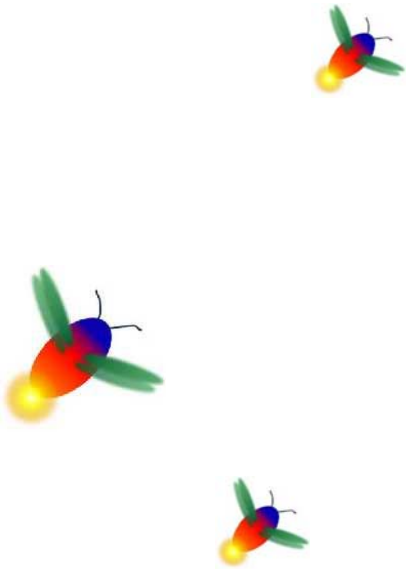
美國罕病組織(NORD)運作模式探索



委託單位：財團法人罕見疾病基金會
贊助單位：Genzyme corporation-Taiwan
計畫名稱： 美國罕病組織(NORD)運作模式探索
計畫主持人：唐先梅副教授
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目錄

美國罕病組織(NORD)運作模式探索

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。

除了議題倡導的進展外，罕見疾病基金會的組織以及募款能量也在不斷的提升中，例如其人員規模已成長至 23 位專職人力，同時其年度募款經費也由 2000 年的 1717 萬元成長至 2003 年的 3163 萬元，其中約有 15%來自政府補助、另 15%來自企業捐助，以及 70%來自社會大眾的小額捐款。因此綜合而言，從議題發展、人員成長、以及經費勸募觀察，充分展現罕見疾病基金會已在國內穩健立足，因而有必要未雨綢繆，開始思索國際化未來在罕見疾病基金會發展中的定位，而其中尤為關鍵的問題即是現有罕病相關國際組織中，可資作為罕病基金會借鏡的典範組織為何？若能深入了解該典範組織的運作

模式與發展軌跡，則將減少罕病基金會未來組織發展的摸索期程，同時也能強化罕病基金會與該國際典範組織的合作交流，讓本土的運作經驗因而能與國際接軌，並達到未來人員、議題、方案、與組織管理經驗交流的目標。

在現有罕病相關的國際組織中，美國罕病組織（National Organization for Rare Disorders）無疑是重要的國際典範，也是最足以作為罕病基金會學習參仿的對象，因而有必要深入探索該組織的運作模式與發展軌跡，以作為罕病基金會未來進行國際化的準備基礎。美國罕病組織與罕病基金會具有許多類似的特性，例如（1）、均由病患家長推動設立：美國罕病組織於 1983 年由病患家長 Abbey Meyers 推動設立，當時她有三位子女均被確診為先天遺傳疾病，並面臨孤兒藥取得的障礙;而我國罕病基金會亦由病患家長陳莉茵與曾敏傑於 1999 年經向社會大眾募款後成立。（2）、均著重跨病類的整合協助：美國罕病組織為跨病類的聯合組織，其所屬團體會員涉及數百種單一罕病組織；我國罕病基金會的屬性為財團法人，而非病患組織之聯合團體，但兩者的運作主軸均著重在跨病類的議題之上。（3）、組織運作初期均強調社會立法：美國罕病組織於 1983 成立，並於該年推動美國孤兒藥法案成功立法；我國罕病基金會於 1999 年成立，並於 2000 年成功推動「罕見疾病防治及藥物法」立法通過，將政府角色與罕病

的關聯與以確立。

罕病基金會自成立以來即已加入美國罕病組織，並取得副團體會員的身分（Associate Member），也開始接觸國外罕病跨病類組織的運作，惟由於罕病基金會成立初期致力於國內罕病醫療環境的改造以及推動社會立法，因此並未積極參與罕病國際社群的互動；如今罕病基金會經六年來的密集運作，已有重要的成果產生，因此未來自然宜強化與國際社群的交流，一方面觀摩國外團體的運作，同時也可將本土的運作經驗與國外團體產生交流，並共同推動未來國際罕病社群的集體合作。

貳、美國罕病組織（NORD）的運作內容

一、宗旨

美國罕病組織為符合聯邦財政部國稅局（Internal Revenue Service, Department of Treasurer）501(C)3 規定的非營利組織，為協助罹患罕見疾病的個人與相關團體的聯盟性、志願性健康組織，美國罕病組織因此透過教育、倡導、研究、及服務性的方案，來致力於確診、治療、與矯正罹患罕見疾病的病人。

根據美國孤兒藥法案相關的定義，所謂「罕見」疾病或「孤兒」疾病係指病患人數在二十萬以下者，估計符合這項定義的疾病應超過六千項之多，因此若綜合計算，預計將有 2500 萬的美國人口受到影響，也就是美國將有十分之一的人口為罕見疾病的患者。

二、年會舉辦

NORD 每年亦舉辦年會，並以研討會、工作坊、展覽等多元化的方式為內容，2005 年的年會於美國華盛頓特區於 9 月 30 日至 10 月 2 日舉行，主題聚焦在「給付」(Reimbursement）之上，希望幫助病患及其家屬能夠獲得醫療與給付，也協助病患組織的負責人得以幫助會員了解相關問題。這項研討會也受到美國國家健康研究院(National

Institutes of Health）所屬「罕見疾病辦公室」(Office of Rare Diseases）的贊助，演講的成員來自包括政府、病患組織及產學界。

NORD 2005 年的年會議程如下：

9/30/05	**Bioethics and Rare Disease Research** Doris Zallen	**general**
	Financing of Clinical Trials Bill Aliski TKT Alan Rosenberg Scott Janis	**general** Company perspective Wellpoint Insurance NIH
	Private Insurance: Affect of Reimbursement on the Rare Disease Community Michael Russo Laureen Kelley	**general** The Bruckner Group LA Kelley Communications
	What is a Tissue/Blood/Brain Bank? Why Go This Route? John Lonsdale Jayne Gershkowitz Henrietta Hyatt-Knorr	**general** NDRI Advocacy Group NIH
	Importance of Medical Privacy **Medical Privacy Project at Georgetown**	**general**

	Univ Emily Stewart	
	Report on Model System for Genetic Testing for Rare Diseases Andrew Faucett	**general**
10/1/05	**Reasons Why Medicaid and Medicare Are Important to You!**	**general**
	Maria Hardin	NORD
	Dan Schriener	CMS Perspective
	Steve Grossman	Advocacy Activities
	Don Moran	Components of MMRD
	Track One Sessions	**breakout**
	Brian Finn	Estate Planning for Special Needs
	Maria Hardin/Christin Engelhardt	Choosing Health Care Plans for Medicare Part D
	Steve Sternberg Mike Laris Lori Russo	Who Will Listen to My Story? Getting the Attention of the Media
	Doris Zallen Steve Groft	Pros & Cons of Clinical Trials
	Track Two Sessions	**breakout**
	Maria Hardin Christin Engelhardt (Document 1, Document 2) Kimberly Symonds	How to Equip Organizations with the Expertise to

	Complete Medicare Applications
Estelle Benson Jayne Gershkowitz Jim Broatch	Coping Mechanisms for Patient Services Personnel
Diane Dorman Steve Grossman Josephine Grima	Influencing Public Policy

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Understanding Your Rights as a Patient Patty Delaney	**general**

Track One Sessions	**breakout**
Brian Finn	Estate Planning for Special Needs
Healthbridge	Choosing Health Care Plans for Medicare Part D
Steve Sternberg Mike Laris Lori Russo	Who Will Listen to My Story? Getting the Attention of the Media
Doris Zallen Steve Groft	Pros & Cons of Clinical Trials

Track Two Sessions	**breakout**
Maria Hardin Christin Engelhardt Kimberly Symonds	How to Equip Organizations with the Expertise to Complete Medicare Applications

Diane Dorman Steve Grossman Josephine Grima	Influencing Public Policy
Mary Dunkle Dana Rawding Rob Tomanio	How to Increase Your Website Traffic

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10/2/05 Advocacy: Affecting Public Policy **general**

Abbey Meyers
Diane Dorman
Steve Grossman

三、頒贈榮譽

NORD 每年亦頒贈榮譽給有功於罕病社群的人士與組織，包括國會議員、研究者、藥廠、個人、以及其他有助於改善罕病病人生活者。2005 年的頒贈宴會於 5 月 23 日在華盛頓特區舉行，並有數種不同類別的獎項，如「國家健康領導獎」、「公共衛生領導獎」、「企業獎」、「治療成就獎」等。2005 年所頒贈的獎項與受獎者如下：

National Health Leadership Award

The Honorable Jeff Bingaman
United States Senator, New Mexico

Public Health Leadership Award

Elizabeth Dickinson
Associate Chief Counsel for Drugs
U.S. Food and Drug Administration

