

Newborn Screening: Claiming Victory Then and Now

October 20, 2010

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Newborn Screening and Genetics Resource Center

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Jana Monaco, BS

Parent Advocate, Organic Acidemia Association

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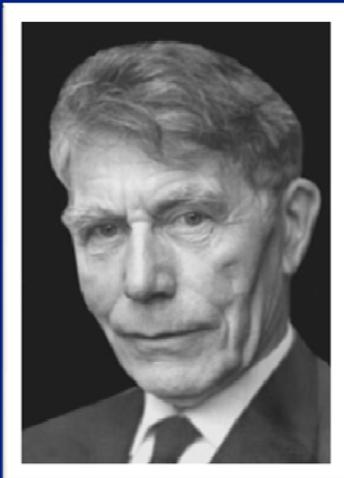


Newborn Screening History

The Early Days



- 1902 – Garrod - Originated the phrase “Inborn Error of Metabolism”



- 1934 – Følling - Identified PKU as an inborn error associated with mental retardation - diaper test - ferric chloride - presence of phenylpyruvic acid in urine

Newborn Screening

History of PKU

- ◆ 1930's: dietary treatment was proposed
- ◆ 1950's: dietary treatment became available
 - greatest cognitive improvement seen in youngest patients



Fig 19. Contrast—untreated and treated phenylketonurics. The 11-year-old boy is severely retarded, whereas his 2½-year-old sister, diagnosed in early infancy and promptly treated with the mind-saving diet, is normal.¹⁷

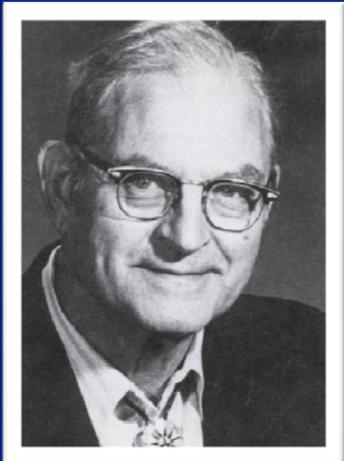
Pediatrics, 105:89, 2000.

Newborn Screening History

The Early Days



➤ 1953 - Bickel - PKU dietary treatment - Published the results of dietary therapy and formula treatment developed by himself, Evelyn Hickmans, John Gerrad, and Louis Woolf in the medical journal Lancet.

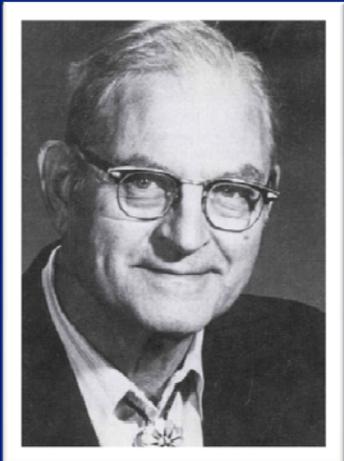


➤ 1959 - Guthrie - Filter paper test for PKU - Developed a simple, inexpensive test with blood on filter paper for early detection of PKU (and other disorders). Early detection and treatment prevents the harmful effects of PKU.

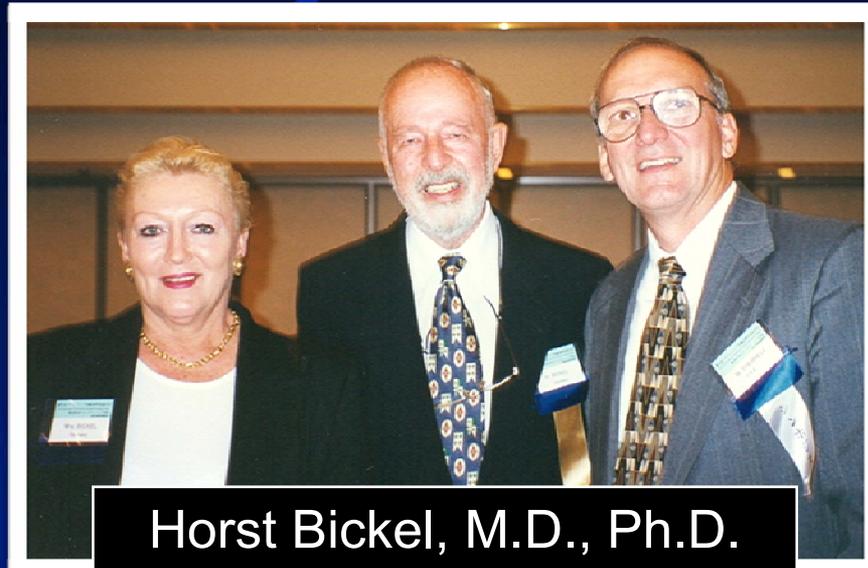
Newborn Screening History



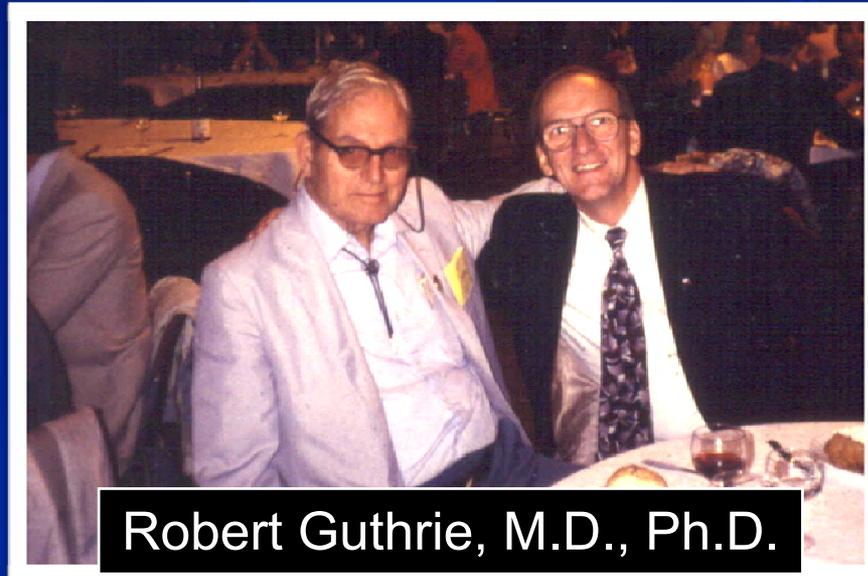
1918-2000



1916-1995

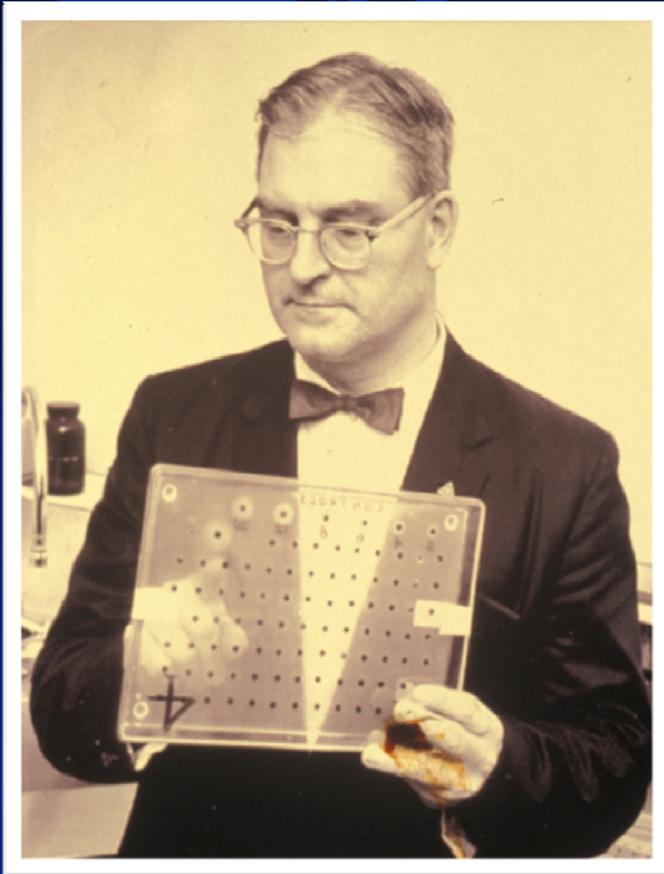


Horst Bickel, M.D., Ph.D.



Robert Guthrie, M.D., Ph.D.

Newborn Screening History



1962 – Children’s Bureau funded study on efficacy of newborn screening for PKU

1962 – Children’s Bureau provided funding support for voluntary newborn screening program at MA Department of Health

1963 – Guthrie – Reported his bioassay for PKU using dried blood collected on filter paper

First Joseph P. Kennedy International Award in Mental Retardation – Dec. 6, 1962

Source: Centerwall and Centerwall: Pediatrics 2000; 105: 101.



