Department of Obstetrics and Gynaecology

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FetalSeq v1.0 Consent Form

Test description:

The DNA from the fetus/proband undergoes library construction and sequencing to identify variants via comparing with the human reference genome. This test is able to determine whether the tested individual has any missing or extra copies of part of the chromosomes. It is important that both biological parents to submit blood sample to help the interpretation of results.

Patient Label

Test reporting:

1. No copy number change detected
2. Clinically significant abnormality detected*(known to be associated with a genetic condition)
3. Variation of uncertain significance detected in the fetus/proband*

* Further genetic counseling is recommended.

Limitation of the test:

1. FetalSeq v1.0 can detect chromosomal numerical disorders, microdeletions and microduplications. However, this test cannot detect all genetic abnormalities such as balanced translocations, inversions, uniparental disomies (UPD), low-level mosaicisms and point mutations.
2. Even if the FetalSeq v1.0 test result is normal, there are still chances that the fetus/proband is affected by other genetic diseases that are not detected by FetalSeq v1.0.
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 consent to undergo FetalSeq v1.0. Yes ☐
- I consent for materials from this sample to be stored or used anonymously for relevant research. Yes ☐

(Name of Patient) (Signature of Patient)

(HKID / Passport Number) (Date)

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

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FetalSeq v1.0 胎兒測序同意書

測試說明:

Patient Label

FetalSeq v1.0 胎兒測序可在文庫建構和測序後比較胎兒/患者的

DNA 序列信息與參考基因組以判斷染色體的數目與劑量的異常增減。

父母的樣本可以讓我們了解胎兒/患者染色體劑量異常增減的重要性。

結果將報告為：

1. 基因劑量正常
2. 基因劑量異常並與已知遺傳性疾病有關聯*
3. 基因劑量異常但目前其與已知疾病的關聯還未清晰*

*我們建議進一步遺傳諮詢

測試限制:

1. FetalSeq v1.0 胎兒測序可以檢測染色體數目異常、基因微缺失、微重複變異。測試不能檢測所有類型的基因組變異，包括染色體平衡易位、倒位、單親二倍體、低比例嵌合型及點突變。
2. 即使報告結果正常，嬰兒亦有機會存在其他 FetalSeq v1.0 胎兒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釋亦可能會隨時間而改變。

- 本人同意進行 FetalSeq v1.0 胎兒測序。 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 ☐

(病人姓名) (病人簽名)

(香港身份證 /護照號碼) (日期)

(醫生/護士姓名) (醫生/護士簽名)