Corporate Awards

Bausch & Lomb
for the development of Retisert for the treatment of chronic non-infectious uveitis

Pharmion Corporation
for the development of Vidaza (azacitidine) for treatment of subtypes of myelodysplastic syndrome

Synthes Corporation
for the development of the vertical expandable prosthetic titanium rib

Therapeutic Achievement Award

Dr. Robert Campbell, Jr.
for the development of the vertical expandable prosthetic titanium rib

Dr. Josep Torrent-Farnell
Chairperson of the European Union's Committee on Orphan Medicinal Products (COMP)

四、重要演說與證詞的搜集

NORD 同時就其主要負責人參與各項演說或公聽會相關之陳述加以彙整，並置入其網站中供各界參閱，以便 NORD 對於政策、規則、或相關議題的主張可以被各界清楚了解。相關的資訊如下：

- Presentation at PDUFA Stakeholders' meeting at National Institutes of Health
  Presented by Diane E. Dorman, November 15, 2006

- Wiley Lecture for 2005 presented to FDA Alumni Association
  Presented by Abbey S. Meyers, April 6, 2005
- Testimony regarding the Food and Drug Administration's (FDA) approval process, drug safety and the concerns of patients before the U.S. Senate Health, Education, Labor, and Pensions Committee
  Presented by Abbey S. Meyers, March 1, 2005

  Responses to follow-up questions posed by the HELP Committee chairman
- Testimony regarding funding of the Office of Rare Diseases at the NIH presented to the House Appropriations Labor, Health and Human Services Education and Related Agencies Subcommittee
  Presented by Diane E. Dorman, May 14, 2003
- Speech at Workshop on Ultra-Orphan Genetic Disease Therapeutics
  Presented by Abbey S. Meyers, May 8, 2003
- NORD's Testimony to the U.S. House of Representatives regarding FY2004 appropriations for the FDA's Orphan Product Research Grants program.
  The same testimony was submitted to the U.S. Senate Agriculture Appropriations Committee. April 2003
- Introductory Remarks, NORD Generic Biologics Conference
  Presented by Abbey S. Meyers, March 19, 2003
- FDA Testimony, Fabry's Disease
  Presented by Abbey S. Meyers, January 13 & 14, 2003
- A Tribute to Roscoe Brady, MD
  Speech delivered at NIH Symposium by Abbey Meyers, October 2002
- Italian Rare Disease Federation
  Speech delivered in Venice by Abbey Meyers, Sept. 2002
- Human Research Subject Protection
  Written comments prepared for the Senate HELP Committee. May 2002. Abbey S. Meyers

- In support of a funding increase for FDA Orphan Products Research Grant Program in FY 2003
  Testimony submitted for the record, March 2002, Diane E. Dorman, NORD Vice President for Public Policy
- Public Meeting on the Reauthorization of the Prescription Drug User Fee Act
  Testimony submitted before the House Energy and Commerce Subcommittee on Health, December 7, 2001, Diane E. Dorman
- Unfulfilled Promises and Stem Cell Research
  Presented to MIT, November 20, 2001, Abbey S. Meyers
- Evaluating the Effectiveness of the Food and Drug Administration Modernization Act
  Testimony submitted before the House Energy and Commerce Subcommittee on Health, May 3, 2001, Abbey S. Meyers
- FY 2002 Appropriations for the Orphan Products Research Grant Program at the Food and Drug Adminstration (FDA)
  Testimony submitted to the Senate Appropriations Subcommittee on Agriculture, Rural Development and Related Agencies, April 2001, Abbey S. Meyers
- Testimony Before the House Appropriations Labor, Health and Human Services, Education and Related Agencies Subcommittee
  March 14, 2001, Diane E. Dorman
- Access to Dietary Supplements for Valid Medical Uses in Children
  February 2001, Abbey S. Meyers
- Analysis by and Recommendations of the Special Emphasis Panel of the NIH on the Coordination of Rare Diseases Research
  Summary ?January 2001, Abbey S. Meyers
- Orphan Drug Development Conference: "Understanding the History of the Orphan Drug Act"
  September 2000, Abbey S. Meyers
- Prescription Drug User Fee Act (PDUFA)
  September 15, 2000
- Subcommittee on Health and Environment

September 15, 2000

- The Patient Advocate Perspective. Preparing for FDA Advisory Committee Presentations. Views from Four Perspectives
  Abbey S. Meyers, President NORD. June 30, 2000
- HHS Enhances Patient Protections: NIH and FDA to Cooperate
  HHS Press Release of May 23,2000
- New Initiatives to Protect Patients in Gene Therapy Trials
  Abbey S. Meyers, President NORD
- Collaboration Between the Pharmaceutical Industry and Patient Organization: A Delicate Balance
  Abbey S. Meyers, President NORD. February 17, 2000. Spain: International Conference of Rare Diseases and Orphan Drugs.
- History of the American Orphan Drug Act
  Abbey S. Meyers, President NORD. February 18, 2000. Spain: International Conference of Rare Diseases and Orphan Drugs
- Exploring the Role of Patient Organization in the Development of Orphan Products
  Abbey S. Meyers, President NORD. September 17, 1999

五、政策與立場文件的呈現

NORD 針對特定事件的立場說明，或是針對與罕病有關的政策主張，或是對於某一特定議題的倡導等，均以立場文件或是信函的方式呈現，同時議題涉及的範圍也極為廣泛，如對於基因治療、病患權益、孤兒藥管理政策、臨床實驗中的病患保障等。相關資訊如下：

- Letter to CMS Administrator Mark McClellan Regarding Excluding Some Orphan Drugs From the CAP Program
  September 6, 2005

- Comments on the Draft Guidance on CMS Formulary Review for the Medicare Prescription Drug Benefits
  December 30, 2004
- Letter to NIH Director Regarding Public Access to Research Information
  By Abbey Meyers, NORD President, October 2004
- NORD Position on Fetal Stem Cell Research
  By Abbey Meyers, NORD President, September 2004
- Position Paper: Patients' Bill of Rights
  By Abbey S. Meyers, President NORD. November 2000
- Position Paper: Genetic Patenting
  By Abbey S. Meyers, President NORD. September 25, 2000
- Position Paper: Patient Protection in Clinical Trials
  By Abbey S. Meyers, President NORD. August 14, 2000
- Position Paper: Genetic Discrimination
  By Abbey S. Meyers, President NORD. August 11, 2000
- Position Paper: Insurance Reimbursement for Orphan Drugs
  By Abbey S. Meyers, President, NORD. August 6, 2000
- Position Paper: Drug Patent Extensions
  By Abbey S. Meyers, President, NORD. August 6, 2000
- NORD Policy Regarding Drug Price Competition & Patent Term Restoration Act of 1984

六、重要新聞搜錄

針為與罕病有關的相關報導或文件，NORD 也經由同意轉載的方式，將相關重要新聞與文件列入其網站中，供各界掌握與相關的新聞報導。例如：

Previous Articles & Letters

Orphan Drugs Save Lives, But Who Pays?

Many Docs Get Newborn Screening Test Results Late

Letter in Support of Increased FDA Funding

Comments Regarding FDA's Regulations, Guidances, Policies and Practices

Letter to the FDA Regarding Pediatric Labeling

FDA Office of Orphan Products Development FY2001 Accomplishments

Secretary's Advisory Committee on Regulatory Reform

New Initiatives to Protect Patients in Gene Therapy Trials

New York Times Article on Rare Disorders

HHS Enhances Patient Protections: NIH and FDA to Cooperate

七、提供研究獎學金

NORD 也提供研究獎學金給醫生進行研究，如 NORD/Roscoe Brady 獎學金即提供經費供年輕醫師研究溶山體疾病（Lysosomal Storage Diseases；LSD)，這項獎助的金額約介於美金五萬至七萬元，至多可以補助兩年，並以 Dr. Roscoe Brady 為名紀念，Dr. Roscoe Brady 係發現 LSD 疾病並發展出酵素療法的傑出科學家，如對 Fabry 和

Gaucher 疾病的治療。這項獎學金於 2001 年春天開始設立，由位於美國麻州的生技公司 Trauskaryotic Therapies（TKT）公司贊助 NORD 一百萬美元設立，以鼓勵對於 LSD 疾病的研究；同時這項獎金部份則由 Geuzym 公司與個人捐款贊助進行。

2003 年 NORD/Roscoe Brady 獎學金得主如下：

Dr. Catharina Whybra of the Johannes Gutenberg University of Mainz, Germany will study the pathomechanisms of Gb3 induced kidney damage in Fabry disease and the effect of Enzyme Replacement Therapy with algalsidase alfa on autonomic cardiac function and late potentials in signal averaged electrocardiograms in patients with Fabry disease. Her work will contribute to the understanding of the pathophysiology of organ damage in the kidney and the effect of Enzyme Replacement Therapy concerning the heart.

