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• 計畫中文名稱	台灣無精蟲或精蟲極度不良的男性不孕病人 Y 染色體基因缺損研究---臨床和病理的對照分析(II)
• 計畫英文名稱	Clinical and Pathological Correlation of the Microdeletion of Y Chromosome for the Patients with Azoospermia and Oligoasthenospermia in Taiwan (II)
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• 中文關鍵字	不孕症；染色體病變；基因突變；無精蟲症；基因缺失
• 英文關鍵字	Infertility；Chromosome disorder；Gene mutation；Azoo spermia；Gene deletion
• 中文摘要	在本年度計畫中，我們從 334 位男性不孕病人中篩檢出 32 位 Y 染色體基因缺損的病人，做臨床、病理、整體染色體異常的分析；並把 32 位病人缺損區域分成四類，將缺損情況和臨床、病理、整體染色體異常對照，發現：(1)Y 染色體上基因缺損的區域有其特性；在 AzFc 區域內的缺損影響製造精蟲功能最小；3 位病人中有 2 位精液中尚有精蟲。AzFc 到 AzFd 區內的缺損則尚有 52.6%在精液內有精蟲，AzFc 到 AzFb 區內的缺損則祇有 12.5%在精液內有精蟲，兩位 Y 染色體完全缺損的病人則全無精蟲。(2)臨床以睪丸大小和血清中 FSH 濃度來評估製造精蟲功能，結果也和 Y 染色體上缺損的區域相稱。從第一類 AzFc 區內缺損、第二類 AzFc 到 AzFd 區內缺損、第三類 AzFc 到 AzFb 區內缺損到第四類 Y 染色體完全缺損，睪丸大小和血清中 FSH 濃度不正常的比率明顯變高。(3)在 32 位 Y 染色體缺損病人中整體染色體異常率(31.3%)要高於 334 位男性不孕病人的染色體異常率(15.9%)。(4)11 例做病理切片病人的結果，以及在任何一區缺損均有不同表現個案來看，Y 染色體上基因缺損絕非影響製精功能的單一因素；還有其他可能基因、染色體缺陷和外在因素影響睪丸的製精功能。
• 英文摘要	Up to this year of our study, we screened out 32 cases of microdeletion of Y chromosome from 334 patients with male infertility. The clinical informations, chromosomal anomalies and pathological pictures of these 32 cases were collected for a correlative study to evaluate the severity of the spermatogenesis defect. We divided the cases of microdeletion of Y chromosome into 4 groups with deletion in 4 different areas; AzFc, from AzFc to AzFd, from AzFc to AzFb and the whole segment of Y chromosome deletion. Based on our observation, there are some remarkable

difference among the 4 groups as the followings: (1) When the deletion area is within AzFc, it has less affect to the spermatogenesis. Two of the 3 patients still have sperm appeared in the semen. If the deletion area is from AzFc to AzFd, 52.6% patients still have sperm appeared in the semen; however, when the deletion area is from Azfc to AzFb, only 12.5% patient is oligozoospermia, others are total azoospermia. (2) Clinically, the testicular size and semen FSH level are used to evaluate the function of spermatogenesis. It is quite compatible to the wideness of deletion area in the Y chromosome. When the deletion is within AzFc area, 66% of the patients have normal size tests and normal FSH level of the deletion extends from AzFc to AzFd area, 47.4% and 73.7% patients have normal size tests and normal FSH. However, patients with microdeletion of Y chromosome in the area from AzFc to AzFb, only 12.5% patients have normal size tests and 37.5% patients have normal FSH. (3) The overall anomaly rate of chromosome is higher (31.3%) in the 32 patients with microdeletion of Y chromosome than that of the 334 patients with male infertility (15.9%) in this study. (4) Based on the findings that spermatid is still present in the pathological specimen of the patients in the different group of Y chromosome deletion, as well as that variable clinical picture appeared in each group of Y chromosome deletion, we can conclude that the causing factors of spermatogenesis defect is multiple. It's needed further investigation.