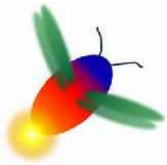
# 二代新生兒篩檢—台灣與國際的接軌



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# 二代新生兒篩檢資料編撰—台灣與國際的接軌

## 壹、前言

罕見疾病基金會自從於 1999 年創立以來，透過議題的倡導以及社會立法與社會政策的參與，對於國內罕見疾病病患的生存環境已有顯著的提升效果，其中最明顯的倡導成果包括有：於 2000 年元月通過「罕見疾病防治及藥物法」、2001 年 10 月修訂「身心障礙者保護法」納入罕見疾病為單一障礙別、2003 年 8 月爭取罕病全數納入健保重大傷病之列、2004 年 7 月爭取於健保總額中將罕病以專款專案方式與醫院自主管理脫鉤，對於罕見疾病病患的生存環境均有重大的助益。

其中另一項正在倡導中的議題為二代新生兒篩檢的擴大實施，其最終目的在促使政府將 Tandem Mass 串聯質譜儀的先進技術與原有第一代新生兒篩檢制度結合，以便增加更多的篩檢項目以保障下一代新生兒的健康。因此罕見疾病基金會於八十九年開始積極推展「二代新生兒篩檢先導計畫」，陸續透過補助一般民眾、偏遠地區、金門地區、原住民、及低收入戶等進行二代新生兒篩檢，並與臺大醫院、台北病理中心、及婦幼保健基金會等全國三大篩檢中心合作，其成效已日漸浮現，惟政府部門至今尚未對第一代新生兒篩檢提出變革，以致罕見疾病基金會有關「二代新生兒篩檢」的倡導議題尚未完成。

爲期能在 2005 年以前達成全面實施「二代新生兒篩檢」的倡導目的，有必要在 2005 年中以記者會型式或其他操作方式訴諸社會大眾，一方面宣導民間有關「二代新生兒篩檢」的合作成果，二方面必需就世界最新趨勢提出回顧與評介，如此方能檢視政府的施政成果以及其與世界趨勢的落差，也促使政府早日實施全方位的 「二代新生兒篩檢」。本研究計畫因此係為罕見疾病基金會 2005 年「二代新生兒篩檢」倡導活動的知識前置作業，透過網路蒐錄及編譯有關美國及國際有關 Tandem Mass 串聯質譜儀應用於新生兒篩檢的最新資訊與發展，期能提供罕見疾病基金會倡導內容所需的必要知識與訊息，使基金會「二代新生兒篩檢」的主張能在專業立足、符合民眾需要、也迎合世界潮流。

## 貳、我國新生兒篩檢的發展

### 一、衛生署的新生兒篩檢狀況

我國政府於民國 73 年起開始規劃建立國內先天性代謝異常新生兒篩檢制度，並於民國 74 年 7 月開始全面實施，其所能篩檢的疾病包括：苯酮尿症、高胱胺酸尿症、蠶豆症、半乳糖血症、先天性甲狀腺低能症等 5 項病症，平均每年約可檢驗出 4,000 至 6,000 名左右的異常新生兒，因而使許多新生兒免於死亡及智力受損等威脅；目前我

國新生兒篩檢率已達 99%以上，在亞洲地區僅次於日本，為長期以來我國重要的婦幼衛生政策。

二、運用串聯質譜儀於新生兒篩檢

然而先天性代謝異常疾病種類繁多，原有的新生兒篩檢技術卻只能篩檢出 5 種病症，為有效解決此一課題，世界各國均不斷投入龐大經費，以期發展出更迅速、敏感度高、篩檢種類更多的儀器，而 Tandem Mass 串聯質譜儀運用在代謝異常疾病的篩檢，便是因應此一需求所研發出來的技術；目前已有歐洲、澳洲和美國部分州，開始運用此種技術進行新生兒篩檢，而日本雖然引進 Tandem Mass 串聯質譜儀的時間較台灣為晚，但是日本政府也已經開始規劃作新的變革，並結合 Tandem Mass 串聯質譜儀的技術與新生兒篩檢制度。

Tandem Mass 串聯質譜儀篩檢技術之所以不同於傳統的新生兒篩檢方式，在於我國傳統的新生兒篩檢只能篩檢出五種疾病，且需依不同疾病進行不同的篩檢方式；而 Tandem Mass 串聯質譜儀可分析血液中各種化合物，藉由微量的血滴，即可篩檢出 20 餘種先天性代謝異常疾病，而傳統的篩檢項目中，苯酮尿症、半乳糖血症以及高胱胺酸血症亦能藉由該儀器篩檢出來，因而極適合進行大量篩檢且整體成本亦較傳統方法為低。

表 2-1 串聯質譜儀所能篩檢的罕見先天代謝疾病

| 分　類 | 胺基酸代謝異常 | | | | |
|---|---|---|---|---|---|
| 疾病英文名 | Phenylketonuria/ Hyperphenylalanine | Maple Syrup Urine Disease | Homocystinuria/ Hypermethionemia | Tyrosinemia Type I | Tyrosinemia Type II |
| 疾病中文名 | 苯酮尿症 | 楓糖尿症 | 高胱胺酸血症/高甲硫胺酸血症 | 酪胺酸血症第一型 | 酪胺酸血症第二型 |
| 疾病簡介 | 簡稱 PKU，是因為人體必需胺基酸中的苯丙胺酸在分解成酪胺酸的代謝路徑中發生障礙，導致苯丙胺酸大量堆積體內，產生許多有毒的代謝物質，造成腦部傷害，甚至嚴重的智力障礙。苯酮尿症可分為食物型與藥物型兩種。食物型的病患要避免吃含苯丙胺酸的食物，舉凡魚、肉、蛋、奶、豆類之食物，都要嚴格控制，病患得靠特殊奶粉來補充營養。藥物型的患者則必須補充一些副作用極大的神經傳導物質，其病症的控制上，較食物型之患者略為困難。 | 楓糖尿症是因為人體中缺少支鏈甲型酮酸脫氫酵素，使得支鏈胺基酸（纈胺酸、白胺酸、異白胺基酸）的代謝無法進行去羧基化反應。通常罹患此症嬰兒，在開始餵食後數天至一周內，會出現嘔吐、嗜睡、食慾減低、呼吸急促、黃疸、抽搐等現象，身上散發焦糖的體味或尿味，嚴重者會意識不清、昏迷甚至死亡。治療原則以限制支鏈胺基酸的攝取，再補充特殊奶粉及維持體內代謝物質之平衡為主。 | 主要成因為胱硫醚合成酵素的功能缺乏，造成高半胱胺酸合成胱胺酸的過程中發生障礙，在體內堆積甲硫胺酸、高胱胺酸、高半胱胺酸及複合雙硫化合物等異常代謝產物。主要症狀為智能不足、骨骼畸型、眼球水晶體脫位、心臟血管疾病及血栓等臨床症狀。治療上可使用高劑量的維生素 B6（VitB6）或限制甲硫胺酸的攝取，再使用特殊奶粉來補充體內所需之胺基酸。 | 酪胺酸是人體一種非必須胺基酸，主要的來源包括飲食攝入及苯丙胺酸代謝所產生的中間產物。遺傳性高酪酸血症主要成因為酪氨酸代謝過程中酵素功能異常所造成，其中第一型為 $\rho$-羥基-苯基-焦葡萄酸氧化酶缺乏所致。急性高酪胺酸血症為快速且猛爆性的病程，若不及時治療，將有死亡的危險，病程通常發作於 1 至 6 個月大的時候，患者常有食慾不振、嘔吐、腹瀉、腹脹及低血糖等病徵，另有肝臟病變及神經方面的症狀。至於慢性高酪胺酸血症多在一歲以後才發展出病狀，包括生長遲緩、腸胃道症狀、進行性肝硬化、多重腎缺損和佝僂症等臨床的表現。目前治療原則以特殊奶粉，並以藥物 NTBC 來治療。 | 遺傳性高酪氨酸血症第二型為酪胺酸胺基轉移酶缺乏症，主要症狀為眼睛和皮膚病變，眼睛的症狀為流淚、畏光且具有灼熱感，而皮膚症狀為手掌或足底有水皰及糜爛的產生，另外有些患者具有神經方面的病變如統合失調及語言遲緩等。目前治療以飲食控制為主。 |
| 特殊奶粉及藥品 | 特殊奶粉 Lofenalac、Phenyl-free(為衛生署公告及補助之特殊營養食品) | 特殊奶粉 MSUD、Ketonex-1（為衛生署公告及補助之特殊營養食品） | 特殊奶粉 Hominex-2 及 Low Met Product（為衛生署公告及補助之特殊營養食品） | 限制苯丙胺酸及酪氨酸之奶粉 | 限制苯丙胺酸及酪氨酸之奶粉 |

| 分　類 | 脂 肪 酸 代 謝 異 常 | | | | | |
|---|---|---|---|---|---|---|
| 疾病英文名 | Very long Chain Acyl-CoA Dehydroxygenase Deficiency | Long Chain Acyl-CoA Dehydroxygenase Deficiency | Medium Chain Acyl-CoA Dehydroxygenase Deficiency | Short Chain Acyl-CoA Dehydroxygenase Deficiency | Long Chain Hydroxy Acyl-CoA Dehydroxygenase Deficiency | Short Chain Hydroxy Acyl-CoA Dehydroxygenase Deficiency |
| 疾病中文名 | 極長鏈醯基輔酶 A 去氫酶缺乏症 | 長鏈醯基輔酶 A 去氫酶缺乏症 | 中鏈醯基輔酶 A 去氫酶缺乏症 | 短鏈醯基輔酶 A 去氫酶缺乏症 | 長鏈羥基醯基輔酶 A 去氫酶缺乏症 | 短鏈羥基醯基輔酶 A 去氫酶缺乏症 |
| 疾病簡介 | 人體由食物或奶粉攝取各種脂肪酸，而脂肪酸必須進入細胞的能量工廠-「粒線體」中以進行分解，分解所產生的能量可提供細胞進行使用。脂肪酸的分解若產生問題，身體許多功能便會失調，特別是肌肉、心臟、腎臟等器官。極長鏈脂肪酸的分解若出現問題，多以低酮性低血糖表現，另外，肝臟及心臟均會發生病變。飲食方面以少量多餐避免飢餓為主，限制長鏈脂肪酸的攝取與補充肉鹼是主要的治療原則。 | 長鏈脂肪酸的分解障礙則會造成餵食困難、肝臟及心臟腫大，另外，腦部的傷害及肌肉無力會陸續發生，若不及時加以治療，會有死亡的危險。目前飲食控制以高碳水化合物低脂肪為主，補充中鏈脂肪酸(MTC oil)、核黃素與肉鹼為治療原則。 | 中鏈脂肪酸分解障礙會造成突然發作的低酮性低血糖、嘔吐、嗜睡及昏迷，症狀會出現在嬰兒及幼童。此病類多發生於白人及北歐地區。治療方式在飲食方面應少量多餐，避免飢餓並限制長鏈及中鏈脂肪酸的攝取，並以肉鹼治療。 | 短鏈脂肪酸分解障礙通常會造成餵食困難及低血糖昏迷，生長可能會出現遲緩，飲食上必須避免飢餓，並以肉鹼來治療。 | 此亦為長鏈脂肪酸分解障礙之疾病，疾病症狀與長鏈醯基輔酶 A 去氫酶缺乏症相似。 | 此亦為短鏈脂肪酸分解障礙之疾病，疾病症狀與短鏈醯基輔酶 A 去氫酶缺乏症相似。 |
| 特殊奶粉及藥品 | | | | | | |

| 分　類 | 脂肪酸代謝異常（續） | | | | |
|---|---|---|---|---|---|
| 疾病英文名 | Carnitine Palmitoyl Transferase Type I Deficiency (CPS-I) | Carnitine/acyl carnitine Translocase Deficiency | Carnitine Palmitoyl Transferase Type II Deficiency (CPS-II) | Glutaria Acidemia Type II (Multiple Acyl-CoA Dehydrogenase Deficiency) | Multiple CoA Carboxylase Deficiency (Biotidase) |
| 疾病中文名 | 肉鹼棕櫚醯基轉移酶缺乏症第一型 | 肉鹼醯基肉鹼轉移酶缺乏症 | 肉鹼棕櫚醯基轉移酶缺乏症第二型 | 戊二酸血症第二型 | 多發性羧化酸酶缺乏症 |
| 疾病簡介 | 肉鹼(或稱卡尼丁)在人體內扮演重要角色，它負責將脂肪酸運送到細胞的能源中樞-「粒線體」,脂肪酸隨即在粒線體中進行分解並產生能量，若肉鹼無法與脂肪酸結合或運輸進入粒線體的功能喪失，都會導致疾病的產生，其症狀大多為心肌病變、低酮性低血醣昏迷以及肌肉無力等。肉鹼棕櫚醯基轉移酶缺乏症第一型是肉鹼無法與脂肪酸結合，第一次發病大多在兩歲左右，以低酮性低血醣昏迷呈現，此外，肝功能及腎功能都會受到影響。 | 此疾病為肉鹼與脂肪酸結合之後無法進入粒線體，通常在新生兒時期即發病，症狀為低酮性低血醣昏迷、心肺功能失調及心律不整等，治愈狀況不佳。 | 此疾病為肉鹼與脂肪酸在共同進入粒線體後，無法進行分離，分為成人型及新生兒型。成人型多因激烈運動之後造成心肌無力，新生兒型則對生命有威脅性，必須及時治療，包括低蛋白、低脂肪及高碳水化合物的飲食治療，應避免飢餓及保持體溫。肉鹼的補充是一種有效的治療方法。 | 此病症主要成因為多發性醯基輔酶A去氫酶缺乏所導致，因而造成脂肪酸及支鏈氨基酸代謝出現問題。主要症狀為新生兒低血糖、酸血症、肌肉無力、肝臟腫大等，另外，腳底會有汗臭味。飲食控制方面以高碳水化合物、低脂肪低蛋白為主，並以少量多餐進行，以補充核黃素與肉鹼為治療原則。 | 此病症是因體內缺乏維生素B中的Biotin，使得許多酵素功能不全。其症狀為癲癇、肌無力、免疫系統失調、皮膚出疹、頭髮掉落、聽力損失及智能障礙。治療原則以口服Biotin，為主，此症若早期診斷可達到極佳的治愈效果。 |
| 特殊奶粉及藥品 | | | | 特殊奶粉<br>Glutarex-2、Provimin、Xlys low try analog（為衛生署公告及補助之特殊營養食品） | |

| 分　類 | 有機酸血症 | | | | | | | |
|---|---|---|---|---|---|---|---|---|
| 疾病英文名 | Propionic Aciduria | Methyl-Malonic Acidemia | Isovaleric Acidemia | 3-Hydorxy-3-Methyl glutary CoA Lyase Deficiency | 3-Methylcrotonyl-CoA Carboxylase Deficiency | 3-Ketothiolase Deficiency | Glutaria Acidemia Type I | Malonic Aciduria |
| 疾病中文名 | 丙酸血症 | 甲基丙二酸血症 | 異戊酸血症 | 白胺酸代謝異常 | 甲基巴豆醯基輔酶A羧酸酶缺乏症 | 2-甲基乙醯基輔酶A酶缺乏症 | 戊二酸血症第一型 | 丙二酸血症 |
| 疾病簡介 | 此病症是有機酸血症的一種，所謂有機酸血症是蛋白質分解途徑出現障礙，許多有害的有機酸便出現在血液中，造成新生兒餵食困難、嘔吐、呼吸急促及昏迷等，若不及時治療，將有死亡的威脅。丙二酸血症的治療原則以降低血酸性為主，若無法以電解質溶液來降血酸，可進行血液透析，另外，可以肉鹼來補充間接性肉鹼缺乏。 | 此症因甲基丙二酸輔酶A變位酶功能異常，導致體內甲基丙二酸、丙酸等有機酸蓄積，造成一系列神經系統損害，嚴重時引起酮症酸中毒、低血糖、高血氨、高甘胺酸血症，新生兒、嬰幼兒期死亡率很高。 | 此病症為異戊酸輔酶A去氫酶缺乏症，發病年齡為0~1歲，急性期症狀包括嘔吐、缺乏食慾、無精打采、嗜睡、神經症狀、體溫低等，通常發作的原因為上呼吸道感染或攝取太多高蛋白食物。治療原則以限制蛋白質攝取及肉鹼和口服甘胺酸(Glycine)治療。 | 患者由於體內無法合成酵素來分解白胺酸，導致體內堆積有害人體的有機酸，若無法及時以藥物治療或食物控制，患者常會因酸中毒而致智障或死亡。發病徵狀如下：持續性嘔吐、四肢無力、盜汗、手腳冰冷、臉色蒼白、呼吸改變、抽筋痙攣、暴躁易怒、昏睡乃至昏迷。 | 此病症亦為白胺酸代謝異常之疾病，疾病症狀包括肌肉無力、癲癇及皮膚方面的病變。治療以飲食控制及補充肉鹼或Biotin為主要原則。 | 此病症之主要症狀為反覆性的酸血症，治療方式以重碳酸鹽及靜脈注射電解質溶液來降低血酸，若嚴重時得以進行血液透析，另外，肉鹼的補充也是重要的。 | 此症之成因為戊二基輔酶A去氫酶缺陷，導致分解離胺酸與色胺酸之代謝途徑有問題，造成有毒的代謝中間產物，如戊二酸等會過量堆積於血液與組織中並排泄到尿液，造成漸進的神經症狀及急性的代謝異常。一般而言，患者在兩歲之前發展正常，可能有無症狀的巨腦，在嬰兒期的晚期呈現出症狀，包括神經症狀如運動困難、漸進式的手足舞蹈症、肌肉低張到僵硬、麻痺、四肢向外翻轉，身體呈弓狀等，也可能會有癲癇或昏睡昏迷的急性發作。目前治療原則以飲食控制及核黃素與肉鹼補充為原則。 | 此病症為丙二酸輔酶A去羧酸酶缺乏，因此造成丙二酸輔酶A無法分解為醯基輔酶A。其症狀為生長遲緩、嘔吐、癲癇、低血糖及心臟病變。飲食控制以低脂肪酸及高碳水化合物為主。 |
| 特殊奶粉及藥品 | 特殊奶粉OS1(為衛生署公告及補助之特殊營養食品) | 特殊奶粉OS1、OS2及P80056(為衛生署公告及補助之特殊營養食品) | 特殊奶粉I-Valex-2(為衛生署公告及補助之特殊營養食品) | 特殊奶粉LEU1(為衛生署公告及補助之特殊營養食品) | | 特殊奶粉Glutariex-2 | | |

| 分　類 | 其　他 | | | | |
|---|---|---|---|---|---|
| 疾病英文名 | Citrullinemia | Arginosuccinic aciduria | Argininemia | Hyperammonemia/ Hyperornithinemia/ Homocitrullinuria syndrome | Non-Ketotic Hyperglycinemia |
| 疾病中文名 | 瓜胺酸血症 | 精胺琥珀酸血症 | 精胺酸血症 | 高血氨/高鳥胺酸血症/低瓜胺酸血症 | 非酮性高甘胺酸血症 |
| 疾病簡介 | 尿素代謝循環是人體內排除氨的主要途徑，尿素代謝循環若發生障礙，則血液中的氨就會大量增加，此類病患出生時並無明顯症狀，在經過餵奶數小時至數天後開始發病，剛開始會有嘔吐、餵食困難、吸吮力變差等現象，緊接著呼吸變得急促、常顯現倦怠感、有時會哭鬧不安及出現痙攣，而意識狀況則是逐漸惡化終至昏迷。若不及時加以治療即會造成智力受損及嚴重的神經系統損害。高血氨症又因基因缺陷的不同而細分為四類，瓜胺酸血症便是其中一項，症狀及如上述，治療原則以降氨藥如安息香酸或苯丁酸鈉鹽為主，另外，特殊奶粉及低蛋白的飲食限制也很重要。另外，補充精胺酸也可改善治療效果。 | 為高血氨症的一種，主要是精胺琥珀水解酶缺乏所致，症狀為嘔吐、餵食困難、吸吮力變差、呼吸急促、嗜睡，嚴重時會有昏迷的現象。治療原則與瓜胺酸血症相似，另外，補充精胺酸也可改善治療效果。 | 為高血氨症的一種，主要為精胺酸酶缺乏所致，疾病症狀及治療原則與瓜胺酸血症相似，均需以特殊奶粉控制蛋白質的攝取。 | 為高血氨症的一種，主要症狀為餵食困難、嘔吐、昏睡、肌肉張力低、痙攣、抽搐、癲癇、呼吸暫停及昏迷等。治療原則以特殊奶粉控制以及低蛋白質食物控制，除了降血氨藥物治療之外，必要時得補充瓜胺酸。 | 此病症為甘胺酸的分解過程障礙，因而造成嚴重的神經症狀，主要症狀包括癲癇、肌肉無力、嗜睡以及昏迷等，目前治療藥物以安息香酸鈉為主，同時給予抗癲癇藥物，低蛋白的飲食限制也是重要的，然而非酮性高甘胺酸血症的治愈機會不佳，通常不易存活。 |
| 特殊奶粉及藥品 | 特殊奶粉 UCD-1、UCD2、P80056(為衛生署公告及補助之特殊營養食品) | | | | |

資料來源：罕見疾病基金會網站（http://www.tfrd.org.tw）

## 三、罕病基金會「二代新生兒篩檢」的推動狀況

罕病基金會自民國八十九年十二月開始推展二代新生兒篩檢，首先是於 89 年 12 月推動二代新生兒篩檢先導計畫，補助臺大醫院及中國醫藥學院附設醫院實驗使用串聯質譜儀進行二代新生兒篩檢，就每

一篩檢個案補助耗材三十元整;90 年 9 月推動罕見疾病遺傳諮詢網，派駐遺傳諮詢員進駐十家醫學中心協助推廣二代新生兒篩檢及國際檢體外送服務；91 年 6 月推動金門縣二代新生兒篩檢先導計畫，與金門縣政府和台北病理中心合作全額補助金門縣每一位進行二代新生兒篩檢的費用；91 年起獲得 ING 安泰人壽 Family Day 贊助二代新生兒篩檢相關推廣費用；92 年 10 月起，推展原住民「二代新生兒篩檢計畫」，與臺大醫院、台北病理中心、衛生保健基金會共同合作，並由罕病基金會全額補助原住民進行二代新生兒篩檢，93 年起並推動「二代新生兒篩檢計畫」，開始全額補助低收入戶進行二代新生兒篩檢。

表 2-2 罕病基金會二代新生兒篩檢各項方案與期程

| 期程 | 方案內容 |
|---|---|
| 89 年 12 月 | 推動二代新生兒篩檢先導計畫（與臺大醫院及中國醫藥學院附設醫院合作） |
| 90 年 09 月 | 推動罕見疾病遺傳諮詢網 |
| 91 年 06 月 | 推動金門縣二代新生兒篩檢先導計畫 |
| 91 年 | ING 安泰人壽 Family Day 贊助二代新生兒篩檢 |
| 92 年 10 月 | 推展原住民「二代新生兒篩檢計畫」(與臺大醫院、台北病理中心、衛生保健基金會共同合作) |
| 93 年 | 開始補助低收入戶「二代新生兒篩檢計畫」 |

罕病基金會經過上述各項有關二代新生兒篩檢方案的推廣，累計

至 93 年 12 月 31 日止，已補助 313 萬元用於二代新生兒篩檢各項的補助，同時受益的新生兒已達五萬餘人，其中尚含弱勢族群原住民人數 2,732 人、偏遠地區人數 1,853 人、低收入戶人數 6 人。

表 2-3　罕病基金會二代新生兒篩檢計畫補助情形

單位：人、元

| 項目 | 中國醫藥學院附設醫院 | 台灣大學附設醫院 | 台北病理中心 | 衛生保健基金會 | 長庚醫院 | 合計 |
|---|---|---|---|---|---|---|
| 一般個案人數 | 2,694 | 45,259 | — | — | | 47,953 |
| 偏遠地區人數 | — | 1,528 | 325<br>(金門地區) | — | | 1,853 |
| 原住民人數 | — | 748 | 1,152 | 797 | 35 | 2,732 |
| 低收入戶人數 | | 2 | 3 | 1 | - | 6 |
| 總補助人數 | 2,694 | 47,537 | 1480 | 798 | 35 | 52,544 |
| 總補助金額 | 80,820 | 2,130,300 | 592,000 | 319,200 | 13,650 | 3,135,970 |

資料來源：罕病基金會二代新生兒篩檢計畫統計(89 年 12 月 22 日至 93 年 12 月 31 日止)。

罕病基金會自民國八十九年十二月推展二代新生兒篩檢至今，成績漸顯，一般民眾自願進行二代篩檢的比例已達至六成，同時，二代篩檢也確實發揮其功能，為新生兒健康把關，截至目前為止已有 15 位病童經篩檢後確診罹患代謝異常疾病，並且獲得適當治療，這些病童都是在原第一代新生兒篩檢中無法檢查出的。目前我國新生兒篩檢的檢體分別由臺大醫院、台北病理中心、及中華民國衛生保健基金會負責，91 年度合計使用第一代新生兒篩檢技術篩檢了 246,506 位新

生兒，其中另行使用第二代新生兒篩檢之普及率已達 52.04%，92 年度一至八月合計第二代新生兒篩檢之普及率已達 60.45%，並且篩檢出楓糖尿症、異戊酸血症、二戊酸血症、有機酸血症、高胱胺酸血症等 15 名病童。

## 參、美國各州擴大新生兒篩檢的推動狀況

### ◎亞利桑納州尋求立法擴大新生兒篩檢至 29 項

亞利桑納州 East Valley 論壇報於 2005 年 2 月 27 日指出，該州 SB1250 法案正尋求立法擴大新生兒篩檢。目前該州記篩檢 8 項新生兒篩檢項目，但是州健康部門與 March of Dimes 基金會正尋求擴大新生兒篩檢至 29 項，以及加入新生兒的強制聽力篩檢。

但是在這之前，負責州公共健康服務業務的助理主任 Rose Conner 指出，必須先保住既有的新生兒篩檢制度，因為持續運作 12 年來只收 40 元檢驗費用將不敷成本的增加，估計在 18 個月至兩年之間該項業務將無法在財務上自給自足，因此無論是要維持或擴大新生兒篩檢，該項費用都必須增加。2004 年該州篩檢了 90,000 新生兒，發現有 5,299 檢驗異常，但最後只有 112 名在臨床上確診異常。

St. Joseph 醫院臨床遺傳學家 Dr. Kirk Aleck 指出，新生兒篩

檢制度若不保，將使亞利桑納州更加落後各州，亞利桑納州過去檢驗 8 項的做法曾領先各州，所篩檢及確診的病患人數也多於其他地區，而他州投資篩檢技術和擴大篩檢項目的做法，也已將亞利桑納州拋在後面。

March of Dimes 基金會人員指出，目前有 39 州篩檢較亞利桑納州為多的項目，其中 12 個州篩檢項目已在 30 個以上。該基金會正致力推動全國新生兒篩檢檢驗的標準化，而未來數州，聯邦一顧問委員會預期將建議篩檢 29 項新生兒篩檢項目。

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East Valley Tribune; February 27, 2005
Bill Seeks Expanded Testing Of Newborns
http://www.eastvalleytribune.com/index.php?sty=37087
BILL SEEKS EXPANDED TESTING OF NEWBORNS
By Jennifer Ryan, Tribune

Deborah Houk never thought her daughter bout with the stomach flu would end in her death. At 19 months old, Lauren appeared healthy and normal, walking six months ahead of her older sister and used to fighting over their toys.

But Lauren couldn fight a genetic disorder her family never knew she had in 1989. The night of her stomach flu, certain stored fats in Lauren body turned toxic, and her father found her dead in her crib the next morning, her lips blue.