Dr. Christopher G. Janson of Cooper Hospital & Robert Wood Johnson Medical School, New Jersey will perform stereotactic gene delivery in conjunction with drug delivery to the brain in a mouse model of Sandhoff disease, as part of "Genetic and drug-based interventions for lysosomal storage disease." Dr. Janson's project will determine the additive effects that are possible when viral-based gene therapy is combined with drug treatments or antisense modalities which simultaneously target the biosynthetic and degradative pathways of gangliosides, in addition to related pathways such as inflammation and apoptosis. These studies will help in the planning of clinical trials for LSD such as Sandhoff and Late-Onset Tay Sachs disease.

Dr. Takeshi Taketani of Shimane Medical University's School of Medicine, Japan, will study to develop a newborn screening test and a monitoring system for Mucopolysaccharidoses (MOS) and mucolipidosis (ML). MPS and ML are progressive lysosomal storage diseases causing multiple problems, including systematic joint and bone disease, hearing and visual loss, and CNS involvement. The undegraded GAGs are stored in lysosomal and/or excreted into blood and urine. To prevent worsening the disease, early diagnosis and early treatment are required. Dr Taketani

will develop the new assay of each GAG in blood and urine and apply onto the screening system.

八、提供小額研究經費

NORD 也有研究經費補助方案，提供科學家開發罕病新的治療與診斷方式，這些研究初期的補助有助於科學家搜集初步的資料，以便後續得以申請其他更高額的經費補助或是吸引企業界開發成為符合美國「孤兒藥」法案獎助的商品，這項產品如藥物、設備、或醫療食物等。NORD 同時在網站中羅列所提供的經費補助資訊、或是其他會員團體所提供的補助資訊等，合計在 2004 年當中，NORD 計已補助 20 個研究獎助。相關得主如下：

NORD research awards for **Adenoid Cystic Carcinoma:**

Mark Darling, MD
University of Western Ontario, London, Ontario, Canada

Barry Ziober, PhD
University of Pennsylvania, Philadelphia, PA

NORD research award for **Cockayne Syndrome:**

Edward Neilan, MD, PhD
Children's Hospital Boston, Boston, MA

NORD research award for **Creutzfeldt Jakob Disease:**
Larisa Cervenakova, MD, PhD
American Red Cross, Jerome H. Holland Laboratory, Rockville, MD

NORD/Ehlers Danlos National Foundation research awards for
Ehlers-Danlos Syndrome:

Peter Byers, MD
University of Washington, Seattle, WA
Richard Wenstrup, MD
Cincinnati Children's Hospital Research Foundation, Cincinnati, OH

NORD research awards for **Giant Hypertrophic Gastritis:**
John Kao, MD
University of Michigan Medical School, Ann Arbor, MI
Andrea Todisco, MD
University of Michigan, Ann Arbor, MI

NORD research award for **Kearns-Sayre Syndrome:**
Jeffrey Towbin, MD
Texas Children's Hospital, Houston, TX

Neurodegeneration with Brain Iron Accumulation (NBIA):

Susan Hayflick, MD
Oregon Health & Science University, Portland, OR

NORD research awards for **Olivopontocerebellar Atrophy (OPCA) and**

Closely Related Neurological Disorders:
Alfredo Brusco, PhD
University of Turin, Torino, Italy
Gwenn Garden, MD, PhD
University of Washington, Seattle, WA
Harry Orr, MD, PhD
University of Minnesota, Minneapolis, MN
Blair Leavitt, MD
University of British Columbia, Vancouver, British Columbia, Canada
Dingbo Lin, PhD
Kansas State University, Manhattan, KS
Mario Mascalchi, MD, PhD
University of Florence, Viale Pieraccini Firenze, Italy
Tohru Matsuura, MD
Baylor College of Medicine, Houston, TX
Puneet Opal, MD, PhD

Northwestern University, Chicago, IL
Ryuji Sakakibara, MD, PhD
Chiba University, Chiba, Japan

NORD research award for **Tyrosinemia Type 1:**
Robert Tanguay, PhD
Universite Laval, Ste-Foy, Quebec Canada

九、研究計畫召募病患參與

NORD 雖然並未針對特定計畫鼓勵病患參與，但仍提供資訊流通的服務，將需要病患參與的研究計畫列在其網站中，供病患了解進行中的研究。相關計畫案例如下：

Fabry Disease

Researchers at NIH are conducting a research study to advance the knowledge of Fabry disease. For information, call (800) 411-1222 (TTY: 866-411-1010).

Platelet Disorders

A study of platelet disorders is being conducted at the National Institutes of Health (NIH). Adults and children with any of the following conditions may be eligible to participate: gray platelet syndrome, Hermansky-Pudlak syndrome, storage pool deficiency, Chediak-Higashi syndrome, Griscelli syndrome, Wiskott-Aldrich syndrome, and thrombocytopenia absent radius syndrome. The researchers hope that study of the causes of these syndromes and related disorders will lead to improved therapies in the future. For information, call the NIH Patient Recruitment Office at (800) 411-1222 (TTY: 866-411-1010).

Gray Platelet Syndrome

Gray platelet syndrome is a rare bleeding disorder that causes excessive bleeding and bruising. Doctors at the National Institutes of Health (NIH) invite those with GPS and their families to participate in a study to identify the gene(s) that cause the disorder. Families with at least one member diagnosed with this syndrome may be eligible. Participants will need to obtain and ship a blood sample to the NIH in Bethesda, MD, for genetic analysis. No travel is required for this study. Shipment costs will be reimbursed. For information, contact Meral Gunay-Aygun, MD, the principal investigator, at (301) 594-4181 (TTY: 866-411-1010) or mgaygun@nih.gov. Information may also be found at http://clinicalcenter.nih.gov.

Hereditary Angioedema

Several study sites in the U.S. and Canada are actively recruiting patients to test a new medication for the treatment of hereditary angioedema (HAE) attacks. HAE is a painful and sometimes life-threatening disease characterized by recurrent local swelling of the subcutaneous tissue and the gastrointestinal tract. Occasionally, swelling occurs in the larynx area, leading to potentially dangerous airway constriction.

Icatibant, a synthetically manufactured substance, is being tested in Phase II/III clinical trials sponsored by Jerini AG, a German company. Icatibant has been designated an orphan drug by the U.S. Food and Drug Administration. For information about the current study, contact Barbara Hobson-Whiting, InClin Inc., at (512) 321-4567 or rmsllc@austin.rr.com. The study is described on www.clinicaltrials.gov.

十、NORD 醫療協助方案（NORD Medication Assistance Program）

從 1987 年開始，NORD 即開始設醫療協助方案，以協助沒有醫療保險或保險不足的病患，得以獲得維生的醫療照護。在美國估計有 5 千萬人未獲有醫療保險，以及日漸增多的個人無法享有給付處方藥的情況下，這項方案更形重要。NORD 與具有人道關懷的藥商與生技公

司合作密切，針對其所得因太高而不符政府 Medicaid 補助，而其所得又因太低而無法購買處方藥的個人與家庭進行協助；NORD 設定了公平、均等、與不偏倚原則，保證病患的隱私，也獲得各界的尊敬。目前與 NORD 合作的廠商，即包括有：

- Acorda Therapeutics
  Zanaflex® Capsules Uninsured Individual Program
  Tizanidine Hydrochloride
  Conditions:
  Spasticity
  Contact:
  Reimbursement Assistance Program at 1-800-464-0097

- Actelion
  Tracleer ® Patient Assistance Program (T.P.A.P.)
  bosentan tablets
  Conditions:
  Pulmonary Arterial Hypertension
  Contact:
  Tracleer Assistance Program (T.A.P.) at 1-866-228-3546

- Allergan, Inc.
  BOTOX?Patient Assistance Program
  (botulinum toxin Type A)
  Conditions:
  Blepharospasm, Strabismus, Hemi-facial spasm, Spasmodic torticollis, Dystonias, Cerebral palsy
  Contact:
  The Botox Reimbursement Hotline at 1-800-530-6680

- Cephalon, Inc.
  PROVIGIL?Assistance Program
  (modafinil)
  Conditions:

Narcolepsy
Contact:
the Provigil?Reimbursement Assistance Hotline at 1-800-675-8415

- Cephalon, Inc.
  TRISENOX® Patient Assistance Program
  (arsenic trioxide) injection
  Conditions:
  relapsed or refractory acute promyelocytic Leukemia (APL), Multiple Myeloma
  Contact:
  Trisenox Reimbursement Advocacy Services, 1-866-261-7730

