



敏兒安 safeT21 express

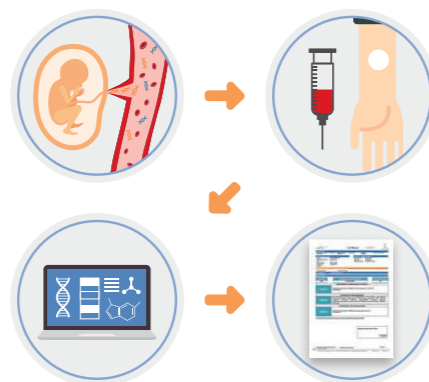
無創性胎兒DNA產前篩檢法



結合香港中文大學科研 香港首創無創性產前檢查技術

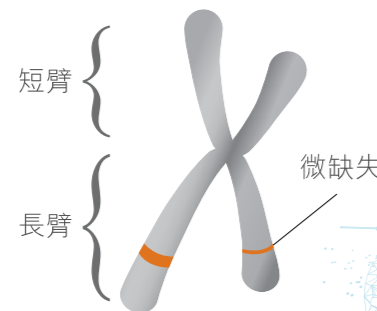
檢測技術

敏兒安 safeT21 express 是一項高敏性胎兒染色體疾病檢測，採用現有專利的無創性產前檢測技術，對媽媽和胎兒的游離DNA進行高通量測序，並將測序結果進行生物信息分析，從中得知胎兒染色體的遺傳信息。透過敏兒安 safeT21 express 可檢測「唐氏綜合症」(T21)、「愛德華氏綜合症」(T18)、「巴陶氏綜合症」(T13)、性染色體異數相關資訊及微缺失或微重複症候群。相較於傳統篩查方法如量度胎兒頸皮或計算媽媽年齡等可靠及準確，所檢測的項目亦相對較多。



微缺失的原理

微缺失是指染色體上微小的缺失片段，需要利用較高精細度的檢測儀器去發現，而敏兒安 safeT21 express 所採用的高通量測序技術，可準確地檢測微小至3Mb的微缺失。



產前檢測比較

	敏兒安 safeT21 express	OSCAR (度頸皮)	染色體核型分析 / 熒光原位雜交 / 基因晶片	
檢測類別	篩查	篩查	診斷性測試	
抽取樣本	血液	血液	絨毛球	羊水
檢測孕週	≥10週 雙胞胎≥12週	11-14週	11-13週	16-22週
導致流產率	無流產 風險	無流產 風險	0.1-0.2%	0.1-0.2%
報告時間 (工作天)	大約5天	1-2天	7-15天	7-15天
測試項目				
染色體三體症	✓	✓	✓	✓
性染色體相關疾病	✓		✓	✓
微缺失症候群	✓		✓	✓



敏兒安 safeT21 express 比傳統早孕期唐氏綜合症篩查準確度更高，有助減少30%香港孕婦因假陽性結果而要進行入侵性程序如抽絨毛或抽羊水的機會^{1,2}，降低孕婦承受額外的流產風險。



標準版檢測項目

染色體三體症

染色體疾病	準確率	病徵可能包括
T21唐氏綜合症	99.65%	先天性心臟/器官缺陷、聽力/視力問題及智力障礙。
T18愛德華氏綜合症	>99.9%	先天性心臟/大腦/器官嚴重缺陷及嚴重智力障礙。嬰兒較少能存活1年以上。
T13巴陶氏綜合症	>99.9%	多種先天性嚴重缺陷及嚴重智力障礙。嬰兒較少能存活1年以上。

性染色體相關疾病

染色體疾病	病徵可能包括
X0 X染色體單體症 (特納綜合症)	心臟缺陷、高血壓、脊椎側彎、聽力/學習障礙。相對於同齡女性，患者較多個子矮小及較遲踏入青春期，而成年患者較多出現不孕情況。
XYY 三體綜合症 (XYY 超雄綜合症/雅各氏綜合症)	腦癱、哮喘、肌肉張力較弱、肌肉/語言發展遲緩、學習障礙、社交困難、情緒失調、行為問題、自閉症及專注力不足。相對於同齡，患者身高高於平均水平。
XXY 柯林菲特氏綜合症	性器官發育不良、肌肉張力較弱及語言發展遲緩。相對於同齡，患者身高高於平均水平，而成年患者較多出現不孕情況。
XXX 三體綜合症 (XXX 超雌綜合症)	學習障礙。相對於同齡，患者身高高於平均水平，而成年患者較多出現不孕情況。