One test at Lauren birth could have saved her life, Houk said. That test, performed on her younger brother, Austin, when he was born, found the same disorder and allowed him to receive a lifesaving treatment, she said.

"As parents, how can we justify losing a child when it can be prevented?" said the Tempe mother. "How can you not test children?"

The question is being put to the test in the Legislature, where SB1250 would pave the way for expanded newborn testing in Arizona.

The state currently tests newborn blood for eight congenital disorders. The state health department and March of Dimes want to expand testing to cover 29 disorders, using the same drop of blood.

They also want to mandate newborn hearing screening and improve tracking of test results. Initial screenings are done voluntarily by hospitals, but 40 percent of babies who fail cannot be identified by the state for follow-up testing, according to the Arizona Commission for the Deaf and Hard of Hearing.

But first, public health officials need to save the newborn screening program they have. A $40 cap on charges for two rounds of tests that have been in the law for the last 12 years can no longer cover required technology upgrades, increased labor costs and the booming number of babies born in Arizona, said Rose Conner, the state health department assistant director for public health services. Also, the program will lose a federal grant in July that has helped support hearing tests for babies.

The department estimates that in 18 months to two years, the newborn screening program will no longer be able to sustain itself financially. The cap needs to be raised for the program to stay afloat and to expand testing, said Conner.

"Eventually there will be a fiscal impact on the department because we won have enough money to continue the current program," she said. "This is a very important public health program. These are disorders that have lifethreatening impacts on babies. The sooner we can detect them, the sooner we can provide interventions and improve the quality of their lives."

Last year, the state screened more than 90,000 infants and identified 5,299 with abnormal results, according to the state health department. Of that number, 112 babies ?2 percent of abnormal results ?were diagnosed with "clinically

significant disorders."

Most of the genetic disorders the state tests for now, and would test for under an expanded program, are rare. But public health authorities and medical providers argue that the costs to treat patients harmed because they were diagnosed late exceed testing expenses. Families like the Houks say it a matter of doing what right to save and improve children lives.

In its original form, SB1250 removed the $40 cap, allowing the health department and other parties to work out an increased charge in the regulatory process. But concerns from health insurers about uncertain costs for increased testing led to an amendment raising the cap to $70. The amendment and the bill were passed last week in the Senate Health Committee. The new cap would increase testing costs by about $2.7 million a year, assuming that 90,000 babies received two rounds of testing. The bulk of testing cost is picked up by private health plans and the state Medicaid program.

"It more about being able to plan for what costs will be," said Regena Frieden, a spokeswoman for Blue Cross Blue Shield of Arizona.

Although the cap is close to the amount the newborn screening program needs, the bill supporters said attaching a monetary amount in the proposed law will require a twothirds vote for approval, which could spell trouble for its passage.

"We e facing an uphill climb as far as the number of votes needed," said Jodi Liggett, a legislative consultant for March of Dimes. "We e still optimistic we can find 20 votes in the Senate, but it tough."

Losing the newborn screening program would put Arizona even further behind, said Dr. Kirk Aleck, director of clinical genetics at St. Joseph Hospital and Medical Center in Phoenix. While the state was once a leader with its eight tests, screening and identifying more people with genetic disorders than almost anywhere else in the nation, investments in new screening technology that broadens the number of tests that can be done have put many other states far ahead of Arizona, he said.

There are 39 states that screen babies for more disorders than Arizona does,

including 12 states that screen for more than 30 disorders, according to March of Dimes officials. The organization is pushing for national standardization of tests, and in the next several weeks, a federal advisory group is expected to recommend that all newborns be tested for 29 medical conditions. Those conditions include everything from sickle cell anemia to rare disorders that many doctors have never before seen in their patients.

"We e in the midst of a genetic revolution. We have all these rare, rare diseases that when you add them up, they aren so rare any more," Aleck said. "It a confluence of technology and increased knowledge of many, many, many more conditions than we had 30 years ago."

The state has one machine and has two more on order that will provide the level of technology needed to expand current genetic screening, said Conner. The technology, called Tandem Mass Spectrometry, is more expensive to run and maintain, but is required to test for conditions like medium chain acyl-Co-A dehydrogenase deficiency, or MCADD ?the disorder that prevented Lauren Houk from breaking down stored fats. Also, the substances used as part of older metabolic testing methods are being phased out, further necessitating the transition to new technologies, said Conner.

With increased testing, however, come greater chances of picking up genetic conditions for which the medical community has little knowledge or treatment options, doctors said. For this reason, March of Dimes has recommended screening only for disorders that can be detected through safe, reliable testing and treated with interventions that cure or significantly improve a patient health, Liggett said.

Still, expanded testing is expected to identify medical conditions in infants who could have varying degrees of illness, said Aleck. Fortunately the treatments, which often involve simple changes to diet or infant formula, will not hurt babies with milder forms of disease, he said.

"We have to start with those things we understand," he said.

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## ◎March of Dimes 基金會對紐約時報一文的聲明

March of Dimes 基金會對紐約時報有關新生兒篩檢的評論，於2005年2月22日發表聲明如下：

「March of Dimes 基金會對紐約時報於2005年2月21日有關新生兒篩檢一文的錯誤與誤導論調感到錯愕。新生兒篩檢是用來檢驗出生缺陷可能導致特定罕見、致命與殘障的一項安全和精確的方法，自1960年代中期以來，新生兒篩檢由於成效良好，以致被美國與世界大多數國家用來作為對新生兒的例行檢查。

紐約時報一文中對於篩檢的偽陽性問題提出過當的警告，事實上醫學上的篩檢需要容納相當數量的人數方能發現罹病的所有個案，即使初期必須付出一些偽陽性的代價，但是在對病患進行實際治療之前，被檢驗出陽性的家長與醫師都會接到再次複檢的通知，以便確定真正的結果。2000年紐約州有關新生兒篩檢的一項報告，已經否定新生兒篩檢出苯酮尿症（PKU）後因不當治療致死的說法，該報實不應重複該項從來不成立的說法。

美國境內每一州別與地區各自規劃與管理自我的新生兒篩檢制度，但是很遺憾的，這些制度在篩檢的數目與類型間存在極大差異，眼前我們面對的真正問題是，如何就最佳的新生兒篩檢和事後追蹤建立全國的統一標準，以確保所有新生兒的公平性。

March of Dimes 基金會支持美國基因醫學會（American College

of Medical Genetics）專家們的建議，這項建議也受到健康與人民服務部長有關新生兒與兒童遺傳異常與基因疾病顧問委員會的支持（Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children)，我們督促健康與人民服務部長接受訂定全國新生兒篩檢標準的建議。

March of Dimes 基金會也督促各州政策制定者，能確保他們的新生兒篩檢能涵蓋研究、開發、及驗證新的檢驗方法來確診和治療這些異常疾病，同時就受影響家庭提供即時的追蹤與諮詢服務。所有的新生兒篩檢都應提供高品質的檢驗，使其具備最新進的技術、訓練有素的人員、和資源來進行即時追蹤與方案評估。March of Dimes 基金會支持有關新生兒篩檢和品質確保有關的項目擴增，以及規範、政策、和程序的開發。

March of Dimes 基金會各州分會與其夥伴將繼續與州長們、州議員、和健康部門緊密合作來提升各州的新生兒篩檢，我們也敦促各州應儘可能的知會所有家長有關擴大新生兒篩檢的潛在利益與可及性，我們也支持家長有權充分知悉有關其新生兒的篩檢結果，我們也支持擴大對醫療提供者有關新生兒篩檢的教育。」

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March of Dimes; February 22, 2005
The New York Times Article on Newborn Screening

http://www.marchofdimes.com/aboutus/14458_15137.asp

THE NEW YORK TIMES ARTICLE ON NEWBORN SCREENING

The March of Dimes is dismayed by the inaccuracies and misleading tone of The New York Times February 21, 2005 article on newborn screening. Newborn screening is a safe and accurate way to test babies for certain rare but deadly or disabling birth defects. Since the mid-1960s, newborn screening programs have been so successful that screening is now routine for millions of babies born each year in the United States and in much of the world.

Undue alarm is raised in the Times article about false positive screening test results. Medical screening programs must cast a wide enough net to identify all affected individuals, even at the cost of some initial false positives. But the family of any newborn who screens positive for a disorder, along with the pediatrician, would receive a notice to bring the baby in for further testing to confirm the diagnosis before any treatment would begin. The New York State Task Force on Life and the Law Newborn Screening report in 2000 rebuffed claims regarding lethal effects from inappropriate treatments for PKU diagnosed through newborn screening. The Times should not have repeated claims that have never been substantiated.

Each state or region in the U.S. designs and operates its own newborn screening program, and, unfortunately, these programs vary widely in the number and type of conditions for which they screen. The real issue before us now is how to obtain equity for all babies by creating a uniform national standard of best practices for newborn screening and followup. The March of Dimes supports the recommendations made by the experts of the American College of Medical Genetics, and endorsed by the HHS Secretary Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. We urge the Secretary of Health and Human Services to accept these recommendations as a national standard for newborn screening for the states.

The March of Dimes urges state policy makers to ensure that their newborn screening programs include research, development, and validation of methods to detect and treat disorders, as well as prompt follow-up and counseling for affected families. All newborn screening programs should provide high quality screening tests with state-of-the-art technology, trained personnel, and

resources for timely follow-up and program evaluation. The March of Dimes supports expansion of capacity and development of standards, policies, and procedures for newborn screening programs and quality assurance.

March of Dimes state chapters and their partners will continue to work closely with governors, state legislators, and health departments to improve state newborn screening programs. We urge the states to inform all parents prospectively about the potential benefits and availability of comprehensive newborn screening. We also support parents' rights to be fully informed about their baby's screening results, and we support the expansion of health care provider education about newborn screening.

March of Dimes News Desk
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**◎Mayo 對串聯質譜儀檢驗的新發現**

美國明尼蘇達州 Mayo 醫院小兒遺傳醫師 Piero Rinaldo 於 2005 年二月於華盛頓所召開的 American Association for the Advancement of Science (AAAS)年會中指出，經過使用串聯質譜儀的雙重分析後，將可以擴展到檢驗約 40 種新生兒篩檢的項目，遠較目前美國各州檢驗的項目為多。Piero Rinaldo 指出，目前串聯質譜儀約可檢驗 40 種新生兒篩檢的項目，但在許多州中尚未引用，由於各州做法不一，以致部份州的新生兒將可能導致確診太遲的問題；他指出這些絕大多數的基因疾病個案，只要早期確診並給予治療將能得到治癒或改善，但部份個案若無法於出生後數天診察，將發生腦部傷

害甚至死亡。

除了各州做法不一的一致性與公平性問題外，Dr. Rinaldo 也指出串聯質譜儀操作上的差異，也導致品質參數的顯著差異，例如偽陽性的比率和陽性的判斷值等。他指出為改善檢驗的品質，未來可以使用生化或分子生物的方式就同一檢體加作另一項檢查，如此也可以避免因向初期檢驗陽性的家庭再次收集血液樣本而造成壓力與焦慮。Mayo 醫院已經與明州衛生部門合作進行串聯質譜儀的雙重分析，已經使偽陽性比率由 2003 年的 0.5% 降為 0.06%，此舉不僅可以改善篩檢的品質，也更具成本效益。Dr. Rinaldo 也指出未來的發展，篩檢的制度將從新生兒期繼續擴大而納入 Wilson 疾病與 glycosylation 代謝異常等。

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WISTV-Channel 10 (Columbia, South Carolina); February 23, 2005
Fed Panel Wants More Screening For Newborns Rare Diseases
http://www.wistv.com/global/story.asp?s=2989431&ClientType=Printable

FED PANEL WANTS MORE SCREENING FOR NEWBORNS RARE DISEASES

(National-NBC) Feb. 23, 2005 - Stephen Monaco went to the hospital four-years-ago with a stomach virus. His mother Jana Monaco says overnight her son became severely brain damaged and permanently disabled, "In 24 hours, you know, we nearly lost him."

The virus had triggered a rare genetic disorder called isovaleric acidemia or IVA.

Stephen's two-year-old sister Caroline has the same disorder. She was tested at birth and Jana says through medication and a special diet, she's Ok, "A $25 test would have made a world of difference for us."

The test isn't required in Virginia, where the Monacos live. In fact, newborn screening varies widely from state to state.

Dr. Michael Watson with the American College of Medical Genetics says a federal advisory panel is recommending that all states test for 29 rare diseases, "You avoid the very bad outcome that may result in many of the cases. And you are also able to avoid the futile therapies that may be put in place for an infant who is affected."

There are concerns. Some tests have a high rate of false positives, causing needless worry and causing children to be treated for conditions they never had.

Dr. Norman Fost at the University of Wisconsin points to early problems and even some deaths with the now-popular PKU test, "Years later we found out they were killing normal children, making children brain damaged who were at risk for nothing."

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## ◎聯邦一顧問委員會將建議篩檢 29 項新生兒篩檢

紐約時報於 2005 年 2 月 21 日指出， 某一極具影響力的聯邦顧問委員會將計畫於未來數週建議政府對新生兒篩檢 29 個罕見的疾病，這些疾病只被極少數的醫事專業所熟悉，同時影響的家庭也極少。

這項建議預期也將引發激辯，支持者認為這些疾病極為嚴重，唯有早期發現方能救命，當出生時未進行檢查，家長後續將面對繁複的檢驗過程，但當確診實往往已錯失最佳的治療時機。

反對者則認為除了五、六項疾病較為明確外，其餘多數篩檢的疾病尚無法得知治療是否有助益，或是得知到底有多少病童有驗出陽性但卻未發病的情形；反對者認為有一項風險，即是輕微的疾病有可能被視為極為嚴重而接受無謂的與積極性的治療，以致影響健康。

支持與反對的雙方也都同意， 該項篩檢除了有意找出 29 項疾病外，也同時會發現 25 種異常的檢驗結果，但尚無人知道這些異常的數據究竟意義為何？也尚無法得知是否與疾病有關聯，若有關聯，則影響為何也無法得知；該聯邦顧問委員會建議這些情形也應告之家長，只是這項告知也仍有爭議。

雖然家長有權拒絕這項篩檢，但目前家長多未被知會即逕行篩檢，而這些約介於 70 至 120 美元的費用，便直接列入帳單中。該委員會的主席也是建議的起草者，邁阿密大學的 Dr. Howell 則建議未來將統整各州有一致的標準，目前有些州只篩檢 4 項，有些州則篩檢 35 項之多。

威斯康辛大學小兒科的教授與醫學倫理中心的主任 Dr. Norman Fost 則提出批評，指出過去苯酮尿症（PKU）錯誤歷史；苯酮尿症為歷史上第一項新生兒篩檢，目前各州也都進行此項篩檢，也被視為拯救新生兒與預防殘障的典範，但 Dr. Norman Fost 則指出數十年前並非如此。該病發生率約為十四萬分之一，每年約影響三百個小孩，特

殊奶粉可以預防智障與神經傷害；1959 年 University of Buffalo 的 Dr. Robert Guthrie 因侄子罹患苯酮尿症而積極研發該病的血液檢驗，並成功游說各州強制篩檢苯酮尿症。

Dr. Norman Fost 指出，這項檢驗過去假定陽性即代表嬰兒帶有疾病，以及特殊飲食計畫是有效且安全的；但結果卻是大錯特錯，因為部分檢驗陽性的嬰兒並未帶有該病，同時特殊飲食計畫也可能有相同的危險。由於若對正常的小孩限制攝取 phenylalanine，不僅對腦部有傷害更使每一細胞的養分攝取不足，以致正常小孩也變成智障。因此在 1960 年代中期，美國小兒科醫學會（American Academy of Pediatrics）發函健康、教育、暨福利部長，指出醫學上無法區分偽陽性與陽性，並建議停止強制篩檢；但後續並無下文。直到 1970 年代，偽陽性的問題才獲解決，並成為重要的典範。Dr. Norman Fost 也批評當醫學倫理家要求再多進行一些臨床實驗時，這些病童的家長團體、家長團體、倡導團體往往認為不能再浪費時間，而急著推動。

但事實上，來自家長的真實故事卻有不同的說法。威斯康辛州 Micki Gartzke 於 1996 年出生的女兒 LeA，即因罹患 galactocerebrosidase 酵素缺乏的罕見基因疾病，因未在早期階段確診，經兩年多的折騰以及耗費了約 25 萬美元診治後，仍不治身亡。Gartzke 太太認為若能早期診斷，就有活命的機會。

March of Dimes 基金會的醫學主任 Dr. Nancy S. Green 則認為，該基金會所建議的 29 項新生兒篩檢項目都具有合理介入的理由，在醫學上也都有根據，也是許多醫是專家集思廣益的結果。由於這些疾病都極為罕見，Dr. Nancy S. Green 也因此認為要求就這些治療與檢驗進行嚴格的科學研究並不確實際，同時每一項疾病每年都只有數十個病患被發現，因此要收集這些資料可能都要數十年，醫師專家們也因此別無選擇，僅能就專業所知告訴家長有關的檢驗與諮詢。不過 Vanderbilt University 的法律與小兒科教授 Dr. Ellen Wright Clayton 則認為，在有關評估治療方面，委員會多依賴最低程度的證據，即只有一是專家與倡導團體的個人意見。

猶他大學小兒科與倫理學的教授，Dr. Jeffrey Botkin，也是美國小兒科醫學會倫理委員會的主席，則提出在規劃的 29 個項目中，只有苯酮尿症（PKU）和或許五種項目前已知治療有效。他認為有問題的檢查包括瓜胺酸血症（citrullinemia）和 arginosuccinic 酸血症，這些都會累積阿摩尼亞而導致昏迷和死亡，而酪胺酸血症（tyrosinemia）則由於缺乏酵素所致，除非進行肝臟移植，否則也可能致死，而每一種代謝異常一年影響的小孩都少於一百人。由於相關新生兒篩檢的情況未必充分理解、治療的情況也不十分清楚，Dr. Botkin 因此也不確定人們是否能接受這項篩檢。在這種情況下，就

使得檢驗出陽性而可能未發病的問題更受注視，他指出通常篩檢都能找出許多這種案例，她們治療即使並不太困難，也可能終生被標籤化為未來將發病，不過這也許是拯救生命的另一種代價，至少你不是沒在拯救生命，或是根本不知正在拯救生命。

目前建議報告雖未公開發表但已經由委員於會中發表，下一步即是正式發行報告，Dr. Watson 說將會繼續收集各種評論，但是公眾的評論將不會改變結論，未來的正式結論將發表於小兒科期刊，未來就看健康與人類服務部長的決定。Dr. Watson 認為統一的新生兒篩檢攸關平等的問題，各方的對話可能延後，直到因新生兒出生在錯的州而診斷太遲時，那時就不得不有行動了。

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New York Times; February 21, 2005
Panel to Advise Testing Babies for 29 Diseases
http://www.nytimes.com/2005/02/21/health/21baby.html

PANEL TO ADVISE TESTING BABIES FOR 29 DISEASES

By GINA KOLATA
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An influential federal advisory group plans to recommend in the next few weeks that all newborns be screened for 29 rare medical conditions, from the well known, like sickle cell anemia, to diseases so obscure that they are known to just a handful of medical specialists and a few dozen devastated families.

But while no one argues with the idea of saving babies, the proposed screening

is generating fierce debate.

The dispute centers on how useful the test findings would be. Would going ahead with the full list of tests result in more good than harm, physically and emotionally? Or would it be better to forgo most of them?

Proponents say that the diseases are terrible and that an early diagnosis can be lifesaving. When testing is not done, parents often end up in a medical odyssey to find out what is wrong with their child. By the time the answer is in, it may be too late for treatment to do much good.

But opponents say that for all but about five or six of the conditions, it is not known whether the treatments help or how often a baby will test positive but never show signs of serious disease. There is a danger, they say, of children with mild versions of illnesses being treated needlessly and aggressively for more serious forms and suffering dire health consequences.

And both sides agree that the tests unintentionally pick up about 25 other conditions, in addition to the 29 that the screening is intended to find. These additional conditions show up as abnormalities, but no one knows what they mean. It is not known whether they are associated with a disease or, if so, what the effects will be.

The federal advisory group recommended informing the parents of such results. But that advice, too, is controversial.

"Giving parents the result, saying, 'Here's the mutation; we are not sure what the outcome will be,' is better than not telling," said Sharon Terry, president and chief executive of the Genetic Alliance, an advocacy group for people with genetic disorders. Ms. Terry said it was paternalistic for doctors to presume that it was better for parents not to know.

Dr. R. Rodney Howell, a professor of pediatrics at the Leonard M. Miller School of Medicine at the University of Miami and the chairman of both the committee that wrote the report and the federal advisory group, agreed.

"Do I feel it will be difficult for physicians and caretakers to deal with this?" Dr. Howell said. "The answer is yes. But I just don't think it is proper for us to

have information about an abnormality without conveying it."

But Dr. Lainie Friedman Ross, a pediatrician and medical ethicist at the University of Chicago, said: "We don't know if they are medical conditions. We don't know what to do with the information. Reporting test data for which there are no systems in place for follow-up testing and treatment is not rejecting paternalism, but it is patient abandonment."

In any event, Dr. Howell said, noting that states were plunging into testing programs: "It's not really a question of, 'Should we expand newborn screening?' It's happening. It's going like a house on fire."

In most states today, parents are not asked if they want their babies tested, though they have the right to decline it; it is simply done, with the cost, about $70 to $120, built into their hospital bills. Dr. Howell said the idea of the new recommendations was "to try to organize the programs and to try to be consistent from state to state."

"Some states screen for four conditions; others screen for 35," said Dr. Michael S. Watson, the federal project's director and the executive director of the American College of Medical Genetics. "A family can have their first child in one state where 25 conditions are screened and then move to another where only four are screened."

Yet, critics say, the fact that testing is happening does not mean that it should be expanded. The history of newborn screening, they say, is filled with cautionary tales.

"The majority of newborn screening tests have failed," said Dr. Norman Fost, a professor of pediatrics and director of the program in medical ethics at the University of Wisconsin. Over the years, Dr. Fost said, "thousands of normal kids have been killed or gotten brain damage by screening tests and treatments that turned out to be ineffective and very dangerous."

To those who ask what is wrong with simply doing every available screening test, Dr. Fost tells what happened with PKU, the first genetic screening test for newborns. Today every state tests for PKU, or phenylketonuria, and it is widely acknowledged as the perfect example of screening that saves lives and prevents

disability. But Dr. Fost says that a few decades ago, the situation was not nearly so rosy.

The disease, affecting one in 14,000 babies, or about 300 each year, involves a missing or defective enzyme that metabolizes the amino acid phenylalanine. Untreated, it leads to mental retardation and neurological damage. But a special diet low in phenylalanine can prevent those consequences.

PKU and its cause were understood by the 1950's, but little could be done. By the time anyone knew there was a problem, the baby was several weeks old and the damage was done.

In 1959, Dr. Robert Guthrie, a microbiologist at the University of Buffalo whose niece had PKU and who was passionate about stamping it out, developed a simple blood test for the condition. Then he began to lobby and soon every state had a law mandating that newborns be tested for PKU.

The testing made two assumptions: that a positive test meant a baby had the disease, and that the special diet was safe and effective.

"Both were stone cold wrong," Dr. Fost said.

Some babies testing positive did not have PKU. Although no one knew it at the time, these babies had a different mutation that was of no clinical significance.

And the special diet could be as dangerous to these normal babies as a regular diet was to babies with PKU.

"If you give a normal kid a diet without enough phenylalanine, not only is there brain damage but every cell in the body is malnourished," Dr. Fost said. "Normal kids became brain-damaged. Many died."

In the mid-1960's, the American Academy of Pediatrics wrote a letter to the secretary of health, education and welfare, Dr. Fost said.

"They said: 'There is a big problem here. We don't know what a true positive test means. We can't distinguish a true positive from a false positive, and we don't know what the right dose of the diet is. Mandatory screening programs

should be stopped.' "

But nothing was done, Dr. Fost said, adding, "This train had left the station, and no one wanted to say we had screwed up."

Finally, by the 1970's, the problems were sorted out. Researchers learned to distinguish true PKU from the innocuous condition and how best to prevent the consequences of the disease with a diet. "Now it is an exemplary program," Dr. Fost said of PKU screening.

PKU was not the only screening test that ran into problems.

It happened, for example, with a test for acidic blood in premature babies, whose immature lungs have difficulty excreting carbon dioxide. The gas combines with water in blood to form carbolic acid. Eventually blood can be acidic enough to be lethal. The treatment seemed obvious: neutralize the acid with intravenous bicarbonate of soda. So for more than a decade, from the 1960's until the mid-1970, every hospital in the country routinely screened premature babies for acidic blood and treated them with bicarbonate of soda.

"It had a religiosity to it," said Dr. Michael Simmons, a professor of pediatrics at the University of North Carolina. "You see blood that was acidic, and you have a drug that can fix it."

But with Dr. Frederick Battaglia, now an emeritus professor at the University of Colorado, Dr. Simmons and his colleagues discovered that babies who got bicarbonate of soda actually did worse and, in particular, had more brain hemorrhages, which can cause devastating brain damage.

Their study, published in 1974, changed medical practice. And Dr. Simmons said it taught him a lasting lesson: "Don't begin therapy until you know it will work."

Dr. Fost said that lesson was all too often forgotten.

"In all these cases of newborn screening gone haywire, there is usually some understandably zealous group of parents of sick kids, patient groups, advocacy groups saying 'Let's get on with it,' " Dr. Fost said. "Some ethicists asked for

clinical trials, but these groups said, 'We don't have time to waste.' "

But parents of children with genetic diseases often tell a different sort of story.

Micki Gartzke of Shorewood, Wis., gave birth to a girl, LeA, on Oct. 14, 1996. When LeA was just a few months old, her body became rigid, she would not eat and she cried inconsolably. She was found to have a rare genetic disorder that made her deficient in an enzyme, galactocerebrosidase, needed in the early stages of brain development. After two years of suffering and a quarter of a million dollars in medical bills, LeA was dead.

Early diagnosis, Ms. Gartzke said, could have led to a lifesaving transplant of umbilical cord blood.

The question posed by the new screening recommendations is whether they will lead to the kind of dangers that followed early PKU testing, or whether they will correctly identify babies like LeA who might have been saved.

Medical specialists say it is difficult to know.

Some, like Dr. Nancy S. Green, the medical director of the March of Dimes, say that each of the 29 conditions "has a reasonable intervention."

The treatments make medical sense, Dr. Green said, and experts in the diseases advise using them. "Keep in mind that there was considerable expert input," she said.

The disorders are extremely rare, she and others said, making it unrealistic to demand the most rigorous scientific studies of the tests and their treatments. With just a few dozen babies born with a condition each year, it could take decades to get such data. Medical experts have little choice but to use their informed judgment about the tests and the therapies.

But Dr. Ellen Wright Clayton, a professor of law and pediatrics at Vanderbilt University, said that in assessing treatments, the committee had relied mostly on "the lowest form of evidence": the personal opinions of medical specialists and advocacy groups.

Dr. Jeffrey Botkin, a professor of pediatrics and medical ethics at the University of Utah and chairman of the ethics committee of the American Academy of Pediatrics, asserted that only PKU and perhaps five other conditions among the 29 had treatments that were known to work.

Tests he considers problematic include those that identify disorders like citrullinemia and arginosuccinic aciduria, which lead to an accumulation of ammonia and can result in coma and death; and tyrosinemia, caused by a missing enzyme, which can be lethal unless the child has a liver transplant. Each disorder affects fewer than 100 babies a year.

"The conditions are not well understood, the spectrum of the disease is not well understood, it is uncertain how efficacious the treatments are, and it is uncertain how well people can tolerate the treatments," Dr. Botkin said.

When little is known about the effectiveness of screening and treatments, Dr. Botkin said, that raises concerns about babies who test positive but have a mild form of a disorder. He added that screening tests typically picked up substantial numbers of such babies.