- Medtronic, Inc.
  ITB?Therapy Patient Assistance Program
  Lioresal?Intrathecal (baclofen injection)
  Conditions:
  Severe spasticity
  Contact:
  NORD at 1-800-999-6673 e-mail nnadig@rarediseases.org

- Orphan Medical, Inc.
  CYSTADANE?Patient Assistance Program
  (betaine anhydrous for oral solution)
  Conditions:
  Homocystinuria
  Contact:
  NORD at 1-800-999-6673 e-mail bnavarette@rarediseases.org

- Orphan Medical, Inc.
  XYREM?Patient Assistance Program
  (sodium oxybate)
  Conditions:
  Narcolepsy with cataplexy
  Contact:
  Xyrem Success Program at 1-866-997-3688

- QOL Medical, Inc.
  Sucraid® Patient Assistance Program

(sucradose)
Conditions:
Congenital sucrase isomaltase deficiency (CSID)
Contact:
ivpcare at 1-800-424-9002

- Questcor Pharmaceuticals, Inc.
  ACTHAR® GEL Patient Assistance Program
  (ACTH)
  Conditions:
  Infantile spasms, multiple sclerosis, Crohns disease, Adrenal insufficiency, etc.
  Contact:
  NORD at 1-800-459-7599 e-mail nnadig@rarediseases.org

- Rare Disease Therapeutics, Inc.
  ORFADIN?Patient Assistance Program
  (nitisinone)
  Conditions:
  Hereditary Tyrosinemia Type I
  Contact:
  NovaFactor at 1-888-454-8860

- Sanofi - Aventis
  RILUTEK?Patient Assistance Program
  (riluzole)
  Conditions:
  Amyotrophic lateral sclerosis
  Contact:
  NORD at 1-800-459-7599 or e-mail nnadig@rarediseases.org

- Sanofi Pasteur, Inc.
  Imovax®/Imogam® Patient Assistance Program
  (Human Diploid Cell Vaccine)
  Conditions:
  Rabies Vaccine
  Contact:
  NORD at 1-877-798-8716 or e-mail nnadig@rarediseases.org

- Sanofi Pasteur, Inc.
  TheraCys® Patient Assistance Program
  [BCG Live (Intravesical)]
  Aventis Pasteur, Inc.
  Conditions:
  Bladder Cancer Therapy
  Contact:
  NORD at 1-877-798-8716 or e-mail nnadig@rarediseases.org

- Sanofi Pasteur, Inc.
  MENOMUNE?Patient Assistance Program
  (meningococcal vaccine)
  Conditions:
  Meningitis
  Contact:
  NORD at 1-877-798-8716 or e-mail nnadig@rarediseases.org

- Serono Laboratories, Inc.
  SEROSTIM?Patient Assistance Program
  [somatropin](rDNA origin) injection
  Conditions:
  AIDS wasting, cachexia
  Contact:
  NORD at 1-888-628-6673 e-mail bgoodbou@rarediseases.org

- Sigma-tau Pharmaceuticals, Inc.
  CARNITOR® Drug Assistance Program
  (L-Carnitine or levo carnitine)
  Conditions:
  Primary systemic carnitine deficiency, dialysis, chronic fatigue syndrome (CFS), several rare metabolic disorders
  Contact:
  NORD at 1-800-999-6673 e-mail bnavarette@rarediseases.org

- Sigma-tau Pharmaceuticals, Inc.
  MATULANE?Drug Assistance Program
  (procarbazine hydrochloride)
  Conditions:
  Hodgkins disease (Stage III and IV) and certain other lymphomas

Contact:
NORD at 1-800-999-6673 e-mail bnavarette@rarediseases.org

- Teva Neuroscience, Inc.
  COPAXONE® Medication Assistance Program COPAXONE® Premium / Co-Payment Assistance Program
  (glatiramer acetate, for injection)
  Conditions:
  Multiple sclerosis (relapsing-remitting)
  Contact:
  Shared Solutions at 1-800-887-8100

- Teva Pharmaceuticals Inc. USA
  Tev-Tropin® Patient Assistance Program
  Tev-Tropin® Co-Payment Assistance Program
  Somatropin (rDNAorigin) for injection
  Conditions:
  growth failure due to inadequate secretion of normal endogenous growth hormone
  Contact:
  Growth Solutions at 1-866-838-8767

- Ucyclyd Pharma, Inc.
  BUPHENYL?br> (sodium phenylbuterate)
  Conditions:
  Urea cycle disorders
  Contact:
  Ucyclyd Reimbursement Hotline at 1-800-711-0811

- **NORD MS Premium /Co-Payment Assistance Program**
  Conditions:
  Multiple Sclerosis
  Contact:
  1-800-634-7207

- **NORD CPP Premium /Co-Payment Assistance Program**
  Conditions:
  Chronic Plaque Psoriasis (CPP)
  Contact:

1-800-634-7207

- **NORD Intrathecal Therapy for Pain Management Co-Payment Assistance Program**
  Conditions:
  Intrathecal Therapy for Pain Management
  Contact:
  1-888-744-2581

十一、建置病患網絡

NORD 同時也建置了隱密的病患交流網絡，這個目標是鼓勵病患交換資訊及相互協助，一旦病患加入這個網絡，即能享有與其他相同疾病病人交流的管道，以促進病人間的相互協助。

參、美國罕病組織（NORD）的會員制度

一、個人會員（Individual Membership）

個人可選擇適合的類別經交付年費後，參與成為美國罕病組織的個人會員，這些所收集的會費將作為支持美國罕病組織所有服務的經費來源，因此不能針對特定疾病使用。個人除了可以因參與幫助罕病病人而獲得成就感以外，同時也享有下列各種不同的優惠：

1、基本個人會員（Basic Individual Membership）資格優惠：（每年美金 30 元）

＊ 年度會員期間每年獲贈三份美國罕病組織的報導。

＊ 可以參與美國罕病組織所運作的「網絡方案」（Networking Program），該方案之目的在促使會員交換資訊及相互扶持，參與的會員將可以與罹患相同疾病的病人互動，該方案也鼓勵會員組成互助團體，相關的運作細節可聯繫 Networking Coordinator, NORD, PO Box 1968, Danbury, CT 06813。

＊ 參加美國罕病組織年會享有折扣。

＊ 可以有機會參與美國罕病組織的「公共政策方案」（Public Policy Program）作為志工。

＊ 可訂閱「罕見疾病新知」（Orphan Diseases Updated）通訊。

2、友誼個人會員（Friend Membership）資格優惠：（每年美金 50 元）

＊ 年度會員期間每年獲贈三份美國罕病組織的報導。

＊ 可以參與美國罕病組織所運作的「網絡方案」（Networking Program）。

＊ 參加美國罕病組織年會享有折扣。

＊ 可以有機會參與美國罕病組織的「公共政策方案」（Public Policy Program）作為志工。

＊ 可訂閱「罕見疾病新知」（Orphan Diseases Updated）通訊。

3、個人贊助會員（Contributing Individual Membership）資格優惠：（每年美金 100 元）

＊ 年度會員期間每年獲贈十份美國罕病組織的報導。

＊ 可以參與美國罕病組織所運作的「網絡方案」（Networking Program）。

＊ 參加美國罕病組織年會享有折扣。

＊ 可以有機會參與美國罕病組織的「公共政策方案」（Public

Policy Program）作為志工。

＊ 可訂閱「罕見疾病新知」（Orphan Diseases Updated）通訊。

4、專業會員（Professional Colleague）資格優惠：（每年美金 75 元）

現任職於相關機構的醫療專業人員、遺傳諮詢員、社工人員、護士、教師、圖書管理人員等，可以參加成為專業會員。該項會員類別主要在協助相關專業人員瞭解罕病社群的最新發展，因此往來聯繫均係以服務機構的地址為限。

＊ 年度會員期間每年獲贈六份美國罕病組織的報導。

＊ 購買NORD Resource Guide可享九折優惠。

＊ 參加美國罕病組織年會享有折扣。

＊ 可訂閱「罕見疾病新知」（Orphan Diseases Updated）通訊。

二、團體會員資格（Organizational Membership）

1、成為團體會員的條件：

美國罕病組織是一個容納眾多組織的聯盟團體，其團體會員均具有協助罕病病患的共同信念，雖然彼此間的宗旨在細節上可能不同，但彼此均認同相互合作可以發聲。團體會員將因美國罕病組織「公共政策副總裁」（Vice President for Public Policy）辦公室在華盛頓特區

