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## 一、中文摘要

鑑於基因辨認在乳癌、卵巢癌治療上的發展，已引起先進國家對此問題相關的社會、病人心理與醫療倫理等議題的熱烈爭議，本研究從回顧西方有關基因辨認在乳癌、卵巢癌治療之醫療、社會心理與醫療倫理文獻，以瞭解目前的發展現況，以及所衍生的種種問題來加以探討，期能對臺灣目前擬發展的基因科技提出若干參考性的建議。

## 二、英文摘要

The identification of BRCA1 and BRCA2, two breast-ovarian cancer susceptibility genes, has brought many ethical and social issues to the forefront. Many geneticists, ethicists, psychologists, and persons at risk for cancer are concerned about the potentially psycho-socio effects of genetic testing on tested persons and their families. Along with the possibilities for the application of gene therapy, complex safety, ethical, and financial issues that will require resolution. The purposes of this study is to review the current development about breast-ovarian cancer gene therapy in developed countries.

## 三、計劃緣由與目的

利用電腦搜尋發表至1998年十二月之前的相關文章。評論所有搜集到與乳癌與卵巢癌有關的基因治療之參考文獻，以瞭解基因科技對乳癌與卵巢癌治療的貢獻。

## 四、結果與討論

BRCA1(maker=185delAG)確實可以解釋許多家族性乳癌的發生。若具有BRCA1的人(gene marker)50歲便具有50%的罹患率，70歲有70%的罹患率。一般學者相

信卵巢癌和乳癌具相同基因來源

(chromosome 17q)。BRCA1的發現確然將癌症研究與防制帶入一個新紀元，而BRCA2雖再次支持基因檢驗之重要但是否僅僅依賴基因之檢驗便可以預防或控制乳癌的發生?除了基因研究本身內含複雜性與不確定性以外，例如，BRCA1之基因marker具有100000 bases，因此可能變形有很多了，且無法完整求出其完全變形。導致乳癌的基因可能還有其他基因marker。癌症的發生乃同時涉及基因與環境兩個層面。基因篩選費用龐大且相關基因序列太複雜，因此無法將基因研究有效延伸進入一個更大的醫療架構中。基因研究可以幫助瞭解與預防乳癌，但基因研究或防制卻無法根本解決乳癌所造成的衝擊，原因有以下幾個：1)與遺傳相關的乳癌比例並不高(約5-10%)；2)乳癌之醫療有其他非基因諮詢的面向，如乳房切除，乳癌復發等；3)乳癌引發心理與社會變化可能會因為基因研究介入變得複雜，而非簡化。但基因諮詢有其風險與利益，其優點不外乎提供某種程度的確定性，但它也可能嚴重衝擊到受訪者之家族關係與社會生活。基於上述種種顧慮，在建立相關研究室/實驗以及研究計畫，應採高標準加以限制。再者，基因研究距離實際臨床仍有一段路途，而且相關醫療干預並未建立，因此大規模的實際醫療提供必須警慎。利用基因科技確實可以測知癌症罹患的先天/遺傳傾向，但基因科技有其基本的問題，如測量精確的問題、實際法律、倫理社會的問題。

綜合各國討論有關基因檢驗可能涉及的法律與倫理問題可從以下幾個層面來討論：

1.關於使用基因檢測公平性的問題：

€ 無法真正落實公平的可能 – 資訊的資源分配不均。

2.關於保障生育權

€ 基因研究可以作為墮胎的一個參考點，但個人是否能自主地/自由

地選擇墮胎？基因研究之地位是否如此明確？

### 3.關於隱私權：

€ 基因檢測引發新的思考架構(關於風險、病理學等)但個人隱私權也形成問題,相對的問題則是不信任與基因資訊的使用。

### 4.關於檢測結果的揭露

€ 檢測結果,雖應告知受試者,但有程度與種類的差異,再者有必要考量心理影響層面。

### 5.是否告知其他親屬

€ 個人隱私權與疾病防治的衝突。  
€ 大多醫生在此立場不一。  
€ 病人若未達法定成年年紀,問題更複雜。

### 6.自願與強制施測

€ 一般國家均採自願檢測,但針對無自主能力之人則可採強制施測/不施測,例如胎兒。自願檢測必須基於「醫生必須充分告知」。

### 7.不引導原則(non-directive)

€ 醫生不應誘導病人作基因檢測,醫生僅提供相關諮詢與提供選擇項目,決定仍在病人手上。

### 8.優生學

€ 是否將基因檢測納入優生學的考量將引起爭議。

就目前發展的狀況而言,基因研究速度快於相關的立法與倫理研究,同時,雖然基因檢查/治療可以解決一些遺傳疾病之治療與檢查,基因治療並不能完全確定疾病的發生與爾後的治療。另,醫療諮詢亦不夠專業。就其倫理面而言,雖然關於基因治療,專業人士具有保密共識(confidentiality)減少病人所受到之傷害,基因治療/諮詢必須基於充分告知後取得的同意(informed consent),但因為基因的知識太複雜,同時相關問題未能解決(例如:如何減低可能傷害,如因果基因的相關資料若為保險公司所用,可能影響個人及其家屬,法律如何規範?其次,如何界定病患與家屬的關係、如何利用基因知識的保密達到防止可能的傷害、何時/基於何種理由