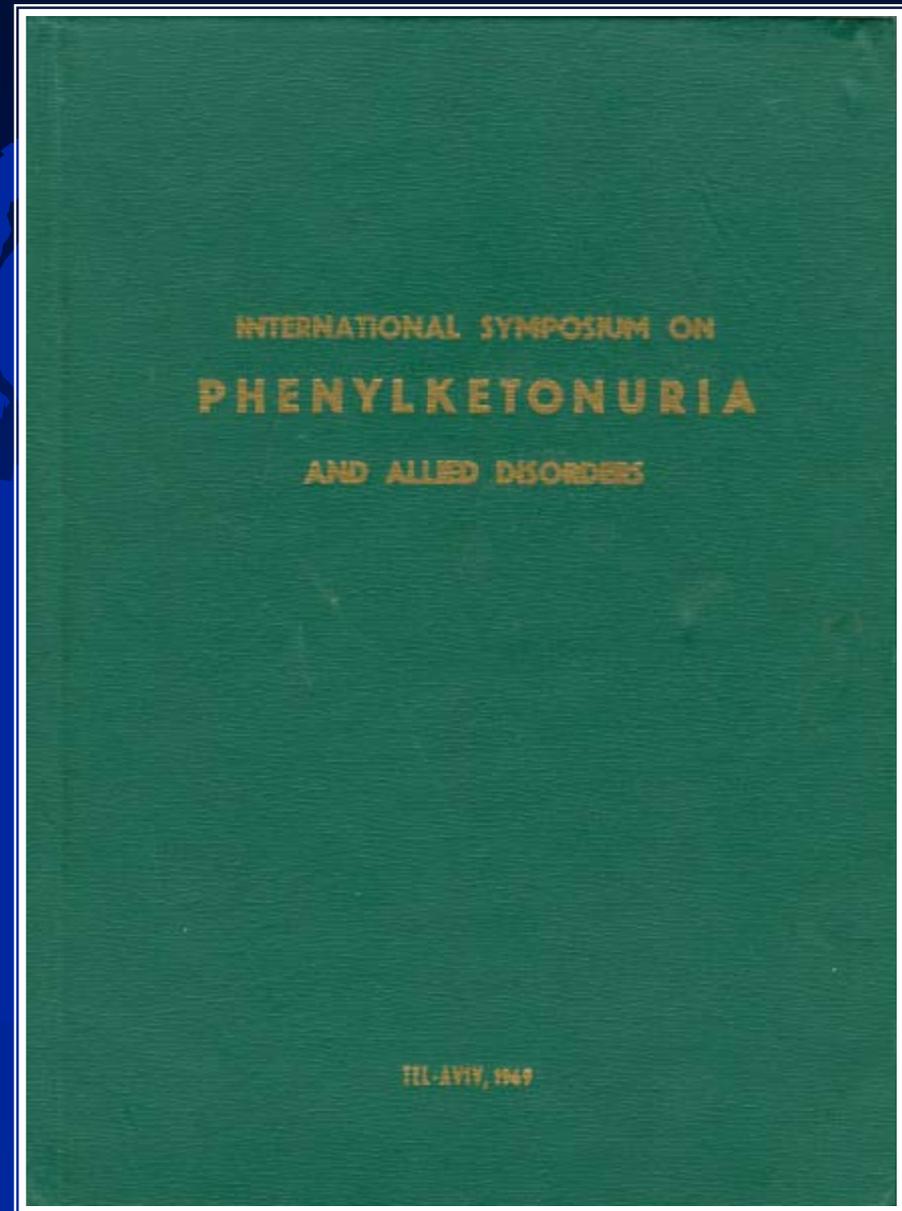
- Dr Murray L. Barr (Barr-Body) in cytogenetics (Canada)
- Dr Samuel A. Kirk, in special education of the mentally retarded (US)
- John Fittinger, President of the US National Association for Retarded Citizens
- John F. Kennedy, President of the United States
- Dr Ivar Asbjörn Fölling, discoverer of PKU (Norway)
- Dr Jerome Lejeune, discovered chromosome abnormality in Down syndrome (France)
- Dr Joe Hin Tjio, co-discoverer of correct chromosome count in humans (Indonesia).

**1966 - 1st Conference on Inborn
Errors of Metabolism –
Dubrovnik, Yugoslavia**

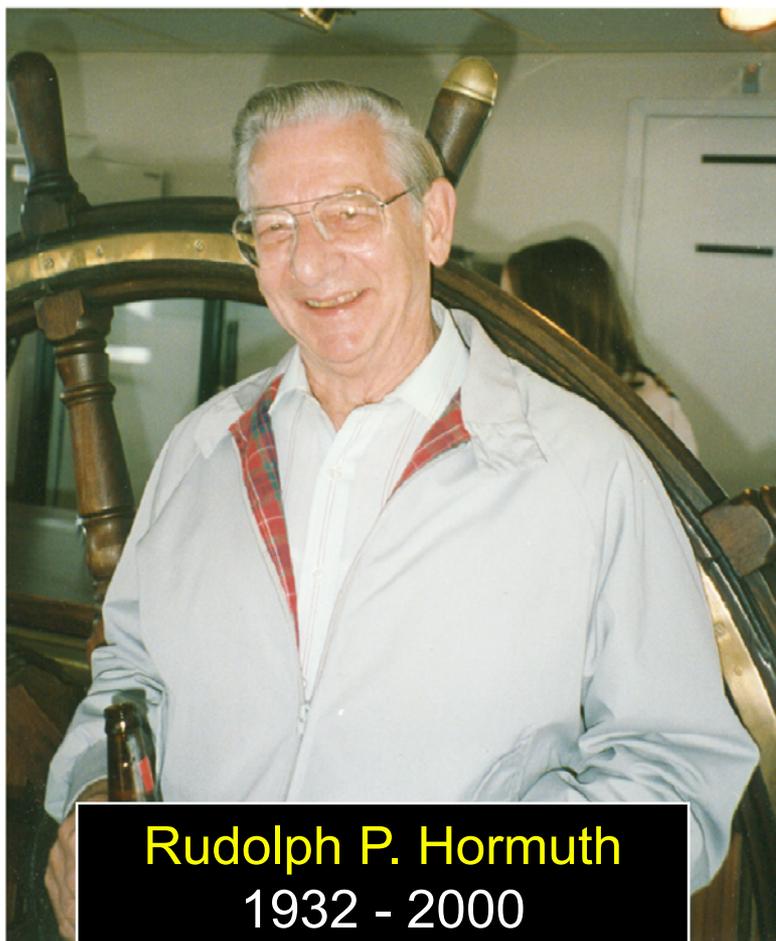
Sponsored by Children's
Bureau, US Dept. of Health,
Education, & Welfare
13 Different Countries

**1969 - International Symposium
on Phenylketonuria and Allied
Disorders – Tel Aviv, Israel**

Sponsored by Maternal and
Child Health Service, US Dept.
of Health, Education, & Welfare
– Special Thanks to Rudolph P.
Hormuth



HRSA – Early and Continuous Support of Newborn Screening



Rudolph P. Hormuth
1932 - 2000



Special Recognition - APHL
Seattle, WA - June 1994

Review

Newborn Screening History

1960's

- 1959 – First PKU screening report - letter
- 1961 – First DBS newborn screening – Erie and Niagara Counties – First case in Niagara after 800 screens
- 1963 – Guthrie Publication on DBS PKU Test
- 1966 – 1st International meeting -Dubrovnik
- 1966 – First Asian program (Japan - PKU)
- 1968 – Guthrie Develops Other Screens
- 1969 – 2nd International meeting - Tel Aviv

Bob Phillips

Horst Bickel



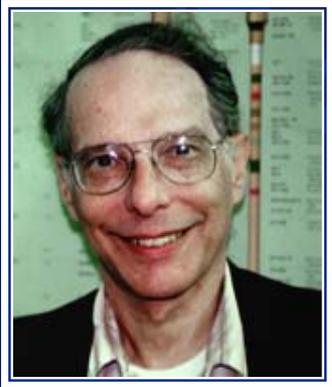
Punching Blood Spots



Newborn Screening History



- 1973 - *Dussault -Thyroxine (T4) determination in dried blood by radioimmunoassay: a screening method for neonatal hypothyroidism. Union Med Can 1973;102:2062-4.*



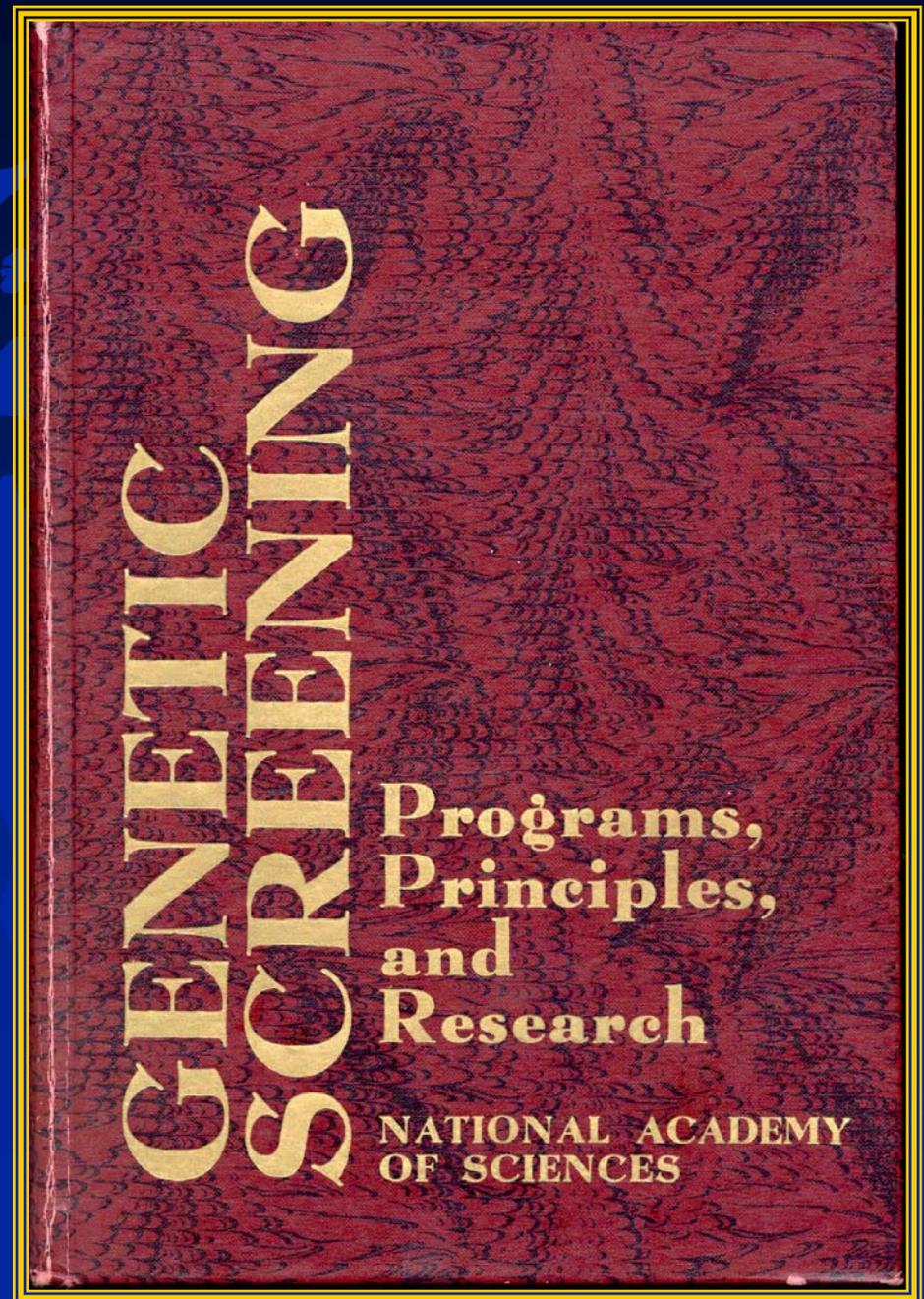
- 1973 - *Garrick - Sickle cell anemia and other hemoglobinopathies: procedures and strategy for screening spots of blood on filter paper as specimens. N Engl J Med 1973; 288:1265-8.*



- 1977 - *Pang - Micro filter paper method for 17-hydroxy-progesterone radioimmunoassay: its application for rapid screening for congenital adrenal hyperplasia. J Clin Endocrin Metab 1977;45:1003-8.*

1975 National Academy of Sciences Report

To: review current practices, identify problems, give guidance and maximize effectiveness of screening programs.



