

Genetic Disorders Carrier Screening Informed Consent

ZG - PGZ - 2014 V1.0

Monogenic diseases are genetic disorders caused by a single defective gene. The defective gene can be inherited from our parents or occur in ourselves during germline development. Monogenic diseases, although individually rare, have a high total prevalence because there are many of them. They are major causes of birth defect, early mortality and disability. Research has shown that each person can potentially carry on average 2.8 disease-causing mutations (0-7). Therefore, carrier testing for genetic disorders before or during pregnancy can help people to make informed decision about their reproductive health, and it is an effective way to control the prevalence of monogenic diseases.

Method for Genetic Disorders Carrier Screening: extracting DNA from the peripheral blood, and performing next-generation sequencing and bioinformatics analysis on the target gene regions. This method detects mutations in the target region with high sensitivity, specificity and reliability, and it provides a direct evidence for the onset of monogenic diseases from the genetic level.

Statement : This test is intended for the general population screening, it analyzes the coding regions of the known disease-causing genes included in this panel and thereby providing risk evaluation and a basis for genetic counseling and birth planning. This test is not intended to change the incidence of the diseases under consideration. For more information related to the disease, please consult your healthcare provider or genetic counselor.

Limitations and potential risks

- 1 The report relate only to the sample(s) tested. The results are only for the purpose of genetic mutation screening and do not represent final diagnosis. They are for clinical reference purposes only.
- 2 Re-sampling may be required at no further cost if the sample fails to meet the criteria for testing and that such failure is the result of Acts of God.
- 3 The technology used in this test is based on a standardized target region capture sequencing platform for exon sequencing. It is applicable to SNVs and within 20bp small InDels on exons and splicing regions. Also some mutations on introns and promoter regions are included. There are kinds of mutations cannot be detected by this test, including large duplication and deletion, balanced translocation, inversions, ploidy changes, uniparental disomy, methylation alterations, germ cell mosaicism, and de novo mutation of germ cell. Accuracy of this test could be affected by previous bone marrow transplant, a recent blood infusion or any other therapies that may introduce exogenous DNA into the blood sample.
- 4 Due to the limitation of the sequencing technology used in this test, a negative result does not totally exclude the possibility that you are a carrier of the disease being tested, and the test may detect mutations with unknown clinical significance.
- 5 Zentrogene / BGI DX is not responsible for any anxiety or psychological issues that may arise during the test or from knowing the results.

Informed consent

- 1 I understand the nature of this test, its expected outcome, risk, and purpose.
- 2 I agree to participate in this genetic test voluntarily and I guarantee that all information provided is true and reliable.
- 3 I understand that the test is not 100% accurate and that the risk mentioned in this report is specific only to the tested samples.
- 4 I understand that my personal information will be kept confidential; I authorize the Laboratory to use my samples and test result for not-for-profit research purposes, provided that all my personal data are removed.
- 5 I understand that this test is a carrier screening for
 - PGZ012 Genetic Disorders
 - PGZ600 Genetic Disorders
 and it is intended to assist clinicians in evaluating my reproductive risk. Further tests including prenatal diagnosis may be necessary if I am screened to be a carrier of a genetic disease and would like to know whether my offspring suffer from this disease. I will be fully responsible for the consequences if I decided not to have further tests in such circumstances.

Test applicant statement

I understand all the above information, I am willing to do the carrier screening test for

- PGZ012 Genetic Disorders PGZ600 Genetic Disorders

I agree to participate in any follow-ups and I know that I'm liable to all risks related to the test.

Test applicant/legal guardian signature : _____ Dated (dd/mm/yyyy) : _____

Relationship to the test subject (for the legal guardian) : _____

Physician statement

I have explained the details of the genetic test requested to the test applicant (or their legal guardian) regarding the test nature, expected purpose, risks and limitations. I have answered their questions (or their legal guardian's). I have obtained consent from the test applicant (or their legal guardian) for this testing.

Physician's signature: _____ Dated (dd/mm/yyyy) : _____

Printed name: _____