Even if the treatment is not onerous, these people "will go through a lifetime of being labeled with a condition they might never have gotten sick with," Dr. Botkin said. "That may be a price that is acceptable if you are saving lives," he said, "but not if you are not saving lives or if you don't know if you are saving lives."

The next step will be to issue the report, which has been presented by panel members but has not been made public. Then comments will be solicited. But, said Dr. Watson, "the public comments won't change our report," which he said would be published in the journal Pediatrics. The comments, he said, will be used by Michael O. Leavitt, the secretary of health and human services, in deciding what to recommend.

Dr. Watson says uniform newborn screening is a matter of equity if nothing else. A diagnosis can be delayed until it is too late simply because the baby was born in the wrong state.

But equity, once again, depends on who is asked.

"Fairness is only an issue when you talk about benefits," Dr. Fost said.

And the benefits, he added, are largely unknown.

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## ◎Florida 進行二代新生兒篩檢 35 項

美國佛羅里達州自 2005 年二月開始，將引用串聯質譜儀開始從該州東北部進行 35 種項目的新生兒篩檢。該州自 2002 年起即任命一個工作小組進行新生兒篩檢業務的評估，事後並作成決議建議該州政府將新生兒篩檢的項目由五項擴增為 30 項，時間上較 2004 年秋天美國基因醫學會（American College of Medical Genetics；ACMG）對於新生兒篩檢的報告，與 March of Dimes 基金會所建議的 29 項檢驗為早。合計原來佛羅里達州已篩檢的項目以及新增的項目，將有 35 種項目列在新生兒篩檢中。

原先估計擴大新生兒篩檢將需要三百萬美元預算，如今在醫院付與州所屬檢驗單位每位新生兒 15 美元檢驗費，以及聯邦 Medicaid 補助差額後，如今州政府已毋需額外經費支出。

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Bradenton Herald (Bradenton, FL); February 19, 2005
Expanded Baby Testing Coming Soon To This Area
http://www.bradenton.com/mld/bradenton/living/health/10929580.htm

# EXPANDED BABY TESTING COMING SOON TO THIS AREA

KATIE POWERS
Special to The Herald

Iread in the paper last year that Florida was going to start testing babies for 30 metabolic tests starting in February of this year. I took my newborn baby in for testing last week and they only did 5. When will the state start testing for 30 metabolic disorders?

The state of Florida has obtained the tandem mass machines that are capable of doing the expanded tests. We have learned from other states that have expanded their programs that before you start testing you need to have the proper follow up care available in case a baby does test positive. Lois Taylor at the Department of Health in Tallahassee told me she wants to make sure that "we do it right to benefit those that will need care." Florida is being very methodical in the advancement of the testing.

In February of this year the expanded testing was started in northeast Florida.

In 2002 a taskforce was convened to determine the efficacy of Florida's Infant Screening program. The taskforce recommended that Florida expand it's testing from 5 tests to 30 and that it change the way it charged hospitals to do that test.

Florida had great timing. We had looked at our metabolic screening program and realized that changes needed to be made.

In the fall of 2004 the American College of Medical Genetics and the March of Dimes recommended that all states test for a minimum of 29 metabolic disorders. This represented the first step towards developing national standards for infant metabolic screening. Florida was already screening for some of the tests recommended and was also testing for some that were not on the recommended list. By combining what Florida was already doing and what was proposed that needed to be done Florida will be actually be testing for 35 disorders.

The expense of expanding the screening program was estimated to be around

3 million dollars. Prior to last year hospitals paid the state lab $20 for the first metabolic screening done on each baby. The problem was that hospitals only had to pay up to 3000 patients. Some hospitals were delivering many more babies than that but didn't have to pay for those babies. Now every hospital pays $15 for each baby no matter how many babies they deliver. Medicaid also agreed to help supplement that payment which allowed the lab to draw down some Federal funds. Because of those changes no money was needed from the state's budget.

If you have a family history of a metabolic disorder you may want to take your baby to a private lab and ask them if they test for that disorder. Screening will still be available in private labs until the state lab is testing 100 percent of the babies born in Florida.

Katie Powers, R.N., is a board-certified lactation consultant and perinatal educator at Manatee Memorial Hospital's Family BirthPlace. Her column appears every other week in WellBeing. Contact her at katie.powers@mmhhs.com.

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## ◎Oklahoma 新增兩項新生兒篩檢並逐漸朝向 30 項

美國奧克拉荷馬州自 2005 年 2 月 14 日情人節起，新增 cystic fibrosis (CF)和 congenital adrenal hyperplasia (CAH)兩項新生兒篩檢項目，同時確診後所提供的後續追蹤服務，也將確保治療以保障新生兒健康與避免嚴重殘障，同時 2005 年下半年預計再增加 medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD)一項，而奧克拉荷馬州府也已購置串聯質譜儀，預期逐漸篩檢 30 項代謝異常的項目，朝向 March of Dimes 基金會所建議的目標。

在 2004 年奧克拉荷馬州合計篩檢了約五萬名新生兒，並發現了 32 名基因異常的新生兒，新增的兩項篩檢，預計每年將發現 17 名 cystic fibrosis (CF)以及 2 名 CAH 的病童。Dr. Pam King 指出，篩檢的成本遠低於無法及時發現致病嬰兒的成本。錯過這些檢驗，醫師將無法及早發現病患，而將造成後續經常的進出醫院與繁複的檢查。研究發現，後續每一位病童進出醫院和診察的成本即輕易的超過 50 萬美元，而因未及時發現所致的殘障服務與收容機構的費用，甚且每年超過一百萬美元；但儘管如此，尚無法估算因失去親人的重大成本。州健康部的官員 Dr. Michael Crutcher 並說，這也是爲甚麼我們對於州健康部門的委員能如此致力提升本州兒童健康十分感激的原因，而擴大新生兒篩檢能順利完成，也是有賴州議員、家長與醫療提供者的多元助力，而提升新生兒篩檢也是預防殘障、改善健康、與拯救生命的途徑。

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The Ponca City News ;February 11, 2005
Valentine's Day Marks Expansion Of Newborn Screening in Oklahoma
http://www.poncacitynews.com/cgi-bin/LiveIQue.acgi$rec=63323?Lifestyle

VALENTINE'S DAY MARKS EXPANSION OF NEWBORN SCREENING IN OKLAHOMA

Valentine's Day, Monday, Feb. 14, will have special relevance for Oklahoma families of newborns born that day and afterward. Effective that date, the Oklahoma State Department of Health will expand its newborn screening

program to include screening for two additional disorders: cystic fibrosis and congenital adrenal hyperplasia. State public health officials say these tests and the provision of follow-up services to ensure treatment within the first weeks of life for affected newborns will help saves lives and prevent severe disability.

To commemorate the expansion of the newborn screening program, selected public health officials and newborn screening program staff will be wearing a bright blue wristband on Valentine's Day that says, "Breathe." While the simple act of breathing is something most persons take for granted, it can be extremely difficult for someone living with cystic fibrosis (CF). Testing for CF in newborns will allow for early detection of the disorder so complications can be prevented or improved. Newborns will also be tested for congenital adrenal hyperplasia (CAH), a disorder that can lead to death if not treated within the first weeks of life.

The addition of these two tests is an important step in moving Oklahoma toward meeting March of Dimes recommendations for uniform national newborn screening. In addition to funding the expansion of the newborn screening program to include CF and CAH screening, the State Board of Health has added a third test for implementation later this year. That test will be for the metabolic disorder medium-chain acyl coenzyme A dehydrogenase deficiency (MCAD). The Oklahoma State Department of Health has purchased new equipment to allow for MCAD testing as well as 30 other metabolic disorders.

Oklahoma is one of nine states offering CF screening to all newborns, and is the 39th state to add CAH screening. Oklahoma's newborn screening program also screens for the disorders of phenylketonuria (PKU), congenital hypothyroidism, classic galactosemia, and sickle cell disease. In 2004, the Oklahoma State Department of Health screened more than 50,000 newborns for genetic disorders and identified 32 newborns with a disorder. Expanded screening is expected to identify 17 newborns with CF and two with CAH each year.

In addition to screening, Oklahoma newborns identified with a disorder receive comprehensive follow-up program services to ensure optimal healthy outcomes, a unique service when compared to newborn screening efforts provided by other states.

"The consequences of not detecting these conditions early are devastating. Without early detection and subsequent intervention, newborns may suffer profoundly, and for some of the disorders, there is a risk of death," said Pam King, director of Genetics. "The cost to screen an infant is far less than the cost of failing to promptly identify affected infants."

Without the screening test, doctors are often unable to identify these disorders quickly. Failure to screen one affected infant can result in an odyssey of frequent hospital stays and invasive procedures trying to determine a diagnosis. Studies have shown costs for hospitalizations and diagnostic procedures can easily exceed $500,000 for one infant. The cost for disability services and in some cases institutionalization for profoundly affected infants who did not receive prompt treatment can exceed $1 million annually.

"Even so, these costs are immeasurable when compared to the effect that the loss of an infant's life has on a family," said State Health Commissioner Dr. Michael Crutcher. "That's why we are indebted to the Board of Health and its many partners who were committed to improving the health of Oklahoma's children."

Crutcher said expansion of the newborn screening program was successful due to diverse support from legislators, families and health care providers.

"Enhancing newborn screening services is a way to prevent mental retardation, improve health, and save lives. When we improve the quality of life for our children, we also improve the quality of life for Oklahoma's future," Crutcher said.

For more information about newborn screening services, contact Pam King, director of Genetics, Oklahoma State Department of Health, 405/271-6617.

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## ◎新英格蘭新生兒篩檢計畫

加拿大知名的 Tm Bioscience 公司與新英格蘭地區新生兒篩檢計畫（The New England Newborn Screening Program）於 2005 年一月 27 日發布合作聲明，將由 Tm Bioscience 公司提供新英格蘭地區新生兒篩檢計畫有關 Tag-It(TM)基因突變檢驗服務。

新英格蘭地區新生兒篩檢計畫係由美國麻州大學醫學院所管理，為針對麻州、緬因州、新罕布什爾州、羅德島州、佛蒙特州、賓州新生兒的公共健康篩檢計畫，每天約篩檢五百位新生兒。該計畫主任 Dr. Anne Marie Comeau 指出，基因檢驗已快速成為照護新生兒的主要工具，能藉著及早診斷極不易發現的疾病而給予適當照護，而對兒童健康有重要的正面影響，Tm Bioscience 公司快速、精確且彈性的基因檢驗極適合新英格蘭地區的新生兒篩檢計畫。

新英格蘭地區新生兒篩檢計畫位於麻州的 Jamaica Plain，自 1997 年 7 月 1 日起由麻州大學醫學院所管理，該計畫雇用了 12 位實驗室的技術員和五位督導員，都受過密集訓練與符合聯邦 Clinical Laboratory Improvement Amendments (CLIA)從事複雜檢驗的規範，此外尚有三位博士與六位醫學博士確保檢驗品質，及提供醫療社群有關新生兒篩檢所發現罕見疾病的後續協助。

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Tm Bioscience Press Release; January 27, 2005
Tm Bioscience to Supply Tests for New England Newborn Screening Program
http://biz.yahoo.com/prnews/050127/to111_1.html

Press Release
Source: Tm Bioscience

Tm Bioscience to Supply Tests for New England Newborn Screening Program

- Tag-It(TM) kits to be used in comprehensive public health screening program for newborns -

TORONTO, Jan. 27 /PRNewswire-FirstCall/ - Tm Bioscience Corporation (Toronto, Ontario; TSX: TMC), an emerging leader in DNA-based diagnostic testing, today announced an agreement to supply The New England Newborn Screening Program (Jamaica Plain, Massachusetts) with the Company's Tag-It(TM) mutation detection tests.

The New England Newborn Screening Program, operated by the University of Massachusetts Medical School, is a comprehensive public health screening program for newborns in the states of Massachusetts, Maine, New Hampshire, Rhode Island, Vermont and Pennsylvania - about 500 babies every day. The program provides high quality, timely, low-cost laboratory screening, clinical follow-up and research to prevent or minimize the effects of disorders that can lead to death, mental retardation and life-compromising conditions in newborns.

"Genetic testing is rapidly becoming an essential component in the care of newborns. It can have a dramatic positive impact on a child's health by identifying otherwise undetectable diseases so that appropriate care can be provided," said Dr. Anne Marie Comeau, Director of The New England Newborn Screening Program. "The flexibility, speed and accuracy of Tm Bioscience's genetic testing platform are ideally suited to the requirements of our program."

"The newborn screening market represents a key opportunity for Tm Bioscience, and we have advanced our underlying technology significantly in order to best serve this market," said Greg Hines, President and CEO of Tm

Bioscience. "We look forward to working with The New England Newborn Screening Program to supply its patient population with the most accurate and up-to-date genetic tests on the market."

ABOUT THE NEW ENGLAND NEWBORN SCREENING PROGRAM

The New England Newborn Screening Program, located in Jamaica Plain, has been operating under the U Mass Medical School since July 1, 1997. The program employs over a dozen laboratory technicians and five technical supervisors, all highly trained, all of whom meet federal Clinical Laboratory Improvement Amendments (CLIA) regulations for high complexity testing. In addition, three Ph.Ds and six M.D.s assure quality analysis of the laboratory technology, testing algorithms, and treatment protocols and provide support to the medical community, who welcome accurate information about the rare disorders included in newborn screening.

ABOUT TAG-IT(TM) GENETIC TESTS

Tm Bioscience's product menu is focused in the fields of human genetic disorders, pharmacogenetics and infectious disease. The Company has already commercialized a series of Tag-It(TM) tests for coagulation genes (Factor V, Factor II, MTHFR)(x), Cystic Fibrosis gene (CFTR 40+4(x) and ASR(xx) format), and drug metabolism genes (Tag-It(TM) P450-2D6, P450-2C9, P450-2C19)(x). All genetic tests from Tm Bioscience are based on the Tag-It(TM) Universal Array platform, which utilizes a proprietary universal tag system that allows for easy optimization, product development and expansion. Assays from Tm operate on the Luminex xMAP(R) system, a well-established bead based instrument. Combined, the Universal Array and Luminex instrument enable the rapid production of flexible, highly accurate, high-throughput, low-cost DNA-based tests.

(x) For Research Use Only. Not for use in diagnostic procedures.

(xx) Analyte Specific Reagent. Analytical and performance characteristics are not established.

ABOUT TM BIOSCIENCE - PUTTING THE HUMAN GENOME TO WORK(TM)

Tm Bioscience is a DNA-based diagnostics company developing a suite of genetic tests. Tm Bioscience's product pipeline includes tests for genetic mutations related to hematology, cystic fibrosis, drug metabolism and other debilitating genetic disorders. Additional information about Tm Bioscience can be found at www.tmbioscience.com.

New England Newborn Screening Program
University of Massachusetts Medical School
www.umassmed.edu

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## ◎肯德基州將進行二代新生兒篩檢項目超過 20 項

肯德基州的報紙 Lexington Herald-Leader 於 2005 年 1 月 27 日指出，Fletcher 州長支持擴大新生兒篩檢的法案。Fletcher 州長的行政團隊已經將擴大新生兒篩檢的法案置於優先提案中，以處理這些未來耗費成本且威脅生命的疾病。

成人與兒童健康促進中心（adult and child health improvement）的主任 Dr. Steve Davis 在州議會的聽證中指出，如果將來這項法案獲得通過，健康與家庭服務的部門（the Cabinet for Health and Family Services）將於七月一日需要相關實驗室的篩檢設備。健康與福利委員會的主席 Julie Denton 指出，該委員會將於二月七日投票，曾於 2005 年大會中提出相關法案的州議員 Denton 則指出，時間十分重要。

現有肯德基州 60 家接生醫院只檢驗四種新生兒篩檢項目，而其他 38 州則依照聯邦的指引和 March of Dimes 基金會的建議，已經篩檢了三十種項目。在 2003 年肯德基州合計篩檢了 53,708 位新生兒，並檢驗了 hypothyroidism、sickle cell anemia、galactosemia 、phenylketonuria 四項疾病，並轉介了 392 位進行後續追蹤。州議員 Ernesto Scorsone 指出，這些疾病許多都與食物消化有關，透過檢驗和飲食計畫即能避免問題，長期也能減少成本。州議員 Kathy Stein 也同樣支持這項法案。

目前新生兒篩檢的費用為 14.50 美元，新增的檢驗則為 50 美元，肯德基大學小兒醫學專家 Dr. Charlton Mabry 則希望保險公司可以擴大新生兒篩檢的部分成本；聯邦 Medicaid 計畫則可以補助肯德基州一半的新生兒進行擴大新生兒篩檢。州政府初期投入的篩檢成本預期將被事後治療成本的降低所抵銷，州健康部門的 Dr. Davis 則指出擴大新生兒篩檢後一年將可節省四百萬美元的成本。加州 Kaiser Permanente 保險計畫（Kaiser Permanente insurance plan in California)2002 年一項針對擴大新生兒篩檢成本效益的研究指出，基於該計畫歷年平均約 32,000 位新生兒的資料分析，早期診斷代謝異常的新生兒具有顯著的成本效益，殘障的情形較不嚴重且住院較少，顯示新生兒期以後確診的代謝病童的醫療成本是新生兒期所篩檢

出病童的兩倍，而就醫次數也是兩倍。

州議員 Ernesto Scorsone 指出，未來將花一百萬美元購置血液檢驗設備，及進行擴大新生兒篩檢前的專業訓練，他指出從一滴血中即能在出生時檢驗超過 20 種疾病。肯德基州所有醫院的檢體將送至 Frankfort 進行檢驗，而只要幾天檢驗結果便能知道。

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Lexington Herald-Leader (Kentucky); January 27, 2005
Fletcher Backs Bill To Expand Newborn Tests

http://www.kentucky.com/mld/heraldleader/news/state/10744876.htm

By Jack Brammer

FRANKFORT - The Fletcher administration has put on a fast track legislation to expand the testing of newborns for life-threatening diseases and conditions that could require costly treatment if discovered later.

If lawmakers approve the expanded screening, the Cabinet for Health and Family Services could have necessary lab equipment in place by July 1, Dr. Steve Davis, the director of adult and child health improvement, told a state Senate hearing yesterday.

The Health and Welfare Committee will vote on the measure Feb. 7, said its chairman, Sen. Julie Denton, R-Louisville. "Time is of the essence," said Denton, who is sponsoring one of three similar testing bills in the 2005 General Assembly.

Currently, Kentucky's 60 birthing hospitals test newborns for four disorders. Thirty-eight other states test for 30 disorders, as federal guidelines and the March of Dimes recommend.

In 2003, Kentucky tested 53,708 newborns for the metabolic disorders of hypothyroidism, sickle cell anemia, galactosemia and phenylketonuria and referred 392 of them for follow-up testing.

"Many of these problems concern food digestion and can be avoided with adequate testing and an appropriate diet," said Sen. Ernesto Scorsone, D-Lexington. "Testing can help newborns avoid years of problems and save dollars in the long run."

Scorsone and Rep. Kathy Stein, D-Lexington, are also sponsoring newborn-testing bills this session.

Current testing costs $14.50 a child. The additional testing would cost about $50. Dr. Charlton Mabry, a University of Kentucky specialist in pediatric medicine, said he hopes insurance companies would absorb some of the cost of expanded testing in the births they cover.

Medicaid pays for screening about half the newborns in the state.

But Denton said the initial cost to the state of expanded tests probably would be "a wash," offset by savings on treatment later. Davis, the Health Cabinet doctor, said about $4 million a year could be saved by expanding testing.

A 2002 study by the Kaiser Permanente insurance plan in California showed that hospitalization costs for children diagnosed with metabolic disorders later on were twice those of children diagnosed as newborns. Those diagnosed later also had twice as many medical visits.

It would cost Kentucky about $1 million to buy the blood testing equipment and for the professional training necessary to start the program, Scorsone said.

He said more than 20 disorders can be detected from a small blood sample taken during the first few days of life. Kentucky hospitals would send the samples to Frankfort for testing. Davis said results could be back to the hospitals in a few days.

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## ◎喬治亞州新增第十項新生兒篩檢項目 MCADD

March of Dimes 基金會喬治亞州分會於 2005 年一月 27 日發布新聞稿指出，該基金會成功的與州健康部門所屬公共健康局（DHR's Division of Public Health）合作新增 Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)於新生兒篩檢項目中。該項疾病為脂肪酸代謝異常的一種，將導致嚴重疾病、殘障、甚或死亡，如今透過串聯值譜儀（tandem mass spectrometry；MS/MS）從血滴中所篩檢的 30 項疾病中即能檢驗得知，事實上該州自 2005 年 1 月 4 日開始篩檢此病以來，一位病童即因早期篩檢而獲致正常與健康的生活。

March of Dimes 基金會支持擴大新生兒篩檢的計畫，並與喬治亞州健康部門所屬公共健康局自 2001 年起合作，並鎖定將 MCADD 列為新增項目的主要目標，如今這項新增計畫也顯示 March of Dimes 基金會喬治亞州分會的重要成果。除此項疾病以外，喬治亞州法律過去尚強制進行九項新生兒篩檢的項目，包括有 Phenylketonuria, Congenital Hypothyroidism, Maple Syrup Urine Disease, Galactosemia, Tyrosinemia, Homocystinuria, Congenital Adrenal Hyperplasia, Biotinidase Deficiency, Medium-Chain Acyl-CoA Dehydrogenase Deficiency and for Sickle Cell disorders 等。

喬治亞州健康部門負責婦幼健康服務的主任 Rosalyn Bacon 指出，很高興與 March of Dimes 基金會建立合作關係，March of Dimes 基金會一直致力於倡導和補助使用串聯值譜儀擴大新生兒篩檢，也扮演提升公眾知悉新生兒篩檢的重要角色。

罹患 MCADD 的病童一般可在出生後二月至二年間診斷出來，但卻可能最早在出生後兩天發病，甚或至成人期才發病，初期並無症狀，直到因長期飢餓或生病而觸發症狀。MCADD 的病童有 20-25%在第一次發病時死亡，一般相信嬰兒猝死症（Sudden Infant Death Syndrome, SIDS） deaths）中約有百分之一是因 MCADD 所致。只要能早期診斷並避免飢餓，罹患 MCADD 的病童預期都能健康成長。

喬治亞州健康部門負責新生兒篩檢的主管 Mary Ann Henson 指出，MCADD 過去被視為罕見疾病，全國估計約有十五萬分之一的發生率，約與苯酮尿症相當，估計喬治亞州一年約可篩檢出 9 位病童，透過早期診斷與治療將可拯救她們的生命。

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March of Dimes Georgia Chapter; January 27, 2005
March of Dimes and DHR Announce Latest Success for Newborn Screening in Georgia
http://biz.yahoo.com/prnews/050127/clth026_1.html

Press Release Source: March of Dimes Georgia Chapter

March of Dimes and DHR Announce Latest Success for Newborn Screening

in Georgia
DHR's Division of Public Health Adds New Genetic Test of Newborns

ATLANTA, Jan. 27 /PRNewswire/ -- The March of Dimes and DHR's Division of Public Health announced today that Georgia's newborns are now being tested for Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), one of the fatty acid oxidation disorders that causes severe illness, disabilities, or even death. MCADD is a genetics test that uses the new tandem mass spectrometry (MS/MS) that features detection of up to 30 disorders using the same blood spot. In fact, since the screenings began on January 4, 2005, one child has already tested positive for MCADD and due to early detection, will be able to lead a normal, healthy life.

March of Dimes supports comprehensive newborn screening for all babies and established a collaborative partnership with DHR's Division of Public Health in 2001 to expand newborn screening for Georgia's newborns and specifically target the implementation of a tenth screening test, MCADD.

"The implementation of the MCADD screening test is an important victory for the March of Dimes and the state of Georgia," said Brian Ziegler, State Director, March of Dimes Georgia Chapter. "The March of Dimes policy is to support screening for specific conditions when there is a documented benefit to the child and there is a reliable test that enables early detection. We will continue our efforts to further expand newborn screening in the state of Georgia."

As a result of this targeted partnership, MCADD was added to Georgia's series of Newborn Screenings because Georgia law (OCGA 31-12-6 & 31-12-7) and Rules and Regulations (Chapter 290-5-24) now require that every live born infant have an adequate blood test for nine treatable metabolic disorders (Phenylketonuria, Congenital Hypothyroidism, Maple Syrup Urine Disease, Galactosemia, Tyrosinemia, Homocystinuria, Congenital Adrenal Hyperplasia, Biotinidase Deficiency, Medium-Chain Acyl-CoA Dehydrogenase Deficiency) and for Sickle Cell disorders.

"We are pleased that DHR's Division of Public Health collaborative partnership with the March of Dimes Georgia Chapter and the Centers for Disease Control and Prevention (CDC) has enabled Georgia law to be

amended to allow the addition of MCADD to Georgia's newborn screening panel," said Rosalyn Bacon, DHR's director of maternal and child health services. "Our partners have been instrumental in advocating for funding to support the new tandem mass spectrometry equipment. They are also key players in promoting awareness of the importance of newborn screening."

MCADD is generally diagnosed between two months and two years of life, but can present as early as two days of life and as late as adulthood. Affected children are healthy and usually asymptomatic until prolonged fasting or an illness that causes them to not want to eat, like the flu, a cold, or ear infection triggers symptoms. The prolonged fasting can lead to hypoglycemia, vomiting, lethargy, seizures, coma, apnea, cardiac arrest, or sudden unexplained death. About 20-25% of these patients die from the first sick episode. MCADD is believed to account for about 1 out of 100 Sudden Infant Death Syndrome (SIDS) deaths. Once this disorder is diagnosed and treatment is begun, children with MCADD can expect to live normal, healthy lives, with normal growth and development. Treatment is effective and focuses on preventing long fasts.

"MCADD was once thought to be a rare condition," said Mary Ann Henson, director of DHR's Newborn Screening for Metabolic and Sickle Cell Disorders Program. "We don't know yet the disorder's exact prevalence in Georgia, but national estimates indicate that one in every 15,000 babies may be affected, making MCADD nearly as common as PKU (Phenylketonuria), a disorder that, if not identified and treated early, causes severe irreversible mental retardation. We believe that the new MCADD test may identify nine babies a year in Georgia with the disease whose lives may be saved through early diagnosis and treatment."

To learn more about Georgia's Newborn Screening for Metabolic and Sickle Cell Disorders Program, visit http://health.state.ga.us/programs/nsmscd/.

For the March of Dimes official policy on newborn screening, please visit http://www.marchofdimes.com/georgia.

The March of Dimes is a national voluntary health agency whose mission is to improve the health of babies by preventing birth defects and infant mortality. Founded in 1938, the March of Dimes funds programs of research, community services, education, and advocacy to save babies.

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Source: March of Dimes Georgia Chapter

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## ◎猶他州首家醫院進行二代新生兒篩檢

猶他州 KSL 電視台於 2005 年 1 月 24 日報導，St. Mark 醫院將成為猶他州首家醫院開辦篩檢 30 項新生兒篩檢項目。目前全國已有三十個以上的州篩檢 30 項新生兒篩檢項目，但猶他州目前僅篩檢 PKU 等四個項目，雖然使用同一血滴在出生時即可增加篩檢其他項目，但猶他州目前尚未強制增加篩檢。

小 Kali Bullock 雖然只有兩天半大，但已經增加篩檢了 30 種新生兒篩檢項目，這對 Bullock 家庭來說是欣慰的事，因為小 Kali Bullock 家裡還有一位姐姐患有無法消化醣和澱粉的疾病，當時等到確診時已經兩歲了。許多類似新陳代謝異常的小孩可能會有發展停滯、心智障礙、感染、心臟病、和肝功能異常的情形，一般相信嬰兒猝死症中約有百分之五到十是由於這 30 種新陳代謝疾病所致。

St. Mark 醫院自三年前即開始實驗擴大新生兒篩檢的檢查，如今已證實更有效率和迅速；猶他州健康部門正在檢討既有的新生兒篩檢政策，很有可能將在幾個月後強制篩檢這 30 種新陳代謝的疾病。

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KSL-TV Channel 5 (Utah); January 24, 2005
Hospital to Test Newborns for 30 Metabolic Disorders
http://tv.ksl.com/index.php?nid=46&sid=146318

HOSPITAL TO TEST NEWBORNS FOR 30 METABOLIC DISORDERS

Ed Yeates Reporting

As of today, St. Mark's Hospital will become the first medical center in Utah to begin testing all newborns for more than 30 metabolic disorders as a required standard of care. More than 30 states have already mandated these additional tests. But Utah is not among them.