性染色體相關疾病分析準確率：99.82%

微缺失症候群

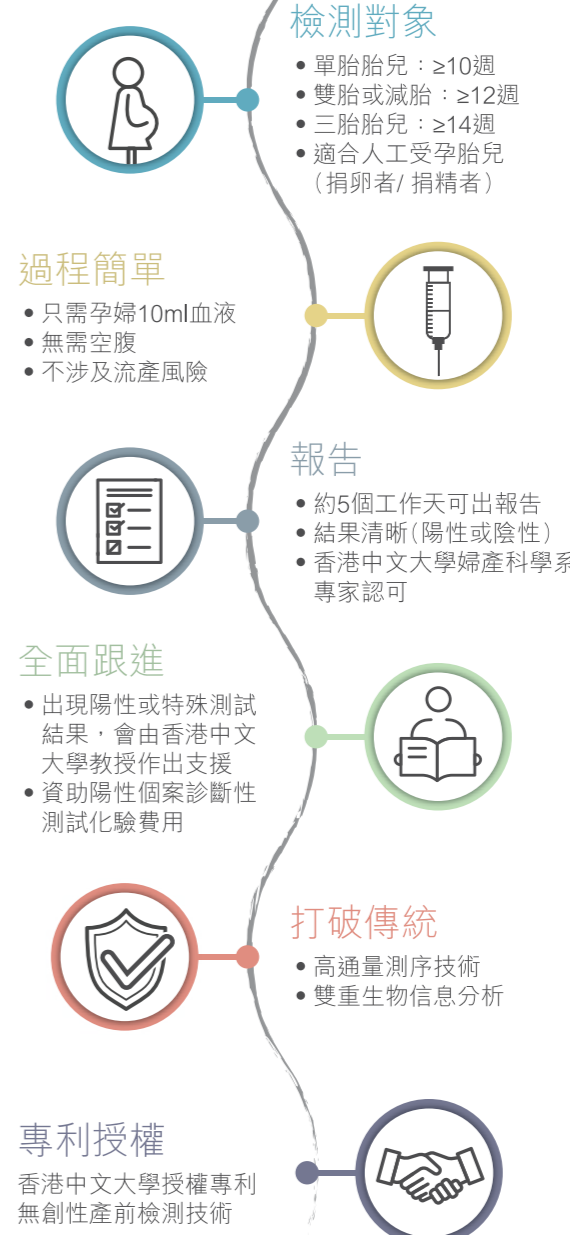
染色體疾病	病徵可能包括
1p36缺失綜合症	心臟/其他先天性缺陷、肌肉張力弱、智力障礙及行為問題。約一半患者會有腦癱症。
2q33.1缺失綜合症	生長發育遲緩、嚴重營養困難、髭裂及行為問題。
15q11.2缺失綜合症/天使綜合症	生長發育遲緩、腦癱問題、嚴重智力障礙、語言/平衡/行動障礙。
5p缺失綜合症/貓哭綜合症	餵養/呼吸困難、頭小、出生體重輕及肌肉張力弱、中度至嚴重智力障礙。
22q11.2缺失綜合症/迪喬治綜合症*	心臟缺陷、免疫系統/腎臟問題、餵養困難、腦癱症及輕度至中度智力障礙。
8q24.1缺失綜合症/毛髮-鼻-指骨綜合症	鼻鬆軟呈球莖狀、長的上唇/人中、頭髮稀疏、骨骼異常及輕度智力障礙。相對於同齡，患者較多個子矮小。
15q11.2缺失綜合症/普瑞德威利綜合症/小胖威利綜合症	餵養困難、肌肉張力弱及智力障礙。部分患者於兒童及成長期食慾大增，或有痴肥問題。

*迪喬治綜合症分析準確率：94.1%

*敏兒安 safeT21 express 只能檢測微小至3Mb長度的微缺失及微重複，而上述的微缺失卻有機會小於3Mb。由於部分微缺失的發生率較低，因此未獲大型數據驗證。



服務簡介



關於雅士能

雅士能基因科技有限公司自2014年成立，提供獨家授權專利的全港首創無創性產前檢測服務。此技術源自香港中文大學創新的科研成果，並獲得世界各地相繼採用。由創立至今，雅士能藉著不同的基因檢測、先進的實驗室設備、專業的化驗技術及人員，獲得大眾信任。未來雅士能將秉持嚴謹、誠懇的態度，專注提供高品質服務，同時透過基因科技及研發新技術，致力帶動更多更全面的分析和檢測，繼續守護大眾健康。

