GeneSyte™

Genea

GENETIC SCREENING FOR COMMON TRISOMIES

Genea GPO Box 4384, Sydney NSW 2001 **p** 1300 652 687 **f** (02) 9229 6400

Informed Consent for Non-Invasive Prenatal Screen for Fetal Chromosome Aneuploidy Disease

Test Description: Screened disease

- Fetal Trisomy 21 (Down syndrome), Trisomy 18 (Edwards syndrome) and Trisomy 13 (Patau syndrome), Sex chromosome aneuploidies (including Turner syndrome).

Sample

- Maternal blood collected from the pregnant woman.

Test method

- Analysis by high-throughput next generation DNA sequencing and bioinformatics.

Limitation and risk

1. This process is a highly sensitive and specific screening test and can detect aneuploidy for chromosomes 13, 18 and 21 with sensitivities of 97.98%, 97.23% and 99.49% respectively. Aneuploidies involving the X and Y chromosomes may also be identified.
2. This screen may not identify disorders caused by the following conditions:
   A. Chromosome monosomy, tetrasomy, and trisomy other than trisomy 21, trisomy 18 and trisomy 13.
   B. Partial trisomy 21, partial trisomy 18 and partial trisomy 13.
   C. Chromosome translocation, inversion, micro-deletion and micro-duplication.
   D. Other chromosomal abnormalities, genomic abnormalities and gene mutations, including uniparental disomy (UPD), abnormal gene imprinting, and single gene disorders.
   E. Infection, drugs, radiation and other environmental factors.
3. Other factors which may affect the result of the non-invasive screen:
   A. A very small percentage of trisomy diseases are caused by mosaicism. In these cases, only a portion of the fetal cells are trisomy cells. Detecting mosaicism by high-throughput next generation sequencing is difficult because of the uncertain proportion of normal and abnormal cells and hence the proportions in the cell free DNA fraction.
   B. Detecting fetal trisomy 21, trisomy 18 and trisomy 13 of twins or multiple gestations poses some unique problems and so carries a level of uncertainty. The risk for missing a fetal trisomy case exists when applying the technology to multiple implantations

(continued over).

Acknowledgement and consent

I acknowledge that I have read the terms in the Informed Consent. I request the service of non-invasive prenatal screen for fetal chromosome aneuploidies. I shall bear the risks associated with this test. I hereby confirm that all the information I provided here is true. I agree to participate in follow-up pertaining to the test results. My results will be provided to my requesting doctor.

Patient details

Surname: _____ Date of birth: _____

Given name: _____

I, _____ request and consent to the GeneSyte™ screen.

Signature: _____ Date: _____

C. When the pregnant woman or her husband is a patient or a carrier of chromosomal disease(s), the accuracy of this screen cannot be guaranteed.

D. If the pregnant woman has undergone allogeneic blood transfusion, or transplant operation, or cell therapy or she is carrying a tumor, the accuracy of this screen cannot be guaranteed.

E. When the fetus carries chromosomal imbalances other than trisomy 21, trisomy 18 and trisomy 13, the accuracy of the screen cannot be guaranteed.

F. If the estimation of gestational age is incorrect, the accuracy of this screen cannot be guaranteed. Contact the testing laboratory to qualify the results.

G. When a low percentage of cell free fetal DNA exists in plasma, the accuracy of this test cannot be guaranteed. This occurs in a low but identifiable proportion of women. A high BMI may also impact on the percentage of cell free DNA in the plasma and reduce accuracy of the screen.

H. The non-invasive prenatal screen for fetal chromosome aneuploidies is available for all singleton pregnancies with gestational age of more than 10 weeks. Completion of the test and the final report may, at times, require up to 10 working days due to experimental procedures or for purposes of confirmation.

I. Reports may be delayed due to unexpected events, such as transportation delays. In some cases, a second sample may be requested to repeat the procedure and this may cause delay in finalising the screen.

J. Incidental findings of other chromosome aneuploidies that are not clinically validated may be reported under consultation with the referring clinician.

Other considerations

1. The procedure is offered on a screening basis only.
2. At all times it is recommended that you follow your physician's advice about your pregnancy.
3. The screen report does not suggest, nor warrant any follow-up actions, which is beyond the scope of testing and is a matter for your doctor to consult on. Genea provides the screen and bears no responsibilities over the follow-up actions and any ethical issues that may rise from the report.
4. Genea is committed to treating the result and report confidentially and will not provide it to a third party without the prior consent of the patient or the referring doctor.
5. As part of our quality assurance and teaching program we may use data obtained from this testing to further develop and publish research.
6. The remaining patient specimen/materials will be kept for an appropriate interval of time according to the government regulation guidelines. The patient agrees that a written request is not needed for such treatment of the remaining materials.

PAYMENT REMITTANCE ADVICE **GENESYTE SCREEN PRICE: $400 (AUD)**

(Please print clearly)

Patient name(s): _____

Suburb: _____ Post code: _____

Daytime contact number: _____

PLEASE COMPLETE DETAILS BELOW

Please tick your method of payment and fill out your credit card details below.

☐ Visa ☐ Mastercard ☐ American Express

Full name of Cardholder: _____

Card number: _____ Expiry: _____

Amount: _____ Signature: _____ Date: _____

ATTACH COMPLETED FORM TO PATHOLOGY REQUEST FORM AND SEND WITH PATHOLOGY SAMPLE.

Postal address: Genea CSR level 3, 321 Kent Street, Sydney NSW 2000
Genea Genetics Pty Limited ABN 82 074 545 681 atf Genea Genetics Unit Trust ABN 97 564 816 652

MKT01810 | Feb 2022