Utah currently requires newborn tests for PKU and three other diseases, but does not mandate additional screenings, even though they can be done from that same single blood stick of the baby's heel taken shortly after birth. But as of today, St. Marks is requiring it.

Little Kali Bullock is only two and a half days old, but as of this morning she was tested for 30 additional metabolic disorders. Having the tests routinely now is comforting to Teisa Bullock since one of her other daughters was born with a condition where the body couldn't digest sugar and a form of starch. The girl was two-years old before she was finally diagnosed.

Teisa Bullock: fter two years of dealing with what we e had to deal with with my daughter, it definitely--after we finally found out and what we knew we could do to control it, and what we could do to help her ˈlr t definitely is peace of mind. It would have been nice to know at birth for sure.?

Many of these metabolic disorders can lead to stunted growth, mental retardation, infection, heart disease and liver failure. It's believed five to ten percent of sudden infant death syndrome cases might actually be one of these 30 undiagnosed conditions.

Colin Kelly, M.D., Pediatrician, St. Mark's Hospital: nd so just by discovering them early and changing the diet or intervening in other ways

makes a big difference.?

As we reported almost three years ago, ARUP labs began a pilot program on the screenings back then, and has now shown the additional tests can be done quickly and efficiently.

The State Health Department is now reviewing its policy and most likely will mandate the additional 30 screenings for all hospitals within the next several months. Pediatricians would like even more tests added to the list.

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## ◎猶他州將推動二代新生兒篩檢至 30 項

猶他州 Deseret Morning 報於 2005 年一月 24 日指出，猶他州法律目前強制新生兒進行聽力與四項代謝疾病的篩檢，如 PKU、galactosemia、congenital low thyroid、hemoglobin diseases，但州健康部門發言人 Steve McDonald 指出，基於州政府內部遺傳與健康顧問委員會的建議，目前正規劃擴大新生兒篩檢至 30 項；目前已有三十個州實施擴大新生兒篩檢，同時 March of Dimes 基金會在去年的報告中也批評猶他州未實施擴大新生兒篩檢。。

St. Mark 醫院發言人 Deb Reiner 指出，MCADD (medium chain acyl-CoA dehydrogenase deficiency)即是其中較常見的一項，因對脂肪燃燒有障礙，可能因飢餓而昏迷和致死，研究指出嬰兒猝死症（sudden infant death syndrome）中約有 5 至 20% 為該項疾病。猶他大學 ARUP 實驗室的 Noriko Kusukawa 指出，約有三千分之一的

嬰兒罹患這些可被篩檢的代謝疾病，如果及早正確診斷，她們都是可以治療的；由於發病後的治療極為高昂，因此即使篩檢只有三千分之一的疾病發現率，仍是合乎成本效益的考量。

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Deseret Morning News (Utah); January 24, 2005
Babies To Be Tested For Metabolic Illness
http://deseretnews.com/dn/view/1,1249,600106973,00.html

BABIES TO BE TESTED FOR METABOLIC ILLNESS
St. Mark's first in state to screen in standard care

By Lois M. Collins
E-mail: lois@desnews.com

St. Mark's Hospital will begin today to routinely screen all newborn babies for 30 different metabolic disorders, the first hospital in the state to offer the tests as part of its standard care.

The metabolic conditions are rare but can have devastating ?even deadly ?consequences if not detected and treated early.

Current state law requires that babies be screened for four metabolic disorders and have a hearing test. The Utah Department of Health is in the process of amending its newborn screening regulations to include the expanded panel, based on recommendations from both its genetics and health advisory committees, according to spokesman Steve McDonald.

Thirty states already require expanded newborn screening, and a March of Dimes report last year criticized Utah for not requiring the tests.

The Supplemental Newborn Screening by tandem mass spectrometry detects more than 30 metabolic disorders, which impact how or if the body breaks down compounds such as proteins, fats and carbohydrates to be used as energy or to promote growth or healing. They include amino acid disorders, organic

acid disorders and fatty acid oxidation disorders.

The best-known is MCADD (medium chain acyl-CoA dehydrogenase deficiency). A baby with MCADD cannot burn fat reserves for energy. "The infant runs out of food to burn for energy and can go into a coma and die," said Deb Reiner, spokeswoman for St. Mark's Hospital.

Studies indicate between 5 and 20 percent of sudden infant death syndrome cases are due to MCADD, she said. If parents know an infant has it, they simply wake their baby during the night for a feeding.

One in about 3,000 babies is born with one of the metabolic disorders, said Noriko Kusukawa, assistant vice president of ARUP Laboratories at the University of Utah. They're all treatable if diagnosed early and correctly. Left alone, they can result in mental retardation, damage to the liver, heart or brain, or even death.

Kusukawa knows of one infant with a metabolic disorder who wasn't diagnosed early enough and now needs a liver transplant.

"Effective early treatment has been proven," she said.

"The cost of treating these, when they do get sick, is so high that it is cost-effective to do this even with a 1-in-3,000" chance of finding a problem.

Last summer, University Hospital began providing the expanded screening to newborns, with parental consent, as part of a pilot project between the hospital, ARUP Laboratories and the health department. The goal was not to vet test efficacy, long established, but rather to see if the expanded testing could be introduced on a large scale in a cost-effective and efficient way, Dr. Nicola Longo, a professor of pediatrics at the U. and director of metabolic services, told the Deseret Morning News at that time.

Although each condition is relatively rare, no one knows which child to screen for which condition until screening answers that question. So testing for all of them makes sense, he said.

From a baby's perspective, it's nothing extra. Babies already have a heel prick to

test for phenylketonuria (PKU); the expanded screening is done with blood from the same prick.

Utah currently mandates tests for PKU, galactosemia, congenital low thyroid and hemoglobin diseases such as sickle cell. Health officials are now working with lawmakers and others to expand the requirements.

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## ◎紐約州將進行二代新生兒篩檢至 44 項

紐約第十號電視台於 2005 年 1 月 20 日報導，紐約州長 George Pataki 可能將要求擴大新生兒篩檢上路，就在九年前紐約州立法讓愛滋寶寶獲得自動且免費的篩檢，當時便使得家長警惕早期治療的重要，而這也是紐約州將擴大實施新生兒篩檢的同樣道理。當這項新生兒篩檢的項目擴大到 44 項時，州長說這將使得紐約州免費的新生兒篩檢成為全美最完整的方案。新生兒的父母將從州健康部門獲得更多有關新生兒篩檢的資訊，去年只篩檢了 11 項代謝疾病，但是今年春天開始，即將變成 44 項之多。

州長同時要求健康部門組成工作小組嘗試開發新的檢驗項目，他特別希望能將 Krabbe 疾病列入，這是罕見但威脅生命的神經疾病，大多數病患發現時已經太遲而無法康復。遺傳學家 Dr. Joan Pellegrino 說，雖然州長想加入的名單中仍有些我們無法百分百加以治療的疾病，但我想只要越來越多的病患被發現，將會鞭策研究者

開發創新的療法。目前紐約州正在開發治療 Krabbe 疾病的試驗療法，或許將需要一年的時間，如果效果良好，州長決定要推動讓紐約州所有的新生兒都獲得篩檢的機會。

在 2004 年開始，紐約州的新生兒篩檢檢驗了 11 項疾病，包括：

Phenylketonuria (PKU)

Maple Syrup Urine Disease (MSUD)

Homocystinuria

Galactosemia

Biotinidase deficiency

Medium chain acyl Co-A dehydrogenase deficiency (MCADD)

Congenital hypothyroidism (CH)

Congenital adrenal hyperplasia (CAH)

Cystic fibrosis (CF)

Sickle cell disease (SSD)

HIV-1 exposure

在擴大實施新生兒篩檢之後，2004 年歲末將增加篩檢下列疾病：

3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)

3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)

Argininosuccinic acidemia (ASA)

Carnitine palmitoyl transferase II deficiency (CPT-II)

Carnitine uptake defect (CUD)

Carnitine-acylcarnitine translocase deficiency (CAT)

Citrullinemia (CIT)

Cobalamin A,B cofactor deficiency (Cbl A,B)

Glutaric acidemia type I (GA-I)

Isovaleric acidemia (IVA)

Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)

Methylmalonyl CoA mutase deficiency (MUT)

Mitochondrial acetoacetyl-CoA thiolase deficiency (BKT)

Mitochondrial trifunctional protein deficiency (TFP)

Multiple acyl-CoA dehydrogenase deficiency (MADD, also known as GA-II)

Multiple carboxylase deficiency (MCD)

Propionic acidemia (PA)

Short-chain acyl-CoA dehydrogenase deficiency (SCADD)

Tyrosinemia (TYR)

Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)

而 2005 年春天開始，又將增加篩檢下列疾病：

2-Methyl 3-hydroxy butyric aciduria (2M3HBA)

2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)

3-Methylglutaconic aciduria (3MGA)

Argininemia (ARG)

Carnitine palmitoyl transferase deficiency II (CPT-II)

Carnitine palmitoyltransferase Ia deficiency (CPT-1A)

Dienoyl-CoA reductase deficiency (DE REDUCT)

Hypermethioninemia (MET)

Isobutyryl-CoA dehydrogenase deficiency (IBG)

Malonic aciduria (MAL)

Medium chain ketoacyl-CoA thiolase deficiency (MCKAT)

Medium/short-chain hydroxy Acyl-CoA dehydrogenase deficiency (M/SCHAD)

Methylmalonic academia (Cbl C,D)

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News 10 Now (New York); January 20, 2005
Newborn Screening Program Expanding
http://news10now.com/content/all_news/Default.asp?ArID=35170&SecID=83&

NEWBORN SCREENING PROGRAM EXPANDING

By: Tammy Palmer, News 10 Now Web Staff

The great news is the Governor may ask for even more screening down the road. Just 9 years ago the Baby AIDS law paved the way for automatic and free AIDS screening for all newborns in New York State. The result was an early warning for parents and early treatment for the babies.

That's the idea behind the latest expansion of the state's newborn screening program. Blood is drawn from every newborn for various tests. As the number of tests grows to 44, the Governor says the expansion will make New York's free screening program, the most comprehensive in the nation.

The Governor is also calling on a Health Department task force to look for new tests to add to the list. Specifically, we know he hopes to add screening for Krabbe disease. That's a rare and life threatening condition that affects the nervous system. Unfortunately, most parents find out about it too late for a fair recovery.

There's no specific treatment for Krabbe disease right now that would be 100% effective, but doctors say early screening could make the difference between life and death.

"Even some of the disorders that we're looking at on the list that the Governor has addes...we may not have 100% treatment for all of those either. But, I think the idea is that as more and more infants are identified early, that is going to spur researchers to try to come up with new innovative ideas that we can't even fathom right now," said Dr. Joan Pellegrino, geneticist.

Right now, New York is in a pilot program, testing the effectiveness of Krabbe screening. That process could take a year. But, if it is determined to work, the Governor indicated that he's determined to have every infant in New York screened for the disease.

MORE INFO

At the start of 2004, the New York State Health Department newborn screening program tested for the following 11 conditions:

Phenylketonuria (PKU)
Maple Syrup Urine Disease (MSUD)
Homocystinuria
Galactosemia
Biotinidase deficiency
Medium chain acyl Co-A dehydrogenase deficiency (MCADD)
Congenital hypothyroidism (CH)
Congenital adrenal hyperplasia (CAH)
Cystic fibrosis (CF)
Sickle cell disease (SSD)
HIV-1 exposure

Under the expansion, the following metabolic disorders were added by the end of 2004:

3-Hydroxy-3-methylglutaryl-CoA lyase deficiency (HMG)
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
Argininosuccinic acidemia (ASA)
Carnitine palmitoyl transferase II deficiency (CPT-II)
Carnitine uptake defect (CUD)
Carnitine-acylcarnitine translocase deficiency (CAT)
Citrullinemia (CIT)
Cobalamin A,B cofactor deficiency (Cbl A,B)
Glutaric acidemia type I (GA-I)
Isovaleric acidemia (IVA)
Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
Methylmalonyl CoA mutase deficiency (MUT)
Mitochondrial acetoacetyl-CoA thiolase deficiency (BKT)
Mitochondrial trifunctional protein deficiency (TFP)
Multiple acyl-CoA dehydrogenase deficiency (MADD, also known as GA-II)
Multiple carboxylase deficiency (MCD)
Propionic acidemia (PA)
Short-chain acyl-CoA dehydrogenase deficiency (SCADD)
Tyrosinemia (TYR)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD)

The remaining tests will be added as part of the expansion to be completed by Spring 2005:

2-Methyl 3-hydroxy butyric aciduria (2M3HBA)
2-Methylbutyryl-CoA dehydrogenase deficiency (2MBG)
3-Methylglutaconic aciduria (3MGA)
Argininemia (ARG)
Carnitine palmitoyl transferase deficiency II (CPT-II)
Carnitine palmitoyltransferase Ia deficiency (CPT-1A)
Dienoyl-CoA reductase deficiency (DE REDUCT)
Hypermethioninemia (MET)
Isobutyryl-CoA dehydrogenase deficiency (IBG)
Malonic aciduria (MAL)
Medium chain ketoacyl-CoA thiolase deficiency (MCKAT)
Medium/short-chain hydroxy Acyl-CoA dehydrogenase deficiency (M/SCHAD)
Methylmalonic academia (Cbl C,D)

WATCH THE VIDEO
(visit website link above to watch the video of the broadcast)

Newborn screening

New parents in New York State will get more from the Health Department's free newborn screening program. Just last year, the state tested infants for 11 diseases and disorders. By, spring of this year, that number will grow to 44. News 10 Now's Tammy Palmer reports.

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## ◎密蘇里州將進行二代新生兒篩檢至 25 項

密蘇里州聖路易郵報（Saint Louis Post）於 2005 年 1 月 2 日指出，該州新生兒篩檢即將擴大至 25 項，該州公共衛生實驗室

（Public Health Laboratory）的助理主任 Larry Evert 指出，這項篩檢預計自 2005 年初春時實施。該州 2001 年的立法規定強制篩檢，但並未提供補助。這將使得密蘇里州列為全國約 12 的州一般，如同伊利諾州使用先進的儀器，進行 25 項以上新生兒篩檢，這些項目都是可治療的疾病，且無及早診斷將有嚴重後果。

聖路易大學遺傳醫學中心主任 Dr. Gary S. Gottesman，同時也是州新生兒篩檢顧問委員會的委員指出，這項措施將帶領我們進入 21 世紀，密蘇里州將在新生兒發病前幫助他們，以減少未來的住院與罹病率，儘管這些疾病極為罕見，許多只有五萬至十萬分之一，甚至更低，但是一但篩檢的項目增加至 20 幾項，發現的病童人數即會更多。

強制篩檢將使得機會更加均得，不會只受限於恰巧聽聞過或是有能力負擔的家庭，通常過去透過私人檢驗所或外州機構的篩檢費約為美金 25 至 100 元之間。Dr. Gary S. Gottesman 則建議大眾，只要經濟許可最好能進行擴大的新生兒篩檢，早期發現異狀，做了還是值得的。他也覺得很沮喪，因為伊利諾州已實施擴大的新生兒篩檢，且費用已由公立或私人保險給付。目前密蘇里州全年篩檢了 75,000 兒，一但實施擴大的新生兒篩檢，預計將發現 50 個基因異常的新生兒。

目前各州新生兒篩檢的項目介於 4 至 34 項之間，去年十月聯邦健康與人類資源部的顧問委員會即建議應評估進行 30 項新生兒篩檢的必要性，目前各州的狀況不一，部分原因係由於篩檢設備的建置費用。像 Pennsylvania, Nebraska and Mississippi 等州則是將該項業務委外由位於賓州 Bridgeville 的 Pediatrix 公司負責，該公司也是全國提供新生兒篩檢服務最大的公司，而部分州則是自行購置串聯質譜儀進行篩檢，設備約在 20 萬美元以上。

密蘇里州法律規定所有強制篩檢的健康檢驗都集中於 Jefferson City 的公共實驗室進行，但由於州的經費缺乏，並未補助新生兒篩檢經費，直到 2004 年才由健康與老人服務部門（Health and Senior Services Department）中提撥聯邦的一項補助，用於補助新生兒篩檢的前置作業成本，如人員、設備與軟體。Larry Evert 指出無疑的許多人對此感到失望，包括家長的倡導團體，她們眼見自己小孩受苦於這些可治療的疾病而推動新生兒篩檢的立法，以及像我們等從工作中清楚的知道這事該做但未做。Larry Evert 指出擴大新生兒篩檢將需要 3.75 百萬美元，將由檢驗費用中給付，在原來的五項篩檢費用 25 元以外，再加收 50 元作為串聯質譜儀檢驗 25 項擴大新生兒篩檢的費用，州法律規定公共與私人保險應提供給付。

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Saint Louis Post-Dispatch; January 2, 2005
Babies Will Be Tested For 25 Disorders
http://www.stltoday.com/stltoday/news/stories.nsf/sciencemedicine/story/74A63F2480C64CB886256F7E001954AF?OpenDocument&Headline=Babies+will+be+tested+for+25+disorders+&highlight=2%2CBabies%2Cwill%2Cbe%2Ctested%2Cfor%2C25%2Cdisorders

BABIES WILL BE TESTED FOR 25 DISORDERS

By Rachel Melcer of the Post-Dispatch

PHOTO CAPTION: Nurse Katie Bremer draws blood from the bottom of a newborn foot in the nursery at St. Johns Mercy Hospital Wednesday morning. (Laurie Skrivan/P-D)

Soon, but not as soon as many health care advocates had hoped, Missouri newborns automatically will be screened for 25 potentially devastating genetic disorders.

The testing should start by early spring, said Larry Evert, assistant director of Missouri's Public Health Laboratory.

The program was mandated, but not funded, in 2001 by the Legislature. And the rollout will come a few months later than the Jan. 1 start date some physicians and observers had expected.

Still, it will place Missouri on a short list of about a dozen states, including Illinois, that use advanced technology to detect at least 25 inherited diseases. All of these conditions can be treated, but without early screening they go unnoticed until serious - and sometimes irreversible - symptoms set in.

"It's bringing us into the 21st century," said Dr. Gary S. Gottesman, director of medical genetics at St. Louis University and a member of the state's Newborn Screening Advisory Committee.

Missouri will be "helping children before they really become ill, hopefully

preventing a lot of inpatient hospital days and morbidity," he said. "Although the disorders are rare - many of them have a 1 in 50,000 or 1 in 100,000 or lower risk - with all of the 20 or so new disorders that we're going to look at, the numbers get bigger."

Mandatory screening will equalize a process that has been available for a few years, but only to those who happen to hear about it - and who can afford to pay for testing at a private or out-of-state university lab. Most charge $25 to $100.

"Right now it's offered to people who might have the financial means," said Brenda Davidson, clinical director of the mother/baby unit at St. Mary's Health Center in Richmond Heights. "That is generally not how we deliver any other health care, at least within our system. We want to deliver all the same health care to all individuals."

Jean Schroeder, director of laboratory services at St. John's Mercy Medical Center in Creve Coeur, said she noticed a groundswell in parents' awareness of and interest in expanded newborn screening about a year ago. It followed a flurry of media stories, including a segment on "The Oprah Winfrey Show."

That demand, coupled with requests from neonatologists, prompted St. John's to contract with Pediatrix Screening of Bridgeville, Pa., the nation's largest provider of newborn screening services.

The hospital stocks test kits, but "we don't promote tests unless it's requested by a family or physician," Schroeder said. As a result, the screenings primarily are conducted on high-risk babies who need greater attention.

Yet the genetic disorders in question can occur in apparently healthy babies, and in families with no prior history of disease.

"I would certainly recommend that people get the supplemental screen done, if they can afford it. The likelihood of finding something is low, but if they do ... it is worthwhile," Gottesman said.

He said he has been frustrated because patients who live in Illinois automatically are screened, and the tests are covered by private or public

insurance. Meanwhile, babies born in Missouri have been left wanting.

About 75,000 babies are born each year in Missouri. Once the expanded screening becomes mandatory, a genetic disorder will be found in about 50 of them, Evert said.

"That's a pretty small needle in a rather big haystack," he said. "But each year, a few babies in Missouri die of these conditions because we are not testing for them. ... And we're going to change babies' lives. They're going to be just as normal as you and I" instead of suffering from developmental or physical disabilities that can result from an untreated disease.

While genetic disorders do not go away, all of those included in the expanded screening can be readily treated. Some simply require a change in diet to bolster nutrition or avoid a trigger, Evert said.

All states test newborns for between three and 34 genetic diseases. In October, an advisory committee to the U.S. Health and Human Services Department endorsed a study from leading geneticists that called for a 30-disease check.

Yet disparities remain, partly because of the expense of setting up screening programs.

Some states, including Pennsylvania, Nebraska and Mississippi, contract with Pediatrix for the work. Others invest in the necessary technology, a tandem mass spectrometer that costs more than $200,000. These machines can use a few drops of blood, obtained by a prick of a baby's heel, to simultaneously test for many different types of molecules that can indicate disease.

Missouri law requires that all mandated health tests be conducted at the public laboratory in Jefferson City, Evert said. But the state, facing budget shortfalls, has not allocated money for expanded newborn screening. Finally, in 2004, the Health and Senior Services Department identified a federal grant to cover start-up costs, including staff, equipment and software to manage results.

"There's no doubt that it was very frustrating to a lot of different people," Evert said. They include parent advocates, who pushed for the screening law after watching their own children suffer from treatable diseases, as well as

"those of us in the business who are close enough to know we really should be doing this, and we're not."

The ongoing cost of the program, about $3.75 million a year, will be covered by test fees, Evert said. The cost to patients will rise to about $50 for a 25-disease test using tandem mass spectrometry from the $25 currently charged for a five-disease screen. Under state law, private and public insurers must cover the cost.

Once the program is in place, Evert's department also will manage follow-up care. It has established a network of pediatric genetic specialists, one of whom will be notified along with a family's pediatrician each time there is a positive result.

"The same day, the same hour that we find a child with one of these conditions, we're on the phone," Evert said. Initial screening results must be followed by more-definitive testing and treatment, and the state will track outcomes.

"We don't want to miss any of these babies," he said. "And there's not much value in finding (them) if you're not also in the position to do something about it."

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**◎維吉尼亞州推動立法擴大新生兒篩檢**

維吉尼亞州 Potomac 報於 2004 年 12 月 26 日指出，聯邦健康與人民服務部一顧問委員會正草擬一份規範各州新生兒篩檢的指引，雖然各州並未被強制接受。而根據維吉尼亞州健康照護聯席委員會(The Virginia General Assembly's Joint Commission on Health Care)

秘書長 Kim Sneed 的說法，該委員會正依據聯邦這項指引準備立法擴大新生兒篩檢。

維吉尼亞州已經購置三部新的串聯質譜儀（Tandem Mass Spectrometers），準備用來作為擴大新生兒篩檢之用，目前估計這項新增篩檢約每位收取 20 至 40 美元。明尼蘇達州 Mayo 醫院的 Dr. Piero Rinaldo，他本身也是聯邦該顧問委員會的委員，已經有 20 多年診治代謝異常病童的經驗，他提到無論是倫理或財務上的理由，都已經足以合理的推動各州擴大新生兒篩檢，他說再繼續無謂的辯論是否擴大新生兒篩檢，只是讓人們更混亂，目前這種檢驗已經可以做出 54 種疾病的檢查，而且新的資訊仍在增加中。他希望聯邦能提供經費作為各州擴大新生兒篩檢的誘因，他指出重要的是新生兒篩檢的公平性、一致性、和普同性。他認為這是全國早就應該實施的做法，面對完全正常的新生兒突然在幾小時內死亡，我的工作就是告訴家長她們小孩死亡的原因，只因為生錯州了。

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Billings Gazette (Montana); December 29, 2004
Patients Fear Insurance Hikes And Hide Genetic Conditions

http://www.billingsgazette.com/index.php?display=rednews/2004/12/29/build/health/40-insurance.inc

PATIENTS FEAR INSURANCE HIKES AND HIDE GENETIC

CONDITIONS

By April Lynch
Knight Ridder News

SAN JOSE, Calif. - Even as genetic tests for more conditions become widely available, patients are taking some of medicine's latest advances undercover.

Afraid that they will be denied care in an increasingly cutthroat health-care market if insurers know too much about them, patients are getting genetic tests without telling insurers, or even their doctors.

Tests for a wide range of genetic conditions, including a common genetic iron disorder and inherited cancer risk, are now available online or by mail for consumers to use without involving a physician or insurance company. Some patients, with their doctors' help, are tested under assumed names. Others may avoid tests altogether.

Genetic science offers vast promise in diagnosing disease, but still has little ability to cure the problems it uncovers. That leaves many patients fearful of stigma. Will a genetic diagnosis tarnish their medical records, cost them insurance or a job or wind up in their children's files?

These fears, once just fodder for science fiction, are shaping a covert system centered on self-reliance - and a culture of secrecy doctors find risky.

People "aren't getting adequate counseling to make difficult decisions," said David Magnus, co-director of the Stanford Center for Biomedical Ethics. "These fears are definitely having an impact."

SCREENING COMMON

The effects are only likely to grow as genetic screening becomes more common. Genetic medicine began moving into the mainstream about 10 years ago and today some tests are widely used.

Almost all newborn babies in California, for example, now undergo tests for gene-based diseases such as phenylketonuria. There are tests for about 1,000

genetic disorders, and more are coming.

"If you think widespread screening for genetic risk factors is coming soon, and I think it is, it can't happen in a vacuum," said Dr. James Zehnder, a blood disorder and gene analysis specialist at Stanford University.

Insurance industry representatives question such tests, saying the public's fears are overblown.

"Companies are not asking for this information, and they don't have plans to," said Anne Eowan, vice president of governmental affairs for the Association of California Life and Health Insurance Companies.

LACK OF TRUST

But many patients find it hard to trust insurers. After all, they reason, look how some insurance companies or employers handle pre-existing medical conditions today. People applying for individual health insurance are sometimes turned down if they have long-term illness. And privacy and discrimination protections vary widely from state to state.

A federal law guards group health insurance from genetic bias, but not those seeking individual insurance. The U.S. Senate approved broader national protections last year, but the bill stalled in the House of Representatives.

So patients may be left in a Catch-22: If a person deliberately withholds information from an insurance company, he may be guilty of insurance fraud. But if he discloses a condition, will he face higher premiums or possibly be unable to get coverage at all?

"The public doesn't feel safe around this issue," said Sharon Terry, president of the nonprofit advocacy group Genetic Alliance.

Many doctors worry that patients' concern over privacy gets in the way of needed care.

"To get a test result and not have an explanation of what that means - that can be alarming," said Dr. Willis Navarro, a blood disorder specialist at the

University of California-San Francisco Medical Center. "Our role, all our training, is to help people."
Many physicians also grapple with handling genetic information. Some help test patients covertly. Others won't.

Kaiser Permanente's Northern California division, decided on full disclosure because doctors there felt the alternative would be a "disservice" to the patient in the long run.

