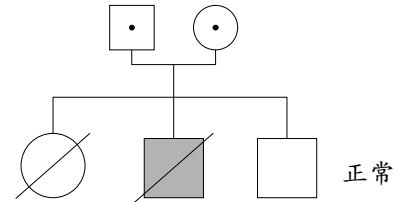


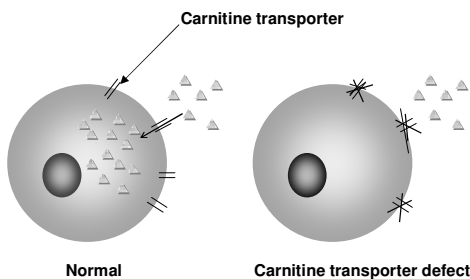
卡尼丁缺乏症 Primary Carnitine Deficiency

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父親 隱性基因攜帶者
母親 隱性基因攜帶者



Carnitine transporter defect



卡尼丁缺乏症 Primary Carnitine Deficiency

- 病徵
- 在嬰兒時候，出現急性病徵，不省人事
- 嚴重的可導致死亡
 - Acute presentation in infancy
- 發大性心臟肌肉病變
 - Cardiomyopathy

OCTN2 基因研究 (1)

- 隱性遺傳病
 - Autosomal recessive inheritance
- 自一九七五年以來，各地的研究人員都在尋找這個致病的基因
 - Researches are searching for the causative gene since 1975
- 病人從父母雙方各遺傳一個病變卡尼丁運载体基因
 - Patient inherits one defective carnitine transporter gene from each of the parent

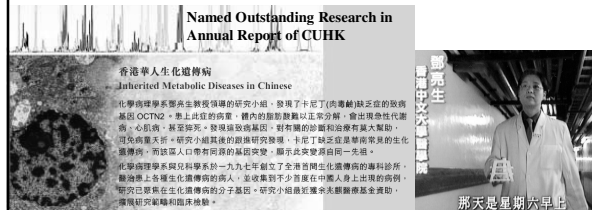
基因突變 (Mutation)

正常
[APPLE][BOY][CAT][DAVID][EGG][FISH]
....

突變
[APPLE][BOY][CAT][DAWID][EGG][FISH]
....

OCTN2 基因研究

- 一九九九年, 中文大學的研究發現 OCTN2 為卡尼丁運輸體基因
- 同時日本及美國的研究 亦獲得相同的發現
- Among the first in identifying the carnitine transporter gene, OCTN2



More patients with primary carnitine deficiency

- 發現卡尼丁缺乏症是一個在華南地區較常見的生化遺傳病, 同時亦發現華南人口攜帶一個同源的基因突變, 顯示此突變是源自同一個先祖.
- cases were diagnosed in Hong Kong, Macau, Taiwan; Sharing a common mutation

HUMAN MUTATION Mutation in Brief #524 (2002)

A Founder Mutation (R254X) of SLC22A5 (OCTN2) in Chinese Primary Carnitine Deficiency Patients

Nelson L.S. Tang¹, W.L. Hwu², Rachel T. Chan³, L.K. Law³, L.M. Fung¹, and W.M. Zhang¹

¹ Department of Chemical Pathology, Faculty of Medicine, The Chinese University of Hong Kong, Hong Kong; ² National Taiwan University Hospital and National Taiwan University College of Medicine, Taiwan; ³ Department of Pediatrics and Neonatology, Centro Hospitalar Conde S. Januário, Macau.

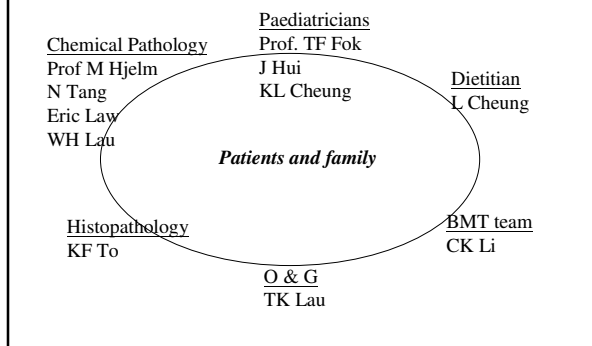
研究成果的應用 (1)

- 準確地診斷: 卡尼丁缺乏症之患者者
- Early diagnosis of patients with primary carnitine deficiency
- 早期產後診斷
- 患病嬰兒長期服食卡尼丁
- 可防止患有該病者猝死

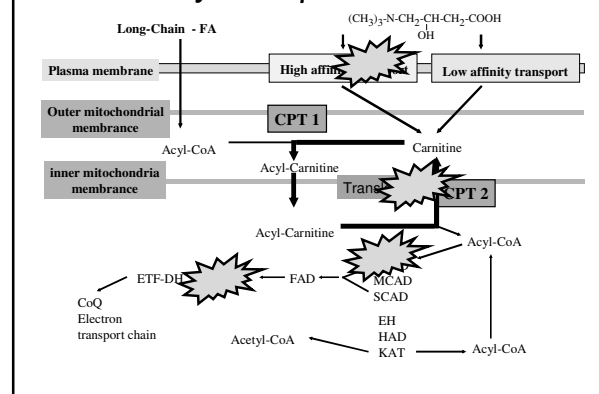
研究成果的應用 (2)

