沛利德®唐氏綜合症分子篩查檢測報告

Prelite Down Syndrome Molecular Screening Test Report

(唐氏症低風險報告範本)

Patient Name:	**Hospital ID:**
DOB:	**Referring Physician:**
Date Collected: 2022/ /	**Specimen: Finger Touch Exosome DNA**
Date Received: 2022/ /	**Lab No.**

Anticoagulant : EDTA
Gestation Age/LMP:

Indication/ Porosity:

RESULTS

C21*orf*105 Epialleles	Epialleles AX1	Epialleles A50	Epialleles B321	Epialleles B333	**Total**
Result	UP	UP	DOWN	DOWN	（N）= 2

The test uses four specific probe duplex derived from the open reading frame sequence of the C21orf105 locus to target four major epigenetic alleles of the C21orf105 locus present in the maternal peripheral blood during midterm pregnancy. The probes specific to the four C21orf105 epigenetic alleles are separately impregnated in each tube of the four-well strip tube. The probe helix in its duplex form can be activated by adopting a transient paranemic configuration at an oil-water interfacial surface. Once the activated probe duplex is triggered into interfacial crossover chain hybridization (iCCH) by a compatible sample, a nucleic acid conglomer with intermingled phosphoribose chains will be formed, which in turn will release hydrogen ion into the aqueous phase. The change of acidity in the aqueous phase will disrupt the pH-sensitive molecular lattice of oil and cause the buoyant density of the oil phase to reverse. The test result can thus be determined visually by inspecting the density reversion of the oil phase in the reaction tube.

INTERPRETATION

本篩查檢測採用雙液相界面遞增檢測方法(BLICH)對孕婦外週血中 C21*orf*105 表觀等位序列(Epialleles) 進行平行檢測,適用於妊娠早期及中期（11-20 孕週）唐氏綜合症的篩查. 樣品測試結果根據下表決定:

結果	條件
高風險	**數量（N）= 4**
低風險	**數量（N）< 4**

N is the number of tubes with aqueous phases located at the bottom of the tube.

本試劑盒經 1000 例臨床試驗，結果如下：

結果	21 三體[4]	正常 [996]
高風險	4	1
低風險	0	995

① 靈敏度：> 99%；
② 特異性：> 99%；
③ 精密性：> 99%；
④ 重覆性：> 99%。

EXTREMELY LOW RISK FOR DOWN'S SYNDROME PREGNANCY

The profile of C21*orf*105 epiallele markers found in the patient's blood is consistent with that of a normal pregnancy free of trisomy 21 risk.

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本檢測结果顯示受檢者為 21 三體低風險，其唐氏妊娠風險趨近零。
受檢者血液中的 C21*ori*105 表觀等位基因標誌物的多點形式，與正常妊娠無 21 三體風險表現的形式一致。

RECOMMENDATION

- 試驗结果顯示的高風險或低風險是提示 21 三體綜合症發生的概率，概率高，則可能性大；低風險的結論，並不能完全排除患有 21 三體綜合症及染色體異常的可能性，該結果只是表明與其他胎兒相比，患有 21 三體綜合症胎兒的機會相對較低。高風險結論時，建議做進一步產前診斷。
- 本**分子檢測**的預期用途為產前篩查，提供孕婦的醫學信息僅為風險概率，而非胎兒疾病的診斷。故高風險結論不一定是 21 三體綜合症妊娠，而低風險結論也不一定無 21 三體綜合症妊娠風險。

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Sign: *BING LING, MD* Date: 2022/ /