"They would be at a disadvantage for their medical care," said Dr. Ronald Bachman, chief of genetics for Kaiser's Oakland, Calif., division and a founder of the Kaiser genetics program.

Some patients with genetic experience agree, and advise others to weigh their fears against their family's health.

"My ancestors didn't keep health records, and it almost cost me my life," said Joe Litel of San Jose, a hemochromatosis patient who is part of Brick's support network. "If you intend to stay alive in this world and see your children grow up, and for your children's health, don't hide from these things."

MORE INFORMATION

Genetic Alliance, based in Washington, D.C., lobbies federal lawmakers on a range of gene-related issues. Find them online at

http://www.geneticalliance.org/ or call (202) 966-5557.

The American Hemochromatosis Society offers information on independent testing and can be found online at http://www.americanhs.org/ or by calling (888) **655-4766.**

For questions on genetic issues and employers, contact the federal Equal Employment Opportunity Commission at http://www.eeoc.gov/ or (800) 669-4000.
Source: Mercury News research

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## ◎奧瑞岡州擴大新生兒篩檢項目

依據奧瑞岡州健康服服務部門網站資料，2004 年 6 月該州使用串聯質譜儀擴大新生兒篩檢項目如下，該州新生兒篩檢亦屬於「美國西北地區新生兒篩檢計畫」（The Northwest Regional Newborn Screening Program; NWRNSP）的一部份：

Amino Acid Disorders

- Phenylketonuria
- Homocystinuria
- Tyrosinemia

Urea Cycle Disorders

- Arginase Deficiency
- Argininosuccinate lyase deficiency (ASA)
- Citrullinemia
    - Classic Citrullinemia
    - Citrullinemia Type II

Organic Acidemias

- Beta-Ketothiolase deficiency
- Glutaric aciduria, Type I (glutaryl-CoA dehydrogenase deficiency)
- HMG-CoA lyase deficiency (3-hydroxy-3-methylglutaryl-CoA lyase deficiency)
- Isobutyryl CoA dehydrogenase deficiency
- Isovaleryl-CoA dehydrogenase deficiency (Isovaleric acidemia)

- Malonic aciduria
- Maple Syrup Urine Disease
- Methylmalonic acidemia (MMA)
    - Methylmalonic aciduria, vitamin B-12 responsive
    - Methylmalonic aciduria, vitamin B-12 nonresponsive
    - Vitamin B12 metabolic defect with methylmalonicacidemia and homocystinuria
- Multiple carboxylase deficiency
- Propionic acidemia (PA)
- 2-Methyl-3-hydroxybutyryl CoA dehydrogenase deficiency
- 2-Methylbutyryl CoA dehydrogenase deficiency
- 3-methylcrotonyl-CoA carboxylase deficiency
- 3-methylglutaconyl-CoA hydratase deficiency
    - 3-methylglutaconyl-CoA aciduria TYPE I
    - 3-methylglutaconyl-CoA aciduria TYPE II
    - 3-methylglutaconyl-CoA aciduria TYPEIII
    - 3-methylglutaconyl-CoA aciduria TYPE IV

Fatty Acid Oxidation Disorders

- Carnitine uptake/transporter defects
    - Carnitine-acylcarnitine translocase deficiency
    - Carnitine transporter defect
    - Carnitine palmitoyl transferase I deficiency (CPT I)
    - Carnitine palmitoyl transferase II deficiency (CPT II)
- Glutaric aciduria, Type II (multiple acyl-CoA dehydrogenase deficiency (MADD))
- Very long chain acyl-CoA dehydrogenase deficiency (VLCADD)

- Long chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
- Short chain acyl-CoA dehydrogenase deficiency (SCADD)

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## ◎明尼蘇達州 2003 年新生兒篩檢項目

**Hemoglobinopathy**
___Sickle cell disorders
___Thallasemias

**Endocrine Disorders**
___Hypothyroidism - Congenital
___Adrenal Hyperplasia - Congenital

**Amino Acid Disorders**
___Phenylketonuria
___Argininosuccinic Aciduria (ASA Lyase Deficiency)
___Citrullinemia (ASA Synthetase Deficiency)
___Hypermethioninemia,
___Homocystinuria (Cystathione synthase deficiency)
___Hyperornithinemia
___Hyperammonimemia
___Hyperhomocitrullinuria
___Maple Syrup Urine Disease (MSUD)
___Tyrosinemia

**Organic Acid Disorders**
___2-Methylbutryrl-CoA Dehydrogenase Deficiency (2MBCD or SBCAD)
___3-Methylcrotonyl-CoA Carboxylase Deficiency (3MCC Deficiency)
___3-Methylglutaconyl-CoA Hydratase Deficiency
___3-Hydroxy-3 Methylglutaryl-CoA Lyase Deficiency (HMG)
___Glutaric Acidemia-Type I (GA 1)
___Isobutyryl Acidemia (IVA)
___Methylmalonic Acidemia (MMA)
___Mitchondrial Acetoacetyl-CoA Thiolase Deficiency (3-Ketothiolase

Deficiency),
___Multiple CoA Carboxylase Deficiency
___Propionic Acidemia (PA)

Fatty Acid Oxidation Disorders
___Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
___Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
___Medium Chain 3-Ketoacyl-CoA Thiolase Deficiency (MCKAT)
___Multiple Acyl-CoA Dehydrogenase Deficiency
___Carnitine Transport Defect
___Carnitine Palmitoyl Transferase Deficiency-Type I (CPT-I)
___Neonatal Carnitine Palmitoyl Transferase Deficiency - Type II (CPT-II)
___Carnitine/Acylcarnitine Translocase Deficiency (CACT)
___Trifunctional Protein Deficiency (TFP Deficiency)
___3-Hydroxy Long Chain Acyl-CoA Dehydrogenase Deficiency (LCHAD)
___Very Long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD),
Argininemia,

Other Disorders
___Biotinidase Deficiency
___Galactosemia
___Glucose 6 Phosphate Dehydrogenase Deficiency (G6PD)

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## ◎加州成為新生兒篩檢的先驅

加州 Contra Costa 時報於 2004 年 12 月 7 日指出，加州過去新生兒篩檢的項目遠落後於其他州，但州政府戲劇性的計畫於 2005 年八月擴大新生兒篩檢，使加州成為美國新生兒篩檢的先驅。

加州政府健康服務部門所屬基因疾病業務的主任，George Cunningham 指出，目前加州政府網站與手冊中已羅列外州有關新生

兒篩檢的資訊，州法律規定醫療提供者需將這些資訊轉交給懷孕婦女，加州政府同時已發出 18,000 信件給婦產科醫生、小兒科醫生、與婦產科醫院，鼓勵她們將新生兒篩檢的資訊提供給未來的家長們；但是一項調查發現，約有 30% 的醫療提供者自己承認並未轉交新生兒篩檢的手冊給家長。

串聯質譜儀在 1990 年代被引進作為新生兒篩檢的先進科技，它同時能篩檢數十種疾病，其中許多疾病只要早期確診並改變飲食計畫即可治療，但部份州則在購置這項設備上進度緩慢，這項設備約值 30 至 40 萬美元，同時也需要重新訓練人員和更換昂貴的電腦系統。過去長久以來，加州只篩檢四項疾病，並集中於八個區域實驗室進行檢驗。目前各州新生兒篩檢的差異極大，2004 年十月聯邦一顧問委員會已建議各州應至少篩檢 30 項基因異常的疾病，而這些也都具有可行的治療方式，March of Dimes 基金會也隨即呼應這項建議。

加州目前也在正確的方向發展，去年州長 Arnold Schwarzenegger 已簽署一項法案提供 2.7 百萬美元經費，作為加州篩檢超過七十種出生缺陷疾病之用，並已簽訂合約租賃 15 部串聯質譜儀，政府人員也正在設置這些設備和提升電腦系統。March of Dimes 基金會估計，加州每年預期將因此多發現 140 位罹患可治療的新生兒疾病，也將因此節省 1.4 億元的後續醫療支出與服務費用。

家長若在外州實驗室進行這項篩檢，將花費約 25 至 100 美元。

有關這項篩檢的訊息尚可參閱下列網站：

Baylor at 800-4BAYLOR (422-9566) or
www.bhcs.com/MedicalSpecialties/MetabolicDisease

Mayo at 800-533-1710 or
www.mayoreferenceservices.org/mml/mml-sns-intro.asp

Pediatrix at 866-463-6436 or www.pediatrixscreening.com

CARES at www.caresfoundation.org

Alliance for Genetic Disorders at www.geneticalliance.org

Star G Project at www.newbornscreening.info

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Contra Costa Times; December 7, 2004
Danville Parents Stress Importance Of Screening After Infant's Deficiency Went Undetected

http://www.contracostatimes.com/mld/cctimes/living/health/10358256.htm?1c

DANVILLE PARENTS STRESS IMPORTANCE OF SCREENING AFTER INFANT'S DEFICIENCY WENT UNDETECTED

By Sandy Kleffman

DANVILLE - Every day, David and Cindy Wyvill have a visual reminder of what might have been.

Twenty-month-old Nathan darts around the house, grabbing his favorite toys, displaying a strong independent streak.

His twin brother, Zachary, has an infectious smile but cannot walk, sit up or roll over. His parents feed him through a tube in his stomach.

The two boys, with their blond curly hair and long eyelashes, are mirror images in appearance but a world apart in their abilities.

Zachary suffered brain damage a few months after his birth because he never received an expanded newborn screening test that would have detected his rare enzyme deficiency. Nathan does not have the condition.

Zachary's life might have been very different if he had been born in Mississippi or one of several other states. There he would have received the needed test. With a modified diet and vitamins, he might have led a normal life.

But California now ranks behind nearly every other state in the disorders it screens for in its tests. The state plans to dramatically expand its testing by August, moving it to the forefront.

But the Wyvills and others are upset that state officials aren't doing more in the interim to inform parents about how to get tests from out-of-state laboratories.

They also question the wisdom of a decision by the former Davis administration in June 2003 to abandon an 18-month expanded testing pilot program because of budget constraints.

"You're talking about 300 kids who will end up just like my son because they let it expire," David Wyvill said.

California now includes information about the out-of-state labs on its Web site and in brochures that health care providers are required by law to hand out to all pregnant women. The state sent more than 18,000 letters to obstetricians, pediatricians and maternity hospitals encouraging them to inform prospective parents about such options, said George Cunningham, chief of the genetic disease branch of the state Department of Health Services. But a state survey last year revealed spotty compliance. Nearly 30 percent of providers reported that they never distribute the brochures.

David Wyvill noted that to obtain the expanded tests, parents often must

contact an out-of-state laboratory, get a kit, take it with them to the delivery and tell hospital officials to take a second blood prick when they do the standard test.

"This puts a lot of pressure and a lot of the responsibility on the parents -- parents who are largely uneducated about this," Wyvill said.

Cunningham said he lobbied hard to save the pilot program, but state officials were wrestling with huge state budget deficits.

"We just couldn't get their attention to act," Cunningham said.

Although Zachary was born during the pilot program, his parents say they were never offered the expanded test and did not understand the limitations of the regular tests.

The Wyvills are part of a national movement pushing states to expand their testing.

New technology introduced in the 1990s, called tandem mass spectrometry, makes it possible to screen for several dozen conditions at once.

Many of the disorders are treatable with a change of diet or medication, if caught early. But some states have been slow to acquire the machines, which cost from $300,000 to $400,000. The technology often requires retraining and an expensive revamp of a state's computer system.

For years, California has screened for just four categories of disorders. Hospital workers take a few drops of blood from a newborn's heel for analysis at one of eight regional laboratories.

All states provide some form of newborn screening but the extent varies widely.

In October, a federal advisory committee recommended that states screen for at least 30 genetic disorders, all of which have available treatments.

The March of Dimes quickly endorsed the idea.

California is headed in that direction. Last summer, Gov. Arnold Schwarzenegger signed a bill providing $2.7 million for the state to test for more than 70 birth defects. California has signed a contract to lease 15 of the tandem mass machines. Officials are rushing to install them and to upgrade the state's computer system by August.

It should detect treatable conditions in an additional 140 babies each year, saving $140 million in medical treatments and services, the March of Dimes estimates. But it will come too late for Zachary.

Zachary and Nathan spent nearly a month in the intensive care nursery at John Muir Medical Center in Walnut Creek after they were born prematurely at 31 weeks.

Everything seemed fine until about four months later when the Wyvills noticed Zachary's head grow large and become misshapen.

A neurosurgeon at Children's Hospital Oakland told them it was probably hydrocephalus, a condition characterized by extra fluid on the brain. The doctor said he would continue to monitor Zachary and sent them home, David Wyvill said.

A couple of months later, flu brought down the entire household. Nathan got over it quickly, but not Zachary.

With her husband out of town on business, Cindy Wyvill rushed Zachary to the emergency room at John Muir and then back to the neurosurgeon at Children's Hospital, where she was told the boy probably was having trouble getting over the flu.

"Little did we all know that he was 21/2, maybe three days into a neurologic crisis, which was giving him brain damage," his father said.

Zachary didn't improve and his parents rushed him back to the hospital.

"He was really inconsolable," his father said. "He just would not stop crying."

When doctors at Children's Hospital couldn't help him, they searched for other

causes. Eventually they diagnosed Zachary with glutaric acidemia type 1, a rare genetic disorder in which his body has difficulty breaking down amino acids, causing toxic chemicals to build up in his blood and tissues.

By then, Zachary had suffered irreversible brain damage.

Cindy Wyvill got a vivid reminder of how Zachary's life might have been different when she spoke at a state hearing on newborn testing. Just one month after Zachary's birth, a Modesto woman had a boy with the same rare disorder, but he was diagnosed almost immediately under the pilot program.

Doctors quickly put him on a special diet with vitamins, and today he is a healthy 1-year-old.

Although John Muir was participating in the pilot program at the time of Zachary's birth, his parents say they were never offered the expanded testing, which the state provided free to parents who consented.

Last month, the Wyvills filed a medical malpractice lawsuit against John Muir, Children's Hospital Oakland and several doctors. Officials at the two hospitals declined to comment, citing federal patient confidentiality laws.

But John Muir spokeswoman Patty Hefner said the hospital provides information about the out-of-state labs.

The Wyvills, meanwhile, take pleasure in Zachary's small signs of progress. Medical experts believe his cognition is good, but he cannot make his body do what he wants it to do.

"No one is willing to tell us exactly what's going to happen, I guess because they just don't know," Cindy Wyvill said.

Zachary smiles often and seems to delight in regular visits from occupational and physical therapists.

"We're not giving up on any possibilities for Zach," his mother said. "The best thing he has working for him now is his own motivation. He is a sweet little kid and he laughs every day. He's just a joy to be around."

SCREENING TESTS

Parents can obtain newborn screening tests from out-of-state laboratories. Fees usually run from $25 to $100.

Parents should discuss supplemental testing with their physicians.

Information can be obtained by contacting these laboratories:

?Baylor at 800-4BAYLOR (422-9566) or www.bhcs.com/MedicalSpecialties/MetabolicDisease

?Mayo at 800-533-1710 or www.mayoreferenceservices.org/mml/mml-sns-intro.asp

?Pediatrix at 866-463-6436 or www.pediatrixscreening.com

Information is also available at these Web sites:

?CARES at www.caresfoundation.org

?Alliance for Genetic Disorders at www.geneticalliance.org

?Star G Project at www.newbornscreening.info

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## ◎德州尋求擴大新生兒篩檢立法中

在 March of Dimes 基金會德州分會的倡導下，將於 2005 年 1 月的會期之前推動倡導擴大新生兒篩檢。March of Dimes 基金會德州分會公共事務負責人 Jorey Berry 指出，他們正尋求法案的支持者，

以便使未來德州得以購置串聯質譜儀來篩檢 27 項疾病，現有德州僅篩檢 March of Dimes 基金會所建議 29 項中的 8 項疾病。

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Save Babies Through Screening Foundation has been asked to share this message with Texas families. Please contact Jorey Berry directly if you are able to participate.

My name is Jorey Berry and I am the State Director of Public Affairs for the Texas Chapter of the March of Dimes. We are leading the effort to advocate for expanded newborn screening in Texas, specifically by advocating before the Texas Legislature during the next legislative session, which begins in January 2005. We are securing bill sponsors to carry a bill that would allow our health department to purchase Tandem Mass Spectrometry and begin screening for several more disorders than we currently do. (Texas screens for 8 of the 29 disorders in the March of Dimes core group. Our legislation would allow Texas to screen for 27 of the 29.)

I am trying to build an advocacy base of Texas families affected by newborn screening disorders, who would be willing to lend their voice to our cause. Are you aware of any families in Texas who would be willing to help?

Thank you very much,

Jorey Berry
State Director of Public Affairs
March of Dimes, Texas Chapter
901 South Mopac, Suite 195
Austin, TX 78746

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## ◎美國基因醫學會建議擴大新生兒篩檢的項目

美國聯邦健康與人民服務部（Department of HHS）預計在數月

後，將發布各州新生兒篩檢的國家準繩。這項準繩係基於美國基因醫學會（American College of Medical Genetics ; ACMG）爲健康與人民服務部所研撰一份報告中的一部份，這分報告雖然尚非最後結論，但是美國基因醫學會在與健康與人民服務部所屬「新生兒與兒童遺傳異常與基因疾病顧問委員會」（the HHS Advisory Committee on Heritable Disorders & Genetic Diseases in Newborns & Children）的一項初稿會議中，已於 2004 年 9 月 23 日中投票接受結論所作建議。該 29 項建議篩檢名單如下：（其中蠶豆症（Glucose-6-phosphate dehydrogenase deficiency）原列於名單中，但經過進一步證據的檢視後被排除）

ORGANIC ACID METABOLISM DISORDERS
Beta-ketothiolase deficiency
Glutaric acidemia type I
Hydroxymethylglutaric aciduria
Isovaleric acidemia
3-Methylcrotonyl-CoA Carboxylase deficiency
Methylmalonic acidemia, Cbl A and Cbl B Forms
Methylmalonic acidemia, mutase deficiency form
Multiple carboxylase deficiency
Proprionic acidemia

FATTY ACID OXIDATION DISORDERS
Carnitine uptake defect
Long-chain hydroxyacyl-CoA dehydrogenase deficiency
Medium-chain acyl-CoA dehydrogenase deficiency
Trifunctional protein deficiency

Very-long-chain acyl-CoA dehydrogenase deficiency

AMINO ACID METABOLISM DISORDERS
Argininosuccinic acidemia
Citrullinemia
Homocystinuria
Maple syrup urine disease
Phenylketonuria
Tyrosinemia type I

HEMOGLOBIN DISORDERS
Hb S/Beta-thalassemia
HB S/C disease
Sickle cell anemia

OTHERS
Biotinidase deficiency
Congenital adrenal hyperplasia
Congenital hypothyroidism
Cystic fibrosis
Galactosemia
Hearing deficiency

[Glucose-6-phosphate dehydrogenase deficiency was originally included on this core list, but was dropped after additional evidence was examined.]

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Association of Public Health Laboratories; November 16, 2004
HHS To Recommend Uniform Panel of Newborn Screening Tests For All States
http://www.aphl.org/article.cfm?ArticleID=80

HHS TO RECOMMEND UNIFORM PANEL OF NEWBORN SCREENING TESTS FOR ALL STATES
Impacts on State Public Health Laboratories Will Vary

Within the next few months, the US Department of Health and Human Services (HHS) is expected to release national guidelines detailing a minimum set of newborn screening tests recommended for inclusion in all state newborn screening programs.

According to Peter Van Dyck, director of the Maternal and Child Health Bureau within the HHS Health Resources and Services Administration (HRSA), a major impetus for the guidelines is the wide disparity in the number of conditions now included in state-mandated newborn screening programs. For example, Kansas, Kentucky and Arkansas test for just four or five conditions each, while Iowa tests for more than 40. Said Van Dyck, e e concerned that there is not equity for parents across states, and we feel that we should move in that direction.?

The guidelines will be based in large part on a report prepared for HHS by the American College of Medical Genetics (ACMG). Although the report is not yet final, the ACMG shared an early draft with the HHS Advisory Committee on Heritable Disorders & Genetic Diseases in Newborns & Children, which voted on September 23 to ccept and recommend?its conclusions.

THE PROCESS

The ACMG study group considered 84 congenital conditions and, based on scientific data and expert opinions from a range of newborn screening stakeholders, classified them into three categories:

1) Conditions recommended for state-mandated screening, reporting, and follow-up (the uniform condition panel).
2) Secondary report only conditions.
3) Conditions not indicated for newborn screening at this time.

ACMG Executive Director Michael Watson said that the primary considerations for placement in the uniform condition panel were the availability of a reasonably accurate test to detect the condition and an efficacious therapy to treat it. However, he noted that the criteria were ot entirely objective.?For example, the study authors considered such questions as ow easy is it to provide a (therapeutic) diet to a baby with PKU??

Watson also noted that the ACMG considered two alternate recommendations for the core panel: o argue that (government) mandated newborn screening is the only way or to say that . . . it should be the standard of care that all babies be screened, basically putting responsibility on the pediatrician. e opted for the first choice,?he said.

The 25 report only conditions ncluding hemoglobin variants, argininemia, malonic acidemia, galactokinase deficiency and MCKAT, to name a few re conditions that are generally detected as part of the differential diagnosis of disorders in the core panel, but ranked low in the ACMG scoring process because they have poorly understood natural histories, currently have no treatments and/or are relatively benign. The ACMG recommends that these secondary findings be reported to parents and healthcare providers, but, said Watson, e don presume the state to be obligated to really monitor these patients long-term.?

Twenty-nine additional conditions ncluding insulin-dependent diabetes mellitus, fragile X syndrome, creatine transport defect and lysosomal storage diseases ere not indicated for newborn screening as reliable tests are not currently available. Newborn screening recommendations for three infectious diseases IV, toxoplasmosis and cytomegalovirus ere deferred.

## REACTION TO THE ACMG REPORT

George Cunningham, who oversees California newborn screening program, lauded many aspects of the ACMG report. The study group, he said, ade a pretty reasonable case?for all of the conditions in the core group. Moreover, he found the model decision matrix included in the report to be ery useful?for states as they consider expanding the core panel as new testing technologies become available. Cunningham said the proposed standards to evaluate the quality of newborn screening programs should be implemented nationally o we e comparing apples to apples (across states).?

But Cunningham objected to the scoring system used to rank conditions, finding it rbitrary and somewhat subjective.?He said, tates should not accept the scoring system as the final arbiter of what they should add to their screening programs.?He also disagreed with the designation of report only

conditions. ll of those conditions,?he said, eed to be followed up, and states need to collect and pool data nationally. If you find something in your screening, you have to follow up on the few you find.?

Cunningham said the standard in California is that nything we report, we follow up.?The state has 14 metabolic centers where families are referred for confirmatory tests, diagnosis and treatment of rare infant disorders, and relevant case data is reported back to the newborn screening program.

A basic, unresolved issue that goes beyond the ACMG report, said Cunningham, is how to count newborn screening tests to enable objective cross-state comparisons. For example, phenylketonuria can be considered one condition or as many as seven, involving different tests, different prognoses and different treatments. e need professional agreement,?he stated.

WHAT IMPACT WILL THIS HAVE?

Before HHS releases final recommendations, Van Dyck said the agency will consider the input of its genetics committee, state maternal and child health programs, professional associations (including APHL), public health laboratory directors and others. He noted that HHS has o authority to demand that states comply with whatever recommendations we come out with,?but that the agency will offer whatever technical assistance it can to facilitate compliance. Earlier this year, HHS funded seven regional newborn screening centers to share best practices among states and develop regional strategies to optimize the full gamut of newborn screening services from educating parents to assuring access to biochemical geneticists and other medical specialists.

William Becker, medical director of the Ohio State Department of Health and a member of the genetics committee, noted in an email that state responses to final HHS recommendations ill vary from the need for legislation enacted to consideration of a fee structure (or modification of an existing fee) to (changes in) the panel of disorders screened to the practice patterns of diagnosis and follow up to issues of monitoring, quality assurance and quality improvement.?

Specific impacts on public health laboratories hich oversee all current public newborn screening programs in the United States and perform the vast

majority of the testing ill depend on the extent to which states expand their screening panels in response to national guidelines. Harry Hannon, chief of CDC Newborn Screening Branch, said that one concern is the need for public health laboratories to acquire expensive new technologies, such as tandem mass spectrometry, to enable them to perform many of the tests recommended by the ACMG. Even though some authorities advocate regional models in which states with low birth counts share testing resources, Hannon said a state natural inclination is to be self-sufficient. o you have to have a car or can you take the bus??he asked rhetorically.

## RESOURCES NECESSARY TO EXPAND SCREENING PROGRAMS

Other concerns are the need for additional staff training and perhaps even additional staff members to handle an increased test load. o resources are available to enable all states to comply with the (ACMG) recommendations,?said Cunningham. Currently, 45 states charge fees to cover the laboratory costs associated with newborn screening. But fees may be insufficient to support greatly expanded screening programs and are not always fully covered by private health insurance or Medicaid.

APHL, CDC, HRSA, and the National Newborn Screening and Genetics Resource Center co-sponsor newborn screening training programs for laboratory scientists through the National Laboratory Training Network and at Duke University Biomedical Center and the Baylor Medical University Institute of Metabolic Disease. APHL also co-sponsors the Newborn Screening Quality Assurance Program (NSQAP), which assesses testing proficiency in state newborn screening laboratories. Hannon said the NSQAP is hampered by the lack of commercially available chemicals to use for quality control and proficiency testing purposes for some of the conditions on the proposed uniform screening panel; LCHAD, for example. Lacking actual biomarkers of interest, the program uses markers that closely resemble those biomarkers so that the NSQAP can conduct proficiency testing for the full range of disorders for which states might screen.

Despite concerns about resources, however, all of those interviewed for this article viewed expanded screening favorably. CDC Hannon, for example, said, a firm supporter that every baby should have access to testing for everything that available (meeting criteria).?

California has already passed legislation that will enable the state newborn screening program to begin a tandem mass spectrometry program sometime next summer. t that time,?said Cunningham, e will be screening for most of those 29 conditions (on the proposed core panel) and will add others soon after,?including many not on the list.

The March of Dimes, an influential infant health advocacy group whose president serves on the HHS genetics committee, has revised its newborn screening policy to include all of the disorders listed in the ACMG report core panel and will base its periodic evaluation of states?newborn screening performance on at least those conditions. The group associate medical director, Siobhan Dolan, said that amilies have had tragic experiences feeling the inequities of the (current newborn screening) system.?All children, she said hould benefit from the wonderful possibilities inherent in newborn screening.?The group will work through its local chapters to stimulate public advocacy to bring state policies into accord with national March of Dimes recommendations.

In the meantime, the HHS genetics committee has met only twice. he committee work is just beginning,?said Becker. Future recommendations will likely deal with funding issues as well as consideration of a national newborn screening process. Becker noted that experts predict the technology to screen for many genetic disorders is ust over the horizon.?This tough issue,?he said ill need much more discussion.?

UNIFORM CONDITION PANEL RECOMMENDED BY AMERICAN COLLEGE OF MEDICAL GENETICS
(As of October 2004)

ORGANIC ACID METABOLISM DISORDERS
Beta-ketothiolase deficiency
Glutaric acidemia type I
Hydroxymethylglutaric aciduria
Isovaleric acidemia
3-Methylcrotonyl-CoA Carboxylase deficiency
Methylmalonic acidemia, Cbl A and Cbl B Forms
Methylmalonic acidemia, mutase deficiency form

Multiple carboxylase deficiency
Proprionic acidemia

FATTY ACID OXIDATION DISORDERS
Carnitine uptake defect
Long-chain hydroxyacyl-CoA dehydrogenase deficiency
Medium-chain acyl-CoA dehydrogenase deficiency
Trifunctional protein deficiency
Very-long-chain acyl-CoA dehydrogenase deficiency

AMINO ACID METABOLISM DISORDERS
Argininosuccinic acidemia
Citrullinemia
Homocystinuria
Maple syrup urine disease
Phenylketonuria
Tyrosinemia type I

HEMOGLOBIN DISORDERS
Hb S/Beta-thalassemia
HB S/C disease
Sickle cell anemia

OTHERS
Biotinidase deficiency
Congenital adrenal hyperplasia
Congenital hypothyroidism
Cystic fibrosis
Galactosemia
Hearing deficiency

[Glucose-6-phosphate dehydrogenase deficiency was originally included on this core list, but was dropped after additional evidence was examined.]

