Lab Report SAMPLE

Final Report

Sample collection date: 01/07/2018 (DD/MM/YYYY)
Sample arrival date: 02/07/2018 (DD/MM/YYYY)
Reported date: 08/07/2018 11:00 am (DD/MM/YYYY)

Patient Information

Name:	CHAN YUEN MIN
HKID/Passport No:	Z123456(7)
Sex:	F
Date Of Birth:	29/07/1980
Xcelom Lab Number:	18H00001A

Referral site Information

Referral Site:	HK Hospital
Clinic ID:	12345678
Referral Doctor:	Dr LEE TAI MAN

Clinical Information from Referring Physician

Gestational Age: 13 weeks 0 days

Test Method safeT21express™

Test Result

Fetal sex	Multifetal Pregnancy Information	Total fetal fraction
Female	Singleton	5.5%

Test Results for Chromosomes 21, 18 and 13	
Negative	Increase in the amount of DNA from chromosomes 21, 18 and 13 not detected.

Test Results for Microdeletion(s)	
Negative	The test includes the search for microdeletions of at least 3Mb in size within the chromosomal regions responsible for 1p36 deletion syndrome, 2q33.1 deletion syndrome, Cri-Du-Chat syndrome, Langer-Giedion syndrome, Angelman syndrome/Prader-Willi syndrome and 22q11 deletion syndrome (include DiGeorge syndrome)

Test Results for Sex Chromosome(s)	
Negative	Quantitative aberration in DNA from chromosome X not detected.

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Signed by registered Part I MLT:

Xcelom Limited
10/F, Metropole Square, 2 On Yiu Street, Shek Mun, Sha Tin, N.T., H.K. T: 3955 1230 F: 3565 0112

Test Performance

Table 1: Test performance summarizes the detection rates and false positive rates of safeT21express™ for singleton pregnancies based on safeT21express™ internal validation data.

	Detection Rate	False Positive Rate
T21	99.65%	0.03%
T18	>99.9%	0.01%
T13	>99.9%	0.02%
Sex Chromosome Aneuploidies	99.82%	0.16%

Based on safeT21express™ internal validation data, 22q11 deletion syndrome (include DiGeorge syndrome) has 94.1176% detection rate, 0.0009% false positive rate and 99.9991% specificity. Limited data are available for other microdeletions. safeT21express™ search for microdeletions of at least 3 Mb in size as the resolution unit, therefore can only be used to detect microdeletions more than 3Mb.

Methodology of the test

DNA isolated from the maternal blood, which contains placental DNA is sequenced by shortgun sequencing technology. Sequencing data is analyzed by safeT21express™'s proprietary algorithm to determine the fetal copy number for all chromosomes including 13,18,21,X and Y, thereby identifying whole chromosome abnormalities at these locations, and the microdeletion panel will identify microdeletions at the specific loci. If a sample fails to meet the quality threshold, no result will be reported for the specified chromosome(s). The test requires sufficient fetal fraction to produce a result. Fetal fraction is determined by using a proprietary algorithm incorporating data from chromosome Y-specific[1], size-based[2] and statistical modeling-integrated count-based method[3].

Limitations of the Test

safeT21express™ is a non-invasive prenatal screening test. The test results do not provide a definitive genetic risk in all individuals. A patient with a positive test result should seek obstetricians' or clinical geneticists' advice and consider invasive prenatal diagnosis, such as chorionic villus sampling or amniocentesis, for confirmation of test results. A negative test result does not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to a number of reasons, including but not limited to mosaicism, or other causes. The referral doctor is responsible for the use of this information in the management of patients. This analysis assumes that the cfDNA tested is of the individual being tested. Despite all professional efforts to minimize confusion of patient samples and to ensure accurate test results, there remains a remote possibility of human error.
Chromosomal and sub-chromosomal findings which are more than 3Mb other than the aforementioned abnormalities might be reported. The advanced chromosomal and sub-chromosomal findings are rare and complex, insufficient validation may result in lower specificity. Consultation for advice from doctor(s) would be required for such findings (Only applicable to Advanced Panel.)
It is recommended that the test results be interpreted and correlated with other clinical findings.

Reference

1. Yu S.C.Y., *et. al.*, PLoS ONE. 2013;8(4)

2. Yu S.C.Y., *et. al.*, PNAS, 2014;111(23)

3. Kim S.K., *et al.*, Prenat. Diagn. 2015;35(810–815)

End of Report

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Laboratory No.:	18H00001A	***Sample collection date*** *(dd/mm/yyyy)* **:**	01/07/2018
Name:	CHAN YUEN MIN	***Sample arrival date*** *(dd/mm/yyyy)* **:**	02/07/2018
ID No.:	Z123456(7)	***Clinic ID:***	12345678
DOB *(dd/mm/yyyy)* **:**	29/07/1980 Sex: F	***Clinic / Hospital:***	HK Hospital
Gestational age:	13 weeks 0 days	***Referring Doctor:***	Dr LEE TAI MAN
		Sample type:	Blood

08/07/2018

safeT21express^{TM} Report Cover Letter

Dear Doctor,

The safeT21expressTM test data of this report has been reviewed by the Department of Obstetrics and Gynaecology. **The result is NEGATIVE.**
Total fetal fraction is 5.5 %.

The SafeT21express non-invasive prenatal screening test results for Trisomy 21, 18 and 13 are negative, increase in the amount of DNA from chromosome 21, 18 and 13 not detected.

The test includes the search for microdeletions of at least 3Mb in size within the chromosomal regions responsible for 1p36 deletion syndrome, 2q33.1 deletion syndrome, Cri-Du-Chat syndrome, Langer-Giedion syndrome, Angelman syndrome/Prader-Willi syndrome and 22q11 deletion syndrome (include DiGeorge syndrome). Quantitative aberration of the aforementioned regions is not detected.

The test result is consistent with a female fetus, quantitative aberration in DNA from chromosome X is not detected.

Please feel free to contact the Prenatal Genetic Diagnosis Centre, Department of Obstetrics and Gynaecology if further discussion about this case is needed.

Reviewed by: Prof. TY LEUNG & Dr. Richard CHOY
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