Department of Obstetrics and Gynaecology

PGD Lab, 4/F, Old Block (K Wing), Prince of Wales Hospital,
The Chinese University of Hong Kong
Shatin, N.T., Hong Kong SAR
Tel: (852) 3505 1557 | Fax: (852) 3505 4810 | www.obg.cuhk.edu.hk

ChromoSeq Consent Form

Patient Label

Test description:

ChromoSeq is a genetic test which applies next generation sequencing technology on long DNA fragments. Superior to FetalSeq, it can also identify chromosomal structural rearrangements and absence of heterozygosity (AOH).

Possible test result(s):

1. No clinically significant chromosomal abnormality associated with phenotype was detected
2. Clinically significant deletion(s) and/or duplication(s) (>50kb) associated with phenotype was detected*
3. Clinically significant chromosomal structural rearrangement (>100kb) associated with phenotype was detected*
4. Variant of uncertain significance (deletion/duplication and/or structural rearrangement) possibly associated with phenotype was detected*
5. Clinically significant absence of heterozygosity region(s) detected (*e.g.,* uniparental isodisomy)*

* Further genetic counseling is recommended.

Limitations of the test:

1. ChromoSeq can detect chromosomal numerical disorders, genome-wide microdeletions/duplications, chromosomal structural rearrangements, and absence of heterozygosity. However, this test cannot detect all genetic abnormalities, such as Robertsonian translocations, translocations/inversions mediated by palindromic AT-rich repeats (PATRRs), low-level mosaicisms (<20%), single nucleotide variant, small insertions/deletions, and deletions/duplications and structural rearrangement below the resolution limit of ChromoSeq.
2. Even if the ChromoSeq test result is normal, it is still possible that the fetus/proband can be affected by other genetic diseases that are not detectable by ChromoSeq.
3. Genetic testing is highly accurate. However, as with all testing, some inaccuracies may occur. Knowledge in genetics is constantly updated, therefore the interpretation of the test results may change over time.

A) I provide informed consent to undergo ChromoSeq.	Yes ☐
B) I understand that the test may possibly reveal incidental findings implicating diagnoses that are unrelated to the original indications of testing, including hereditary cancer syndrome, carrier status of autosomal recessive disorders (not reported in prenatal samples), late onset neurological disorders, etc. Such results may potentially affect Participant(s) and/or family members in terms of insurance, job and academic application, psychological and social issues.	Yes ☐
C) I consent that the contents mentioned in B) would not be provided if not opted.	Yes ☐
D) I choose to **OPT-IN** ☐ or **OPT-OUT** ☐ of knowing the contents mentioned in B) above.	

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基因檢查同意書 Consent for genetic testing

我明白基因測試的目的。 I have been informed about the purpose of this genetic test.	Yes 是☐
我明白基因測試的結果可能涉及我和家人的健康、心理或保險問題。 I understand that the results of this genetic test can involve possible medical, psychological or insurance issues for me and my family.	Yes 是☐
我明白只有上述基因/ 突變/ 基因位點會被測試。 I understand that only the above-mentioned gene/mutation/locus will be tested.	Yes 是☐
我明白我/我的孩子可能需要提供第二個樣本，以確認第一個樣本的初步測試結果。 I understand that I / my child may need to give a second sample to confirm initial finding in the first sample.	Yes 是☐
我明白在某些情況下，基因測試結果是無法確定一個可能存在的基因突變。這可能是由於目前缺乏完整的基因結構知識，或由於所使用的技術不能檢測某些類型的基因變化。此外，突變可能發生在其他已知或未知的致病基因。 I understand that in some cases, genetic testing is unable to identify an abnormality, even though an abnormality may exist. This may be due to the current lack of knowledge of the complete gene structure, or inability of the technology used to identify certain types of changes in genes. In addition, a mutation may not be detected because the mutation may occur in an alternative gene.	Yes 是☐
我明白基因測試通常是準確的。不過，正如所有測試，偏差有可能會出現。由於基因知識不斷更新，這個基因測試結果的解釋亦可能會隨時間而改變。 I understand that genetic testing is usually accurate. However, as with all testing, some inaccuracies may occur. Genetic testing is ongoing and the interpretation of the test results may change over time.	Yes 是☐
本人同意進行已向本人解釋過的基因測試。 I consent to be tested for genetic test which have been explained to me.	Yes 是☐
本人同意測試中抽取的樣本可被儲存或不具名地用作其他有關的研究。 I consent for materials from this sample to be stored / used anonymously for relevant research. 如果您不希望您的血液及DNA樣本被保留作以上用途，請在這裡劃上剔號: ☐ If you don't want your sample to be kept for these purposes please tick here: ☐	Yes 是☐
我提出的問題已經被回答。 Any questions that I have asked have been answered.	Yes 是☐