The Association of Public Health Laboratories (APHL) works to safeguard the public's health by strengthening public health laboratories in the United States and across the world. In collaboration with members, APHL advances

laboratory systems and practices, and promotes policies that support healthy communities.

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## ◎陸易斯安那州篩檢 10 項新生兒疾病

陸易斯安那州 Lafayette Daily 報於 2004 年 11 月 30 日指出，該州健康部門所屬實驗室自 2004 年 11 月 1 日起開始篩檢 10 項可能致命的新生兒基因異常疾病。該州基因計畫（Louisiana State Genetics Program）負責篩檢所有出生的新生兒，而不像其他的州，該計畫尚且供應後續的飲食奶粉及大多數的藥物，事實上這些特殊奶粉往往就等於是她們的藥物，而聯邦的 Medicaid 計畫則給付照護的費用。

這 10 項所篩檢的疾病發生率極低，分別介於一萬至十萬分之一，而由於每年陸易斯安那州有 65,000 新生兒，因此所篩檢的罹病人數也在增加中。在 2004 年 11 月 1 日以前，陸易斯安那州已篩檢 3 項基因異常疾病和 sickle-cell anemia、under-performing thyroid gland，預期將逐漸朝向 30 種新生兒疾病的篩檢。目前州健康部門所屬實驗室每天能篩檢約 500 項檢體，都是拜自動化的串聯質譜儀所賜。

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The Lafayette Daily Advertiser; November 30, 2004
Family Benefits From New State Testing That Is Already Saving Lives
http://www.acadiananow.com/news/html/87E566F8-15A0-40C3-AF72-0242C0A104FB.shtml

FAMILY BENEFITS FROM NEW STATE TESTING THAT IS ALREADY SAVING LIVES

OPELOUSAS ?Laney and Shane Smith of Opelousas have a lot to be thankful for this holiday season. Their new daughter, Macey, is alive and well, thanks to a new state policy that allowed doctors to detect a genetic disorder that would have killed her.

On Nov. 1, Louisiana state health lab began checking for eight potentially deadly genetic disorders in the blood of all newborns. Macey Smith was the 57th baby to receive the tests and the first to benefit.

Macey tested positive for citrullinemia, a rare genetic disorder that would have killed her within weeks.

t first, it was so scary,?Laney Smith said. t was overwhelming.?
Now, thanks to the tests and prompt action by doctors, Macey is thriving.

According to the Smiths, by all rights, Macey should have died. She was born Oct. 24, a week before the new testing program was to begin.

hank God they found it,?Laney Smith said. he wasn even supposed to be looked at.?
Hayward Genetics Center, part of the Tulane University Health Sciences Center, handled Macey case.

ust because the state was behind in its work load she was saved,?Fran Simon of Tulane HSC said. therwise, the baby would have gotten sleepier and sleepier and died.?
Citrullinemia prevents a baby body from properly processing protein, causing a buildup of ammonia that leads to sluggishness and, eventually, coma and death.

Like many of the other disorders on the state testing list, citrullinemia has no easily detectable symptoms such as a rash or high fever.

here are really no signs,?Laney Smith said. he just looked like a sleepy baby. I had no idea anything was wrong with her.?
A call from Dr. Hans Andersson with the Hayward Center is credited with saving Macey life.

a clinical geneticist,?Andersson said. e are the referral center for new babies who test positive. We do the confirmatory testing.?
He called the baby doctor, who took further tests at Opelousas General Health System, where Macey was born. The next day, Macey was transferred to Tulane University Hospital, where she spent 36 hours in neonatal intensive -care.

er ammonia level was at 263 ?anything above 300 starts causing brain damage,?Laney Smith said. hey caught it just in time.?
According to Andersson, anything above 70 is considered abnormal. He said the disease would not normally be noticed until the child is catastrophically ill.

his is what is great about this screening program. You can start treatment before there is damage,?Andersson said.

t gives me chills when I think how close she was,?Laney Smith said. he doing much better. She fighting for her bottle. She staying up more.?
Andersson praised his entire team with helping to save Macey life.

t takes a great deal of time and energy ?it a team effort,?Andersson said.

He also had praise for the Louisiana State Genetics Program, which screens the blood of all newborns. He said the state program is one of the best. Fifteen states don do any testing at all.

nlike many other states, the LSGP pays for all the diet and most of the medicines. This is extremely important,?said Andersson, who along with Amy Cunningham ?a metabolic nutritionist on his staff ?has developed a drug and diet regimen for Macey. Medicaid is paying for her care.

n cases like this, the food is really the medicine,?she said.

While Macey will need to watch her diet for her entire life, Andersson said there is no reason she cannot lead a healthy, full life.

he baby will be followed once a month in our clinic in Lafayette. We want to make sure she stays healthy,?Simon said.

The incidence of each of the 10 disorders now being tested is extremely low, ranging from one in 100,000 to one in 10,000. Still, the state has about 65,000 newborns each year, so the numbers add up.

Even before Nov. 1, the state was already testing for three genetic disorders plus sickle-cell anemia and an under-performing thyroid gland. It hopes to increase those tests to include 30 genetic diseases in coming years.

Andersson said the increase will be done gradually.

he state lab already does 500 blood tests every day. We need to do it gradually to keep from overwhelming the lab,?Andersson said. What makes the increased testing possible is an automated screening process known as Tandem Mass Spectrometry.

his is a new technology that has come along. This allows us to analyze for additional indicators,?Andersson said.

Andersson added that, at least for now, testing will be limited to treatable conditions.

e don screen for diseases that we can do anything for,?he said.

Today, Macey is home with her parents, who are both Opelousas natives. Laney is a stay-at-home mom and Shane a carpenter. They also have a 3-year-old son, Brennon.

e loves his new sister. He getting good with her, wanting to hold her,?Smith said.

(John Pope with The (New Orleans) Times-Picayune contributed to this story.)
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## ◎科羅拉多州規劃擴大新生兒篩檢項目

科羅拉多州 Rocky Mountain 報於 2004 年 11 月 20 日指出，新的科技創新預期將每年將篩檢出該州 20 名以上罹患先天代謝疾病的病童。如果這項方案經過立法通過，採用能篩檢 34 種不同疾病的串聯質譜儀將是科羅拉多州的重要進展；而這項創新也將取代原來只篩檢 7 項疾病的舊有計畫，收費將介於 53.25 至 59 美元之間，也將完全免費，預期將於 2006 年 1 月 1 日全面實施。

新生兒篩檢在科羅拉多州並不是新鮮的事，因為科羅拉多州自 1963 年起即開始篩檢苯酮尿症，隨後也陸續加入 cystic fibrosis 和 sickle-cell anemia 等。現有的新生兒篩檢尚需事先經家長同意，而科羅拉多州公共健康與環境局（Department of Public Health and the Environment）則指出，98%的家長均表示同意。

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Rocky Mountain News; November 20, 2004
Saving Babies' Lives
http://www.rockymountainnews.com/drmn/opinion/article/0,1299,DRMN_38_3341821,00.html

SAVING BABIES' LIVES

New technology may mean new life for up to 20 Colorado babies who are

born each year with one of dozens of rare, inherited metabolic diseases.

This is an important step for Colorado to take. If the program is approved by the legislature, tandem mass spectrometry, as it's called, will screen for 34 different conditions. The screening is done twice, once almost immediately after birth and the second time seven to 10 days later. Each requires only a spot of blood on a card. The system will replace a screening program that checks only seven conditions. The fee - the screening is entirely cash-funded - will rise from $53.25 to $59. It would be fully implemented by Jan. 1, 2006.

Newborn screening is nothing new. Colorado began testing for a condition known as phenylketonuria in 1963. Over time, other serious conditions have been added, including cystic fibrosis and sickle-cell anemia. Not all can be treated with complete success, but the harmful consequences can usually be reduced if treatment is started early.

Newborn screening should be nothing controversial, and for the most part it isn't. Parents are asked to consent, and the Colorado Department of Public Health and the Environment says 98 percent do. Some people do object to the counseling that can follow a diagnosis, perhaps on the grounds that it might encourage parents to consider abortion in a later pregnancy if the condition happened again. But that is hardly the only possible outcome. They may decide not to have any more children, or to adopt instead.

In any case, concern about what might happen in a future pregnancy is not a very good reason to refuse testing on a child who has already been born. It means taking a risk with that child's health or even life, and few parents will want to do that.

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## ◎科羅拉多州將擴大新生兒篩檢列為最優先健康政策

科羅拉多州 Rocky Mountain 報於 2004 年 11 月 16 日報導，科羅拉多州公共健康與環境局的局長 Doug Benevento 指出，科羅拉多州

將擴大新生兒篩檢列為最優先健康政策，預計 2006 年將把新生兒篩檢項目提高至 34 項，是過去檢驗項目的五倍以上之多，檢驗費用也由 5.75 美元提高為 53 美元，約可多發現 20 位可治療的罹病嬰兒。

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Rocky Mountain News(Colorado); November 16, 2004
Testing Newborns A Top Priority In Colorado
http://rockymountainnews.com/drmn/local/article/0,1299,DRMN_15_3331820,00.html

TESTING NEWBORNS A TOP PRIORITY
Health department to add on to exam to find birth defects

By Bill Scanlon, Rocky Mountain News
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Old and new faces on the Joint Budget Committee grumbled Monday about the slow pace of lifting Colorado out of last place in childhood immunizations.

Doug Benevento, executive director of the Colorado Department of Public Health and Environment, told the panel that so far a $1.4 million effort to help boost the state's immunization rate has focused on information gathering.

Three years ago, when Colorado had the 35th-best rate in the nation, the Joint Budget Committee asked the state health department to gather information so the rate could be improved, noted Rep. Tom Plant, D-Boulder. "Here we are in 2004, and we're actually lower. It's frustrating."

Sen. Abel Tapia, D-Pueblo, chairman-elect of the Joint Budget Committee, said, "It's odd to hear that you're just developing a plan. Immunization is an important issue on the campaign trail. If you don't immunize, you're going to spend a lot more dollars later on health problems."

Benevento said he doesn't want to launch a full-scale effort to immunize kids until he knows where the rates are lowest and what the roadblocks are.

According to the National Immunization Survey, Colorado ranks last in the nation, with 63 percent of its 2-year-olds fully immunized.

Lawmakers have blamed Colorado's tough Medicaid eligibility rules and the difficulty rural families have reaching a clinic.

But another reason came up on Monday. Two years ago, when two manufacturers pulled out of the market, leaving a shortage of doses for diphtheria, tetanus and whooping cough, some states ponied up the money to buy doses at inflated prices, Benevento said. Colorado did not.

Colorado was facing a budget crisis and had depleted its infant immunization fund for other purposes.

"Frankly, it was expensive. We didn't have the dollars to buy them," he said.

Some shots, like the ones for whooping cough, are designed primarily to protect the individual from the population, said Dr. Ned Calonge, Colorado's chief medical officer. Others, like the ones for measles, are designed to protect the population from a single exposed person. Measles can spread quickly through a neighborhood or school, especially a preschool.

That's why it's important to raise the overall rate of vaccination.

Plant said immunizations are a high priority, so as soon as the health department comes up with a way to reach the under-immunized kids, the money will be found.

Panel members praised the department for acquiring and appropriating the funds needed to get drugs to the 350 HIV-positive Coloradans who'd been on a waiting list.

Newborns next year will be tested for 34 treatable birth defects - five times more than this year's Colorado babies were treated for, Benevento told the panel.

"It's our top priority," he said.

Adding $5.75 to the $53 test will mean 20 additional babies a year will be found with defects that are treatable, Calonge said.

Seven Colorado babies a year are born with medium-chain Acyl-CoA Dehydrogenase Deficiency, a disorder that greatly increases the risk of sudden infant death syndrome, Calonge said. He expects the new test to save the life of one baby suffering from the disorder each year. The additional tests will improve the lives of several others each year.

Nurses will prick the babies' heels a few days after birth to draw blood, then smear it on a card. Health department technicians will run the card through scanners to detect problems from cystic fibrosis to sickle-cell anemia.

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**◎加州自 2005 年 8 月起擴大新生兒篩檢項目**

加州聖地牙哥 KFMB 新聞台於 2004 年 11 月 15 日指出，加州將自 2005 年 8 月起擴大新生兒篩檢項目，目前加州僅篩檢四項代謝疾病，但同樣的血片卻可作為擴大檢驗數十種新生兒篩檢項目之用，現階段家長可經由醫師諮詢自行尋求這項擴大的新生兒篩檢。

州參議員 Dede Alpert 指出，目前加州的新生兒篩檢遠遠落後各州，因此他支持修正加州落時的新生兒篩檢立法；他指出新生兒因未適當篩檢導致昏迷、腦部傷害、殘障、甚至死亡，但只要篩檢後飲食控制即可避免上述的嚴重後果。他也主持一項公開資訊的服務，讓家長們知道最新的新生兒篩檢訊息，雖然要花費 25 元，但卻是你所花過最值得的 25 元；這項新增的篩檢，自 2005 年 8 月起也將成為加州標準的新生兒篩檢流程。

州參議員 Dede Alpert 指出加州過去在此議題上行動不夠迅速，主要是因為成本考量，但是有一項研究已指出，我們每花費一元在新生兒篩檢上，即能在未來節省三元的支出，而每一項所篩檢的疾病雖然都十分罕見，但所有超過 30 項的新生兒篩檢，預計一年將可在加州發現 140 位罹病的小孩。

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KFMB-TV News (San Diego, California); November 15, 2004
Additional Screening For Babies
KFMB.COM
ADDITIONAL SCREENING FOR BABIES

(11-15-2004) - Starting in August of 2005, babies born in California will begin being screened for several preventable illnesses. Now until that time, parents can give their babies added protection, but they  l need to know what questions to ask.

With a quick knick of a heal, newborns in California are tested for four metabolic disorders. But that same drop of blood could be tested for about 10 times that number of disorders.

e're really lagging behind the rest of the country,?said State Senator Dede Alpert.

Alpert championed the cause to revise California's outdated testing of newborns.

ou see babies that have gone into comas, again babies who've suffered irreparable brain damage, physical damage as well and in the worst cases death,?continued Alpert.

And parents could prevent the horrible outcomes of these disorders just by changing a baby's diet.

Senator Alpert created a public service announcement to make parents aware that updated testing is available, but right now you have to ask your doctor for it.

It about $25, but Alpert thinks it could be the best $25 you ever spent.

Starting in august of 2005, it will be the standard practice in California.

Alpert says the only reason our state didn act sooner was because of the perceived cost.

But this may be minimal compared to the state programs that are needed to provide for these children.

e've had a study done that indicates you probably save $3 for every $1 that's spent,?explained Alpert.

All of the more than 30 disorders that will be tested are very rare. It's estimated only 140 babies will be saved by these tests.

California Dept. of Health - Newborn Screening: (510) 412-1502

Save Babies Through Screening Foundation - www.savebabies.org

California DHS ?Genetic Disease Branch - www.dhs.ca.gov/gdb/

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## ◎俄亥俄州開始擴大新生兒篩檢至 30 項

俄亥俄州哥倫布士派遣報（The Columbus Dispatch）於 2004 年 8 月 6 日指出，俄亥俄州將自 2004 年 8 月起擴大新生兒篩檢至 30 項。該州於 1990 年起要求篩檢 5 項，2002 年起要求強制篩檢 13 項及 16

項自行決定篩檢與否，若家長決定篩檢則提供免費補助；而 2004 年 8 月起擴大新生兒篩檢後，也將使俄亥俄州成為全國篩檢項目最多的 19 個州別之一，而只要一滴血即可檢驗數十種疾病的檢查。

負責管理州屬新生兒篩檢實驗室業務的 Dr. William Becker 指出，過去兩年中，已經有約三十萬新生兒接受篩檢，約占過去兩年出生新生兒的 96% 。他說新發明的串聯質譜儀讓這項多元的檢驗變得可行，俄亥俄州大學醫學中心的醫院協調員 Gail Johannes 指出，我們希望警告愈少的人，但同時又不能錯過每一個可能的生命，因此偽陽性的問題雖然會發生，卻是必要之惡。

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the Columbus Dispatch; August 6, 2004
Ohio Newborn Screening To Expand This Month

By: Sarah Frank

NEWBORN SCREENING TO EXPAND THIS MONTH
Routine blood scan will now check for 30 serious disorders

When Candy Kush adopted 5-week-old Kelsea, she knew her daughter had a rare amino-acid disorder that would require dietary vigilance for life.

Kelsea tested positive at birth for phenylketonuria, or PKU, one of only a few diseases Ohio hospitals screened for when she was born 16 years ago.

This month, the Ohio Department of Health will start requiring hospitals to screen babies for 30 disorders. In the 1990s, there were five required tests.

Since 2002, 13 have been mandatory and 16 optional.

The change will make Ohio one of 19 states that require that many newborn screenings.

One blood draw is enough to run all the tests.

Kelsea's disease makes it difficult for her to digest an amino acid found in meats and milk. Her treatment is a strict lowprotein diet of mostly vegetables. It affects about one in 17,000 newborns.

If the disease isn't found at birth, it can cause brain damage and mental retardation.

Kush considers her daughter lucky.

"I guess it's too late for the kids who were born with those rare diseases," said Kush, a 48-year-old West Side resident, referring to the tests. "But it's just in time for babies being born today."

Kush is a member of the Newborn Screening Advisory Council, a group that makes recommendations to the state Health Department.

Among the newly mandated tests are a number of metabolic disorders and an enzyme disorder called biotinidase deficiency.

Some of the diseases occur in less than one in 1 million babies. They can lead to brain damage or immune system failure. Common symptoms can include weight loss, jaundice and vomiting.

Some symptoms don't show for more than a year.

In 2002, the state created a program in which parents could opt to have their newborns tested for the additional 16 diseases at no cost.

About 300,000 babies - or 96 percent of those born during the past two years - were tested, said Dr. William Becker, who oversees the state's lab.
Becker said a mass spectrometer has made it possible to perform multiple tests.

In 1996, Ohio tested for five diseases and Health Department officials said the cost of adding others outweighed the benefits. That attitude has changed, Becker said.

"Right now, the philosophical camp that's winning the tug of war is that camp that supports screening for everything that's possible," he said.
Hospitals purchase screening kits from the state for $33.75 apiece. That is expected to rise to $45.16 this month to cover the cost of testing for biotinidase deficiency.

Gail Johannes, hospital coordinator for the Ohio State University Medical Center, said false positives will occur but are a necessary evil.
"You want to alarm as few people as possible, but you don't want to miss anyone," she said.

Email: sfrank@dispatch.com
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**◎美國各州新生兒篩檢項目差異大**

西維吉尼亞州 The Herald-Dispatch 報於 2004 年九月 22 日指出，根據美國全國新生兒篩檢暨基因資源中心（U.S. National Newborn Screening and Genetics Resource Center）資料顯示，全國現有 15 個州的新生兒篩檢項目在五項或五項以下，其中包括西維吉尼亞州和肯德基州，而篩檢超過 25 項遺傳疾病的州則有 19 個，其餘的州則篩檢數目介於兩者之間；而上週俄亥俄州篩檢數目增加為 30 項疾病，提升到第 13 名。

最近聯邦的一項分析顯示，由於各州新生兒篩檢項目的差異，每年因此有超過 1,000 位以上尚未被診斷的罹病新生兒，而新發明的串

聯質譜儀則可透過一滴血檢驗超過 40 種以上嚴重或致命的基因疾病。美國目前每一位新生兒都被篩檢兩項若不治療會導致心智障礙的罕見疾病，包括 hypothyroidism 和 hypothyroidism（PKU），而大多數的州也都篩檢了 sickle cell anemia。

未來的家長被建議接受該州強制的新生兒篩檢項目，若該強制的篩檢項目少於 30，則被建議尋求民間的檢驗實驗室補作不足的篩檢，費用約介於 25 至 100 美元之間。當大家在辯論各州是否應篩檢 30 項新生兒疾病時，本週聯邦一顧問委員會則將建議去除區域間的差異加以統一；部分州多因預算不足而限制篩檢項目，這項擴大的新生兒檢驗一般花費 50 美元，並由保險給付，但首先各州便需先自購或與臨州共享一部約 40 萬美元的機器。

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The Herald-Dispatch (West Virginia); September 22, 2004
Newborn Testing Varies By State
http://www.herald-dispatch.com/2004/September/22/LNspot.htm

NEWBORN TESTING VARIES BY STATE

Genetic diseases often diagnosed with a single drop of blood -- but screenings could cost extra

By CHRISTINA REDEKOPP

Ryann Davis, 2, rests her head beside her 3-month-old sister, Rylee Davis at their home in Barboursville Thursday. Rylee Davis was born with Nonketotic Hyperglycinemia, a rare genetic metabolic disorder. Deborah

Davis, Rylee and Ryann mother. would like to educate people about options pregnant women have for comprehensive newborn testing. West Virginia is one of the states that do the least amount of testing.

BARBOURSVILLE -- Rylee Davis looks like a typical baby napping peacefully nestled beneath a blanket on a couch at her mother side.

The 3-month-old always appears this way because she has nonketotic hyperglycinemia, a rare genetic disease. If her parents, Aaron and Deborah Davis of Barboursville, lived in another state, the disorder might have been detected during a newborn screening.

Although Rylee Davis?condition is untreatable regardless of early testing, she might have been spared some of the tests she had to undergo later on to be diagnosed, Deborah Davis said.

"There no question that different states screen for different disorders," said Dr. Charlotte Jones, pediatric neurologist at University Pediatrics at the Marshall University Medical Center. "A child living in West Virginia is screened for different disorders than a child screened in Arizona, Alaska or Alabama. The poorer the state, the less they screen for."

In Rylee Davis?case, after she was sent home from the hospital, her parents returned within 24 hours because she was not responding as their firstborn, Ryann, had. At first, hospital staff speculated dehydration and meningitis. After a series of tests, she was diagnosed with the fatal disorder that causes retardation and lethargy among other symptoms.

A more comprehensive screening may have taken several days to come back and Rylee still may have had to undergo such tests as spinal taps, blood work and MRIs, Deborah Davis said. But Davis wants to at least get the word out that parents have more screenings available to them -- they may just be at an extra cost.

Fifteen states, including West Virginia and Kentucky, require testing for five

or fewer of rare inherited diseases, according to the U.S. National Newborn Screening and Genetics Resource Center. Nineteen states test for 25 diseases or more; the rest fall in the middle. Last month, Ohio began requiring screening for 30 diseases, up from 13.

A recent federal analysis suggests that more than 1,000 babies a year may go undiagnosed because of state testing variation. New technology called tandem mass spectrometry can analyze a single drop of blood for more than 40 other serious, sometimes life-threatening, genetic diseases.

Today, every U.S. baby is tested for two rare diseases that can cause retardation if untreated: hypothyroidism and the metabolic disease phenylketonuria, or PKU. Most also are tested for sickle cell anemia, a blood disease.

Expectant parents are advised to check what their state requires and if it

fewer than 30, to consider a private screening lab, which may cost $25-$100.

This week, a government advisory committee is expected to move to end the geographic disparity, as it debates whether every state should test every newborn for 30 genetic illnesses.

States often cite tight budgets for limiting testing. Per person, the tests cost roughly $50, covered by insurance. But first, the state must buy, or share with a neighboring state, a $400,000 machine to do the analysis.

Unfortunately, some of the diseases that are tested for do not have a treatment, Jones said. Also, like other health screenings, results have a high rate of false positives.

"This testing is available, you have to be able to pay for it," Jones said.

Deborah Davis said the issue also spurs a debate about whether families want to know about disorders for which symptoms may not appear until later in life.

In the meantime, Davis is glad to live where she does because of the wealth

of specialists in Huntington who are available to the family. She also wants to help other pregnant women and families to be aware about the issue of newborn testing and that kits are available for more comprehensive testing.

"Some disorders the comprehensive screen tests for are treatable if caught early enough -- usually within two weeks after birth," Davis said. "The child would go on to live a healthy and normal life. If left untreated, the child could have retardation and other health issues."

Dr. Joe Evans, a pediatrician at University Pediatrics, sees numerous infants and no parent has ever asked him about more comprehensive testing, he said.

"Obviously, it would be nice if we would test for more, I sure there expense involved," he said.

The four disorders that West Virginia tests for are rare but treatable, he said.

"(Rylee ) really beat the odds," Davis said. "We feel God sent her here for a reason. We feel part of our responsibility is to educate the public."

The Davis family also has a 2-year-old, Ryann, who doesn have the disorder. NKH is caused by a recessive gene carried by both parents so Deborah and Aaron Davis have a one-in-four chance of having a baby with the disease, Deborah Davis said.

Davis said she wants to spread the word about services available to local residents who have children with special needs.

"It important for other families to know you don have to live a defeated lifestyle," Davis said.

In particular, Davis says West Virginia Birth to Three helps with speech therapy and developmental and occupational therapy. Also, pediatric hospice

is available to families.

"My husband and I decided we weren going to live each day of her life waiting for her to die," she said. "We take her to the grocery store and to church. We e just been loving and enjoying her because we don know how long we will have her. We feel we e blessed to have her in our lives."

ON THE WEB
A national database at http://genes-r-us.uthscsa.edu lists what tests states now require. For supplemental screening, that Web site also lists private labs, such as Baylor Medical Center or Pediatrix Medical Group, that sell test kits for parents to bring to the hospital. The nurse who performs the state-mandated blood test can take a drop for the private test -- The Associated Press.

Lauran Neergaard, medical reporter for The Associated Press in Washington, contributed to this report.

