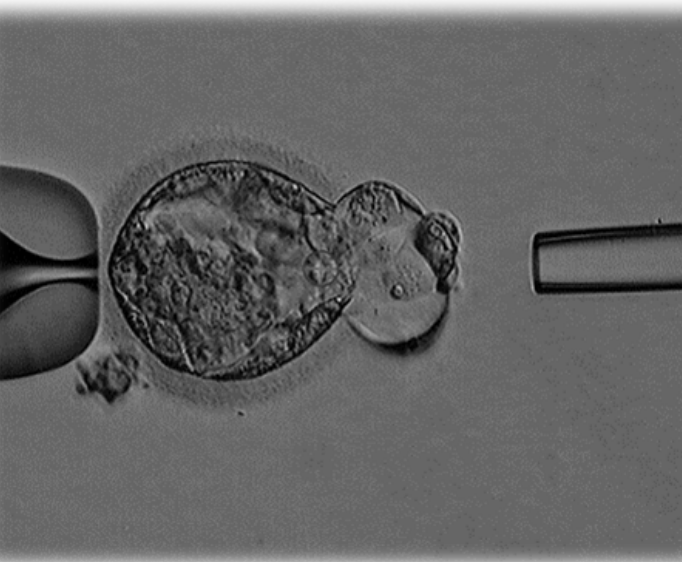


## 香港中文大學 婦產科學系

Department of Obstetrics & Gynaecology  
The Chinese University of Hong Kong



# Preimplantation Genetic Testing (PGT) 胚胎植入前遺傳學檢測

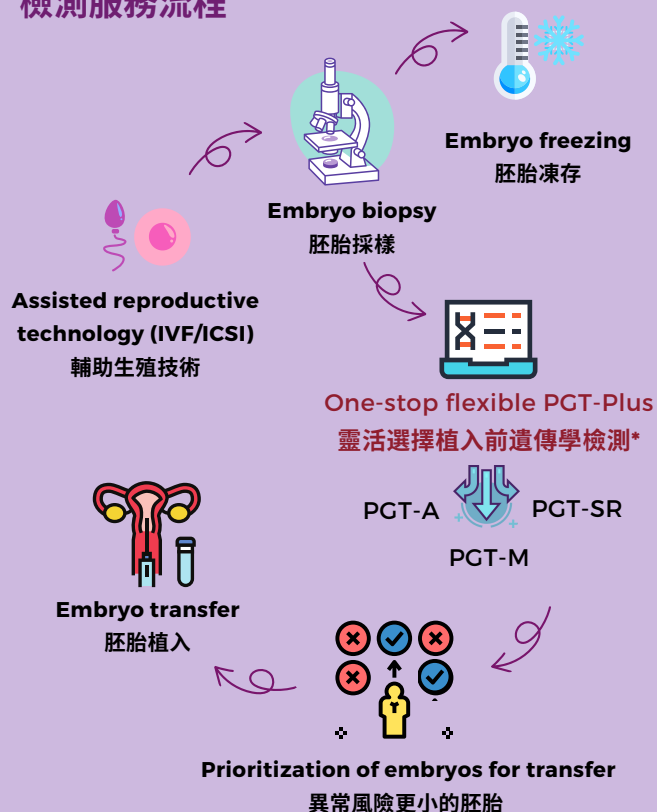
香港中文大學婦產科學系研發全新一站式PGT-PLUS平台。為胚胎植入前遺傳學檢測提供靈活而全面的選擇。

CUHK developed an all-in-one PGT-plus platform. A new one-stop flexible PGT test to be considered prior to embryo transfer.

PGT License No: T3004



## PGT Workflow 檢測服務流程



### \*報告週期 (Turn-Around-Time)

PGT-A: 10 個工作日 (working days)

PGT-M/-SR: 15 個工作日 (working days)

## 聯繫我們 Contact Us

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<https://ivfhk.com/en/pre-implantation-genetic-testing>



## PGT-PLUS

### 一站式全面檢測服務，靈活自選

#### • 植入前染色體數目異常檢測(PGT-A)

染色體數目異常；≥4Mb的染色體片段拷貝數異常；≥30%的染色體嵌合；三倍體；單親同二體。

提高反覆著床失敗/習慣性流產患者的着床/懷孕率  
減低習慣性流產患者早期流產風險  
減低出生缺陷風險  
提高健康嬰孩活產機會  
植入一個正常胚胎，減低植入多胚胎的多胎妊娠風險

#### • 植入前單基因遺傳病診斷(PGT-M)

- 針對性檢測單基因疾病。

#### • 植入前染色體結構異常檢測(PGT-SR)

- 檢測染色體非平衡性重排。  
- 識別攜帶平衡易位的胚胎。

選擇沒有單基因疾病或染色體結構異常（及攜帶者）的胚胎植入子宮，從而減少家族性遺傳病的傳遞風險。



### 本檢測適用於

- 高齡女性(35歲或以上)
- 復發性流產
- 反覆著床失敗
- 男性不育症
- 單基因疾病攜帶者或患者(PGT-M)
- 染色體平衡易位攜帶者(PGT-SR)



### 樣本要求

	胚胎樣本	父母樣本	先證者樣本
PGT-A	✓		
PGT-M+PGT-A	✓	✓	✓
PGT-SR <sup>1</sup> +PGT-A	✓		
PGT-SR <sup>2</sup> +PGT-A	✓	✓	✓