(病人/監護人姓名Name of Patient / Guardian)	病人/監護人簽名 Signature of Patient / Guardian)
(身份證 /護照號碼ID / Passport No.)	日期 Date (dd/mm/yyyy):
(醫生姓名Name of Physician)	(醫生簽名 Signature of Physician)
(部門 /醫院/診所 Department / Hospital/ Clinic)	日期 Date (dd/mm/yyyy):

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ChromoSeq 染色體測序同意書

測試說明:

ChromoSeq 染色體測序是一種基於 DNA 長片段的二代測序技術。與胎兒測序（FetalSeq）相比, 該新技術同時能檢測染色體結構異常和雜合性缺失。

Patient Label

結果將報告為:

1. 未檢測出與患者表型相關的具有臨床意義的染色體變異
2. 檢測到與患者表型相關的致病性缺失或重複（大於 50kb）*
3. 檢測到與患者表型相關的具有臨床意義的染色體結構異常（大於 100kb）*
4. 檢測到缺失或重複或染色體結構異常, 但目前其與已知疾病的關聯還未清晰 *
5. 檢測到與患者表型相關的具有臨床意義的雜合性缺失（例如單親二倍體）*

*建議進一步遺傳諮詢。

測試限制:

1. ChromoSeq 染色體測序可以檢測染色體數目異常、微缺失、微重複、結構異常及雜合性缺失。測試不能檢測所有類型的基因組變異，包括羅伯遜易位、因長片段重複序列引起的易位或倒位、低比例嵌合型（<20%）、單核苷酸突變，微小插入或缺失以及缺失、重複和結構變異等檢測分辨率以下的變異。
2. 即使報告結果正常，胎兒/患者亦有機會存在其他 ChromoSeq 染色體測序沒有涵蓋的基因變異。
3. 基因測試通常是準確的。不過，正如所有測試，偏差有可能會出現。由於基因知識不斷更新，這個基因測試結果的解讀亦可能會隨時間而改變。

A) 本人同意進行 ChromoSeq 染色體測序。 是 ☐

B) 本人明白是次基因檢驗有可能會有偶然發現，這類發現或者會與最初檢測的原因無關。有關這類發現的診斷包括遺傳性癌症、隱性遺傳病的攜帶狀況(產前樣品一般不報道)、或遲發性神經系統疾病等等。以上所述情況可能會對接受檢驗者在保險申請、工作或入學申請、心理、或社交倫理等層面構成影響和負擔。 是 ☐

C) 本人已知，如未勾選，上述 B)中內容將不會在報告中提供。 是 ☐

D) 本人選擇 **獲知** ☐ 或 **不獲知** ☐ 上述 B)中內容。 是 ☐

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我明白只有上述基因/ 突變/ 基因位點會被測試。 I understand that only the above-mentioned gene/mutation/locus will be tested.	Yes 是☐
我明白我/我的孩子可能需要提供第二個樣本，以確認第一個樣本的初步測試結果。 I understand that I / my child may need to give a second sample to confirm initial finding in the first sample.	Yes 是☐
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本人同意進行已向本人解釋過的基因測試。 I consent to be tested for genetic test which have been explained to me.	Yes 是☐
本人同意測試中抽取的樣本可被儲存或不具名地用作其他有關的研究。 I consent for materials from this sample to be stored / used anonymously for relevant research. 如果您不希望您的血液及DNA樣本被保留作以上用途，請在這裡劃上剔號: ☐ If you don't want your sample to be kept for these purposes please tick here: ☐	Yes 是☐
我提出的問題已經被回答。 Any questions that I have asked have been answered.	Yes 是☐

(病人/監護人姓名Name of Patient / Guardian)	病人/監護人簽名 Signature of Patient / Guardian)
(身份證 /護照號碼ID / Passport No.)	日期 Date (dd/mm/yyyy):
(醫生姓名Name of Physician)	(醫生簽名 Signature of Physician)
(部門 /醫院/診所 Department / Hospital/ Clinic)	日期 Date (dd/mm/yyyy):