

PGT 的流程

進入試管嬰兒療程後，當胚胎發展到第五至第六天達至囊胚階段，胚胎專家會在胚胎中抽取4-8個細胞作基因分析，而其胚胎會被冷凍保存。由於囊胚細胞數目多達百個以上，因此活檢並不會對胚胎發展構成重大影響。一般而言，診斷結果需時約2-4星期。報告內的正常胚胎可作移植之用。

How is PGT carried out

At day 5/6 of development, the blastocyst has over a hundred cells and removing 4-8 cells has only a small impact. The biopsied embryo(s) will be cryopreserved while awaiting test outcome. Depending on complexity, the results will be available within 2-4 weeks.

PGT 的適應症

通常，以下5類患者可能會受益於PGT技術：

- 年齡較大的女性 (35歲或以上)
- 慣性流產的女性
- 多次著床 / 受孕失敗的女性
- 單基因遺傳病攜帶者
- 染色體結構異常攜帶者

Who may benefit from PGT?

In general, there are 5 main groups of patients that may benefit from PGT:

- IVF in advanced female age - 35 years old / above or older
- Patients that have had recurrent miscarriages
- Repeated IVF failure
- Carrier of inherited genetic diseases
- Carriers of chromosomal translocations

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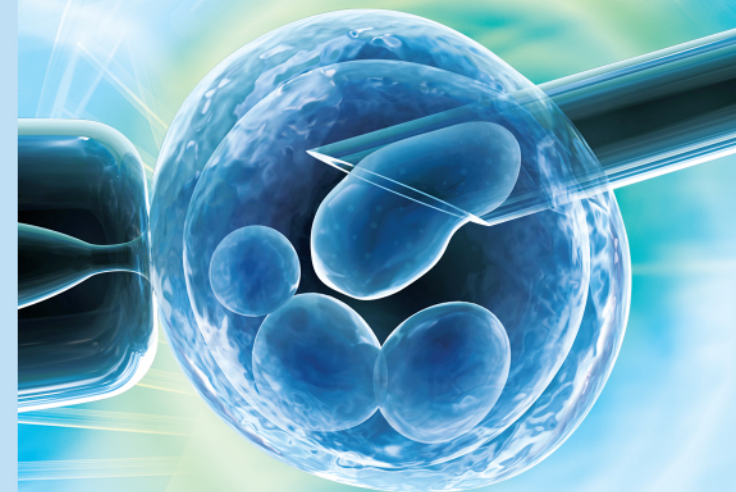
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港鐵 MTR

港鐵尖東站 K 出口，步行約1分鐘
East TST MTR Station (Exit K), approx. 1-minute walk

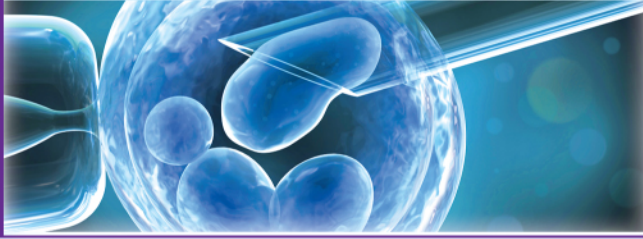
港鐵尖沙咀站 E 出口，步行約3分鐘
TST MTR Station (Exit E), approx. 3-minute walk



胚胎植入前 基因診斷 PGT

胚胎植入前基因診斷

Pre-implantation Genetic Test



簡介

胚胎植入前基因診斷 (PGT) 是一項配合體外受精 (IVF) 的臨床檢測服務，用於檢測胚胎在植入前是否帶着單基因遺傳病或染色體異常。目前有兩種檢測技術，分別是非整倍體性胚胎 (PGT-A) 和單基因遺傳疾病或染色體結構變異 / 重排 (PGT-M/SR) 植入前基因診斷以避免移植有該基因的胚胎遺傳給下一代。

Introduction

Pre-implantation genetic testing (PGT) is a test performed on embryo(s) to identify single gene disorders or chromosomal abnormalities. Pre-implantation Genetic Testing for Aneuploidy (PGT-A) uses complete chromosome studies to evaluate the total number of chromosomes in a single cell and determines any gain or loss of chromosomes. Pre-implantation Genetic Testing for monogenic defects (PGT-M) is used to screen for monogenic disorders. Moreover, Pre-implantation Genetic Testing for structural rearrangement (PGT-SR) is used to screen for chromosomal rearrangements such as translocation or inversion. These screening tests prevent transfer of embryo(s) carrying that particular monogenic disorder and/or structural rearrangement, and avoid the need for termination of pregnancy related to that disease.

