Department of Obstetrics and Gynaecology

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ChromoSeq Consent Form

Patient Label

Test description:
ChromoSeq is a genetic test which utilizes genome-sequencing technology on large 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 detected to be associated with phenotype
2. Clinically significant deletion or duplication detected to be associated with phenotype*
3. Variation of uncertain significance (copy number change) detected possibly associated with phenotype*
4. Clinically significant chromosomal structural rearrangement detected (>100kb) to be associated with phenotype*
5. Absence of heterozygosity region(s) detected (>5Mb; uniparental isodisomy or parental consanguinity)*

* Further genetic counseling is recommended.

Limitations of the test:

1. ChromoSeq can detect chromosomal numerical disorders, microdeletions and microduplications, 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%) and single nucleotide variants.
2. Even if the ChromoSeq test result is normal, there are still possibilities that the fetus/proband is affected by other genetic diseases that are not detectable by ChromoSeq.
3. Genetic testing is usually 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.

- I provide informed consent to undergo ChromoSeq. Yes ☐
- I consent for materials from this sample to be stored and used anonymously for relevant research. Yes ☐ No ☐

_____

(Name of Patient) (Signature of Patient)

_____

(HKID / Passport Number) (Date)

_____

(Name of Doctor / Nurse) (Signature of Doctor / Nurse)

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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 長片段的全基因組測序技術。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*
3. 檢測到缺失或重複，但目前其與已知疾病的關聯還未清晰*
4. 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）*
5. 檢測到雜合性缺失（大於 5Mb；單親二倍體或存在生物學上的父母近親關係）*

*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%）及單核苷酸突變。
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。

- 本人同意進行 ChromoSeq 染色體測序。 是 ☐
- 本人同意從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。 是 ☐ 否 ☐

(患者姓名) (患者簽名)

(香港身份證 /護照號碼) (日期)

(醫生/護士姓名) (醫生/護士簽名)