Role for CDC in Quality Assurance of Newborn Screening

1975 - The Committee for the Study of Inborn Errors of Metabolism, National Academy of Sciences noted that greater quality control of PKU screening was essential and recommended that:

“a single laboratory -- within CDC, for instance -- should be responsible for maintaining the proficiency of the regional laboratories.”

[Source: Genetic Screening – Programs, Principles, and Research, National Academy of Sciences, Washington, D.C. 1975]

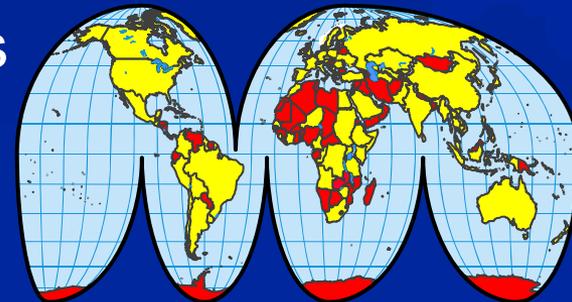
Newborn Screening Laboratory Proficiency Testing

1979 – Memorandum of Understanding signed between HRSA (Dr. Jane Lin-Fu) and CDC (Dr. Harry Hannon) provided funds to establish a newborn screening laboratory proficiency testing program at the CDC.

2008 – Celebrated 30th Anniversary
(Now funded by CDC)



2010 – Serves over 475 laboratories in over 75 countries, over 50 analytes



— Participating countries

1979 - Letter from Dr. Guthrie Assistance offered for developing QA program at CDC

State University of New York at Buffalo



Dr. Harry Hannon

5 January 1979

Dear Dr. Hannon,

I was delighted to receive your call yesterday and to learn that the CDC hopes to begin newborn genetic screening programs. I have waited a long time for this good news from the CDC.

.....

Very sincerely yours,
Robert Guthrie, Ph.D., M.D.

many other programs. We send this report to all the laboratories that contribute data to us. These laboratories are only the ones using our other tests, not the PKU test alone.

1st U.S. National Disease Specific Newborn Screening Meeting

Congenital Hypothyroidism

Atlanta, GA
August 1979



TEST CENTER
for
INFANT HYPOTHYROID METABOLISM

HYPOTHYROID GYU GAI MSUD
 OTHER _____

OR No. 150501

Infant's Name: _____ (Last)

Birth Date: _____ Year _____

Baby's Present Weight: _____ lbs Oz

Baby Feeding: Well Poorly
 Breast Bottle Both

Baby's Doctor: _____

Address: _____ Tel. # _____

COMPLETELY FILL ALL CIRCLES WITH BLOOD
SOAK THROUGH FROM REVERSE SIDE

○ ○ ○ ○

Proceedings of a Conference on a National Model for
**STANDARDIZATION OF
NEONATAL HYPOTHYROID
SCREENING PROGRAMS**

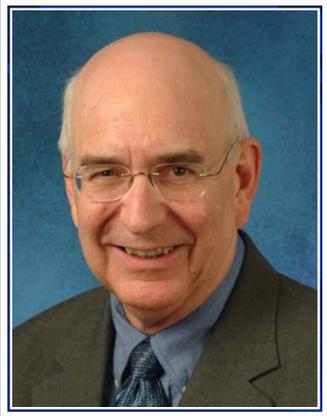
U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES • Public Health Service • Center for Disease Control

Newborn Screening History

1980's

- 1980 – 1st uses of microcomputers in NBS (TX, NY)
- 1981 – 1st National NBS Meeting – Austin, TX
- 1982 – 1st combined metabolic/hypothyroid international meeting (5th International Newborn Screening Meeting - Tokyo, Japan)
- 1986 – 1st formal discussion of international quality assurance (6th international meeting - Austin, TX)
- 1987 – 1st international quality assurance meeting - Nikko, Japan
- 1988 - International Society for Neonatal Screening formed

Brief Newborn Screening History - DNA

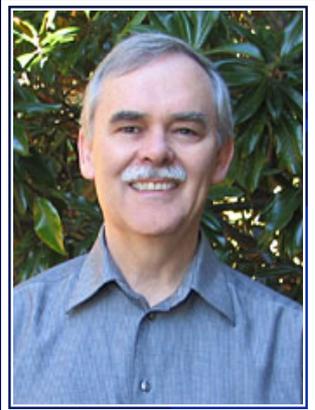


1987 - McCabe - *DNA microextraction from dried blood spots on filter paper blotters: potential applications to newborn screening.*
Hum Genet 75:213-216.



1993 – Farrell - *Application of DNA analysis in a population-screening program for neonatal diagnosis of cystic fibrosis (CF): comparison of screening protocols.*
Am J Hum Genet 52:616-26.

Brief Newborn Screening History – MS/MS



➤ 1990 - Millington - *Tandem mass spectrometry: a new method for acylcarnitine profiling with potential for neonatal screening for inborn errors of metabolism.* J Inher Metabol Dis 13:321-324.



➤ 1993 - Chace - *Rapid diagnosis of phenylketonuria by quantitative analysis for phenylketonuria and tyrosine in neonatal blood spots by tandem mass spectrometry.* Clin Chem 39:66-71.



➤ 1995 - Rashed - *Diagnosis of inborn errors of metabolism from blood spots by acylcarnitines and amino acids profiling using automated electrospray tandem mass spectrometry.* Pediatr Res 38:324-31.

The 1990s Began a Paradigm Shift: “Technology Driven”

Previously: 1 Test, 1 Disorder

Now: Multiplex Assays

1 Test, Many Disorders

Hemoglobins: Isoelectric Focusing, HPLC

Metabolics: Tandem Mass Spectrometry

Lanetta Jordan, MD, MPH, MSPH

Chief Medical Officer,
Sickle Cell Disease Association of America

Director, Sickle Cell Services,
Memorial Healthcare System, Hollywood, FL

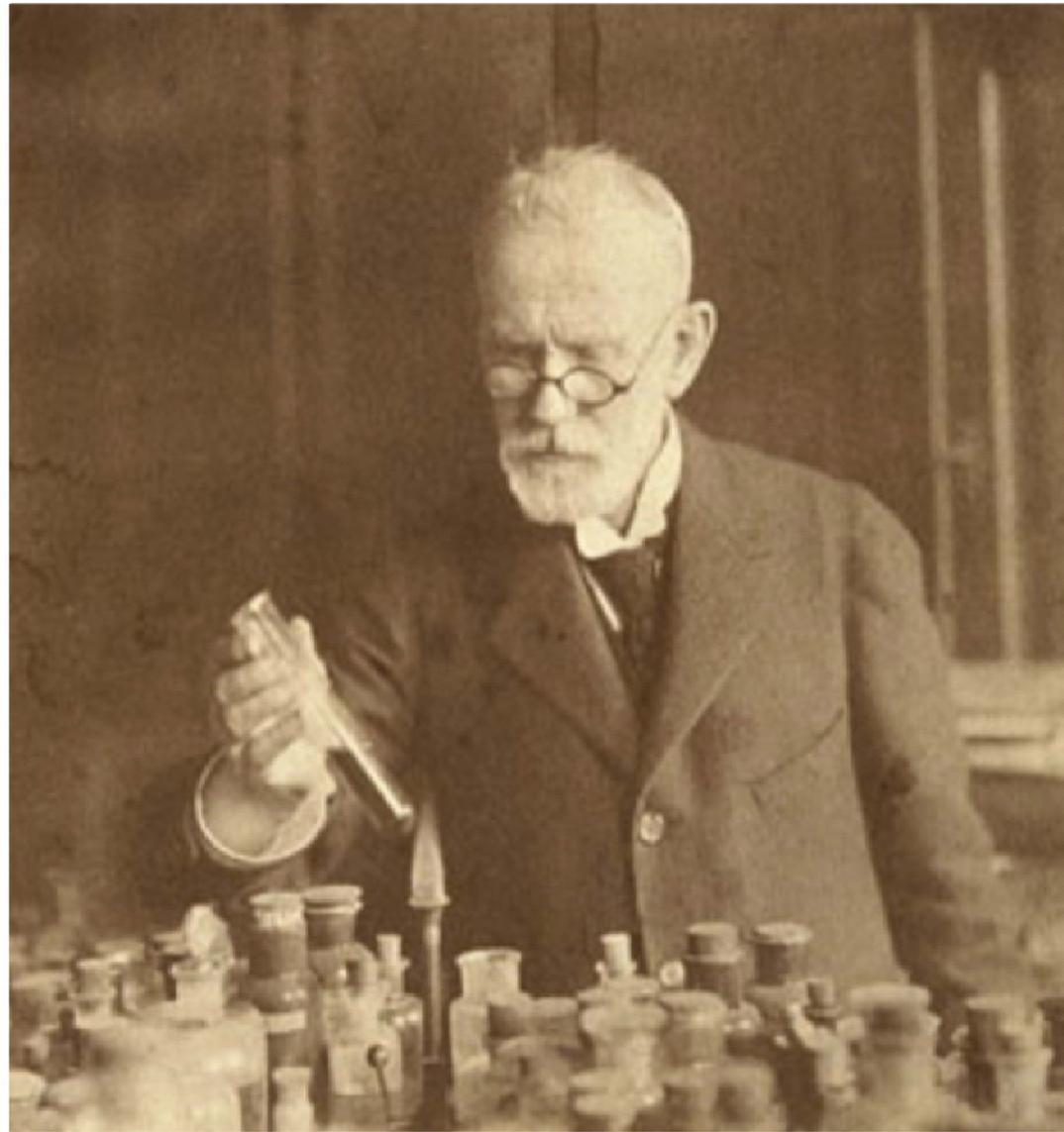


Sickle Cell Disease: 100 Years in America



“transformation from a disease of slaves to an immigrant disease to an American public health challenge”

James B. Herrick, M.D.
(1861-1954)



First Clinical Description 100 Years (1910 – 2010)

“... unusual blood findings, no duplicate of which I have ever seen described. Whether the blood picture represents merely a freakish poikilocytosis or is dependent on some peculiar physical or chemical condition of the blood, or is characteristic of some particular disease, I cannot at present answer”

James Herrick, M.D.,
Chicago, 1910

PECULIAR ELONGATED AND SICKLE-SHAPED RED BLOOD CORPUSCLES IN A CASE OF SEVERE ANEMIA

JAMES B. HERRICK, M.D.