的努力而受益，如收到美國罕病組織有關政策與醫療的經常性簡報、與特定團體會員有關議題的指引等。

美國罕病組織有責任確保其團體會員符合倫理標準及負責任的治理，以維持對慈善團體的公眾信任。而團體會員資格則區分為「全國正式團體會員」（National Organizations）與「副團體會員」（Associate Organizations）兩種。「全國正式團體會員」的條件需要為非營利的志願性組織，運作範圍為全國性且董（理）監事會的組成也需反應這項全國性的特質，如果其董（理）監事會的組成僅限定於一個地區或是一個家庭，則將不符合全國的代表性，因此將不符合「全國正式團體會員」的資格，但是或許可以符合「副團體會員」的資格。

2、「全國正式團體會員」（National Organizations）資格要件：

＊ 具有美國政府財政部的非營利證明（如 501(c)3 的文件）。

＊ 章程符合慈善組織的標準，且非僅限定服務特定政治、宗教、或是種族的對象。

＊ 具有效力的章程。

＊ 董（理）監事會人選的姓名與地址可以反映其全國區位的多樣性，擁有運作的委員會，至少五位或以上的董（理）監事。

＊ 每年至少召開三次過半數董（理）監事參與的董（理）監事

會議，其中至少有一次係面對面的出席會議，其餘則可以採電子或視迅會議進行。

* 擁有至少三位以上組成的醫療顧問。
* 最近一年組織的 IRS 990 or 990EZ 表格與資訊。
* 組織的財務報告及最近一年的財務稽核報告（這些資訊可以應公眾需要而提供）。
* 組織的年度報告（必須對公眾開放）。
* 對社會大眾發行的教育資料（如手冊、通訊）。
* 每年預算需經董（理）監事通過。
* 足夠的隱私權規範以保障病患及家庭。
* 董（理）監事與會務人員的利益迴避規範。
* 組織宗旨和方案需不與美國罕病組織衝突。

3、美國罕病組織的團體會員名單（星號為「全國正式團體會員」）

Acid Maltase Deficiency Association Inc.
★Alagille Syndrome Alliance
★Alpha 1 Association
★Alpha 1 Foundation
ALS Association-Greater Philadelphia Chapter
American Autoimmune Related Diseases Association, Inc.
American Behcet's Disease Association
★American Brain Tumor Association
★American Porphyria Foundation
American Self-Help Group Clearinghouse
★American Syringomyelia Alliance Project, Inc.
★Aplastic Anemia & MDS International Foundation, Inc.
Association CMTC (Cutis Marmorata Telangiectatica Congenita)
★Association for Glycogen Storage Disease
★Association of Gastrointestinal Motility Disorders, Inc. (AGMD)
Ataxia-Telangiectasia Children's Project
★Batten Disease Support and Research Association
★Benign Essential Blepharospasm Research Foundation, Inc.
Canadian Organization for Rare Disorders
CDG Family Network Foundation
★Charcot-Marie-Tooth Association
Children's Cardiomyopathy Foundation
Children's Craniofacial Association
Children's PKU Network
★Chromosome 18 Registry and Research Society
Chromosome Deletion Outreach, Inc.
Chronic Granulomatous Disease Association, Inc.
★Cleft Palate Foundation
CLIMB (Children Living with Inherited Metabolic Diseases)
Coalition for Pulmonary Fibrosis
Consortium of Multiple Sclerosis Centers
Contact A Family
★Cornelia de Lange Syndrome Foundation Inc.
Cushing Support and Research Foundation, Inc.

Cutis Laxa Internationale
Cystinosis Foundation
Dysautonomia Foundation, Inc.
Dystonia Medical Research Foundation
DEBRA of America (Dystrophic Epidermolysis Bullosa Research Association of America)
Ehlers-Danlos National Foundation
Epilepsy Foundation
(The) Erythromelalgia Association
EURORDIS
Families of Spinal Muscular Atrophy
Family Caregiver Alliance
Family Support Network of North Carolina
Fibromuscular Dysplasia Society of America
Foundation for Ichthyosis & Related Skin Types
Freeman-Sheldon Parent Support Group
Foundation Fighting Blindness
GBS/CIDP Foundation International
Genetic Alliance
GOLD, Global Organisation For Lysosomal Diseases
Hereditary Colon Cancer Association (HCCA)
Hereditary Disease Foundation, Inc.
Hereditary Hemorrhagic Telangiectasia (HHT) Foundation International
Hereditary Neuropathy Foundation
Histiocytosis Association of America
Huntington's Disease Society of America
Hydrocephalus Association
Immune Deficiency Foundation
Incontinentia Pigmenti International Foundation
International Fibrodysplasia Ossificans Progressiva (FOP) Association
International Joseph Diseases Foundation, Inc.
International Rett Syndrome Association
Interstitial Cystitis Association of America, Inc.
Klippel-Trenaunay Support Group
Les Turner Amyotrophic Lateral Sclerosis Foundation, Ltd.
Lowe Syndrome Association
Lymphangioleiomyomatosis (LAM) Foundation

- Mastocytosis Society, Inc.
- Melorheostosis Association
- Mercy Medical Airlift
- ML 4 (Mucolipidosis Type IV Foundation)
- Moebius Syndrome Foundation
- Myasthenia Gravis Foundation of America
- Myositis Association
- Narcolepsy Network, Inc.
- National Adrenal Diseases Foundation
- National Alopecia Areata Foundation
- National Ataxia Foundation
- National Foundation for Ectodermal Dysplasias
- National Fragile X Foundation
- National Hemophilia Foundation
- National Kidney Foundation
- National Lymphedema Network
- National Marfan Foundation
- National MPS (Mucopolysaccharidoses/Mucolipdoses) Society
- National Multiple Sclerosis Society
- National Neurofibromatosis Foundation, Inc.
- National Niemann-Pick Disease Foundation, Inc.
- National Organization for Albinism and Hypopigmentation, NOAH
- National PKU News
- National Spasmodic Dysphonia Association
- National Spasmodic Torticollis Association
- National Tay-Sachs and Allied Diseases Association, Inc.
- National Urea Cycle Disorders Foundation
- Neurofibromatosis, Inc.
- Organic Acidemia Association
- Osteogenesis Imperfecta Foundation, Inc.
- Osteoporosis and Related Bone Diseases National Resource Center
- (The) Oxalosis and Hyperoxaluria Foundation
- Paget Foundation for Paget's Disease of Bone and Related Disorders
- Parent to Parent of New Zealand
- Parkinson's Disease Foundation, Inc.
- Platelet Disorder Support Association
- Prader-Willi Syndrome Association (USA)
- Pulmonary Hypertension Association

- Recurrent Respiratory Papillomatosis Foundation
- Reflex Sympathetic Dystrophy Syndrome Association of America
- Sarcoid Networking Association
- Sarcoma Foundation of America
- Scleroderma Foundation, Inc.
- Shwachman-Diamond Syndrome Foundation
- Society for Progressive Supranuclear Palsy
- Sotos Syndrome Support Association
- Stevens Johnson Syndrome Foundation and Support Group
- Sturge-Weber Foundation
- Taiwan Foundation For Rare Disorders
- Tourette Syndrome Association, Inc.
- Trigeminal Neuralgia Association
- United Leukodystrophy Foundation
- United Mitochondrial Disease Foundation
- Vestibular Disorders Association, VEDA
- VHL Family Alliance
- Wegener's Granulomatosis Association
- Williams Syndrome Association
- Wilson's Disease Association International

肆、美國罕病組織（NORD）的交流平台角色

NORD 雖然是美國全國性的罕病組織，但是也同時收納其他國家的罕病組織作為會員或副會員（Associate Member），因此成員非常廣泛。而在其每年的年會中也同時提供有攤位，鼓勵會員組織能在年會中提供展示，以相互交流；此外，在其網站中亦羅列有關相關會員組織的活動資訊，作為跨病類與跨團體間之交流平台。

＊ 與華盛頓特區直接連線：各團體會員組織與美國罕病組織在華盛頓特區的辦公室緊密結合，定期收到有關罕病社群利益有關的最新發展。

＊ 在醫療會議中作為代表：美國罕病組織會務人員每年均參加許多醫療專業會議，各團體會員組織的出版品被鼓勵由美國罕病組織加以協助陳列。

＊ 提供資訊公告平台：在美國罕病組織網頁中的「研究」欄中，各團體會員組織被邀請張貼研究經費與計畫的資訊，以供醫療與科學社群分享相關資訊；美國罕病組織網頁每月約有 14 萬人瀏覽，提供各組織研究經費曝光的絕佳機會。例如：

NORD member organizations may submit copy for this page to webmaster@rarediseases.org. All entries should be submitted in the format used below.