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**◎March of Dimes 基金會對新生兒篩檢報告的聲明（2004 年 9 月 22 日）**

針對美國健康與人類服務部（Department of Health and Human Services）所屬「母子健康局」（Maternal and Child Health Bureau）與美國基因醫學會（American College of Medical Genetics；ACMG）對於新生兒篩檢的報告，March of Dimes 基金會發表如下之聲明：

「無論新生兒的出生地為何，本會支持我國針對所有新生兒進行廣泛的篩檢；只要符合對於新生兒有助益及血滴等的檢驗能準確早期

診斷，本會的政策一向支持新生兒篩檢。我們認同家長有權知道檢驗的全部結果，也支持強化對醫療提供者有關新生兒篩檢的教育。本會所屬各州分會與家長將與州長、州議員、與州健康部門緊密合作，以改善各州的新生兒篩檢業務。

本會十分認同美國基因醫學會（ACMG）報告中所作建議，如提升新生兒篩檢該項領域、確立新生兒篩檢的工作小組、及提供各州的政策指引等；我們支持該報告所作的建議，並敦促健康與人類服務部部長應接受該建議，以作為全國新生兒篩檢的統一準則。

而基於美國基因醫學會（ACMG）的報告，本會對於新生兒篩檢的立場也將有如下的補充：我們將敦促各州政府依據美國基因醫學會（ACMG）報告所建議的篩檢至少三十項代謝異常的項目，這些項目均符合本會過去考量新生兒篩檢的原則，也涵蓋本會過去所建議的九項代謝異常篩檢與聽力篩檢，我們也將修正並納入該三十項項目於對各州政府新生兒篩檢執行成效的評估中。

我們也將督促各州政府針對美國基因醫學會（ACMG）報告中所提25項，雖然檢驗值得信賴但目前卻乏有效治療的項目，提供檢驗的結果；我們未來對於各州政府新生兒篩檢執行成效的評估中也將列入此部分。我們也必須指出，不同的組織對於新生兒篩檢項目的分類與計算也有所不同，也因此篩檢的內涵雖然相同但項目有時仍有些差

異。

我們將敦促各州政府將來能知會家長有關擴大新生兒篩檢的潛在助益和可及性；我們也將建議專業團體如美國小兒科醫學會和政府政策規劃者，發展更佳的系統來教育醫事專業人員有關新生兒的篩檢。我們也敦促政府與民間一起合作來強化新生兒篩檢，包括開發和驗證新的方法來診斷和治療異常疾病，以及確保對於受疾病影響的家庭能提供及時的追蹤與諮商。

新生兒篩檢是項快速變遷的領域，我們深知專家對於新生兒篩檢的意見也將隨著醫療證據的增加而演化，也因此我們隨時準備更新我們對於政府和大眾的建議，此外我們也敦促美國基因醫學會的相關報告能定期更新以因應新的資料產生；同時以美國基因醫學會的建議為模式，未來針對新生兒感染疾病和其他異常項目也應定期檢視，以納入統合的新生兒篩檢項目中。

本會未來將繼續與聯邦及州政府合作，擴大新生兒篩檢項目與確保本國每一位新生兒的均等，也敦促關心此議題的個人、團體、醫療提供者更加緊密合作，以達成共同的目標。」

註 1：美國基因醫學會於 2005 年 1 月將 G6PD（俗稱蠶豆症）從新生兒篩檢建議名單中移除。

註 2：March of Dimes 基金會對過去對於新生兒篩檢的建議項目包括，Phenylketonuria (PKU)、Congenital hypothyroidism、Congenital adrenal hyperplasia (CAH)、Biotinidase deficiency、Maple syrup urine disease、Galactosemia、Homocystinuria、Sickle cell anemia、Medium chain acyl-CoA dehyrogenase deficiency (MCAD)。

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MARCH OF DIMES STATEMENT ON NEWBORN SCREENING REPORT

WHITE PLAINS, N.Y., SEPT. 22, 2004 -- The March of Dimes today issued the following statement on the report on newborn screening prepared for the Maternal and Child Health Bureau of the U.S. Health Resources and Services Administration by the American College of Medical Genetics (ACMG):

The March of Dimes supports comprehensive newborn screening for all babies in this country, regardless of their place of birth. Our policy is to support screening for specific conditions when there is a documented benefit to the child and there is a reliable test that enables early detection from newborn blood spots or other means. We support parents' rights to be fully informed about their baby's screening results, and we support expansion of health care provider education about newborn screening. March of Dimes state chapters and their partners work closely with governors, state legislators, and health departments to improve state newborn screening programs.

The March of Dimes strongly commends the ACMG report for advancing the field of newborn screening, defining a uniform panel for newborn screening, and providing a policy framework for the states. We support the recommendations in this report and we urge the Secretary of Health and Human Services to accept them as a national standard for newborn screening.

Based on the findings of this report, the March of Dimes will expand our policy on newborn screening as follows:

We will urge every state to screen every baby for at least the 30 disorders listed in the ACMG report. These 30 disorders meet our inclusion criteria, and include all of the nine metabolic tests plus hearing screening contained in our previous policy. We will revise our periodic evaluation of states' performance on newborn screening to include at least these 30.

We will urge states to provide test results for an additional 25 "reportable" conditions named in the ACMG report for which there are reliable tests but not yet documented treatments. We also will revise our periodic evaluation of states to include this reporting.

It must be noted that various organizations have alternate ways of classifying and counting many of these disorders and may have different number totals for these same conditions.

We will urge states to inform all parents prospectively about the potential benefits and availability of comprehensive newborn screening.

We will recommend that professional groups, such as the American Academy of Pediatrics, as well as government policy makers, develop better systems for educating health professionals about newborn screening.

We urge public and private entities to work together to strengthen newborn screening programs. This would include developing and validating methods to detect and treat disorders, as well as ensuring prompt followup and counseling for affected families.

Newborn screening is a rapidly changing field. We know that expert opinion on newborn screening will continue to evolve as medical evidence mounts, and we stand ready to update our recommendations to the states and to the public. Likewise, we urge that the ACMG report be periodically updated to accommodate new data and capabilities. In addition, there must be timely review of infectious diseases and other disorders in newborns for possible future inclusion in universal newborn screening, using the ACMG report as a model.

The March of Dimes will continue to work at both the state and federal levels for comprehensive programs and equity in newborn screening for every baby in this country. We urge all concerned individuals, groups, and health care providers to work even more closely together to achieve this common goal.

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## ◎家長應有知情信件的通知

美國健康與人類服務部（Department of Health and Human Services）所屬「母子健康局」（Maternal and Child Health Bureau）副首長 Peter C. van Dyck ，於 2004 年 7 月間正式去函各州健康部門與各州新生兒篩檢業務單位，指出在 2004 年 6 月 7 日所召開「新生兒與兒童遺傳異常與基因疾病顧問委員會」（Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children）會議中，家長與外界提出各州新生兒篩檢方案有必要提供家長，有關其新生兒是否接受各州新生兒篩檢項目以外其他新增項目的廣泛資訊。事實上在 1999 年，由美國小兒科醫學會所召集的工作小組所作的建議中，即已指出家長作為新生兒的代表人，應獲得有關新生兒篩檢的相關資訊，而各州或地區政府也應結合小孩有特殊健康需求的家長，來提供家長有關新生兒篩檢的教育資訊。因此 Peter C. van Dyck 鼓勵各州政府，除應提供教育資訊及該州新生兒篩檢項目以外，也應提供尚未納入各州的新增新生兒篩檢項目相關資訊。

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The following letter was recently sent from the U.S. Department of Health and Human Services Health Resources and Services Administration (HRSA) to the state health officials, maternal and child health directors, newborn screening programs and children with special health care needs directors.

Trish Mullaley, President of the National Coalition for PKU & Allied Disorders, reports that this letter was a direct result of parent testimony at the last HRSA meeting. Your message was loud and clear!

This letter will be a good tool when advocating in your state since the Maternal and Child Health Bureau (MCHB) has recommended to the state newborn programs that they should devise a way to give parents notification about expanded screening (even if the state doesn't offer it at this time) and the option of additional screening not included under the state program. It can be used to help generate expansion, whether through the state or birthing hospitals. Every pregnant mother needs to have access to this information.

Save Babies Through Screening Foundation would like to thank Trish Mullaley for sharing this letter with our list.

*****MATERNAL AND CHILD HEALTH BUREAU LETTER*****

Dear Colleague:

In the past few years, the possibilities for screening newborns have greatly expanded. And while all States may not be utilizing these new technologies, parents need to know about the full spectrum of options available for newborn screening. At the recent inaugural meeting of the Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children, held on June 7-8, 2004, public comments from parents and others indicated a need for State newborn screening programs to more comprehensively inform parents of the potential for their infants to receive additional newborn screening tests that may not be required under State law.

In 1999, the Newborn Screening Task Force convened by the American Academy of Pediatrics recommended that arents should receive information (on behalf of their children) about newborn screening??indicating ach state or region should, with input from families who have children with special health care needs and/or parent information centers, develop and provide family educational materials about newborn screening.?

This need for both educating and informing parents about newborn screening in general and which conditions are screened for in their State, specifically, has also been supported by the Health Resources and Services Administration Maternal and Child Health Bureau (MCHB) recent work with parents in several States. Parents want to know about newborn screening. I encourage you to facilitate the development of educational materials that are easy to read to inform parents of your State newborn screening program and for what conditions you screen in your program. Additionally, I also encourage you to facilitate the development of educational materials that inform parents about the option to have their babies screened for additional conditions that could be routinely tested for in the newborn period but at this time are not covered required by your State program.

If you have any questions, please contact Michele Puryear, M.D., Ph.D., Chief of MCHB Genetic Services Branch, at (301) 443-1080.

Sincerely yours,

Peter C. van Dyck, M.D., M.P.H.
Associate Administrator for Maternal and Child Health

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## 肆、美國各州新生兒篩檢現況

（資料來源：2005/1 National Newborn Screening & Genetic Resource Center Website； http://genes-r-us.uthscsa.edu/）

# Alabama

### Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| William Callan<br>jcallan@adph.state.al.us<br>334-260-3400 ext. 4432 | Belinda Thompson<br>BelThompson@adph.state.al.us<br>334-206-5955 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
58,900

**Major Racial/Ethnic Groups**
White: 67%　　American Indian: <1%
African American: 32%　　Asian/Pacific islander: 1%
Hispanic Ethnicity: 3% (may also be included in race categories above)

**Alabama Statute** (addresses testing, rule making, follow-up and fees)
Ala. Code § 22-20-3 (1991)
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all newborns, with second test requested on all infants at 2-6 weeks.
**NBS Fee:** $139.33

### Disorders

14 disorders mandated

Click here for list of disorders states screen for

# Alaska

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Cheryl Hermerath<br>cheryl.a.hermerath@state.or.us<br>503-229-6576 | Thalia Wood<br>thalia_wood@health.state.ak.us<br>907-269-3499 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
9,800

**Major Racial/Ethnic Groups**
White: 66% American Indian: 25%
African American: 4% Asian/Pacific islander: 5%

Hispanic Ethnicity: 6% ( may be included in race categories above)

**Alaska Statute** Information not available

**Screening Requirements**
Required by law on all newborns, with second test requested if first was done prior to 48 hours.

**NBS Fee:** $55

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Arizona

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Ethel Beraquit<br>beraque@azdhs.gov<br>602-542-1150 | Ruthann Smejkal<br>rsmejka@azdhs.gov<br>602-364-1409 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
87,400

**Major Racial/Ethnic Groups**
White: 88% American Indian: 7%
African American: 3% Asian/Pacific islander: 2%

Hispanic Ethnicity: 41% (may also be included in race categories above)

**Arizona Statute** Ariz. Rev. Stat. Ann. § 36-694 (1993) - addresses establishing NBS program and funding.
Ariz. Rev. Stat. Ann. § 20-2327 (2000) - coverage of medical foods
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all newborns, with second test required if first test done prior to 24 hours. Second test recommended on all infants.

**NBS Fee:** **$**20 for first screen, $15 for second

## Disorders

8 disorders mandated

Click here for a list of disorders states screen for

# Arkansas

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Mani Chidambaram<br>mchidambaram@healthyarkansas.com<br>501-661-2445 | Jackie Whitfield<br>jwhitfield@healthyarkansas.com<br>501-280-4756 |

**Click here for this programs newborn screening information**

**Approximate Births**
36,800

**Major Racial/Ethnic Groups**
White: 78% American Indian: 1%
African American: 20% Asian/Pacific islander: 1%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**Arkansas Statute**
Ark. Stat. Ann. § 20-15-302 (1995) - addresses testing.
Ark. Stat. Ann. § 23-79-129 (1995) - addresses insurance coverage for testing.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Testing required by law on all newborns.

**NBS Fee:** $14.83

## Disorders

**4 disorders mandated**

**Click here for a list of disorders states screen for**

# California

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| John Sherwin, Ph.D.<br>jsherwin@dhs.ca.gov<br>510-231-1728 | Fred Lorey, Ph.D.<br>florey@dhs.ca.gov<br>510-412-1490 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
529,500

**Major Racial/Ethnic Groups**
White: 81% American Indian: 1%
African American: 7% Asian/Pacific islander: 11%
Hispanic Ethnicity: 49% (may also be included in race categories above)

**California Statute**
Cal. Health & Safety Code § 125000 and 125001 (1998) - addresses testing, counseling, information and fees.
Cal. Health & Safety Code § 125000 and 125001 (1998) - addresses insurance coverage for testing.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirement**
Testing required by law on all newborns.

**NBS Fee:** $78.00

## Disorders

>30 disorders mandated (currently screening for 4 - additional disorders to be added by August 2005)

Click here for a list of disorders states screen for

# Colorado

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Jim Beebe<br>james.beebe@state.co.us<br>303-692-3488 | Laura Taylor<br>laura.taylor@state.co.us<br>303-692-2425 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
68,500

**Major Racial/Ethnic Groups**
White: 91% American Indian: 1%
African American: 5% Asian/Pacific islander: 3%

Hispanic Ethnicity: 26% (may also be included in race categories above)

**Colorado Statute**
Colo. Rev. Stat. §25-4-1001, et seq. - addresses testing and fees.
Section 25-4-1004.5(3) C.R.S. - addresses addition of CAH, and testing responsibilities.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Initial and second test required by law on all newborns.

**NBS Fee:** $53.25

## Disorders

7 disorders mandated

Click here for a list of disorders states screen for

# Connecticut

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Katherine Kelley<br>kati.kelley@po.state.ct.us<br>860-509-8513 | Dottie Trebisacci<br>dorothy.trebisacci@state.ct.us<br>860-509-8081 |

**Click here to go to the Connecticut Public Health Laboratory Website**

**Approximate Births**
42,600

**Major Racial/Ethnic Groups**
White: 84% American Indian: <1%
African American: 12% Asian/Pacific islander: 4%

Hispanic Ethnicity: 20% (may also be included in race categories above)

**Connecticut Statute**
Conn. Gen. Stat. § 19a-55 - addresses testing requirements.
Conn. Gen. Stat. § 19a-59a (1997) - addresses medical foods.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all newborns, with second test recommended on all newborns between 7-14 days of age.

**NBS Fee:** $28.00

## Disorders

12 disorders mandated plus Cystic Fibrosis universal pilot on all newborns

Click here for a list of disorders states screen for

# Delaware

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Jane Getchell<br>jane.getchell@state.de.us<br>302-653-2870 | Betsy Voss<br>bvoss@state.de.us<br>302-741-2990 |

Click here to go to the Delaware Public Health Laboratory Website

**Approximate Births**
11,300

**Major Racial/Ethnic Groups**
White: 74% American Indian: <1%
African American: 23% Asian/Pacific islander: 3%

Hispanic Ethnicity: 8% (may also be included in race categories above)

**Delaware Statute**
Information not available.

**Screening Requirements**
Initial and second test required by law on all newborns.

**NBS Fee:** $64

## Disorders

29 disorders mandated

Click here for a list of disorders states screen for

# District of Columbia

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Pediatrix<br>pediatrixscreeninginformation@pediatrix.com<br>866-463-6436 | Joyce Brooks<br>jbrooks@dchealth.com<br>202-727-7540 |

[Click here for the D.C. Maternal and Child Health Division Website]

**Approximate Births**
15,000

**Major Racial/Ethnic Groups**
White: 38% American Indian: <1%
African American: 59% Asian/Pacific islander: 3%

Hispanic Ethnicity: 9% (may also be included in race categories above)

**District of Columbia Statute**
Information not available.

**Screening Requirements**
Required by law on all newborns, with second test required if initial screen was done prior to 24 hours.

**NBS Fee:** None

## Disorders

7 disorders mandated

Click here for a list of disorders states screen for

# Florida

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Lisa Bates<br>lisa_bates@doh.state.fl.us<br>904-791-1641 | Lois Taylor<br>lois_taylor@doh.state.fl.us<br>8850-245-4201 |

Click here to go to the Florida Public Health Laboratory Services website

**Approximate Births**
205,500

**Major Racial/Ethnic Groups**
White: 74% American Indian: <1%
African American: 23% Asian/Pacific islander: 2%
Hispanic Ethnicity: 21% (may also be included in race categories above)

**Florida Statute**
Fla. Stat. Title 29 § 383.14 (1996) - addresses NBS fees.
For more information click on this link: National Conference of State Legislators

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hours.

**NBS Fee:** $15.00

## Disorders

5 disorders mandated and currently tested. Over 25 additional disorders mandated but not yet implemented.

Click here for a list of disorders states screen for

# Georgia

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Muthukrishnan Ramachandran<br>chandran@dhr.state.ga.us<br>404-327-6800 | Mary Ann Henson<br>mahenson@dhr.state.ga.us<br>404-657-6357 |

**Click here to go to the laboratory services website**

**Approximate Births**
134,600

**Major Racial/Ethnic Groups**
White: 64% American Indian: <1%
African American: 33% Asian/Pacific islander: 2%

Hispanic Ethnicity: 9% (may also be included in race categories above)

**Georgia Statute**
Ga. Code Ann. § 31-12-6 and 31-12-7 - addresses establishment of NBS system and disorders to be included. For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hours.

**NBS Fee:** None

## Disorders

10 disorders mandated

Click here for a list of disorders states screen for

# Hawaii

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Cheryl Hermerath<br>cheryl.a.hermerath@state.or.us<br>503-229-6576 | Christine A. Matsumoto<br>chris.matsumoto@fhsd.health.state.hi.us<br>808-733-9069 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
17,500

**Major Racial/Ethnic Groups**
White: 24% American Indian: 1%
African American: 3% Asian/Pacific islander: 73%

Hispanic Ethnicity: 13% (may also be included in race categories above)

**Hawaii Statute**
Hawaii Rev. Stat. § 321-291 (1999) - addresses testing and funding.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $47.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Idaho

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Cheryl Hermerath<br>cheryl.a.hermerath@state.or.us<br>503-229-6576 | Brett Harrell<br>harrell@idhw.state.id.us<br>208-334-5962 |

[Click here to go to the Idaho Department of Health website]

**Approximate Births**
20,400

**Major Racial/Ethnic Groups**
White: 97% American Indian: 1%
African American: <1% Asian/Pacific islander: 1%

Hispanic Ethnicity: 12% (may also be included in race categories above)

**Idaho Statute**
Information not available.

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hours.

**NBS Fee:** $23

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Illinois

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| David Jinks<br>djinksl@idph.state.il.us<br>312-793-1053 | Claudia Nash<br>cnash@idph.state.il.us<br>217-524-4900 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
177,600

**Major Racial/Ethnic Groups**
White: 77% American Indian: < 1%
African American: 19% Asian/Pacific islander: 4%

Hispanic Ethnicity: 21% (may also be included in race categories above)

**Illinois Statute**
Ill. HJR42 (2001) - addresses parental information
Ill. Rev. Stat. ch. 410, § 240/0.01, et seq - addresses the newborn screening system.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $47.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Indiana

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Barb Lesko<br>bglesko@iupui.edu<br>317-278-2502 | Kirstin J. Schwandt<br>kschwand@isdh.state.in.us<br>317-233-1300 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
85,500

**Major Racial/Ethnic Groups**
White: 88%  American Indian: < 1%
African American: 11%  Asian/Pacific islander: 1%

Hispanic Ethnicity: 6%  (may also be included in race categories above)

**Indiana Statute**
Ind. Code § 12-15-5-1 (2001) - addresses addition of tests and fee structure.
Ind. Code § 16-41-17-2 (1999) - addresses disorders to be screened for.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hrs.

**NBS Fee:** $62.50

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Iowa

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Stan Berberich<br>sberberich@uhl.uiowa.edu<br>515-243-0141 | Marcia Valbracht, MHA<br>mvalbrac@uhl.uiowa.edu<br>515-243-0141, ext. 2 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
37,800

**Major Racial/Ethnic Groups**
White: 94% American Indian: 1%
African American: 3% Asian/Pacific islander: 2%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**Iowa Statute**
Iowa Code § 136A, et seq. - addresses newborn screening and a disorder registry.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hours.

**NBS Fee:** $56.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Kansas

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Willie Craft<br>wcraft@kdhe.state.ks.us<br>785-296-1650 | Melanie Warren<br>mwarren@kdhe.state.ks.us<br>785-291-3363 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
39,700

**Major Racial/Ethnic Groups**
White: 89%  American Indian:  1%
African American: 8%  Asian/Pacific islander: 2%

Hispanic Ethnicity: 12%  (may also be included in race categories above)

**Kansas Statute**
Kan. Stat. Ann. § 65-180 (1997) - addresses coverage for medical foods.
Kan. Stat. Ann. § 65-180 (1994) - addresses addition of tests and parental education.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:**  None

## Disorders

4 disorders mandated

Click here for a list of disorders states screen for

# Kentucky

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Vera Foree<br>vera.foree@ky.gov<br>502-564-4446 | Sandy Fawbush<br>Sandy.Fawbush@ky.gov<br>502-564-3761 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
52,700

**Major Racial/Ethnic Groups**
White: 90% American Indian: <1%
African American: 9% Asian/Pacific islander: 1%

Hispanic Ethnicity: 2% (may also be included in race categories above)

**Kentucky Statute**
Ky. Rev. Stat. § 214.155 (2001)- addresses screening (including expanded testing) and fees.
Ky. Rev. Stat. §§ 205.560, 213.141, 304.17A (2000)- addresses follow-up and coverage for metabolic foods and other treatments.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hrs.

**NBS Fee:** $14.50

## Disorders

4 disorders mandated

Click here for a list of disorders states screen for

# Louisiana

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Art Hagar<br>ahagar@dhh.la.us<br>504-568-2554 | Charles Meyers<br>charlie@dhh.la.us<br>504-568-5070 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
65,100

**Major Racial/Ethnic Groups**
White: 58% American Indian: <1%
African American: 40% Asian/Pacific islander: 1%
Hispanic Ethnicity: 3% (may also be included in race categories above)

**Louisiana Statute**
La. Rev. Stat. Ann. § 40:1299.1 (1999) general screening mandate.
La. Acts, P.A. 997 (SB 708) (1993)- addresses galactosemia screening.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hrs.

**NBS Fee:** $18.00

## Disorders

5 disorders mandated and currently being screened for. 5 disorders

detectable by MS/MS are also being screened for in a pilot on all infants.

Click here for a list of disorders states screen for

# Maine

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Roger Eaton<br>roger.eaton@umassmed.edu<br>617-983-6300 | Jennifer Brown<br>jennifer.brown@maine.gov<br>207-287-5351 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
13,400

**Major Racial/Ethnic Groups**
White: 97% American Indian: 1%
African American: 1% Asian/Pacific islander: 1%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**Maine Statute**
Me. Rev. Stat. Ann. tit. 24-A, § 4238 (1995) - addresses metabolic formula and food.
Me. Rev. Stat. Ann. tit. 22, § 1533 (1983)- addresses genetic services.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hours.

**NBS Fee:** $47

## Disorders

9 disorders mandated; 18 disorders universal pilot

Click here for a list of disorders states screen for

# Maryland

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Linda Corcoran<br>corcoran@dhmh.state.md.us<br>410-767-6170 | Susan Panny<br>pannys@dhmh.state.md.us<br>410-767-6730 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
68,800

**Major Racial/Ethnic Groups**
White: 65% American Indian: < 1%
African American: 31% Asian/Pacific islander: 4%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**Maryland Statute**
Information not available

**Screening Requirements**
Voluntary program. Screening recommended on all infants, with a second test recommended if initial screen is done prior to 24 hours.

**NBS Fee:** $42.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Massachusetts

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Roger Eaton<br>roger.eaton@umassmed.edu<br>617-983-6300 | Cecelia McGonagle<br>cecelia.mcgonagle@state.ma.us<br>617-522-3700 ext.6348 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
81,700

**Major Racial/Ethnic Groups**
White: 85% American Indian: < 1%
African American: 10% Asian/Pacific islander: 5%
Hispanic Ethnicity: 11% (may also be included in race categories above)

**Massachusetts Statute**
Mass. Acts, Chap. 176 § 8B - addresses insurance coverage and medical formulas.
Mass. Acts, Chap. 111 § 10A - addresses screening requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $54.75

## Disorders

10 disorders mandated; 19 disorders universal pilot

Click here for a list of disorders states screen for

# Michigan

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Harry Hawkins<br>hawkinsh@michigan.gov<br>517-335-8095 | Bill Young<br>youngw@michigan.gov<br>517-335-8938 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
128,600

**Major Racial/Ethnic Groups**
White: 79% American Indian: 1%
African American: 18% Asian/Pacific islander: 3%
Hispanic Ethnicity: 11% (may also be included in race categories above)

**Michigan Statute**
Mich. Comp. Laws § 5431.1-6 (1999) - addresses screening requirements.
Mich. Comp. Laws § 5431.7-9 (1999) - addresses collection of additional blood sample for storage.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test recommended if initial screen is done prior to 24 hours.

**NBS Fee:** $55.72

## Disorders

11disorders mandated

Click here for a list of disorders states screen for

# Minnesota

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Mark McCann<br>mark.mccann@state.mn.us<br>612-676-5450 | Carolyn Anderson<br>carolyn.anderson@state.mn.us<br>651-281-9979 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
68,000

**Major Racial/Ethnic Groups**
White: 87% American Indian: 2%
African American: 6% Asian/Pacific islander: 5%

Hispanic Ethnicity: 10% (may also be included in race categories above)

**Minnesota Statute**
Minn. Stat. § 144.125, 144.126, 144.128 (1997) - addresses screening requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $61.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Mississippi

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Jerry McClure<br>jmcclure@msdh.state.ms.us<br>601-576-7619 | Jerry McClure<br>jmcclure@msdh.state.ms.us<br>601-576-7619 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
40,500

**Major Racial/Ethnic Groups**
White: 52% American Indian: 1%
African American: 47% Asian/Pacific islander: 1%

Hispanic Ethnicity: 1% (may also be included in race categories above)

**Mississippi Statute**
Miss. Code Ann. § 41-21-201, et seq. (2001) - addresses screening requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hours.

**NBS Fee:** $70.00

## Disorders

40 disorders mandated

Click here for a list of disorders states screen for

# Missouri

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| William Walden<br>waldew@dhss.state.mo.us<br>573-751-2662 | Julie Rayburn<br>raburj@dhss.state.mo.us<br>573-751-6266 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
76,400

**Major Racial/Ethnic Groups**
White: 83% American Indian: <1%
African American: 15% Asian/Pacific islander: 2%
Hispanic Ethnicity: 3% (may also be included in race categories above)

**Missouri Statute**
Mo. Rev. Stat. § 191.332 (2001) - addresses expanded screening.
Mo. Rev. Stat. § 191.331 (1997) - addresses screening program.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.
Second test recommended on all infants.

**NBS Fee:** $25.00

## Disorders

14 disorders mandated

Click here for a list of disorders states screen for

# Montana

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Susanne Norris Zanto<br>szanto@state.mt.us<br>406-444-2839 | Joan DeDycker<br>jdedycker@mt.gov<br>406-444-1216 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
11,000

**Major Racial/Ethnic Groups**
White: 87% American Indian: 12%
African American: <1% Asian/Pacific islander: 1%

Hispanic Ethnicity: 4% (may also be included in race categories above)

**Montana Statute**
Mont. Code Ann. §33-22-131 (1999) - addresses treatment for metabolic disorders.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee: $39.34** (PKU, GAL, CH - **$9.88** CF - **$10.00** MS/MS - **$2.00** BIO - **$4.00** CAH.)

## Disorders

4 disorders mandated; 22 select population, limited pilot or by request.

Click here for a list of disorders states screen for

# Nebraska

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Julie Miller<br>julie.miller@hhss.ne.gov<br>402-471-6733 | Julie Miller<br>julie.miller@hhss.ne.gov<br>402-471-6733 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
25,500

**Major Racial/Ethnic Groups**
White: 91% American Indian: 2%
African American: 5% Asian/Pacific islander: 2%

Hispanic Ethnicity: 11% (may also be included in race categories above)

**Nebraska Statute**
Neb.Rev.Stat.secs 71-519 to 71-524 - addresses requirements for newborn screening program.

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $30.75

## Disorders

6 disorders mandated; 27 select populations, limited pilot or by request.

