Our expanded carrier genetic screening package

A valuable reproductive planning tool

Most individuals carry pathogenic variants in genes that can be responsible for severe conditions, and the absence of a family history does not eliminate this risk.

The majority of individuals at risk do not have a family member with a diagnosis. Expanded carrier screening provides access to options for managing reproductive risk, particularly when carried out before conception.

Testing can be done for all couples or individuals who

- **are planning a pregnancy**
- **are in the fertility process**
- **are using a gamete donor**

LGBTQ+ FRIENDLY

An easier process for you and your patients

ONLINE ACCESS TO THE TEST HERE
without prescription
Patients can order their test directly via our website

BILINGUAL EXPLANATORY WEBINAR
Explanation of the process, test, type of results, limitations, and support for consent

SALIVA COLLECTION AT HOME
A kit will be sent to patients, who will then mail the sample collected to the laboratory

LABORATORY GENETIC ANALYSIS
The test will be carried out in a partner laboratory which will provide **the results in 3 to 4 weeks**

POST-TEST GENETIC COUNSELLING
Results will be disclosed during a telehealth consultation with a genetic counsellor

RESULTS INTERPRETATION REPORT
In addition to their results, your patients will receive a personalized report to ensure continuity of care

Reliable testing performed by an established clinical laboratory

The panel includes up to 569 recessive or X-linked genetic conditions

Panel design includes all pan-ethnic conditions included in most existing guidelines as well as severe conditions for which clinical management could be facilitated by early diagnosis.

Examples of tested conditions
Cystic fibrosis, spinal muscular atrophy, Fragile X syndrome, Duchenne muscular dystrophy, hemoglobinopathies, genetic conditions common in individuals of French Canadian or Ashkenazi Jewish origin, etc.

List of genes tested available here

The laboratory analysis
A next-generation sequencing (NGS) approach to which targeted reflex testing is added when relevant, particularly Sanger sequencing, Southern Blot, MLPA, etc.

- Detection of nucleotide polymorphism in the SMN1 gene associated with spinal muscular atrophy
- Complete sequencing of the CFTR gene associated with cystic fibrosis
- Molecular confirmation of hemoglobinopathies detected by complete blood count and hemoglobin electrophoresis
- Identification of AGG interruptions within CGG repeats in the FMR1 gene associated with Fragile X syndrome for females

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MY GENETIC HEALTH
1-844-440-5454 | info@genolife.ca | genolife.ca