國民健康局科技研究發展計畫原始數據資料庫
資料讀我檔案

計畫名稱：國內新生兒先天代謝異常疾病篩檢項目增減可行性之探討
計畫編號：DOH92-HP-1210
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研究報告中文摘要：

國內新生兒先天代謝異常疾病篩檢從民國七十三年一月起展開先期作業及前瞻性篩檢，七十四年七月正式開始全國性篩檢。國內目前政策下常規的新生兒篩檢項目為苯酮尿症（PKU）、高胱胺酸尿症（HCU）、半乳糖血症（GAL）、先天性甲狀腺低能症（CHT）及 G6PD 缺乏症，近年來篩檢率已超過 99% 。國內自民國 73 年至 91 年篩檢了四百多萬新生兒，CHT、PKU 及 G6PD 缺乏症的發生率較高，分別約為 1/1,800 ~ 1/2,700，1/31,000 ~1/35,000 及 1/60；而 HCU 及 GAL 之發生率則較低，分別約為 1/212,000 ~ 1/263,000 及 1/744,000 ~ 1/1,115,000。

國內外近年來發展出一些其他的篩檢項目及技術，如先天性腎上腺增生症（CAH）及利用串聯質譜儀（Tandem Mass；MS/MS）篩檢的先天代謝異常疾病。但並非每一種可篩檢出的疾病均符合世界衛生組織（WHO）1968 年發表之疾病篩檢指導方針及 1994 年美國 IOM 遺傳風險評估委員會建議的新生兒篩檢展開篩檢的條件。此外國內新生兒篩檢項目及作業系統已執行了二十年，亦宜適檢討是否需刪減或修改篩檢項目。本計畫全面檢討國內的現行的新生兒常規篩檢項目及評估未來可能為適當增加之篩檢項目。
本研究透過網際網路的「搜尋引擎」及「文獻期刊資料庫」，蒐集近年來國內外有關新生兒篩檢、確認診斷方法、治療、監偵及其治療成效的文獻，並將搜尋到的資料建置成「新生兒篩檢項目增減評估研究作業網站」，供後續研究查詢使用。歸納整理蒐集到的相關文獻資料，設計分別適用於「篩檢中心」，「小兒遺傳專科及內分泌次專科醫師」及「遺傳諮詢中心」調查表，用以收集目前國內「非常規」篩檢項目及現行篩檢作業成果，臨床發現相關疾病病患人數及診斷治療經驗與病患之預後，以及對篩檢項目增減之意見。透過「罕見疾病基金會」、「臨床疾患追蹤管理治療中心」、國健局「罕見疾病通報資料庫」，收集與本計畫相關疾病的病患數量、發生率、確診方法、確診單位及治療成效等相關資料。

透過兩次專家學者會議，由國內小兒遺傳及內分泌次專科醫師、各篩檢中心主持人及國內串聯質譜儀專家討論決議，繼續維持原有五項常規篩檢項目，建議納入為新增之常規篩檢項目，包含 CAH、MSUD、MCAD、GA I、IVA、MMA 等六項疾病。建議進行「先趨性全面篩檢」之項目，包含 BD、ASA、CIT、PA、

HMG、2MBCD、BKT、3MCC、MCD、3-Methylglutaconyl-CoA Hydratase Deficiency、 2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency 等十一種疾病。其他可利用串聯質譜儀篩檢之疾病項目，現階段不宜進行全面篩檢，建議先進行「先趨性研究」收集更完善的資料後，再行評估。配合上述調整建議，建立了「新增之常規篩檢」項目及建議進行「先趨性全面篩檢」項目的篩檢作業原則，作為未來執行之依據。