PGT-A 非整倍體胚胎植入前基因診斷

非整倍體胚胎植入前基因診斷是利用染色體分析儀來篩查單一個胚胎細胞內的染色體數目有否增多或缺少。由於高齡及有慣性流產史的患者有較高機率出現染色體數目異常，因此技術主要適用於高齡婦女、慣性流產、進行多次著床或受孕失敗、染色體三倍體胎兒受孕歷史的女性、或男性嚴重精弱。研究顯示，非整倍體胚胎植入前基因診斷對多次著床失敗的女性，可能有效地提高著床 / 臨床妊娠率和活產的機會率*。

PGT-A Diagnostic Test

PGT-A involves screening the embryo for normal chromosome copy number. The main indications for the use of PGT-A in IVF treatments include advanced maternal age (AMA), repeated implantation failure, and recurrent pregnancy loss (RPL). It is well known that the rate of chromosome abnormalities is higher in patients with AMA and RPL. Also, PGT-A has been used in women with previous trisomic conceptions; partners with severe sperm defects and improve implantation in those with repeated implantation failure*.

PGT-M 單基因遺傳疾病植入前遺傳學檢測

單基因遺傳疾病植入前遺傳學檢測是在進行胚胎移植前，從體外受精的胚胎裏抽取數個細胞進行基因分析，從而篩選出沒有遺傳缺陷的胚胎進行移植。PGT-M的優點是透過此基因診斷技術，攜帶致病基因的父或母可以避免把疾病遺傳給下一代，也降低了因反覆懷有罹病的胎兒而選擇性人工終止妊娠的需要。

PGT-M Genetic Test

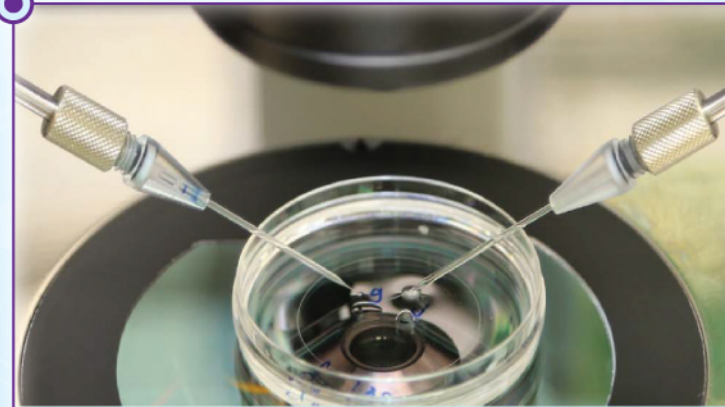
PGT-M involves removing a few cells from an IVF /ICSI embryo(s) to test for a specific genetic condition before transferring the embryo to the uterus. PGT-M for inherited genetic diseases can clearly benefit couples that carry a gene for a serious medical condition likely to affect their future children. It avoids selective pregnancy termination due to the inheritance of the maternal or paternal diseases.

PGT-SR 染色體結構重排植入前遺傳學檢測

除此之外，染色體結構重排植入前遺傳學檢測適用於染色體結構變異（如平衡易位、羅伯遜易位）攜帶者。他們的胚胎一般有較高機率出現染色體異常，而導致流產或發育有缺陷的胎兒。因此通過染色體結構重排植入前遺傳學檢測，能篩選出沒有染色體缺陷的胚胎進行移植，從而大大降低攜帶者的流產率和誕下有缺陷嬰兒的風險。

PGT-SR Genetic Test

PGT-SR can benefit patients that are carriers of a chromosomal arrangement called a balanced translocation. Embryos from these couples have a high percentage of chromosomal abnormalities. These embryos are at very high risk for miscarriage or could result in the birth of a child with birth defects. By having IVF aneuploidy screening and transfer of chromosomally normal embryos, the risk for miscarriage and birth defects is greatly reduced.



* 以上資訊仍待隨機對照試驗驗證。
Please note that these are yet to be confirmed by randomized controlled trials.