CHICAGO

This case is reported because of the unusual blood findings, no duplicate of which I have ever seen described. Whether the blood picture represents merely a freakish poikilocytosis or is dependent on some peculiar physical or chemical condition of the blood, or is characteristic of some particular disease, I cannot at present answer. I report some details that may seem non-essential, thinking that if a similar blood condition is found in some other case a comparison of clinical conditions may help in solving the problem.

History.—The patient was an intelligent negro of 20, who had been in the United States three months, during which time he was a student in one of the professional schools in Chicago. His former residence had been Grenada, West Indies, where he had been born and brought up, one of a family of four children, all living, and all well with the exception of himself. His mother was living and in good health; his father had died of accident. At the age of 10 the patient had had yaws. This was a common disease in the locality where he lived. The lesions, as he described them, had been pustular, with formation of ulcers and scabs. On healing, scars, many of which he pointed out, were left. Some of the ulcers had been as large as a silver quarter of a dollar. The disease lasted about one year and during this time he had felt somewhat weak and indisposed. Most of the ulcers had been on the legs and the patient himself had thought that this location of the lesions might have been due to the bruises and scratches that were frequently produced as he ran about, a barefoot boy, through the streets and the brush. He was sure he had never had ground-itch, though he said it was not uncommon in Grenada. He had attended school up to the age of 17. Since leaving school, that is, for the past three years, he had felt a disinclination to take exercise. For about a year he had noticed some palpitation and shortness of breath which he had attributed to excessive smoking. There had been times when he thought he was bilious and when the whites of the eyes had been tinged with yellow. At such times he had not had any pain, chill or fever. Three years previously he had had a purulent discharge from the right ear lasting six months. He had had no diarrheas and no hemorrhages at any time. He denied syphilis and gonorrhoea. There was never any rheumatism or other joint trouble. On landing in New York in September, 1904, he had a sore on one ankle for which he consulted a physician. Tincture of iodine was applied and in a week the sore had healed, leaving a scar similar to the others on the limbs. For the past five weeks he had been coughing. Two days prior to examination he had “taken cold,” his cough had grown worse and he had had a slight chill, followed by fever. It was this cough and fever for which he wished treatment at the hospital, and of which he chiefly complained, though he mentioned also that he felt weak and dizzy, had headache and catarrh of the nose.

“The unusual character of the blood findings in the case here recorded is alone sufficient to justify its report. When, however, we compare these findings, as well as the clinical history, with two similar cases previously reported, one by James B. Herrick, the other by R.E. Washburn, we are forced to the conclusion that we have in these three cases a group which belongs quite apart from anything heretofore described...these three cases have points of resemblance so characteristic and so constant that they cannot be explained as accidental”

James E. Cook, M.D. and
Jerome Meyer, M.D.
St. Louis, 1915

SEVERE ANEMIA WITH REMARKABLE ELONGATED
AND SICKLE-SHAPED RED BLOOD CELLS
AND CHRONIC LEG ULCER*

JEROME E. COOK, M.D., AND JEROME MEYER, M.D.
ST. LOUIS

The unusual character of the blood findings in the case here recorded is alone sufficient to justify its report. When, however, we compare these findings, as well as the clinical history, with two similar cases previously reported, one by James B. Herrick, the other by R. E. Washburn, we are forced to the conclusion that we have in these three cases a group which belongs quite apart from anything heretofore described. We desire to present the history and findings in our own case, with the abstracts of the cases of Herrick and Washburn, and to show that the three have points of resemblance so characteristic and so constant that they cannot be explained as accidental. Our thanks are due to the above-mentioned authors for the opportunity of examining blood smears of their cases and for other courtesies.

AUTHORS' CASE.—Personal History.—O. B., a mulatto woman, aged 21, entered the Washington University Hospital, Nov. 3, 1914, giving the following history: She was born in St. Louis and had never been out of its immediate vicinity. She had had most of the diseases of childhood, and suffered at intervals, ever since she could remember, with “rheumatic” pains; the joints had never been swollen. Shortly after her marriage, two years ago, she developed “pus-tubes,” which were removed a year later. At the age of 5 an ulcer broke out on her ankle, which healed in about a month, under treatment. This did not recur until two years ago when a similar condition developed which lasted three or four months and was healed with scarlet-red ointment and bichlorid packs. Ten months before entering the hospital the ulcer again appeared. It caused her no pain and has been treated in various ways.

Physical Examination.—Nov. 3, 1914. The patient was a light colored mulatto, with evidence of some loss of weight, conjunctivae and mucous membranes rather pale. The conjunctivae had a greenish tint. The rest of the physical examination, with the exception of the leg condition to be described, was not significant. There was a soft systolic murmur at the apex, transmitted to the left axilla, and heard over the left precordial area, the second pulmonic was louder than the second aortic and definitely accentuated. The systolic blood pressure varied from 105 to 125, the diastolic between 60 and 65. The lungs were clear, the spleen not palpable. On the left ankle, just above the internal malleolus was an ulcer 3 by 2 cm., the edges slightly indurated, the whole being of a light pink color. On the outer side of the ankle there was a scar 7 cm. by 3 cm. The skin in this region was dark and shiny.

* Submitted for publication June 17, 1915.

* From the Department of Internal Medicine, Washington University Medical School.

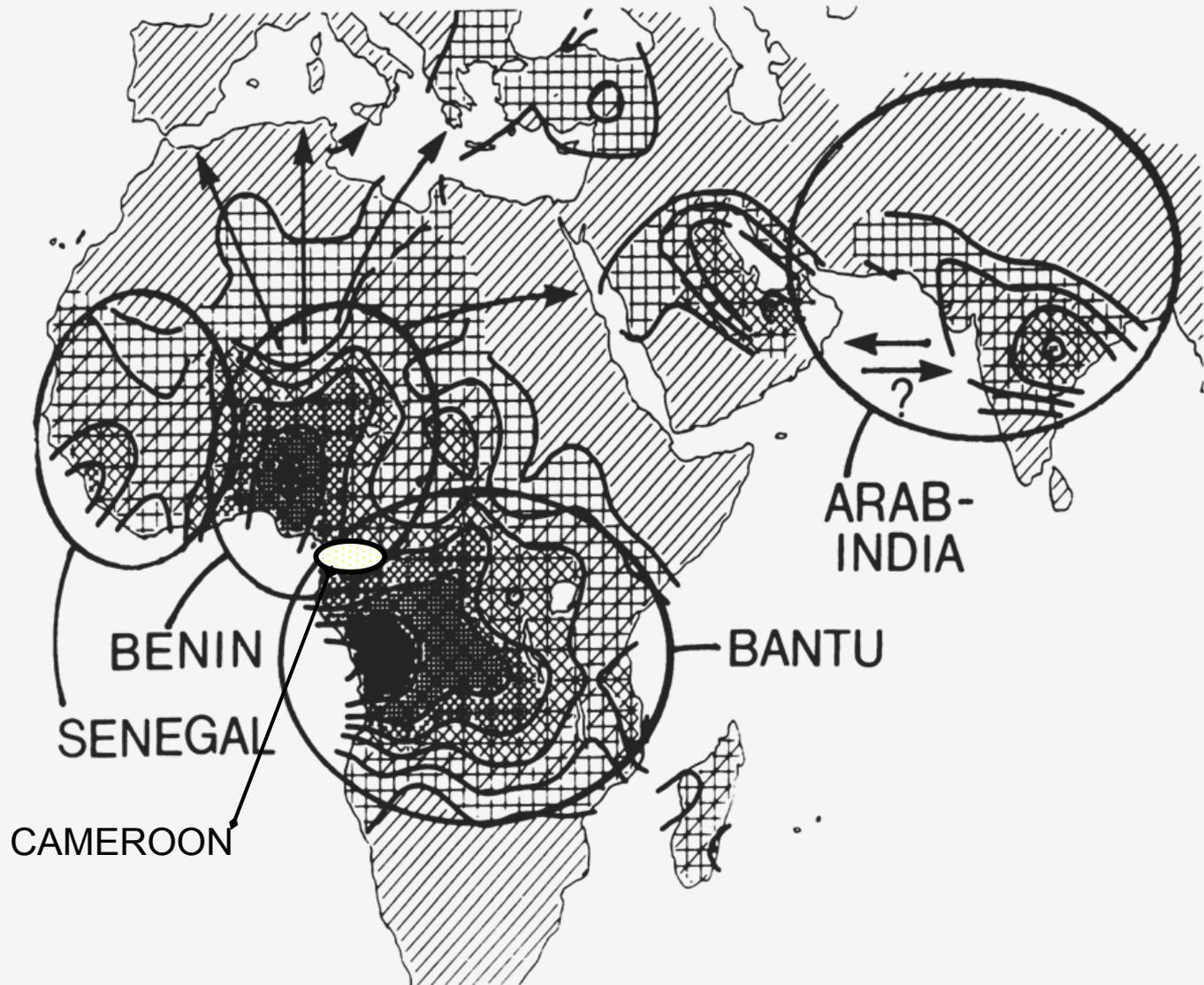
Sickle Cell Disease: 100 Years in America

Sickle Cell Identity

“The most significant feature of sickle cell anemia is not its characteristic bizarre deformation of erythrocytes but the fact that it is apparently the only known disease completely confined to a single race”

Journal of the American Medical Association, 1947

Multicentric Origins of the Sickle Gene



U.S. Newborn Screening Data Ranked by Apparent Incidence



SCD-SS + SC	1:2,474
Primary Congenital Hypothyroidism	1:3,044
Cystic Fibrosis	1:3,924
Toxoplasmosis	1:10,415
Clinically Significant Hyperphenylalaninemia	1:13,947
Classical CAH	1:18,987
Classical Galactosemia	1:53,261
Biotinidase	1:61,319
MSUD	1:230,028
Homocystinuria	1:343,650

Life Expectancy of individuals born with Sickle Cell Disease

☞ Life expectancy - from 14 yrs (1973) to 40-45 yrs in 2000¹.

