• 系統編號	RC8901-0344
• 計畫中文名稱	Mosaic Klinefelter Syndrome 病人睪丸切片以 FISH(Fluorescence in Situ Hybridization)分析其精蟲非雙套染色體
• 計畫英文名稱	Sperm Aneupoid Detection by Apply Fluorescence in Situ Hybridization (FISH) in Testis Biopsy of Mosaic Klinefelter Syndrome Patient
• 主管機關	行政院國家科學委員會
• 計畫編號	NSC88-2314-B038-115
• 執行機構	台北醫學院醫學研究所
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• 使用語言	中文
• 研究人員	江漢聲 Chiang, Han-Sun
• 中文關鍵字	睪丸活組織檢查法；精子；染色體；螢光原位雜交法；遺傳疾病；精細胞
• 英文關鍵字	Testis biopsy；Spermatozoon；Chromosome；Fluorescence in situ hybridization (FISH)；Hereditary disease；Spermatid；Mosaic Klinefelter syndrome
• 中文摘要	Klinefelter 症候群(47XXY)原本是不能治療的男性不孕,目前已有重大突破,在部分 Mosaic XXY 病人可經由睪丸精蟲萃取術來授孕。本研究計畫以21例Klinefelter症候群病人做臨床染色體篩檢,發現有3例Mosaic XXY;在21例中有10例做睪丸切片,9例同時用DXz1和DYz1 Probe 進行染色體 FISH(Fluorescence in situ hybridization)檢測,看 XY 非雙套(Aneuploidy)染色體精蟲或精原細胞的存在與否。結果發現在 10 例睪丸切片中僅有 1 例有精原細胞的製造,但未成熟,8 例的 FISH 檢查均未發現有精原細胞的存在。這個研究顯示 Mosaic XXY 的比例在我們的族群中不高(14.3%),目前也沒發現有 Spermatid 的存在,然而 FISH 的技巧可以正確的篩檢遺傳疾病原本無法生育的個案,將來在 Klinefelter 病人診療規劃、生育遺傳諮詢上是必要的一個流程。
• 英文摘要	Intracytoplasmic injection of single sperm (ICSI) is a big break through of male infertility. For the patients with Klinefelter's Syndrome (47XXY), they were previous intreatable, now could possibly be treated with TESE (Testicular sperm extraction) and following ICSI. Some patients with Mosaic 47XXY do have sperm in the testis. In this project, we collected 21 cases of Klinefelter's Syndrome for a complete chromosome study. Three of them are Mosaic 47XXY. After general evaluation and counseling, testis biopsy was performed for 10 patients and 8 of them simultaneously did dual labeled FISH with DXz1 and Dyz1 probes to detect the existence of the aneuploidy of the sperm or spermatid. In our preliminary result of the 10 cases testis biopsy, only 1 case had "hypospermatogenesis". All of the 9 cases was not found to have Aneuploidy spermatid in the FISH diagnosis. It seems the percentage of mosaic 47XXY is rather low (14.3%) in our Klinefelter's patients'

population. However, we had established the laboratory diagnosis with FISH technique and confirmed by clinical and pathological data. From this way we can elucidate the spermatogenesis in these Mosaic XXY chromosome patients. FISH diagnosis for the evidence of spermatogenesis will be an important information of the treatment protocol and counseling for the patients with Klinefelter's Syndrome.