關於無創性胎兒DNA產前篩檢法

敏兒安 safeT21 express 由香港中文大學團隊研發，採用最新的基因分析科技以確保高準確率及安全性。與傳統入侵性測試相比，此技術準確率高及能更早得悉結果，並不涉及流產風險，從而減低孕婦及其家屬的心理壓力。現時，雅士能基因科技有限公司是唯一獲得香港中文大學專利授權於本港進行測試的機構。

- 參考文獻：
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safe|21^{express}

Non-invasive analysis of fetal DNA for prenatal DNA screening



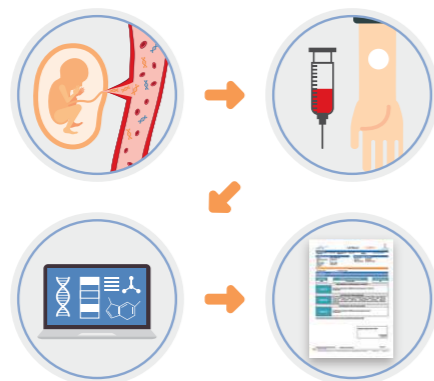
safe|21^{express} belongs to you and me



Non-invasive Prenatal Testing Pioneered in Hong Kong
Exclusive Patent Licensing from CUHK

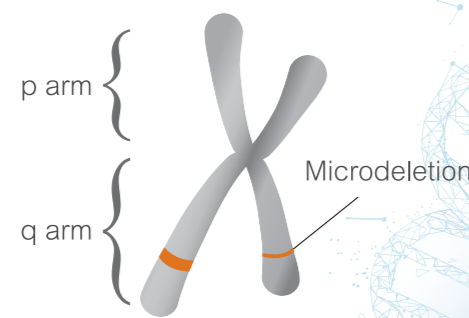
The Test and Technology

safe|21^{express} adopts the latest and patented Non-invasive Prenatal Testing Technology (NIPT) for the screening of fetal chromosomal aneuploidies. The test utilizes Next Generation Sequencing followed by bioinformatics analysis on both maternal DNA and cell free placental DNA found in maternal blood. The detection of fetal chromosomal aneuploidies including Down Syndrome (T21), Edwards Syndrome (T18), Patau Syndrome (T13), Sex Chromosome Aneuploidies and microdeletion/microduplication all in ONE test is now made possible. Compared to the traditional screening methods based on nuchal translucency or maternal age, safe|21^{express} is more sensitive, reliable, accurate and informative.



Microdeletion Syndrome

Microdeletion syndromes are disorders that are caused by deletion of a small chromosomal segment spanning several genes that is yet too small to be detected by conventional cytogenetic methods. It can only be detected by advanced technology with high resolution. The safe|21^{express} use of Next Generation Sequencing enables the accurate detection of microdeletions with a minimum size of 3Mb.



Prenatal Tests Comparison

	safe 21 ^{express}	OSCAR	Karyotyping Analysis / FISH / CMA	
Test Type	Screening Test	Screening Test	Diagnostic Test	
Sample Type	Blood	Blood	Chorionic Villus	Amniotic Fluid
Gestational Age	≥Week 10 Twins ≥Week 12	Week 11-14	Week 11-13	Week 16-22
Miscarriage Risk	No	No	0.1-0.2%	0.1-0.2%
Turnaround Time (Working Days)	About 5 days	1-2 days	7-15 days	7-15 days
Testing Items				
Trisomies	✓	✓	✓	✓
Sex Chromosome Aneuploidies	✓		✓	✓
Microdeletions Screening	✓		✓	✓



Compared to the traditional Down Syndrome screening, safe|21^{express} has a higher accuracy. This has reduced the need for invasive procedures (performed in Hong Kong) such as Chorionic Villus Sampling (CVS) or Amniocentesis (Amnio) by almost 30%^{1,2}, minimizing the unnecessary risk of miscarriages.

-30%

Standard Panel Testing Items

Trisomies

Chromosomal Disorders	Accuracy	Symptoms may include
T21 Down Syndrome	99.65%	Birth defects of the heart or other organs. Hearing or vision problems. Intellectual disability.
T18 Edwards Syndrome	>99.9%	Severe birth defects of heart, brain and other organs. Severe intellectual disability. Life expectancy of less than 1 year.
T13 Patau Syndrome	>99.9%	Many serious birth defects. Severe intellectual disability. Life expectancy of less than 1 year.