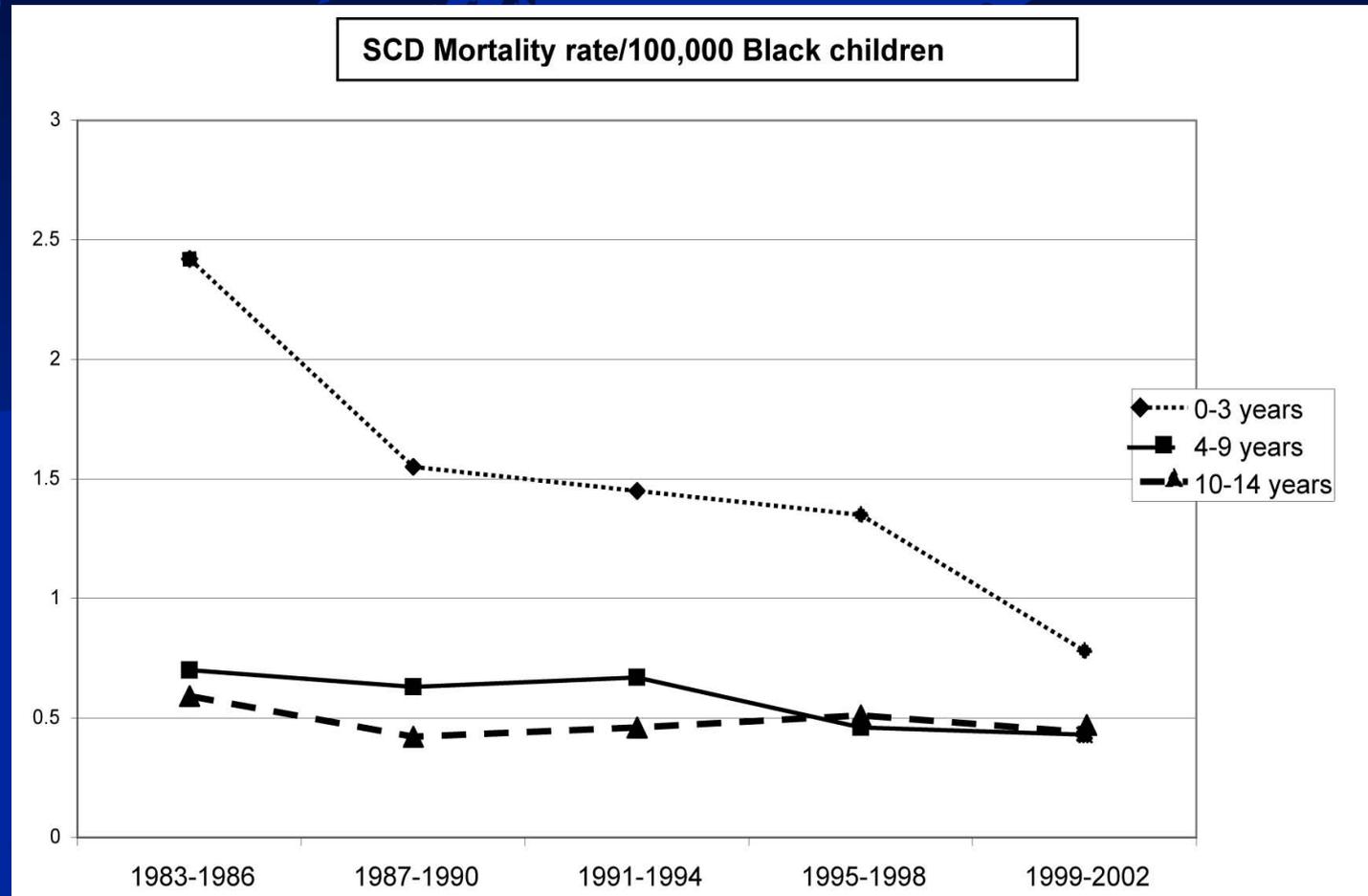
– Treatments Interventions (After 100 Years)

1. Hydroxyurea – only FDA approved medication for the treatment of SS and S β thal^o (underutilized)
2. Transfusions – Acute, Chronic, Episodic
3. Chelation

– Universal Newborn Screening, Penicillin prophylaxis

Impact of PCV7 Vaccine on SCD-related Child Mortality

- ◆ In 2000, PCV7 recommended by CDC for all US children <2 years of age
- ◆ SCD-related deaths among children <4 fell 42%



Advances in Identification and Treatment

1. Newborn screening followed by penicillin prophylaxis and anti-pneumococcal vaccination
2. Modern pain management
3. Stroke prevention strategies: TCD, MRI and MRA
4. Regular red cell transfusion service
5. Hydroxyurea therapy
6. Neuropsychological dysfunction - diagnosis and management
7. Hematopoietic stem cell transplantation
8. Premarital, pre-pregnancy, pre-natal testing, and counseling



The **First** Identified Genetic Disease

Impetus for the Human Genome Project

SCD Complications

Serious and Complex Clinical Conditions

- Hyperhemolysis¹
- Vascular occlusion¹
- Chronic heart failure²
- Primary and recurrent stroke¹
- ACS¹
- Pulmonary hypertension¹
- Osteomyelitis¹
- Osteonecrosis¹
- Splenic infarct and/or sequestration¹
- Papillary necrosis¹
- Renal failure¹
- Severe recurrent pain²
- Complicated pregnancy³

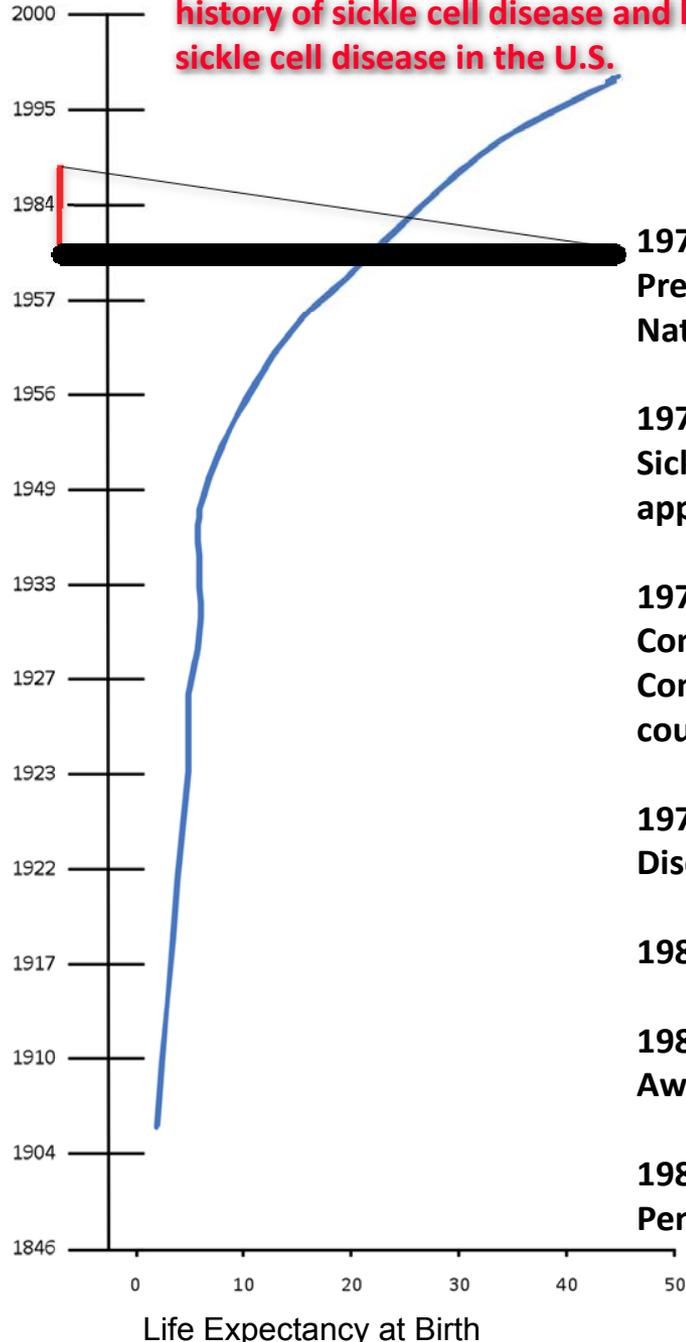
ACS, acute chest syndrome.

1. Lonergan GJ et al. *Radiographics*. 2001;21:971-994. 2. Ohene-Frempong K. *Semin Hematol*. 2001;38(suppl 1):5-13.

3. Josephson CD et al. *Transfus Med Rev*. 2007;21:118-133

Timeline of major scientific, health services and community events in the history of sickle cell disease and life expectancy at birth of persons with sickle cell disease in the U.S.

FDA approves Hydroxyurea



Marotta Confirms Mutation

Ingram suggests substitution of valine for glutamic acid

Ingram demonstrates Hb S

Pauling shows electrophoretic difference between Hb A & S

Diggs distinguishes symptomatic from asymptomatic cases

Hahn & Gillespie identify low oxygen as cause of sickling

Taliaferro & Huck perform pedigree studies

First use of term "Sickle cell anemia"

Emmel demonstrates in vitro sickling

Herrick first published description

Ernest Irons sketched sickled cells

Article "Case of Absence of the Spleen"

1971 – 1987

1971 – African American Community Pressured Congress to make SCD a priority National Association for Sickle Cell Disease

1972 – Sickle Cell Control Act & National Sickle Cell Disease Program (NIH appropriated funds to HRSA)

1978 – National Genetic Disease Act, Comprehensive Sickle Cell Centers, Community Based SCD education, screening, counseling services

1978 – Cooperative Study of Sickle Cell Disease

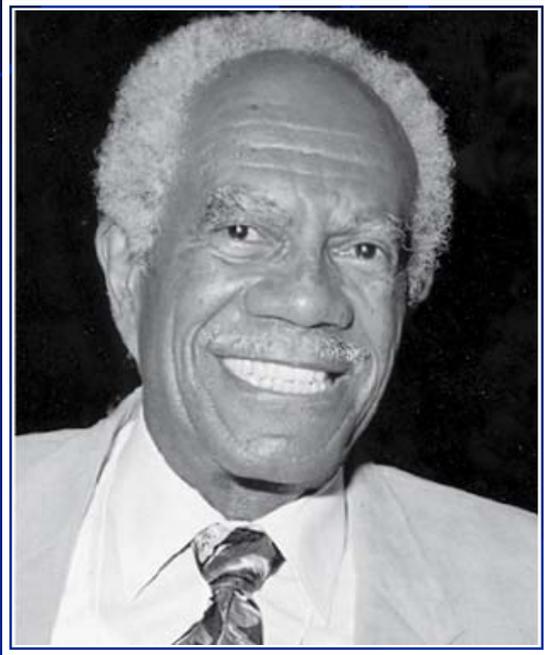
1983 – Prophylactic Penicillin (PROPS I)