CANCER RESEARCH FUND / VHL FAMILY ALLIANCE
171 Clinton Road
Brookline, MA 02445
Phone: 617-277-5667 x4
Fax: 866-209-0288
E-mail: research@vhl.org
Web: http://www.vhl.org

Disease name: Funding available for research on von Hippel-Lindau disease, with special focus on strategies that will translate into therapies that will improve the lives of people with VHL.
Amount: $25,000 to $40,000 (total direct costs only)
Deadline to apply: April 3, 2006
Guidelines and application: www.vhl.org/research

CHILDREN'S CARDIOMYOPATHY FOUNDATION
P.O. Box 547
Tenafly, New Jersey 07670
Phone: 201-227-8852
Fax: 201-227-7016
E-mail: info@childrenscardiomyopathy.org
Web: http://www.childrenscardiomyopathy.org

Disease name: Funding available for biomedical research on pediatric cardiomyopathy (dilated, hypertrophic, restrictive or arrhythmogenic right ventricular cardiomyopathy)
Amount: $25,000 to $50,000 (total direct costs only)
Deadline to apply: October 3, 2005
Guidelines and application:
www.childrenscardiomyopathy.org/site/grants.php

DYSAUTONOMIA FOUNDATION INC.
633 Third Ave., 12th Floor
New York, NY 10017-6706

Phone: 212-949-6644
Fax: 212-682-7625
E-mail: dys212@aol.com
Web: www.familialdysautonomia.org

Disease name: familial dysautonomia
Amount: up to $100,000 per year
Length of study: 2 years
Deadline to apply: TBA

THE HHT FOUNDATION INTERNATIONAL
P.O. Box 329
Monkton, MD 21111
Contact: Connie Anderson, Research Program Administrator
Phone: 800-448-6389
E-mail: research@hht.org
Web: www.hht.org

Disease Name: Hereditary Hemorrhagic Telangiectasia (HHT)
Amount: $50,000
Length of Study: 1 year
Number of Grants: 2
Letter of Intent Deadline: January 14, 2006 11:59PM EST
Application Submission Deadline: March 1, 2006 11:59PM EST

The mission of the Hereditary Hemorrhagic Telangiectasia (HHT) Foundation International is to find a cure for HHT. The HHT Foundation International (HFI) announces its 2006 request for proposals (RFP) for the following two topics:

"HHT: Treatment of Disease"
"HHT: Mechanisms of Disease"

Under this RFP researchers are invited to submit investigator-initiated proposals regarding either of the two topics. One grant (up to $50,000 US) will be awarded in the 2006 competition for each of these topics.
Grant applications may be submitted by international principal investigators. Post-doctoral students or fellows are not eligible to apply as principal investigators for this RFP.
Letters of intent (LOI) are mandatory, and are treated with confidentiality

by the HFI and its reviewers. The letter of intent process will allow the HHT Foundation International staff to estimate the potential review workload, avoid potential conflicts of interest in the review process, and verify contact information. The LOI should be a maximum of 1 page and be submitted electronically to research@hht.org.

The electronic application form can be requested by emailing research@hht.org.

THE LAM (LYMPHANGIOMYOMATOSIS) FOUNDATION
10105 Beacon Hills Drive
Cincinnati, OH 45241-1335
Contact: Dr. Frank McCormack, Chair, Scientific Advisory Board
Phone: (513) 558-4831
E-mail: lam@one.net or frank.mccormack@uc.edu
Web: http://lam.uc.edu (Details and downloadable forms)

Disease name: Lymphangiomyomatosis (LAM)
Amount: See descriptions below
Length of study: See descriptions below
Deadline to apply: September 1

THE LAM (Lymphangioleiomyomatosis) FOUNDATION offers research funding for the study of the cellular and molecular basis of Lymphangiomyomatosis (LAM). The three-year **LAM Post Doctoral Fellowship Awards** and **Established Investigator Awards** provide a maximum of $150,000, renewable for up to two additional years. Established Awardees (faculty level investigators), must have a substantial track record in research and are eligible to receive funding for technician support and supplies. More than 50% of the funds must be used for fellow salary support, and the maximum fellow salaries allowed are based on NIH guidelines for postdoctoral fellows. The balance of the funds may be used for fringe benefits, supplies, or animal costs. No overhead or indirect costs are provided. Funds not spent are to be returned to the Foundation at the end of each funding year. **Pilot Project Awards** are available up to $25,000. Candidates must have at least two years of experience, a PhD, MD, or equivalent degree, and perform the work in a laboratory with established expertise in smooth muscle biology, genetics of tuberous sclerosis or other LAM-related areas.

Examples of competitive proposals include those that focus on the genetic regulation of smooth muscle growth or the development of a smooth muscle cell line that is representative of the LAM lesion. Mechanistic, hypothesis driven approaches of all types are welcomed. Formalin-fixed LAM tissues, dispersed LAM lung cells, genetic probes and other reagents are available.

THE STURGE-WEBER FOUNDATION
PO Box 418 Mt. Freedom, NJ 07970
Phone: 973-895-4445
Fax: 973-895-4846
E-mail: swf@sturge-weber.com
Web: www.sturge-weber.com

Disease name: Sturge-Weber syndrome
Amount: $30,000
Length of study: 1 year
Deadline to apply: September 1. Letter of Intent due 3 months prior.

TOURETTE SYNDROME ASSOCIATION, INC.
42-40 Bell Blvd, suite 205, Bayside, NY 11361
Phone: 718-224-2999 ext. 222
Fax: 718-279-9596
E-mail: grantadministrator@tsa-usa.org
Web: tsa-usa.org

Disease Name: Tourette Syndrome
Amount: up to $75,000; Fellowships: $40,000
Length of study: 1 year.
Deadline to apply: Letter of Intent due October 17, 2005

The Tourette Syndrome Association is requesting research grant proposals in basic and clinical studies on all aspects of Tourette Syndrome. Areas of specific interest include; animal models, basal ganglia physiology, behavioral neurosciences, clinical trials, neurochemistry, neuroimaging and neuropathology. Other relevant fields of interest are biochemistry, epidemiology, genetics, molecular biology, neuroanatomy, neurology, neuropsychology, neurophysiology, pharmacology, psychiatry and psychology. For preliminary screening, a

Letter of Intent briefly describing the scientific basis and relevance of the proposed project is requested. The approximate project funding level should be included. Intent letters should be sent as an email attachment to: grantadministrator@tsa-usa.org. The cover page to accompany each letter can be downloaded from our website: www.tsa-usa.org/research/intent.html.

US IMMUNODEFICIENCY NETWORK
40 W. Chesapeake Ave., Suite 308
Towson, MD 21204
Phone: (800) 296-4433 Ext. 211
Fax: (410) 321-9165
E-mail: tb@primaryimmune.org
Web: www.usidnet.org

Disease Name: Primary Immune Deficiency Diseases
Amount: $50,000 to $150,000 annually including indirect cost
Length of Study: Up to two years
Deadline: No Deadline Dates for Concept Proposals

The US Immunodeficiency Network (USIDNET) is requesting Concept Research Proposals focused on primary immune deficiency diseases. USIDNET will consider Concept Proposals from U.S. and non U.S. citizens. All application submissions must be made online through RAMS proposalCENTRAL website https://v2.ramscompany.com/Login.asp.

WEGENER'S GRANULOMATOSIS ASSOCIATION
P.O. Box 28660
Kansas City, MO 64188-8660
Phone: 800-277-9474
Fax: 816-436-8211
E-mail: wga@wgassociation.org
Web: www.wgassociation.org

Disease name: Wegener's Granulomatosis
Amount: Up to $35,000 per year
Length of Study: 1 year
Deadlines to apply: June 1 and December 1

The Wegener's Granulomatosis Association is requesting proposals to be considered for funding under the Association's Small Grant Program. Proposals are being sought from academic researchers who would be interested in receiving seed money grants to support pilot studies which would lead to much larger multi-year Wegener's Granulomatosis studies that could be funded by the NIH or some other sponsor of large-scale medical research. Proposals are being sought to research the etiology, epidemiology, diagnosis, treatment, and patient relief from the complications of living with Wegener's Granulomatosis.

