FABRY氏疾病—女性帶原者病例報告

陳昌文　吳振光　蕭廣仁

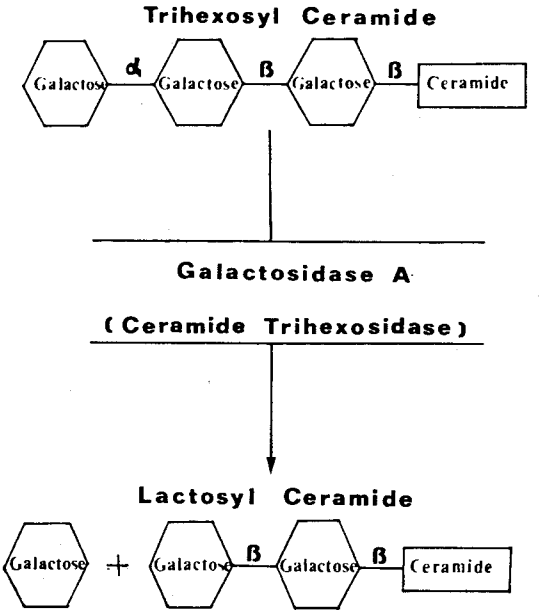
Fabry氏疾病為一種性聯遺傳疾病，其致病機轉為體內缺乏甲種半乳糖苷酶(α-galactosidase A)所致。在男性患者其血漿內完全缺乏甲種半乳糖苷酶之活性，常見以皮膚、心臟血管、腎臟及眼部疾病為臨床表徵，而女性其血漿內甲種半乳糖苷酶之活性僅為正常人之15%至40%，而以角膜漩渦狀表淺性混濁為臨床特異性表徵。本文報告一位35歲女性患者，其角膜上皮呈現典型的漩渦狀色素沉著及甲種半乳糖苷酶之生化測定，證實為一罕見的Fabry氏疾病帶原者，故提出報告。

Key words: Fabry's disease, angiokeratoma corporis diffusum, vortex epitheliopathy, α-galactosidase A

前　　言

Fabry氏疾病又稱為瀰漫性體血管角質瘤(angiokeratoma corporis diffusum)[1]，是屬於一種與性染色體有關的疾病，其致病機轉為體內缺乏甲種半乳糖苷酶(α-Galactosidase A)致使飽合脂質Trihexosyl Ceramide異常貯積引起(如圖一)。在男性常見以皮膚、心臟血管、腎臟及眼部疾病為表徵；女性則往往沒有症狀，僅以角膜表淺性混濁為臨床表徵。

本文報告一位35歲女性，在常規性體檢時，發現其角膜上皮呈現典型的渦輪狀表淺性混濁，經回溯性的追踪檢查，包括甲種半乳糖苷酶之測定，證實為一罕見的Fabry氏帶原者，故提出報告。



圖一：Fabry氏疾病之致病機轉，因體內缺乏甲種半乳糖苷酶(α-galactosidase A)，致使Trihexosyl ceramide無法轉變形成Lactosyl ceramide，導致飽和脂肪質Trihexosyl ceramide異常貯積於全身組織之疾病。

病 例 報 告

林女士，35歲，病人於民國73年10月30日至榮民總醫院體檢室接受常規檢查時，發現角膜特殊表徵，經詳細檢查病患及其家屬之病史如下：

榮民總醫院　眼科

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FABRY'S DISEASE – OCULAR FINDINGS AND ENZYMATIC ASSAY IN A FEMALE CARRIER

Chang-Wen Chen, John-Kuang Wu and Kwang-Jen Hsiao*

*Department of Ophthalmology and Medical Research**
Veterans General Hospital, Taipei, Taiwan, R.O.C.

Fabry's disease is an X-linked inborn error of glucosphingolipid metabobic disorder. The pathogenesis of the disease is due to deficient activity of the ceramide trihexosidase, a specific α-galactosidase A. The activity of α-galactosidase A in plasma is absent in affected males (hemizygotes) who clinically manifest multiple organs involvement. And in female carriers (heterzygotes) who only manifest characteristic corneal epitheliopathy are present at only 15% to 40% of the normal level of enzymatic activity.

A patient (female, 35 years old) with corneal vortex epitheliopathy and a family history of cardiac and renal diseases was diagnosed as heaving heterzygote of Fabry's disease by an enzymatic assay. Her α-galactosidase A in plasma was 5.14 nmol/hr/ml, about 40% of that of normal controls.

To the best of our knowledge, this is the first case of Fabry's disease diagnosed by the enzymatic assay in Taiwan.