為配合新增篩檢項目篩檢時效，建議規定新生兒須在出生後 24 至 72 小時內採血，採集機構採血後須於 24 小時內寄出血片；遞件公司在隔日上午十時前須送達篩檢中心實驗室，實驗室收件後 56 小時內須發出 G6PD、CAH、GAL 及利用串聯質譜儀篩檢項目之陽性通知；陽性個案及轉介醫院應於個案出生後七天 (含假日) 以內接獲通知，進行適當的確診及防治工作。在新增的新生兒常規篩檢項目及先趨性全面篩檢項目正式實施前，應將整體篩檢流程，自採血、寄送檢體、篩檢中心檢測，至轉介醫院確診、治療及後續的治療監偵系統皆完整規劃建立完善，測試完成後，再行全面正式提供篩檢服務。為符合社會的公平與正義，國內所有新生兒不分地區貧富均應接受新生兒「常規篩檢項目」檢測之服務。此外往後「新生兒常規篩檢項目」內容亦應定期檢討評估，以符合時代醫療科技及知識之進步。

中文關鍵詞(至少三個)：先天代謝異常疾病，新生兒篩檢，串聯質譜儀

Research Data Archive, Department of Health, The Executive Yuan, R.O.C.
Readme file

Project Title: ：Assessment for the current neonatal screening program for inborn error of metabolism in Taiwan – The feasibility of adding or deleting screening diseases

Project Number:DOH92-HP-..1210

Executing Institute:National Yang Ming University
Principal Investigator(P.I.): Kwang-Jen Hsiao
P.I. Position Title: Professor
P.I. Institute: Insititute of Genetics

Abstract:

Some of the congenital metabolic disorders have no specific clinical symptoms during neonatal period, if not treated early irreversible damages such as mental retardation will occur. The permanent damages can be avoided if these are able to be detected biochemically by neonatal screening in the early stage of life, and treated immediately with appropriate therapy and intervention.

The nationwide newborn screening for inborn error of metabolism in Taiwan has been started in 1985. Congenital hypothyroidism (CHT), phenylketonuria (PKU), homocystinuria (HCU), galactosemia (GAL), and glucose-6-phosphate dehydrogenase deficiency (G6PD) have been screened routinely. The screening coverage rate in Taiwan has reached 99% since 1996. From 1984.1 to 2002.12, 4,462,600 newborns have been screened. The incidences of CHT, PKU, HCU, and GAL were reported to be about 1/1,800 ~ 1/2,700, 1/31,000 ~ 1/35,000, 1/212,000 ~ 1/263,000, and 1/744,000 ~ 1/1,115,000, respectively. The incidence of G6PD deficiency was estimated to be about 2% (male 3%, female 0.9%) in Taiwan.

In order to review the proper screening items for routine neonatal screening program in Taiwan, we have collected related information and literatures from Internet and databases. A website and literature database http://ns.pmf.org.tw were install for this review and future research/clinical usage. After reviewing that information and collecting information and opinions from metabolic/endocrinological pediatricians, screening centers, and genetics counseling centers, a list of candidate items was produced for the expert panel to review. The panel has made the final adjustment of the list and approved the guidelines for the implementation of those tests and the improvement of the screening system to accommodate the new screening items.

The conclusion from the review has reached following consent recommendations about the adjustment of the items for neonatal screening in Taiwan.

1). the 5 current routine items should be kept, 2). congenital adrenal hyperplasia (CAH), maple syrup urine disease (MSUD), Medium Chain Acyl- CoA Dehydrogenase Deficiency (MCAD), Glutaric Aciduria Type I (GAI), Methylmalonic Aciduria (MMA), and isovaleric academia (IVA) should be included as routine items, 3). biotinidase deficiency, argininosuccinase deficiency, propionic academia, and C5OH-carnitine should be included as routine items for a pilot project, 4). any other disease which could be detected by MS/MS should be considered as a research item only at the present time, 5). any disease incorporated into the routine services, including pilot project items, should have confirmatory diagnosis and follow up treatment system prepared in place before its screening program starts, 6). the positive results of CAH, G6PD, GAL, and MS/MS tests should be referred for follow-up no later than 7 days after birth, 7). the routine screening items should be available to all the newborns non-selectively, 8). the routine screening items should be reviewed periodically.

Keywords: neonatal screening, congenital metabolic diseases, tandem mass spectrometry