* 分享年會與特定活動資訊：各團體會員組織被邀請於美國罕病組織網頁中張貼其補助與舉辦的相關活動，如病患研討會與社會教育活動等。由於罕病相關醫療人員、病患、及研究者經常瀏覽美國罕病組織網頁，因此是宣傳各組織活動的絕佳機會。
* 指導非營利組織的治理：各團體會員組織被鼓勵運用美國罕病組織會務人員的經驗與知識，以提升有效治理非營利組織的經驗。
* 流通資訊與刊物：各團體會員組織定期收到「Orphan Disease Update」及「NORD Online」兩份刊物，後者是一項有關醫療發展的即時資訊。
* 建構網絡：參與美國罕病組織「Networking Program」的病人，被鼓勵與其單一病類組織接觸，同時也被告知他的姓名會與

美國罕病組織所屬相關的團體會員分享，如果這個組織與其疾病極為關聯時；當美國罕病組織被研究者要求張貼臨床試驗計畫給病人參考時，美國罕病組織也會知會相關所屬團體會員有關該試驗計畫。

＊ NORD 的網站中也列有相關團體的網站資訊，透過相互連結，使得使用者可以從 NORD 的窗口中接觸到罕病的相關團體。例如：

- NORD's Database of 2,000 Patient Organizations and Government Agencies
- NORD Member Organizations (130)
- Academic Clinical And/Or Research Centers for Rare Diseases (1)
- Cancer Resources (2)
- Clinical Trials (3)
- Conferences & Presentations (1)
- General Health Sites (9)
- Genetics For Medical Professionals (3)
- Genetics Information (3)
- Government Sites (14)
- Health Policy Information (1)
- Health Privacy Information (3)
- International Sites (8)
- Laboratory Testing
- Medicare/Medicaid Information (8)

- National Family Caregivers Month
- Newborn Screening (3)
- Planning For Children With Special Needs (1)
- Resources for People with Disabilities (1)
- Umbrella Organizations (23)
- Universities/Hospitals (8)
- Women's Health (1)
- Other Sites of Interest (4)

＊ 發行 NORD e-News 如下：

NORD e-News
News from the National Organization for Rare Disorders (NORD)

CDC Warns of Salmonella Risk From Pet Rodents
Officials at the Centers for Disease Control and Prevention (CDC) have issued a warning about the risk for salmonellosis from contact with pet rodents, such as hamsters, mice, or rats. The agency found 15 such infections between 2003 and 2004. This is the first time the infection has been traced to rodents.

Children shouldn't be allowed to engage in behaviors such as kissing their pets, the CDC officials noted. They recommended thorough handwashing after handling the pets and their bedding.

Salmonellosis refers to infection with Salmonella bacteria. This is a group of bacteria that can cause illness in humans with symptoms such as diarrhea, fever, and abdominal cramps. In a small percentage of cases, the disease can become life-threatening.

Research Provides Clues on How Brain-Wasting Diseases Develop
Scientists at the Rocky Mountain Laboratories (RML) are finding clues

that may help explain how brain-wasting prion diseases develop and how they might be delayed or even prevented. The researchers are studying prion proteins, the agents that cause diseases such as Creutzfeldt-Jacob disease in humans and mad cow disease in cattle. Prions are misfolded proteins that can be contagious from one species to another.

Mice exposed to the prion that is known to cause scrapie, a brain-wasting disease of sheep, all developed visible signs of infection, such as twitching and poor coordination, within 150 days. However, mice exposed to a variant form of abnormal prion protein, which lacked the ability to anchor into the host cell membrane, showed evidence of exposure to disease but no disease symptoms.

The work "provides tantalizing clues as to how we might delay or even prevent such diseases by preventing certain cellular interactions," said Anthony S. Fauci, MD, Director of the National Institute of Allergy and Infectious Diseases (NIAID) at the National Institutes of Health (NIH). The research may also have implications for Alzheimer's disease, he noted.

Save The Dates for 2005 NORD Annual Conference

The NORD Annual Conference for 2005 will take place Sept. 30 to Oct. 2 at the Hilton Crystal City at Ronald Reagan National Airport, Arlington, Virginia. An optional Capitol Hill Visit is planned as a pre-conference event for Sept. 29.

The theme will be *Access to Medical Care: Navigating Medicare, Medicaid, and Private Insurance*. There will be two tiers of workshops, one for individual patients and their families, and one for leaders of patient organizations. For information, watch NORD's Web site (www.rarediseases.org) or call the NORD Development Office at (203) 744-0100.

Support Legislation to End Two-Year Waiting Period

NORD is one of many patient advocacy organizations supporting Senator Jeff Bingaman's (D-NM) bill to phase out, over 10 years, a mandated waiting period that leaves many people who have serious disabilities without health coverage at a time when they most need it. The bill, *Ending the Medicare Disability Waiting Period Act of 2005*, would phase out a two-year waiting period between the time when people with disabilities qualify for Social Security Disability Insurance (SSDI), and

when they can begin receiving Medicare benefits. It would also provide a mechanism through which the waiting period could be waived for people with life-threatening illnesses. (Details)

Access to Quality Testing for Rare Diseases
The Office of Rare Diseases (ORD) at the National Institutes of Health (NIH) and the Centers for Disease Control and Prevention (CDC) are planning a conference on access to quality genetic-testing for rare diseases. NORD will be one of several co-sponsors.

The majority of the 6,000 to 7,000 rare diseases known today are considered genetic conditions. As a result, genetic testing is an essential element in diagnosis and disease management. However, the development of tests hasn't kept pace with the explosion in understanding of these diseases. The conference will take place in Rockville, MD, on Sept. 26-27. (Details)

伍、美國罕病組織（NORD）的出版品

一、 NORD 資源手冊（NORD Resource Guide 第五版）

這項資源手冊被廣泛的由機構單位參考使用，包括圖書館、醫院、社會福利機構、學校等，病患及家屬也可以透過這項資源手冊發現相關有用的資訊與激勵。這項手冊第五版係於 2005 年出版，涵蓋 740 頁的內容，搜錄了 1,348 個病患組織、基金會、與協助罕病病患相關組織的資訊，包括該組織的宗旨、服務內容、電話、傳真、地址、網址、電子信箱等；每本訂價為美金 50 元。

二、 NORD 罕病指引（NORD Guide to Rare Disorders）

這本「NORD 罕病指引」的教科書主要是為醫生與醫事人員而出版，係於 2003 年出版，作者為 Lippincott、Williams 和 Wilkins 第三位。內容涵蓋 800 種與罕見疾病有關的資訊，分別由專精該項疾病的醫生與醫事人員撰寫，其中許多則是由發現該項疾病的醫生或研究人員所撰寫。參與的醫生與醫事人員合計超過 600 人，並由一個編審群負責校閱，訂價則是 90 美元。

三、 疾病簡介（Free Guides for Physicians）

NORD 也出版一系列的疾病簡介手冊，醫生與醫事人員可以要求負責提供，NORD 希望這項疾病手冊可以增進病患對於疾病的認識，並鼓勵病患早期診斷以獲得適當的轉介。這個疾病手冊系列已出版六冊，經費則是由與該項疾病有關的病患組織或有興趣提升該項疾病衛教的個人所捐助。

四、 「生物學名藥白皮書」(Generic Biological White Papers)

美國「孤兒藥」的市場中約有超過 20%係屬於生物(biological)製品；換言之，這些藥物係來自於生物的活體組織，當最先上市的「孤兒藥」生物製劑即將面臨專利保障到期之際，對於其他「學名藥」是否能以原廠藥在較低價格下維持相同的安全性與有效性，這項問題便變得十分重要。NORD 曾經針對這項主題在美國華盛頓特區舉行一場專家會議，討論與上述主題相關的科學、法律、規律、與經濟面向的問題，而這些討論便集結成這份白皮書，供各方免費索閱。

五、 「罕見疾病報導」(Orphan Diseases Update)

這是一份由 NORD 每年發行三期的報導雜誌，內容包括有與罕病相關最新的研究、倡導、社會事件、有助於照護者的建議與資源、相關網站、臨床實驗、與經費輔助的機會等。該報導合計為 16 頁的內

容，主要的發行對象為與罕病有關的社群、病患及家屬、醫生、護士、遺傳諮詢員、醫事人員、研究人員、教師、社工人員、政府官員及藥廠人員等。

六、 「NORD 年報」(NORD Annual Report)

該項年報列出 NORD 每年的工作重點、經費來源、經費支出情形，並每年出版，各界可以隨時索閱。

七、 「網路疾病簡介」(Rare Diseases Database)

NORD 同時搜錄各項罕見疾病的簡介，並置於網站中供民眾閱覽，或是供民眾來函索取。其建置內容涵蓋了 1,150 類疾病，包括每類疾病的症狀、類型、相關組織、與結合資訊等。

陸、美國罕病組織（NORD）的經費與支出

一、NORD 紀念花園方案（NORD’s Tribute Garden）

NORD 在網站中在設立「紀念花園」的空間，供民眾追思紀念過逝的家人或朋友，贊助人只要捐贈美金 100 元，即可在此處發表一頁紀念的文字及張貼照片。此外也設有訪客留言簿（Guestbook），只要捐款 25 美元或以上，即可瀏覽「紀念花園」中的「紀念章」，並在其中簽名留念致意。