Click here for a list of disorders states screen for

# Nevada

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Cheryl Hermerath<br>cheryl.a.hermerath@state.or.us<br>503-229-6576 | Gloria Deyhle<br>gdeyhle@nvhd.state.nv.us<br>775-684-4285 |

**Click here to go to the Nevada State Health Division website**

**Approximate Births**
32,200

**Major Racial/Ethnic Groups**
White: 85% American Indian: 1%
African American: 8% Asian/Pacific islander: 6%

Hispanic Ethnicity: 33% (may also be included in race categories above)

**Nevada Statute**
Nev. Rev. Stat. § 442.118 (1989) - addresses sickle cell screening.

**Screening Requirements**
Initial and second screen required by law on all infants.

**NBS Fee:** $64.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# New Hampshire

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Roger Eaton<br>roger.eaton@umassmed.edu<br>617-983-6300 | Marcia Lavochkin<br>MLavochkin@dhhs.state.nh.us<br>603-271-4225 |

**Approximate Births**
13,900

**Major Racial/Ethnic Groups**
White: 97% American Indian: < 1%
African American: 1% Asian/Pacific islander: 2%
Hispanic Ethnicity: 7% (may also be included in race categories above)

**New Hampshire Statute**
N.H. Rev. Stat. Ann. § 132:10 (1999) - addresses newborn screening test profile.
N.H. Rev. Stat. Ann. § 417-D:2-a (1996) - addresses health coverage for nbs collection.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.
Very low birth weight infants ( <1500 gms) retested at 2 weeks of age.

**NBS Fee:** $18.00

## Disorders

6 disorders mandated; Sickle cell disease - selected population, or by request.

Click here for a list of disorders states screen for

# New Jersey

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Alan Bergum<br>abergum@doh.state.nj.us<br>609-292-4811 | Mary Mickles<br>mary.mickles@doh.state.nj.us<br>609-292-1582 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
111,800

**Major Racial/Ethnic Groups**
White: 74% American Indian: < 1%
African American: 19% Asian/Pacific islander: 7%
Hispanic Ethnicity: 19% (may also be included in race categories above)

**New Jersey Statute**
N.J. Executive Order 126 (2001) - addresses expanding newborn screening.
N.J. Rev. Stat. §§ 17:48-6s, 17:48A-7q, 17:48E-35.16 (1997) - addresses health coverage for metabolic formula and foods.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hours.

**NBS Fee:** $71.00

## Disorders

20 disorders mandated; 6 limited pilot

Click here for a list of disorders states screen for

# New Mexico

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Larry Chavez<br>larry.chavez@state.nm.us<br>505-841-2581 | Carla Ortiz<br>carlaa.ortiz@doh.state.nm.us<br>505-476-8858 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
27,300

**Major Racial/Ethnic Groups**
White: 83% American Indian: 14%
African American: 2% Asian/Pacific islander: 1%

Hispanic Ethnicity: 25% (may also be included in race categories above)

**New Mexico Statute**
N.M. Stat. Ann. 7 § 30.6.2, et seq. (1996) - addresses newborn screening program.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Initial and second screen required by law on all infants.

**NBS Fee:** $32.00

## Disorders

6 disorders mandated.

Click here for a list of disorders states screen for

# New York

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Kenneth A. Pass<br>kap03@health.state.ny.us<br>518-473-1993 | Deborah Rodriguez<br>dar05@health.state.ny.us<br>518-486-4949 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
252,300

**Major Racial/Ethnic Groups**
White: 72% American Indian: <1%
African American: 21% Asian/Pacific islander: 7%
Hispanic Ethnicity: 25% (may also be included in race categories above)

**New York Statute**
N.Y. Public Health Law § 2500-a - addresses newborn screening program.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required within 3-5 days if initial screen is done prior to 24 hours.

**NBS Fee:** None

## Disorders

11 disorders mandated and currently being screened for. Greater than 30 disorders detectable by MS/MS are also mandated and are being screened for as a pilot on all infants.

Click here for a list of disorders states screen for

# North Carolina

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Shu Huey Chaing<br>shu.chaing@ncmail.net<br>919-733-3937 | Lara Percenti<br>lara.percenti@ncmail.net<br>919-715-3418 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
118,200

**Major Racial/Ethnic Groups**
White: 72% American Indian: 1%
African American: 25% Asian/Pacific islander: 2%

Hispanic Ethnicity: 9% (may also be included in race categories above)

**North Carolina Statute**
Information not available

**Screening Requirements**
Required by law on all infants, with a second test required within 48-72 hours if initial screen is done prior to 24 hours.

**NBS Fee:** $10

## Disorders

26 disorders mandated

Click here for a list of disorders states screen for

# North Dakota

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Stan Berberich<br>sberberi@uhl.uiowa.edu<br>515-243-0141 | Barbara Scweitzer<br>bschweit@state.nd.us<br>701-328-4538 |

Click here to go to the Maternal and Child Health Division website

**Approximate Births**
8,900

**Major Racial/Ethnic Groups**
White: 88% American Indian: 9%
African American: 1% Asian/Pacific islander: 1%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**North Dakota Statute**
N.D. Cent. Code § 23-01-03.1 - addresses use of nbs specimens for research.
For more information click on this link: National Conference of State Legislators

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $36.00

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Ohio

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| William J. Becker<br>bbecker@odh.ohio.gov<br>614-644-4590 | Ram Chandrasekar<br>rchandra@odh.ohio.gov<br>614-466-5600 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
149,000

**Major Racial/Ethnic Groups**
White: 84% American Indian: <1%
African American: 14% Asian/Pacific islander: 2%

Hispanic Ethnicity: 3% (may also be included in race categories above)

**Ohio Statute**
Ohio Rev. Code Ann. §§ 3701.501 (1996) - addresses newborn screening program requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hrs.

**NBS Fee:** $45.16

## Disorders

29 disorders mandated

Click here for a list of disorders states screen for

# Oklahoma

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Debbie Kline<br>DebbieK@health.ok.gov<br>405-271-5070 | Pam King<br>pamk@health.state.ok.us<br>405-271-9444 X56737 |

Click here to go to the Oklahoma Department of Health website

**Approximate Births**
49,200

**Major Racial/Ethnic Groups**
White: 79% American Indian: 10%
African American: 10% Asian/Pacific islander: 2%

Hispanic Ethnicity: 10% (may also be included in race categories above)

**Oklahoma Statute**
Okla. Stat. § 63-1-533) - addresses newborn screening education.
For more information click on this link: National Conference of State Legislators

**Screening Requirements**
Required by law on all infants, with a second test required within 3-5 days if initial screen is done prior to 24 hours.

**NBS Fee:** $10.50

## Disorders

7 disorders mandated

Click here for a list of disorders states screen for

# Oregon

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Cheryl Hermerath<br>cheryl.a.hermerath@state.or.us<br>503-229-6576 | Leanne Rien, RN<br>leanne.c.rien@state.or.us<br>503-229-5466 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
46,100

**Major Racial/Ethnic Groups**
White: 92% American Indian: 2%
African American: 2% Asian/Pacific islander: 5%

Hispanic Ethnicity: 16% (may also be included in race categories above)

**Oregon Statute**
Or. Rev. Stat. § 431.310 (1993) - addresses newborn screening fees.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Initial and second screen required by law on all infants.

**NBS Fee:** $54.00

## Disorders

26 disorders mandated; 8 disorders universal pilot

Click here for a list of disorders states screen for

# Pennsylvania

## Newborn Screening Contact Information

| | |
|---|---|
| **NBS Laboratory**<br>Barbara Kendro<br>bkendro@state.pa.us<br>610-280-3464 | **Follow-up Program**<br>Karen Espenshade<br>kaespensha@state.pa.us<br>717-783-8143 |

[Click here to go to the Bureau of Laboratories website]

**Approximate Births**
142,950

**Major Racial/Ethnic Groups**
White: 83% American Indian: <1%
African American: 14% Asian/Pacific islander: 2%

Hispanic Ethnicity: 5% (may also be included in race categories above)

**Pennsylvania Statute**
Information not available.

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 36 hrs.

**NBS Fee:** None

## Disorders

6 disorders mandated; >30 disorders select population, limited pilot or by request.

Click here for a list of disorders states screen for

# Rhode Island

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Roger Eaton<br>roger.eaton@umassmed.edu<br>617-983-6300 | Ellen Amore<br>ellena@doh.state.ri.us<br>401-222-4601 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
13,550

**Major Racial/Ethnic Groups**
White: 88%
African American: 8%
American Indian: 1%
Asian/Pacific islander: 3%

Hispanic Ethnicity: 29% (may also be included in race categories above)

**Rhode Island Statute**
R.I. Gen. Laws § 23-13-14 (1995) - addresses newborn screening requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $59.00

## Disorders

9 disorders mandated

Click here for a list of disorders states screen for

# South Carolina

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Tom Hickey<br>hickeytm@dhec.sc.gov<br>803-896-0963 | Kathy Tomashitis<br>tomashk@dhec.sc.gov<br>803-898-0619 _ |

[Click here for the Bureau of Maternal and Child Health website]

**Approximate Births**
52,200

**Major Racial/Ethnic Groups**
White: 63% American Indian: <1%
African American: 35% Asian/Pacific islander: 1%

Hispanic Ethnicity: 3% (may also be included in race categories above)

**South Carolina Statute**
S.C. Code Ann. § 44-37-30 (1994) - addresses newborn screening program.
For more information click on this link: National Conference of State Legislators

**Screening Requirements**
Required by law on all infants, with a second test recommended if initial screen is done prior to 24 hours.

**NBS Fee:** $42.00

## Disorders

30 disorders mandated

Click here for a list of disorders states screen for

# South Dakota

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Lucy Fossen<br>lucy.fossen@state.sd.us<br>605-773-2944 | Lucy Fossen<br>lucy.fossen@state.sd.us<br>605-773-2944 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
11,000

**Major Racial/Ethnic Groups**
White: 84% American Indian: 14%
African American: 1% Asian/Pacific islander: 1%
Hispanic Ethnicity: 2% (may also be included in race categories above)

**South Dakota Statute**
S.D. Code Ann. §§ 34-24-19, et seq. - addresses newborn screening program requirements.
S.D. Code Ann. §§ 58-41-98, 58-40-21, 58-38-23, 58-18-41 and 58-17-62 - address health insurance coverage.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test recommended if initial screen is done prior to 24 hours.

**NBS Fee:** $18.53

## Disorders

3 disorders mandated; 30 disorders select populations, limited pilot or by request.

Click here for a list of disorders states screen for

# Tennessee

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Jim Gibson<br>jgibson@mail.state.tn.us<br>615-262-6303 | Mitzi Lamberth<br>mitzi.lamberth@state.tn.us<br>615-262-6304 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
82,600

**Major Racial/Ethnic Groups**
White: 77% American Indian: <1%
African American: 21% Asian/Pacific islander: 1%

Hispanic Ethnicity: 3% (may also be included in race categories above)

**Tennessee Statute**
Tenn. Code Ann. § 68-5-401 (1997) - addresses newborn screening program requirements.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hours.

**NBS Fee:** $47.50

## Disorders

>30 disorders mandated

Click here for a list of disorders states screen for

# Texas

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Eldridge Hutcheson<br>Eldridge.Hutcheson@dshs.state.tx.us<br>512-458-7430 | Margaret Drummond-Borg<br>margaret.borg@dshs.state.tx.us<br>512-458-7700 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
374,100

**Major Racial/Ethnic Groups**
White: 85% American Indian: <1%
African American: 11% Asian/Pacific islander: 3%

Hispanic Ethnicity: 46% (may also be included in race categories above)

**Texas Statute**
Tex. Code Ann. Health & Safety Code §§ 33.011, et seq. (1991) - addresses newborn screening requirements. For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Initial and second screen required by law on all infants.

**NBS Fee:** $19.50 for each mandated specimen.

## Disorders

5 disorders mandated

Click here for a list of disorders states screen for

# Utah

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Barbara Jepson<br>bjepson@utah.gov<br>801-584-8400 | Fay A. Keune<br>fkeune@utah.gov<br>801-584-8256 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
50,300

**Major Racial/Ethnic Groups**
White: 95% American Indian: 1%
African American: 1% Asian/Pacific islander: 3%

Hispanic Ethnicity: 12% (may also be included in race categories above)

**Utah Statute**
Utah Code Ann. § 26-10-6 (1998)T - addresses newborn screening program and fees. For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Initial and second screen required by law on all infants.

**NBS Fee:** $32.00

## Disorders

4 disorders mandated; 25 additional disorders offered as pilot project on request

Click here for a list of disorders states screen for

# Vermont

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Roger Eaton<br>roger.eaton@umassmed.edu<br>617-983-6300 | Cindy Ingham<br>cingham@vdh.state.vt.us<br>802-951-5180 |

**Click here for the Vermont Department of Health website**

**Approximate Births**
6,100

**Major Racial/Ethnic Groups**
White: 99% American Indian: <1%
African American: 1% Asian/Pacific islander: <1%

Hispanic Ethnicity: 2% (may also be included in race categories above)

**Vermont Statute**
Vt. Stat. Ann. tit. 8, § 4089e - addresses medical foods.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $33.30

## Disorders

21 disorders mandated

Click here for a list of disorders states screen for

# Virginia

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Charlie Stevenson<br>cstevenson@dgs.state.va.us<br>804-648-4480 | Sharon Williams<br>sharonk.williams@vdh.virginia.gov<br>804-864-7712 |

**Click here to go to the Virginia Department of Health Genetics website**

**Approximate Births**
97,400

**Major Racial/Ethnic Groups**
White: 71% American Indian: <1%
African American: 24% Asian/Pacific islander: 5%

Hispanic Ethnicity: 7% (may also be included in race categories above)

**Virginia Statute**
Va. Code § 32.1-65 (2001) - addresses newborn screening testing requirements.
Va. Code § 32.1-65 (2001) - addresses additional testing for confirmation.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 24 hrs.

**NBS Fee:** $32.00

## Disorders

9 disorders mandated

Click here for a list of disorders states screen for

# Washington

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Mike Glass<br>mike.glass@doh.wa.gov<br>206-361-4996 | Sheila Neier<br>sheila.neier@doh.wa.gov<br>206-418-5509 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
78,600

**Major Racial/Ethnic Groups**
White: 86% American Indian: 2%
African American: 4% Asian/Pacific islander: 8%

Hispanic Ethnicity: 18% (may also be included in race categories above)

**Washington Statute**
Wash. Rev. Code § 70.83.040 (1999) - addresses fee for addition services.
Wash. Rev. Code § 70.83.020 (1991) - addresses requirement of newborn screening.
For more information click on this link: **National Conference of State Legislators**

**Screening Requirements**
Required by law on all infants, with a second test recommended between 7 and 14 days.

**NBS Fee:** $64.40

## Disorders

9 disorders mandated

Click here for a list of disorders states screen for

# West Virginia

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Barbara Eckerd<br>barbaraeckerd@wvdhhr.org<br>304-558-3530 | Tara Morris<br>taramorris@wvdhhr.org<br>304-558-5388 |

[Click here to go to the West Virginia genetics website]

**Approximate Births**
21,100

**Major Racial/Ethnic Groups**
White: 96% American Indian: 0%
African American: 4% Asian/Pacific islander: 1%

Hispanic Ethnicity: 1% (may also be included in race categories above)

**West Virginia Statute**
Information not available.

**Screening Requirements**
Required by law on all infants, with a second test required if initial screen is done prior to 48 hours.

**NBS Fee:** 15.85

## Disorders

4 disorders mandated

Click here for a list of disorders states screen for

# Wisconsin

## Newborn Screening Contact Information

| NBS Laboratory | Follow-up Program |
|---|---|
| Gary Hoffman<br>hoffman@mail.slh.wisc.edu<br>608-262-4692 | Karen Kennedy-Parker<br>karen@mail.slh.wisc.edu<br>608-262-5817 |

**Click here to go to this programs newborn screening website**

**Approximate Births**
67,400

**Major Racial/Ethnic Groups**
White: 86% American Indian: 1%
African American: 10% Asian/Pacific islander: 3%

Hispanic Ethnicity: 6% (may also be included in race categories above)

**Wisconsin Statute**
Wis. Stat. § 253.13(1) - addresses newborn screening requirement.
**For more information click on this link:** National Conference of State Legislators

**Screening Requirements**
Required by law on all infants, with a second test recommended if initial screen is done prior to 24 hours.

**NBS Fee:** $65.50

## Disorders

26 disorders mandated

Click here for a list of disorders states screen for

# Wyoming

## Newborn Screening Contact Information

| | |
|---|---|
| **NBS Laboratory**<br>Larry Sater<br>303-692-3672<br>larry.sater@state.co.us | **Follow-up Program**<br>Dorothy Ailes<br>307-777-7166<br>dailes@state.wy.us |

[Click here to go to the Wyoming Department of Health website]

**Approximate Births**
5,800

**Major Racial/Ethnic Groups**
White: 93% American Indian: 5%
African American: 1% Asian/Pacific islander: 1%

Hispanic Ethnicity: 9% (may also be included in race categories above)

**Wyoming Statute**
Wyo. Stat. § 35-4-801 - addresses newborn screening requirements.
**For more information click on this link:** National Conference of State Legislators

**Screening Requirements**
Required by law on all infants.

**NBS Fee:** $45.00

## Disorders

7 Disorders mandated

Click here for a list of disorders states screen for

# 伍、主要國家新生兒篩檢現況

## Australia

在 2002 年以前，澳洲各省的新生兒篩檢項目存在差異，但至少均篩檢了 Phenylketonuria, Primary Hypothyroidism (CH), Galactosaemia, Cystic Fibrosis (CF)等四項疾病，而當時除了昆士蘭省與西澳省以外，各州均已購置串聯質譜儀進行 20 項新增的擴大新生兒篩檢。但昆士蘭省隨後亦於 2002 年中起購置串聯質譜儀，而最後的西澳省亦於 2004 年起購置串聯質譜儀進行擴大新生兒篩檢；目前澳洲各省均已購置串聯質譜儀進行擴大新生兒篩檢，而成為全世界第一個全面進行擴大新生兒篩檢的國家。該項新生兒篩檢為免費，且非強制性，家長得拒絕其新生兒接受篩檢。

目前澳洲的新生兒篩檢分由五個實驗室進行檢驗：Western Australia; South Australia; Victoria; New South Wales and Queensland。而相關的篩檢政策則是由「澳洲人類基因學會」（Human Genetics Society of Australasia）及「澳洲皇家醫學會小兒科小組」（Division of Paediatrics of the Royal Australasian College of Physicians）的聯合會議決定。

澳洲新聞網站（www.news.com.au）於 2004 年 12 月 17 日指出，

感謝篩檢技術的創新，澳洲西部的新生兒將於出生時接受篩檢可能致命的罕見疾病。澳洲健康部門的代理主任 Neale Fong 指出，澳洲西部的醫院已經於本月引進串聯質譜儀，這是近 35 年來的最大創新；這項檢驗只要於新生兒出生兩天於腳跟扎取幾滴血液，即可檢驗澳洲 35 年來傳統所篩檢的苯酮尿症等疾病(phenylketonuria、congenital hypothyroidism、galactosaemia、cystic fibrosis)，而串聯質譜儀使用相同的方法，卻可擴大更多項新生兒篩檢的檢查。

可能導致發展遲緩、昏迷、中風、器官衰竭和死亡的氨機酸、有機酸、和脂肪酸的代謝異常疾病，均可透過串聯質譜儀加以驗出；Neale Fong 指出雖然每一項疾病的人數都很少，但是串聯質譜儀所能偵測出的許多項疾病，將使得澳洲西部每千位新生兒即可篩檢出一位病童，使得澳洲成為全世界少數使用最新科技進行新生兒篩檢的國家之一。

澳洲的新生兒篩檢通常於出生後 48 至 72 小時採取血樣，但篩檢的項目則因各省而有不同。而澳洲的維多利亞省、新南威爾斯省、和南澳省則都是於出生第二天取樣，並透過串聯質譜儀加以分析。維多利亞省也設有「基因支持網絡團體」(Genetic Support Network of Victoria; GSNV)，用來幫助基因異常的病患能夠獲得適當與正確的資訊，同時支持她們能夠因應未來在健康和福利上的挑戰，而該支持網

絡也用來整合相關的資訊、資源與協助。以維多利亞省為例，PKU 自 1971 年開始篩檢、congenital hypothyroidism 自 1977 年起、cystic fibrosis 從 1989 年起、而使用串聯質譜儀篩檢代謝疾病則自 2001 年開始，目前維多利亞省即已有數百名因篩檢而維持健康的兒童。為確保篩檢試紙的安全，自 2003 年起這些 Guthrie cards 已重置於安全且上鎖的設備中，並由健康服務部檢查與核准，同時需無限期加以保存。

## Canada

加拿大的每一位新生兒都必須接受新生兒篩檢，檢查項目至少包括 hypothyroidism 和 phenylketonuria，家長在出生前都會被知會有關這項篩檢的目的，這項做法是依據立法的執行，而篩檢的結果也都會由醫療提供者適當的向家長說明，以確保能及時的照護這些特殊的新生兒以及支持這些家庭。篩檢都在出院前執行，或確保能在出院後的有效時間內進行。

卑詩省則檢驗了四種疾病，包括 Phenylketonuria (PKU) 、Congenital Hypothyroidism (CH)、Galactosaemia (GS)、和 Medium

Chain Acyl-CoA Dehydrogenase Deficiency (MCAD) 等四種，每年約有五萬新生兒接受篩檢。

自 2000 年五月底開始，亞伯特（Alberta）省的新生兒篩檢登錄系統則於十七個區域健康單位開始運作，這項登錄系統能夠更有效的追蹤新生兒的狀況；一般新生兒於出生時即給予一項個人的健康編號（Personal Health Number），篩檢都在出院前執行，但對於提前出院者，則由公共衛生護士或助產士於事後採樣，並由省新生兒篩檢實驗室負責追蹤，以確保每一位新生兒都能得到代謝性疾病的篩檢。

## Denmark

丹麥傳統的新生兒篩檢包含了 phenylke-tonuri (PKU)、congenital hypothyreosis 、 congenital toxoplasmosis 等，但自 2002 年二月一日起，則開始實驗性的運用串聯質譜儀篩檢代謝性的疾病。

## Finland

雖然多數已開發國家都已提供新生兒篩檢，以便對可治療的先天代謝疾病施以及早治療，以避免身心障礙甚至死亡，但芬蘭則僅限於針對 congenital hypothyroidism 進行篩檢，同時由於其種族的特

性，因此對於 phenylketonuria（PKU）亦無需加以篩檢。但由於移民的增加使族群日趨複雜，因此使用串聯質譜儀篩檢代謝性的疾病的討論則已日漸增加。

## Germany

根據德國各醫學會於 2002 年所聯合發表有關德國代謝與內分泌新生兒篩檢指引的內容指出，新生兒的篩檢除了傳統的 Congenital Hypothyreosis（CH）、Congenital Adrenal Hyerplasia（CAH）、Deficiency of Biotinidase、Classical form of Galacosemia（Deficiency of GALT）等四項以外，同時建議串聯質譜儀所能篩檢的其他 11 項疾病亦列為建議名單之中。

Nennstiel-Ratzel U, Liebl B, Zapf A. 等人於 2003 年 3 月第 65 期 Gesundheitswesen 刊物中指出，為建立符合德國國家標準的新生兒篩檢模式，其於 1999 至 2001 年於德國計使用串聯質譜儀篩檢了 35 萬新生兒，篩檢比率由之前的 80% 提高到 98% 以上，並發現 217 位代謝與內分泌異常的新生兒，同時也發現醫事人員與家長都展現高度的支持度。

而在臨床上，以 Giessen 大學給與家長有關新生兒篩檢的說明資料中，則亦將如下疾病列為使用串聯質譜進行新生兒篩檢的項目：

MSUD、Defects of the Carnine Metabolism、Fatty-Acid-Oxidation Disorder Glutaric Acidemia Type I and Type II、Defects of Urea Cycle、Defects of Holocarboxylase-Synthetase、Isovaleric Acidemia、3-Methylcrotonyl-CoA deficiency、Methylmalonic acidemia、PKU、Proprionic acidemia、Tyrosinemia type I、Cystic fibrosis、G6PDH

## Ireland

愛爾蘭共和國自 1966 年起開始進行 PKU 的新生兒篩檢，當時也是世界上第一個這類型的全國性篩檢制度。1971 年起加入 Homocystinuria、1972 年起加入 Classical Galactosemia 和 MSUD、1979 年加入 Congenital Hypothyroidism 。1990 年起健康部長任命一個新陳代謝工作小組（Metabolic Disorders Working Group）來評估新生兒篩檢制度，並於 1993 年提出評估報告，建議應有責任確保每一位新生兒都能獲得代謝異常的檢查、建立完成上述目的之操作工具與方法、以及建立團隊合作的模式等三項建議。依據該新陳代謝工作小組對於樣本收集的操作與流程建議，並於 1999 和 2001 年出版「愛爾蘭新生兒操作守冊」(A Practical Guide to Newborn Screening in Ireland)。2004 年起在健康與兒童部充足經費的支持

下，已成立一個 Congenital Toxoplasmosis 的試驗篩檢小組，同時 Cystic fibrosis 的工作小組也已經成立，準備評估未來納入新生兒篩檢作準備。

## Japan

日本的新生兒篩檢（Newborn Mass Screening Program; NMSP）係由健康部（Department of Health ）所創立，用於早期診斷可治療的疾病並提供照護，目前篩檢的疾病包括 phenylketonuria (PKU)、maple syrup urine disease(MSUD)、 homocystinuria (HCU), galactosemia (GE)、hypothyroidism、21-hydroxyase deficiency 等。自 1970 年起，新生兒篩檢在日本兒童健康照護體系中扮演重要的角色，幾乎所有的新生兒均已接受過新生兒的篩檢，而成為日本重要的健康方案。

## Philippine

菲律賓的新生兒篩檢開始於 1996 年由 Dr. Carmelita Domingo 和 Dr. Carmencita Padilla 所主持的研究計畫，當時只有馬尼拉都會區內的 24 家醫院參與，如今已擴展為全國各地區超過 200 個機構和社區參與的計畫，包括醫院、

鄉村醫療單位、健康中心、診所和生產中心。該計畫提高部分先天疾病於病發前早期確診的機會，目前篩檢的項目有: Congenital Hypothyroidism (CH), Congenital Adrenal Hyperplasia (CAH), Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency, Galactosemia (Gal) and Phenylketonuria (PKU).

## United Kingdom

英國的新生兒篩檢最早開始於 1969 年起篩檢 PKU，1981 年加入 Congenital Hypothyroidism (CHT)，目前新生兒篩檢是英國規模最大的篩檢項目之一，每年平均約篩檢 60 萬個新生兒，同時每年也約發現約 250 位患有 PKU 和 Congenital Hypothyroidism 的病童，得以在未發病前進行有效的治療；每年新生兒中也有超過 99% 接受過新生兒篩檢。2004 年起使用新生兒篩檢同樣的血片，也已陸續階段性的進行 Sickle Cell Disorders 和 Cystic Fibrosis 的篩檢。除此之外，在英格蘭的部分地區以及 UK 其他的國家中也針對不同的情況進行篩檢，例如威爾斯 (Wales) 針對裘馨氏肌肉萎縮症、或是不定期的試驗篩檢等，未來預期將進行擴大新生兒篩檢。2004 年起英格蘭也試驗性的進行 Medium-chain acyl-CoA dehydrogenase

deficiency（MCAD）的篩檢，並將於2007年進行評估。

UK並於2002年成立「新生兒篩檢規劃中心」(UK Newborn Screening Programme Centre; UKNSPC)，其目標在爲新生兒篩檢發展品質確保的方案與管理架構，以提供新生兒與家長高品質的篩檢服務。UK的四個國家分別有自己的健康部門、人民也有不同的健康需求與期待，因此也會使得執行的進度與方式產生差異，因此「新生兒篩檢規劃中心」將透過四個國家間的聯盟策略，以確保新生兒不因出生何地均能獲得一致的高品質篩檢服務。