Sex Chromosome Aneuploidies

Chromosomal Disorders	Symptoms may include
X0 Monosomy X (Turner Syndrome)	Heart defects. High blood pressure. Scoliosis. Hearing problems. Learning disabilities. Shorter than average in height. Delayed puberty. Infertility.
XXY Syndrome (Jacob's Syndrome)	Epilepsy and Asthma. Hypotonia. Delayed development of motor skills and speech skills. Learning disabilities. Social difficulties. Emotional disorder. Behavioral problem. Autism and insufficient concentration. Taller than average in height.
XXY Klinefelter Syndrome	Impaired development of the sexual organs. Hypotonia. Delayed speech development. Taller than average in height. Infertility.
XXX Triple X Syndrome	Learning disabilities. Taller than average in height. Infertility.

Accuracy of Sex Chromosome Aneuploidies: 99.82%

Microdeletion Syndromes

Chromosomal Disorders	Symptoms may include
1p36 Deletion Syndrome	Heart and other congenital abnormalities. Hypotonia. Intellectual and behavioral problems. Half of them also suffer from epilepsy.
2q33.1 Deletion Syndrome	Delayed growth. Significant feeding difficulties. Cleft palate. Behavioral problems.
15q11.2 Deletion/Angelman Syndrome	Delayed growth. Epilepsy. Severe intellectual disability and speech impairment. Problems with balancing and movement.
5p Deletion/Cri-du-chat Syndrome	Feeding and breathing difficulties. Smaller head size, low birth weight, and hypotonia in infancy. Moderate to severe intellectual disability.
22q11.2 Deletion/DiGeorge Syndrome*	Heart, immune system and kidney abnormalities. Feeding difficulties. Epilepsy. Mild to moderate intellectual disability.
8q24.1 Deletion/Langer-Giedion Syndrome	Characteristic craniofacial appearance includes a bulbous nose, an elongated upper lip, a long philtrum and sparse hair. Bone abnormalities. Mild intellectual disability. Shorter than average in height.
15q11.2 Deletion/Prader-Willi Syndrome	Feeding difficulties. Hypotonia. Intellectual disability. Some may develop an insatiable appetite and obesity in childhood.

*Accuracy of DiGeorge Syndrome: 94.1%

*Since microdeletions are rare, limited data is available for validating the detection rate of most microdeletions. Microdeletions may occur in less than 3Mb in size, safe|21^{express} only searches for microdeletions with a minimum size of 3Mb.

Our Services



Suitable for

- Singleton: ≥10 Weeks
- Twins/Fetal Reduction: ≥12 Weeks
- Triplets: ≥14 Weeks
- IVF Pregnancy (Egg/Sperm Donor)

Non-invasive Procedure

- 10ml Blood
- Fasting is unnecessary
- No miscarriage risk



Report

- Around 5 working days
- Positive / Negative results
- Endorsed by CUHK Obstetrics and Gynaecology medical professionals

All rounded care

- Positive or special cases will be supported by CUHK professors
- Sponsorship for positive cases invasive diagnostic test laboratory test fee



Breakthrough Technology

- Next Generation Sequencing
- Combined Count-and-Size-Based bioinformatics

Exclusive Licensee

Exclusive patent from the Chinese University of Hong Kong for the provision of NIPT services in HK



About Xcelom

Founded in 2014, Xcelom Limited is the exclusive licensee and provider of Non-invasive Prenatal Testing (NIPT) services in HK. The technology, an innovation by a world-renowned university research team in Hong Kong, has now been adopted worldwide for an improved screening of fetal chromosomal abnormalities.

Since our establishment, Xcelom has gained extensive support from the public for the services offered by our team of experts and state of the art laboratory. Xcelom will strive to improve and expand the spectrum of services offered, to continue to provide well-rounded care for our client's health.

About Non-invasive Prenatal Testing

safe|21^{express} was invented by a research team from the Chinese University of Hong Kong. It adopts the latest genomics technology, which enables highly accurate and safe prenatal testing. Compared to traditional screening methods, safe|21^{express} is more sensitive, reliable, accurate and informative, thereby reducing the need for unnecessary invasive procedures, minimizing the emotional stress expecting mothers and their families might have to undergo.

Xcelom Limited is the exclusive licensee of the NIPT patented service from the Chinese University of Hong Kong.

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