二、其他捐款方式

民眾只要透過網路購物，如www.iGive.com這個網址，即可指定NORD為受贈單位，而NORD也將獲得一定比率的回饋；同時民眾在 2005 年年底以前加入這個購物網站為會員後 45 天內，只要有購物，NORD 即可再獲得美金 5 元的捐贈。

此外，NORD 亦獲 National Student Speech Language and Hearing Association(NSSLHA)選為 2005 年的受贈單位，該協會的”NSSLHA”募款活動，將動員所屬 300 個散佈在校園中的分會及 12000 名會員進行募款。自從 1990 年以來，NSSLHA 已募得超過 20 萬美元，用來資助服務溝通障礙的全國性組織。

三、NORD 的財務狀況

根據 Charity Navigator 組織對於 3100 個美國慈善團體的評鑑結果顯示，NORD 於 2004 年合計收入為 7.61 百萬美元，支出為 6.97 百萬美元，盈餘為 63 萬 9 千美元。而在所有的經費支出中，有 93.2%用於服務方案支出、4.7%為行政管理費用、2.1%為募款成本。其總裁 Abbey Megers 的年薪為 129.943 美元，佔該組織總支出的 1.86%。

柒、結語與建議

NORD 在 2002 和 2003 年，曾獲 Worth 雜誌選為美國最佳 100 大慈善團體之一，它之所以被選拔上的理由為，它能有效使用捐款且提供的服務真的改變環境，透過小規模但工作勤奮的人力，加上無數醫生的志願投入，使得 NORD 自 1983 年以來，即成為全美罕病最核心的資訊中心，提供病患、家屬與醫療照護者相關協助。在 Worth 雜誌針對 819,000 個非營利組織的評選過程中，NORD 被評選為美國前 100 最佳的非營利組織，在 2004 全年當中，合計有五百萬人次進入 NORD 的網站，也有 30 萬通電話與電子信件，而醫療協助方案也幫助數千個個案，顯示 NORD 為值得參考的罕病組織。

由於各國的環境、文化、醫療、與政策本就存在差異，因此並無存在罕病組織的經營典範可放諸四海皆準，但由於美國 NORD 組織創建於 1983 年，其運作歷史長久，且對於立法、政策、研究發展、與倡導等經驗皆有值得參考之處，因此透過檢視 NORD 的實質運作方式，也可以提供我國罕病基金會自我檢視發展的軌跡，以及學習可資參仿的創意方案設計。

就美國 NORD 與我國罕病基金會的運作相較，也可以發現彼此間存在許多相似之處，例如：

1、均由病患家長主導創立及經營：美國 NORD 係由病患家長 Abbey Meyers 女士創建，並擔任總裁負責經營至今；我國罕病基金會則由病患家長陳莉茵、曾敏傑於 1999 年創立，並由曾敏傑擔任創會後執行長至 2005 年 3 月止。

2、均重視立法與政策倡導：NORD 於 1983 年推動美國「孤兒藥」法案的立法，並長期監測與推動和罕病有關的政策發展，此點與我國罕病基金會亦極為類似，如罕病基金會於 2000 年推動「罕見疾病防治及藥物法」的立法通過，而隨後也修改「身心障礙者保護法」納入罕病，及推動全民健保對於罕病的專案專款保障等，同時也在衛生署罕病藥物審議委員會中積極參與，均在立法與政策施行層次上推動罕病的保障與發展。

3、均提供罕病跨病類的推動：美國 NORD 組織為「聯盟」的運作模式，其會員除以個人名義參加以外，亦容納團體會員參與，而會務實質運作內容亦包容跨病類的推動；我國罕病基金會的運作型式為「基金會」，而非「會員組織」的型式，但實質運作內容則亦著重跨病類的服務及推廣。

4、均與政府維持既倡導又合作的關係：「倡導」強調改變法律、政策、制度與規範等，NORD 的倡導工作即展現試圖改變現狀的嘗試，例如 NORD 即已提前呼籲如何促進罕病學名藥的安全與普及，推動

在孤兒藥保障到期後，加速其普及且又能兼顧安全性；此外，NORD也與政府部門維持合作的關係，在每年的年會中，均可發現政府相關主管單位密切的參與其中。我國罕病基金會係以「倡導」的角色出發，如推動立法、遺傳諮詢師認證、二代新生兒篩檢等，均強調改革現狀的企圖，但在相關方案的合作上，如國際檢體外送服務、國內檢體確診制度建置、罕病宣導等議題上，則與政府亦維持合作的關係。

5、均與醫界維持緊密的友善合作：從NORD所提供的運作內容顯示，從其榮譽頒贈、出版品提供、協尋病患參與研究計畫、提供小額醫師研究經費等，均可發現NORD將對於醫界的合作與服務列為極高優先，其投入的資源與服務尚且超過對於病患的服務之上；我國罕病基金會在運作上亦與醫界維持緊密的友善合作，但服務對象的主體仍以病患及家屬為主，醫界人士則為輔，但合作關係則十分密切。

6、均為受肯定且具責信的非營利組織：美國NORD曾在2002及2003年獲Worth雜誌選為全美前100大慈善團體之列，同時也受到Charity Navigator獨立組織評定為四顆星（exceptional）的評價；我國罕病基金會除獲衛生署頒發有功於罕病藥物引進以外，亦於2002年獲「中華民國公共關係基金會」評選為「最佳非營利

組織公共服務獎」，顯示兩者均為普受肯定且具責信的非營利團體。

美國 NORD 組織雖然與我國罕病基金會有諸多的相似性，但是兩者間也仍在差異性，例如：

1、 美國 NORD 組織似乎較著重間接服務，而我國罕病基金會的業務中，直接服務比重日漸增加，無論是人力配置與經費投入，直接服務應已凌駕於間接服務之上。

2、 美國 NORD 組織對於醫事社群的服務似乎更甚於對病患的服務，而相反的，我國罕病基金會應以病患服務為重點，雖然檢體外送、遺傳諮詢員配置、二代新生兒篩檢的推廣，及部份研究經費提供等，仍以協助醫事人員為對象，但就方案規模、人力配置、及經費使用而言，應仍不及對於病患的投入。

3、 美國 NORD 的出版品雖然較少，但卻更具學術性，例如 Resource Guide 或專業參考手冊等;而我國罕病基金會的出版品雖然較為多元而豐富，例如營養手冊、照護手冊、罕病單張等，但由於使用對象係著重在病患與民眾，因此較具通俗性與社會教育性。

4、 美國 NORD 組織的經費規模顯著高於我國的罕病基金會。以 2004 年為例，NORD 的經費全年已達 7.6 百萬美元，我國罕病基金會則

約達1.5百萬美元，兩者的經費規模仍有顯著差異。但由於NORD早於1983年時創建，而我國罕病基金會則於1999年時創立，成立至今僅約六年，能有此成績，也屬不易。

透過對於美國NORD運作的檢視，仍可發現可資我國罕病基金會參考的方向，我國罕病基金會雖然在病患直接服務的內容上較NORD為廣泛且完整，但NORD的特色也有可資罕病基金會進一步思考之處，例如：

1、如何加強與學術界的合作：從美國NORD所出版的手冊、教科書、疾病簡介、網路疾病資料庫等，都顯示NORD與學術界的合作極為深入與專業，未來罕病基金會也應考慮如何強化與學術界的合作，透過資料的彙整、調查、分析、建置資料庫等方式，運用學術工具與人力，深化對於罕病議題的掌握。

2、強化與醫界的合作：罕病基金會雖然長期以來與醫界維持友善互動，但NORD的運作經驗顯示，其對於新進醫師提供獎學金、小額先驅性研究經費補助、舉辦年會及頒贈榮譽等，也都是未來可考慮的新增方案。務實而言，若短期有承載上的困難，罕病基金會仍可透過與學術性團體合作的方式來協力進行，例如人類遺傳學會、生命教育協會、生命倫理協會、社會工作學會等。

3、 重要文獻的彙整與建置：針對國內有關罕病的重要研究成果、政策宣言、首長談話、重要新聞與專文等，也可參仿 NORD 的方式，加以搜錄建置，使罕病基金會成為建置與管理罕病重要資訊的平台。

4、 思考前瞻性的倡導議題：以 NORD 為例，其已開始專注罕病學名藥的普及與安全性問題；對於基因科技的創新、遺傳倫理的討論、孤兒藥研發的掌握、保險歧視、產前診斷的推動、身心障礙定義的修正、二代健保政策的走向等本土性未來議題，罕病基金會均需有更前瞻性的預先涉入，以維持在倡導上的前瞻性與主動性。