Department of Obstetrics & Gynaecology

REPRODUCTIVE MEDICINE

Preimplantation Genetic Diagnosis (PGD)

What is PGD?

PGD, as offered by Department of O&G (CUHK), is a diagnostic test performed on embryos to identify single gene disorder/ chromosomal abnormalities before implantation. PGD is used to test for a specific monogenic disorder and avoid the passing of the genetic disease to the offspring. The main advantage is that it avoids selective pregnancy termination as this method makes it likely that the embryo transferred will be free of the disease under consideration.

Who is suitable for PGD?

You are eligible for PGD if:

- You have family history of a genetic disease and
- You and/or your partner is a carrier of a genetic disorder
- You or your partner is a carrier of balanced chromosomal translocation.

How is PGD carried out?

- Pre-PGD workup (please refer details below)
- In vitro fertilization
- Embryo culture
- Embryo biopsy: few cells will be removed from each day-5 embryo
- Genetic test will be performed on the cells biopsied
 - > For single gene disease, polymerase chain reaction (PCR) is performed
 - ➢ For translocation, array CGH is used

The test result will usually be available within 2-7 days. All embryos will be frozen before knowing the PGD result. Normal embryos, if available, will be thawed and transferred in a subsequent cycle.

What is Pre-PGD workup?

Single gene disorder varies among individuals, therefore, we have to custom-make PGD tests for every individual couple. Blood will be taken from couple (and other family members, if necessary). Pre-PGD workup will usually take 1-3 months to complete. The workup duration depends on the complexity of the genetic condition. Due to the complexity of different gene disorder is variable, not every pre-PGD workup is guaranteed to be applicable in single cell.

Advantages of PGD

PGD identifies the embryo(s) with a normal targeted gene for intrauterine transfer; the potential advantage is to avoid selective pregnancy termination due to the inheritance of the patient's disease gene.

How much PGD alone costs?

- 1. Pre-PGD workup: \$5,000
- 2. Laboratory administrative fee (once PGD is requested): \$2,000
- 3. Biopsy fee: \$5,000
- 4. PGD fee: \$4,800-20,000 (depends on disease complexity)

For more details, call us at 2632 1455 or visit our website at <u>www.ivfhk.com</u>

植入前基因診斷(PGD)

甚麼是胚胎植入前基因診斷(PGD)?

植入前基因診斷 (PGD) 是中文大學婦產科最新的臨床檢測服務,為植入前的胚胎作單基因診斷。PGD 是 用來診斷單基因疾病以避免將該疾病遺傳給下一代。PGD 的主要優點是它避免了選擇性人工終止妊娠,因 為所移植的胚胎應為沒有該單基因疾病。

誰適合選用 PGD?

我們建議選用 PGD 的病人包括:

- 夫婦任何一方有已知的家族性遺傳病;
- 夫婦任何一方是某種已知的單基因疾病基因攜帶者;
- 夫婦任何一方是已知的染色體平衡易位的攜帶者

PGD 的流程

- PGD的事前準備 (詳情請參閱『PGD 需要什麼事前準備?』)
- 安排病人以及其配偶做體外受精
- 胚胎培養
- 胚胎活檢:我們會在第五天的胚胎中抽取一些細胞作分析
- 所抽取的細胞會進行以下的基因測試:
 - > 若檢測單基因疾病便會選用聚合酶連鎖反應方法(polymerase chain reaction, 即 PCR)
 - ▶ 若檢測染色體平衡異位則會利用晶片全基因組對比分析法 (array comparative genomic hybridization, 即 aCGH)

一般而言,在 2-7 天內會有測檢結果。在未有測檢結果之前,所有胚胎會進行冷凍。報告出來以後,正常的胚胎(如有的話)就可在下一次的周期植入子宮内。

PGD 需要什麼事前準備?

由於不同人有不同的單基因疾病,因此每一對夫妻都需要建立其個人化的 PGD 測試。我們會在 PGD 測試 開始之前先抽取病人與其配偶的血液(有時也需要抽取其他家人的血液一同檢測)。PGD 的事前準備通常 會需要一到三個月的時間才能完成,時間長短通常取決於該疾病的複雜程度。由於不同的單基因疾病有不 同的複雜程度,我們無法保證每個 PGD 測試都能成功地建立並應用於單細胞內。

PGD 的好處

PGD 能診斷沒有單基因疾病的胚胎並植入子宮內,其有利優勢是避免了選擇性人工終止妊娠,因為所移植的胚胎應為沒有該單基因疾病。

PGD 的費用

1. PGD 的事前準備費用:港幣\$5,000

- 2. 化驗室行政費用:港幣\$2,000
- 3. 胚胎活檢費用:港幣\$5,000
- 4. 植入前基因診斷(PGD)費用:港幣\$4,800-20,000 (取決於該疾病的複雜程度)

如欲知道更多資料,請電 2632 1455

或請瀏覽我們的網頁 www.ivfhk.com