1983 – September- National Sickle Cell Awareness Month

1987 – Universal Screening and Prophylactic Penicillin

Life Expectancy at Birth

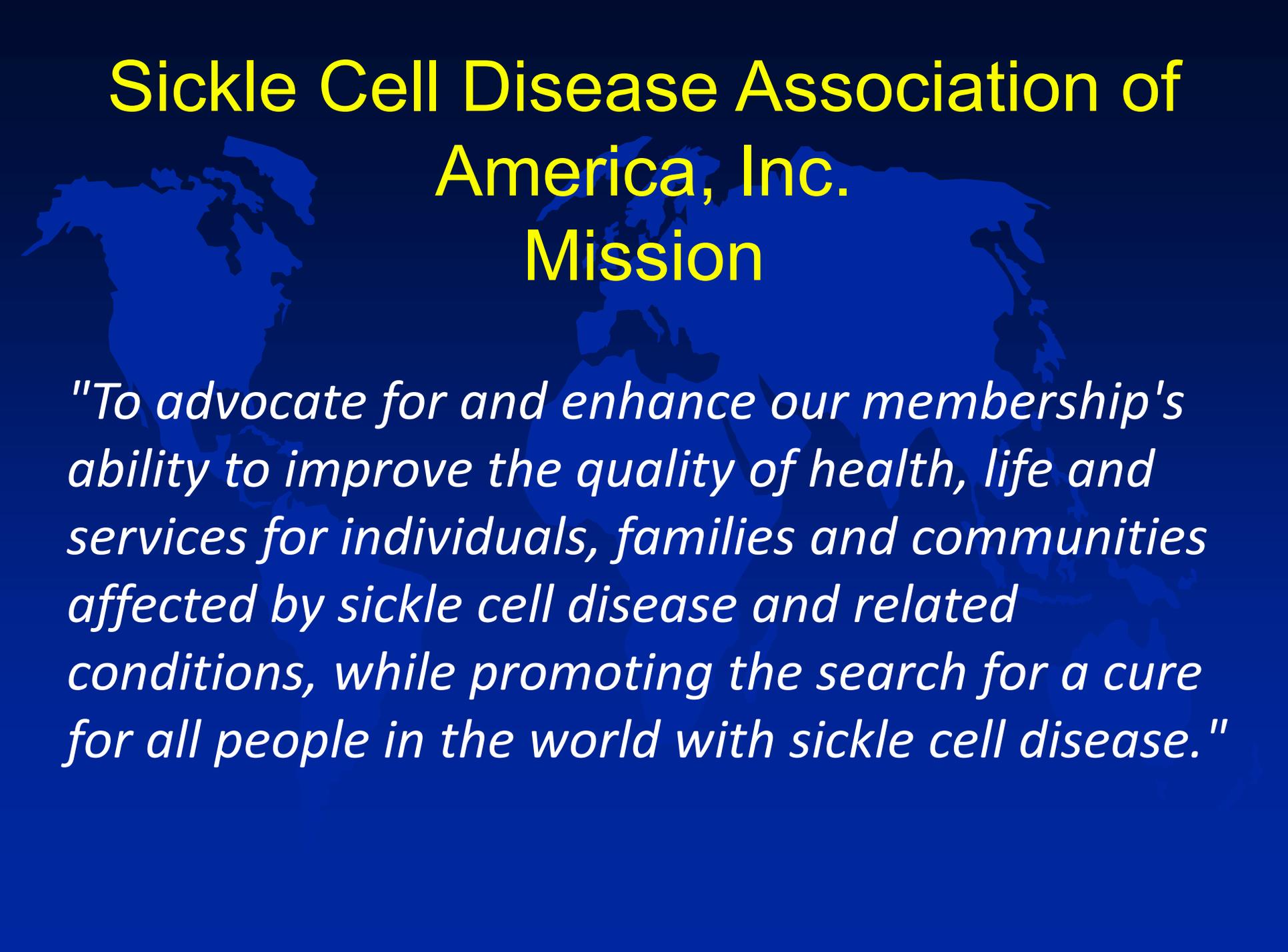
National Sickle Cell Disease Advocacy Organization - formed in 1972



Charles F. Whitten, M.D.
1922-2008
Co-Founder

1971 - African American Community Pressured Congress to make SCD a priority. Vision of a national coordinated approach to address issues related to sickle cell disease was unveiled when representatives of 15 community sickle cell organizations met at “Wingspread,” a Racine, Wisconsin conference center, as guest of Johnson Foundation. Out of that meeting, the **National Association for Sickle Cell Disease** was founded.

1994 – The National Association for Sickle Cell Disease’s name was changed to the **Sickle Cell Disease Association of America, Inc.**



Sickle Cell Disease Association of America, Inc.

Mission

"To advocate for and enhance our membership's ability to improve the quality of health, life and services for individuals, families and communities affected by sickle cell disease and related conditions, while promoting the search for a cure for all people in the world with sickle cell disease."

Public Advocacy

Highest Level of Awareness about
the issue in the Community

Advocacy

1. 50 SCDAAs Member Organizations in 28 States
2. Provide Social Support, Community Awareness, Genetic Counseling, Screening
3. Hill Day
4. Legislative Portfolio – HRSA, NIH/NHLBI, CDC
5. Partner with Sickle Cell Treatment Centers
6. Counseling and Certification Programs
7. Annual Sickle Cell Disease Annual Convention – 38th Annual held September 21-24, 2010
8. National Poster Child