1. 檢測染色體非平衡性重排
2. 檢測染色體非平衡性重排並區分攜帶與非攜帶平衡易位的胚胎

## Test fee 檢測費用

### PGT-A

每個胚胎 HK\$ 3,000

Per embryo\*

\*無胚胎起始檢測數限制

\*No minimum embryo number is required

### PGT-M & PGT-SR

起始4例胚胎 HK\$ 35,000

For the first four embryos

每增加一例 HK\$ 3,000

For each additional one

### 其他費用 Others

基礎化驗費用 HK\$ 2,500

Basic Laboratory fee for PGT

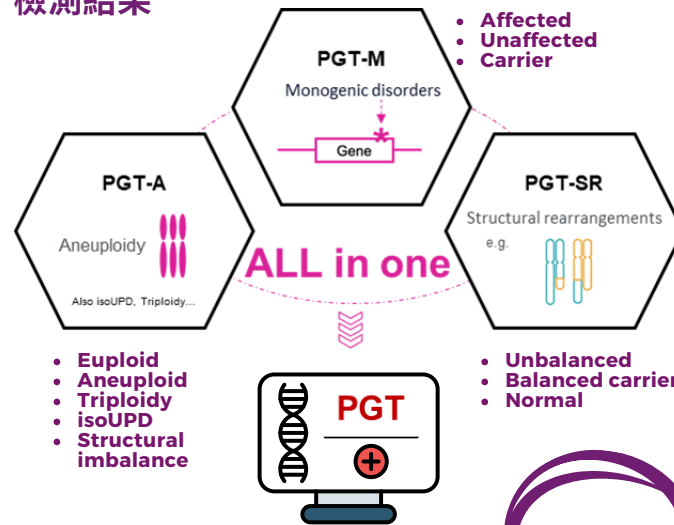
胚胎採樣費用 HK\$ 5,500

Embryo Biopsy fee

基因遺傳學診斷預實驗費用 HK\$ 5,000

PGT-M/-SR workup fee

## Possible results 檢測結果



## How does PGT improve IVF success PGT 如何提高輔助生殖成功率

- When an embryo carries an incorrect number of chromosomes, it often fails to implant or results in early miscarriage. Some chromosomally abnormal pregnancies can survive to term but may lead to abnormal live birth. For example, Down syndrome (Trisomy 21). Besides numerical abnormalities, other genetic alterations include polyploidy, genome-wide uniparental isodisomy can be associated with imprinting defects and molar pregnancies. PGT screens embryos to identify those that are likely to contain the correct number of chromosomes with normal biparental inheritance.
- 胚胎細胞內的染色體數目不正確通常會植入失敗或導致早期流產。有些染色體異常的胚胎可以存活到足月，但可能導致出生缺陷。例如，唐氏症(21號染色體三體)。除了數目異常外，其他遺傳變異包括多倍體、全基因組單親同二體可與印跡缺陷和葡萄胎相關。PGT可輔助篩選更有可能包含來源自雙親的染色體數目正確的胚胎。



## One-stop flexible PGT-Plus

### Preimplantation Genetic Testing for Aneuploidies (PGT-A)

- Gains or losses of chromosomes (aneuploidy) and large chromosome segments of  $\geq 4$  million base pairs (Mb) in size
- $\geq 30\%$  chromosomal mosaicism
- Triploidy & Uniparental isodisomy (isoUPD)

### Preimplantation Genetic Testing for Monogenic Disorders (PGT-M)

- Detection of targeted monogenic disorders

### Preimplantation Genetic Testing for Structural Rearrangements (PGT-SR)

- Reduces the likelihood of transferring an embryo with unbalanced chromosomal rearrangement
- Balanced translocation carrier can be detected

## Advantages of PGT

- Improve implantation rate in those with recurrent implantation failure
- Reduce the subsequent miscarriage risk in couples with recurrent pregnancy loss
- Reduce risk of birth defects
- Reduce risk of multiple pregnancies by single embryo transfer (SET)
- Against known familial/targeted genetic mutations and structural rearrangements for intrauterine transfer.

## Limitations 技術局限性

- PGT-A: It cannot detect sub-microscopic abnormalities less than 4 Mb. In addition, mosaicism may lead to the PGT-A result not being representative of the embryo. 無法檢測小於4Mb的亞顯微異常。此外，嵌合現象可能導致PGT-A結果無法代表胚胎。
- PGT-M/-SR: Misdiagnosis due to amplification bias, allele drop-out, and recombination can never be ruled out. Prenatal diagnosis via amniocentesis is strongly recommended if clinical pregnancy is achieved. 不能排除擴增偏倚、等位基因缺失和重組導致的誤診。強烈建議成功受孕後透過羊膜穿刺術行產前診斷。



## The test is suitable for

- Advanced maternal age (age 35 or above)
- Recurrent miscarriages
- Recurrent implantation failure
- Severe male factor infertility
- Carriers or affected individuals of monogenic disorders (PGT-M)
- Carriers of a balanced chromosome translocation (PGT-SR)



## Sample requirement

	Embryo biopsies	Parental samples	Proband samples
PGT-A	√		
PGT-M+PGT-A	√	√	√
PGT-SR <sup>1</sup> +PGT-A	√		
PGT-SR <sup>2</sup> +PGT-A	√	√	√

- For the detection of genomic imbalance
- For the detection of genomic imbalance and distinguish carriers and noncarriers of balanced translocation