FAMILY*ASSAY*™

SUMMARY

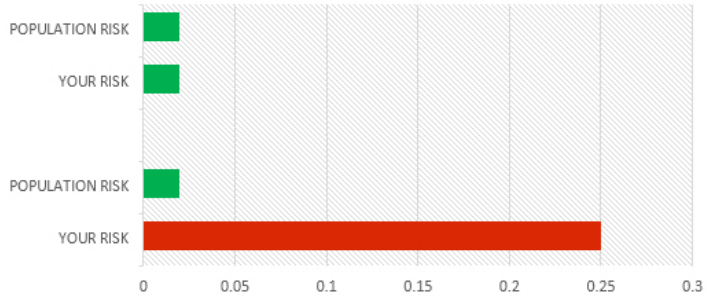
Some couples have family histories of inherited serious diseases and conditions. They want to have children but don't want them to suffer in the same way. So they want to understand their risk of passing those inherited diseases on to their offspring. Genetic counselling can help with this in some cases, but relies heavily on family history: it cannot provide a complete analysis without using detailed genetic information.

Performed **in conjunction with** genetic counselling, **FAMILY*ASSAY*™** is a genetic test, performed on both partners, that assesses the combined risk of passing those diseases to your children, by comparing fragments of your DNA against each other and against a comprehensive database of relevant genetic markers that are found in people carrying those diseases.
This informs you of the risk - IF ANY - of your children either suffering from or even carrying those diseases and so helps you, with your genetic counsellor, to make decisions as you plan your family.

THE ASSAY

The test measures and reports the risk of your child suffering from or carrying inheritable diseases that you are concerned about, by searching for and comparing genetic markers in the DNA of each partner.

Figure 1: Example DONOR / RECIPIENT compatibility report.



Each genetic marker has many forms and certain forms are known to relate to a risk of carrying or suffering from specific diseases. If the test identifies any of these “disease” forms, it will measure the risk of a child being conceived who either suffers from or carries that disease.

1. Samples of cheek cells, which contain DNA, are collected from the female partner by gently rubbing the inside of the mouth using up to 5 non-invasive, cotton-tipped swabs (buccal swabs).
2. The male partner may provide either a sample of cheek cells or a sample of semen.

IMPORTANT NOTE: Changes (mutations) occur in both men and women as they get older or if they are exposed to certain chemicals or radiation. We recommend that the male partner provides a sample of semen for analysis as it contains the most recent form of his DNA that can possibly be passed to his children.

3. DNA is extracted from recipient buccal cells and/or semen sample 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 risk of a child conceived by you suffering from or carrying any of the diseases that you are concerned about, compared with the risk found in couples of a similar age and country of origin (Figure 1).

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. Your samples are labelled by codes that are known only to HEALI and your identity will never be revealed.

VALIDATION AND DEVELOPMENT

FAMILY*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.