Advocacy and Awareness

Highest point of social visibility for
Sickle Cell Disease in America

Movies by African American celebrities

Bill Cosby

To all my Friends on Shore, 1972

Sidney Poitier

A Warm December, 1974

Highlighted the lives of people with

House Resolution 1663

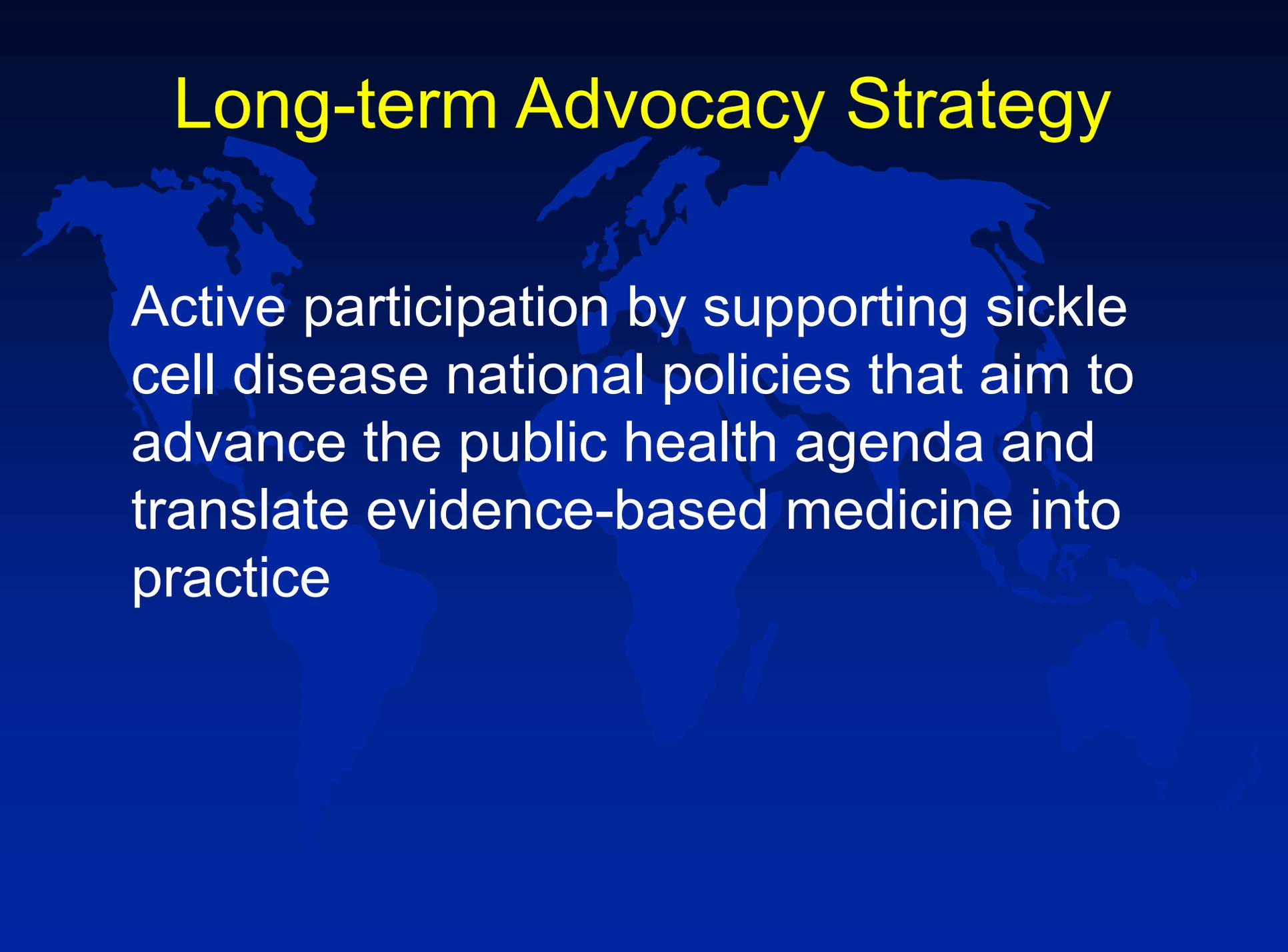
September - Sickle Cell Awareness Month

Introduced by the Congressional Black Caucus

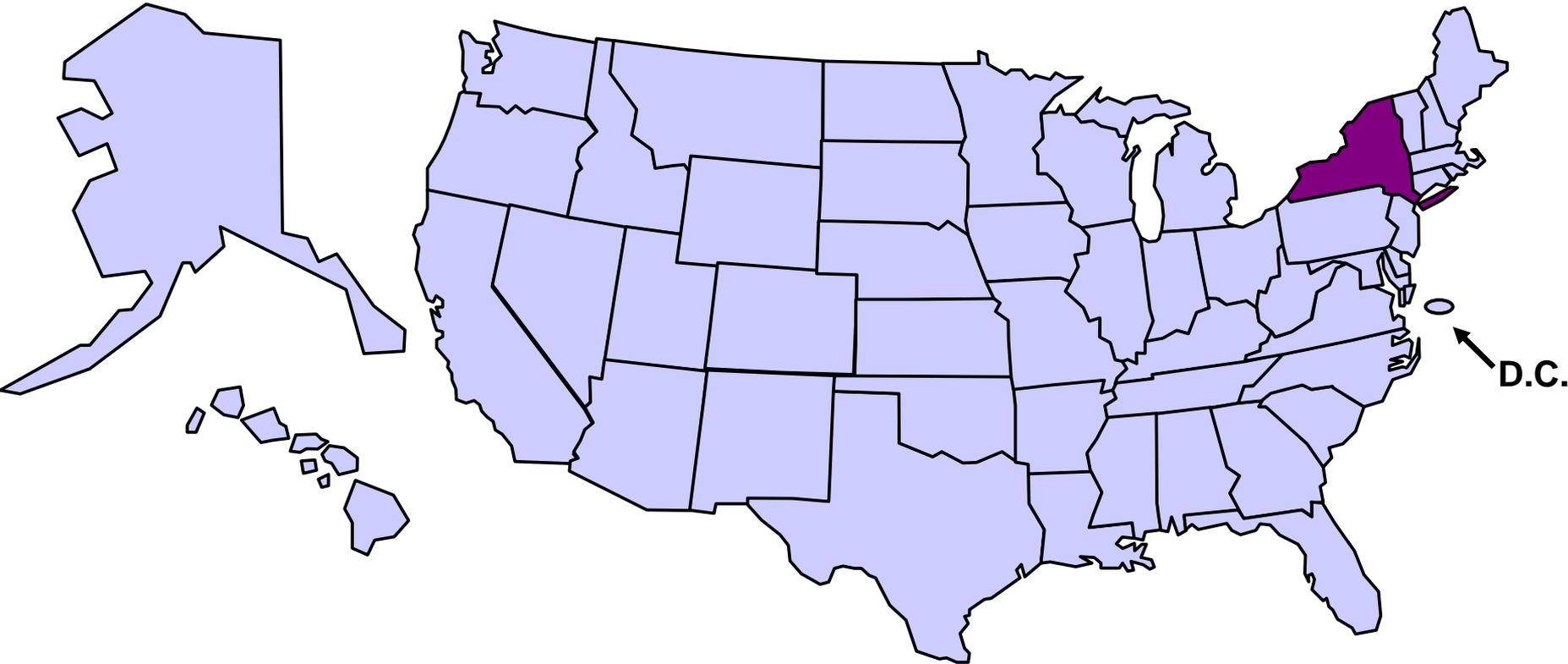
Signed in August 1983

Presidents Carter, Reagan, Bush, Clinton and Obama have greeted the SCDAA National Poster Child

Long-term Advocacy Strategy



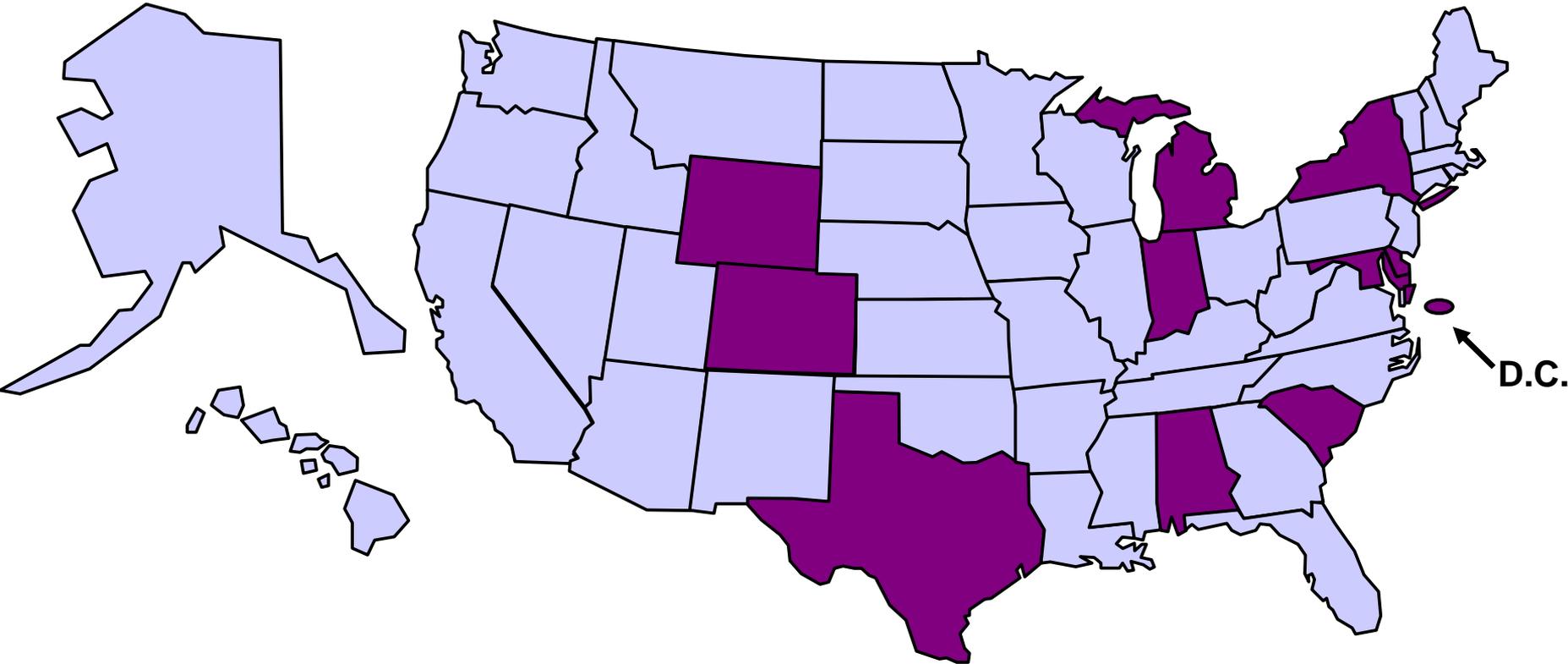
Active participation by supporting sickle cell disease national policies that aim to advance the public health agenda and translate evidence-based medicine into practice



- Universal Newborn Hemoglobinopathy Screening Mandated
- Newborn Hemoglobinopathy Not Universally Mandated

