing Registry; it is updated continually as additional genes are validated.

HEALI welcomes collaboration with Fertility Clinics where this leads to additional validation data.

## ABOUT HEALI

HEALI is a personalised medicine company whose aim is to improve medical decision making for treatment of a range of life changing illnesses and conditions.

Its assay platform, comprising analysis of genetic markers with clinical or physical data and interpretation of the data set using its proprietary decision making algorithm, is applicable, in principle, to any condition where an individual's genetic information can be used to categorise likely outcome or most effective treatment.