醫生可以不遵守保密的原則,透露相關基因資訊,以便防止疾病發生與治療,醫生又如何協助病患與其家屬,並同時考量社會所承當的風險。因此,基因檢測諮詢應該納入病患個人的日常生活與醫生的治療史;基因相關資訊不應擅自披露,除非相關人等有受傷害的可能;專業諮詢制度應建立;醫療人員應有責任歸屬問題。

再就採行基因檢測的受試者與醫療人員可能引起的社會心理問題而言,研究結果發現,大多數醫生支持將DNA檢測引入乳癌的防治,但由於當病人的DNA檢測呈現「陽性」(帶有BRCA1或與乳癌相關的基因),則病人可能呈現高度焦慮,因此在無其他方式保障一病患的情況下,醫生都同意病人的自決才是基本前提。其次,大多數醫生相信基因檢測引入醫療將促發社會區隔(在保險上、法律權益上),因此他們皆以為利用立法保障基因資訊的流通與相關病患才是解決之道。大致上,醫生皆願意實際參與基因檢查,或將基因檢查加入其執業。

就帶乳癌基因者而言,大部份接受基因試驗的原因有:(1)想知道自己的孩子是否在risk範圍內;(2)想好好照顧自己。而大部份不接受基因試驗的原因有:(1)擔心基因試驗不安全;(2)質疑基因試驗的正確性;(3)擔心會有情緒上之反應。

另外研究亦指出,接受基因試驗的婦女中有60-87%的婦女認為醫療單位不應該未經同意,將breast-ovarian cancer的Genetic testing揭發給保險公司或員工知道;95%的婦女同意應該有權利選擇試驗,即使醫師反對;88%的婦女同意父母應同意他們未成年子女接受試驗;同時在不用寫同意書的情況下,醫療單位可將遺傳試驗結果告知親屬;父母應有權為未成年子女決定接受遺傳試驗。

再如就基因檢測對醫療消費者、醫療單位及一般大眾三方面所造成的影響而論,則可分條簡述如下:

### € 醫療消費者

Carrier status 往往會造成心理上羞辱的感覺。如何減輕Risk information 所造成

的心理傷害及影響，是很重要的工作。消費者應有權力充分瞭解基因試驗所帶來的好處及限制，對於社會、經濟及社會文化層面的考量皆不可忽視。

#### € 醫療業者

在醫療業者方面，最重要的是 Confidential genetic information 的問題，對於保險業者公司、醫療工作人員及家屬成員應考量 Confidential 的問題；其次建議醫療業者應提供有效的諮詢服務，使病人瞭解 Genetic Testing 的重要性。諮詢服務的工作人員應接受訓練，即使是醫師本人皆接受心理學的訓練。

#### € 大眾的期待

大眾對 Genetic testing 的態度，無論接受或拒絕，皆與是否被保險公司 Cover，及對試驗結果的態度有關。而疾病本身是否有治療的可能，亦是民眾決定是否要作試驗的一個很重要因素，使民眾瞭解 Genetic Testing 的限制及其潛能，將有助解除民眾對疾病宿命論的觀念。

### 五、計劃結果自評

經過對相關文獻的蒐尋與閱讀，逐漸瞭解癌症的研究已進入基因為基礎(gene-based model)的研究，此將可使學界更加瞭解癌細胞的運作。本人因為閱讀把關文獻而受益斐淺。相關的文獻應廣為流傳，以使研究資源逐漸能轉向以基因為基礎的研究。然而，基因研究雖然可以幫助瞭解與預防乳癌，但基因研究或防制卻無法根本解決乳癌所造成的衝擊，原因有以下幾個：1)與遺傳相關的乳癌比例並不高(約 5-10%)；2)乳癌之醫療有其他非基因諮詢的面向，如乳房切除，乳癌復發等；3)乳癌引發心理與社會變化可能會因為基因研究介入變得複雜，而非簡化。同時，基因諮詢有其風險與利益，其優點不外乎提供某種程度的確定性，但它也可能嚴重衝擊到受訪者之家族關係與社會生活。基於上述種種顧慮，在建立相關研究室/實驗是以及研究計畫，應採高標準加以限制。再者，基因研究距離實際臨床仍有一大段路途，而且相關醫療干預並未建立，因此大規模

的實際醫療的提供須更加警慎。同時，研究者不可忽略基因科技有其基本的問題，如測量精確的問題、實際法律、倫理社會的爭議。

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