- 準確地找出攜帶失效基因者
- Identify carriers in the families
- 1. 多吃肉類 (含有大量卡尼丁)
- 2. 有些藥物要避免使用
- 3. 心臟檢查
- 4. 婚前遺傳檢查, 產後子女嬰兒的檢查

中文大學 - 威爾斯醫院 生化遺傳病專科門診



Fatty acid β oxidation



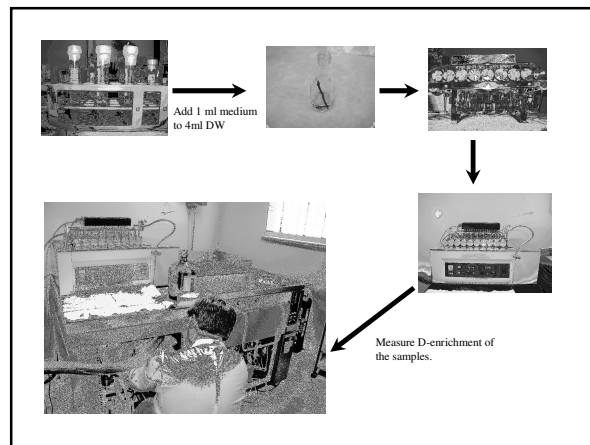
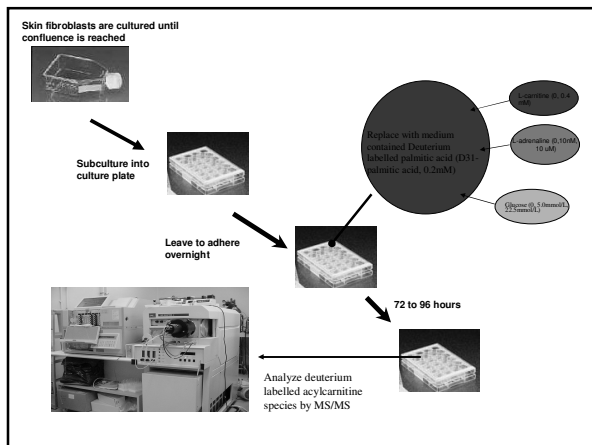
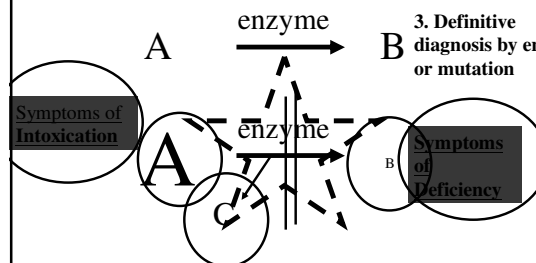
What FOAD are found in Southern Chinese?

1. Primary Carnitine deficiency
2. Glutaric aciduria type 2 (ETF chain) defects
3. Carnitine translocase deficiency
4. VLCAD
5. SCAD

Approach to diagnose Inherited Metabolic Diseases

- 3 steps

1. Clinical Symptoms
2. Metabolites
3. Definitive diagnosis by enzyme or mutation



What is the Chinese term for Carnitine ?

- 肉毒碱 before 1999
- At 1999, I created a new term 卡尼丁 in a press conference.

鄧亮生續稱，目前導致嬰兒猝死的原因也無法解釋，其中有十分一與先天性新陳代謝病有關，「卡尼丁」缺乏症是其中一種，父母兩人同時為這種罕見遺傳病的隱性攜帶基因者，人口比率僅為一百萬分之一，這類父母產下的嬰孩，則有四分之一機會有「卡尼丁」缺乏症。¶

History of Chinese term for Carnitine

Results 1 - 10 of about 3,180 for 卡尼丁 (¶)

1 result stored on your computer - Hide - About
[carnitine in the news.doc - 死因缺卡尼丁【本報訊】嬰兒猝死原因向來是不解之謎，中文大學醫學](#)

eBay 台湾 Jarrow Formulas - Carnitine+CoQ10 (卡尼丁+輔酶Q10) ...
 熱門商品: Jarrow Formulas - Carnitine CoQ10 卡尼丁輔酶Q10 - 50顆在eBay 台湾商品类别居家生活与食品名产, 食品与地方特产, 营养品, 其他
[cgi.tw.ebay.com/Jarrow-Formulas-Carnitine-CoQ10-Q10-50-WDQItemZ110000303997Q10h2007Q10categoryZ19262QQcm2V... - 58k - Supplemental Result - Cached - Similar pages](http://cgi.tw.ebay.com/Jarrow-Formulas-Carnitine-CoQ10-Q10-50-WDQItemZ110000303997Q10h2007Q10categoryZ19262QQcm2V...)

eBay 台湾 Doctor's Best - Acetyl-L-Carnitine (乙醯L-卡尼丁) - 60顆 ...
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[cgi.tw.ebay.com/Doctors-Best-Acetyl-L-Carnitine-60-WDQItemZ110000303997Q10h2007Q10categoryZ19262QQcm2Vie... - 57k - Supplemental Result - Cached - Similar pages](http://cgi.tw.ebay.com/Doctors-Best-Acetyl-L-Carnitine-60-WDQItemZ110000303997Q10h2007Q10categoryZ19262QQcm2Vie...)
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