U.S. History of Hemoglobinopathy Screening

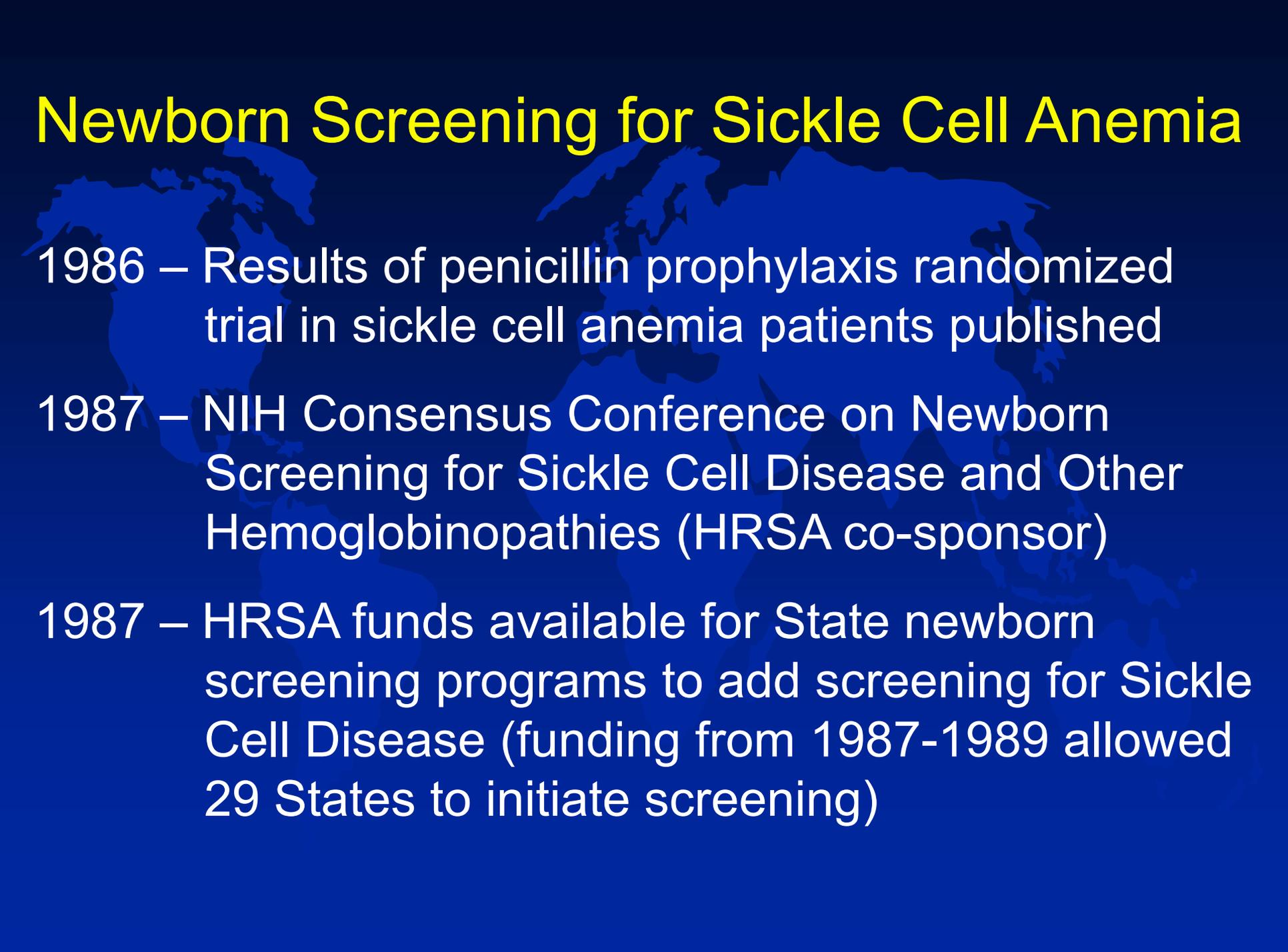
April 1, 1975



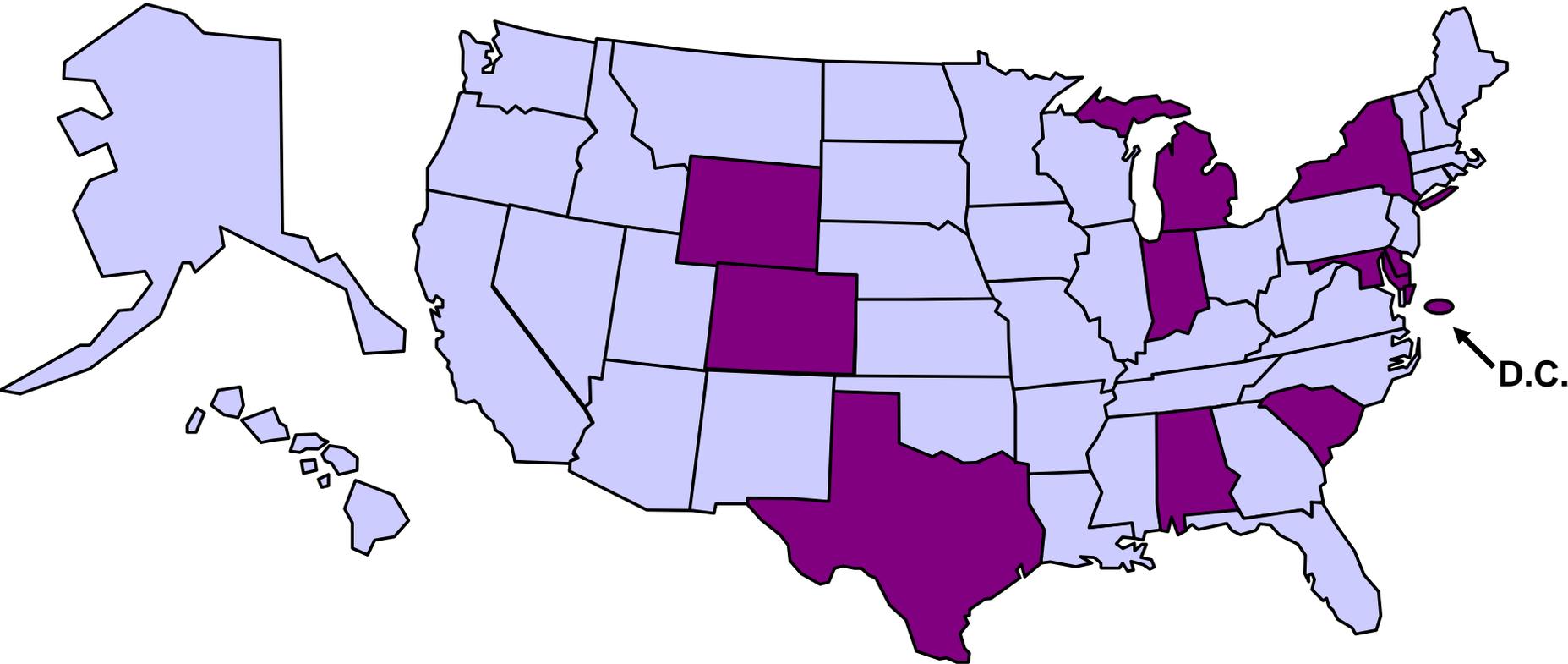
- Universal Newborn Hemoglobinopathy Screening Mandated
- Newborn Hemoglobinopathy Not Universally Mandated

U.S. History of Hemoglobinopathy Screening July 1, 1987

Newborn Screening for Sickle Cell Anemia

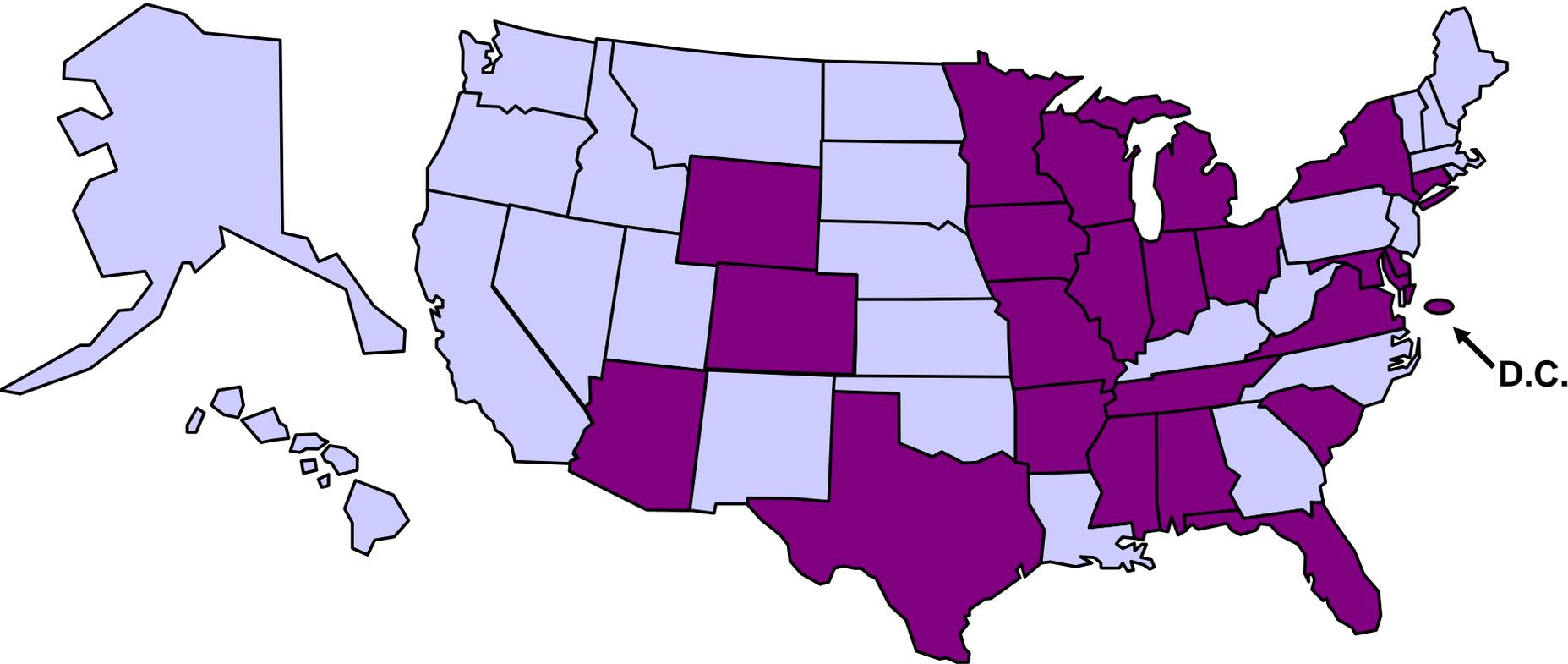


- 1986 – Results of penicillin prophylaxis randomized trial in sickle cell anemia patients published
- 1987 – NIH Consensus Conference on Newborn Screening for Sickle Cell Disease and Other Hemoglobinopathies (HRSA co-sponsor)
- 1987 – HRSA funds available for State newborn screening programs to add screening for Sickle Cell Disease (funding from 1987-1989 allowed 29 States to initiate screening)



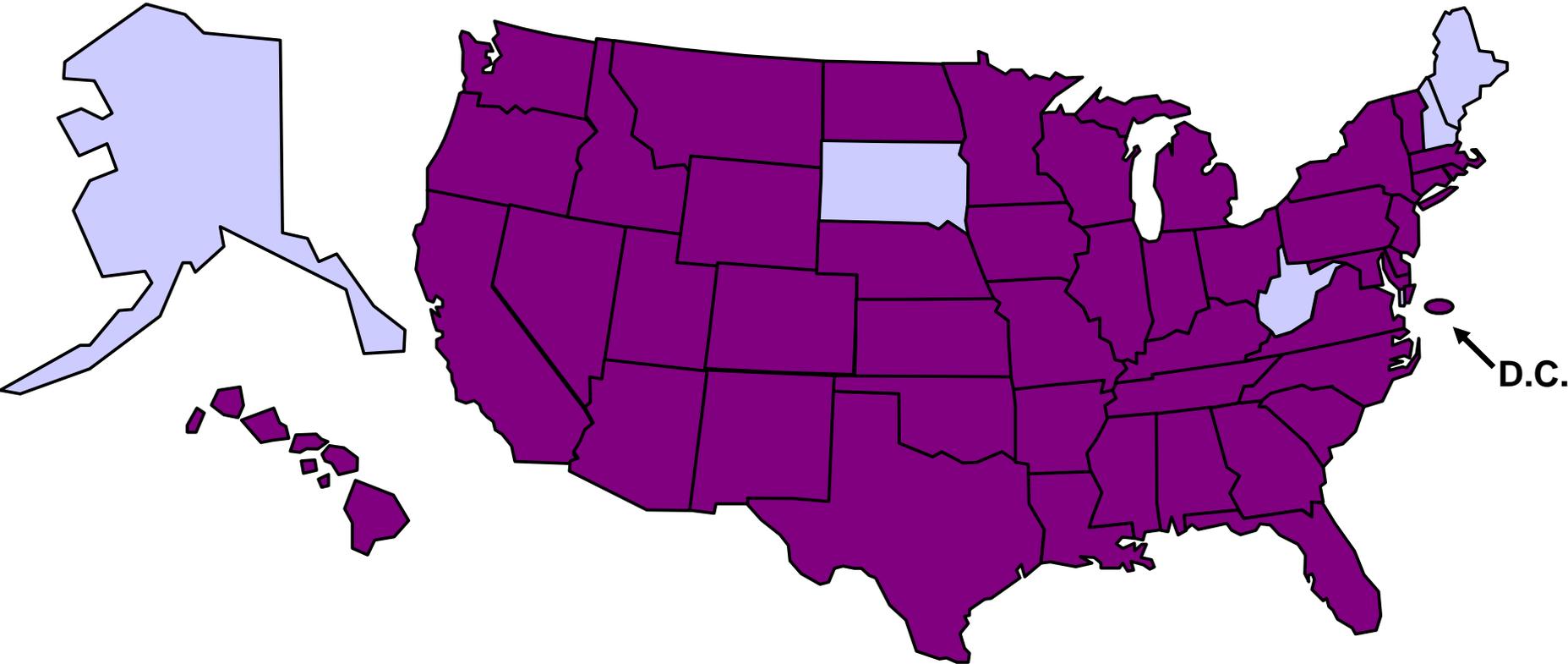
- Universal Newborn Hemoglobinopathy Screening Mandated
- Newborn Hemoglobinopathy Not Universally Mandated

U.S. History of Hemoglobinopathy Screening July 1, 1987



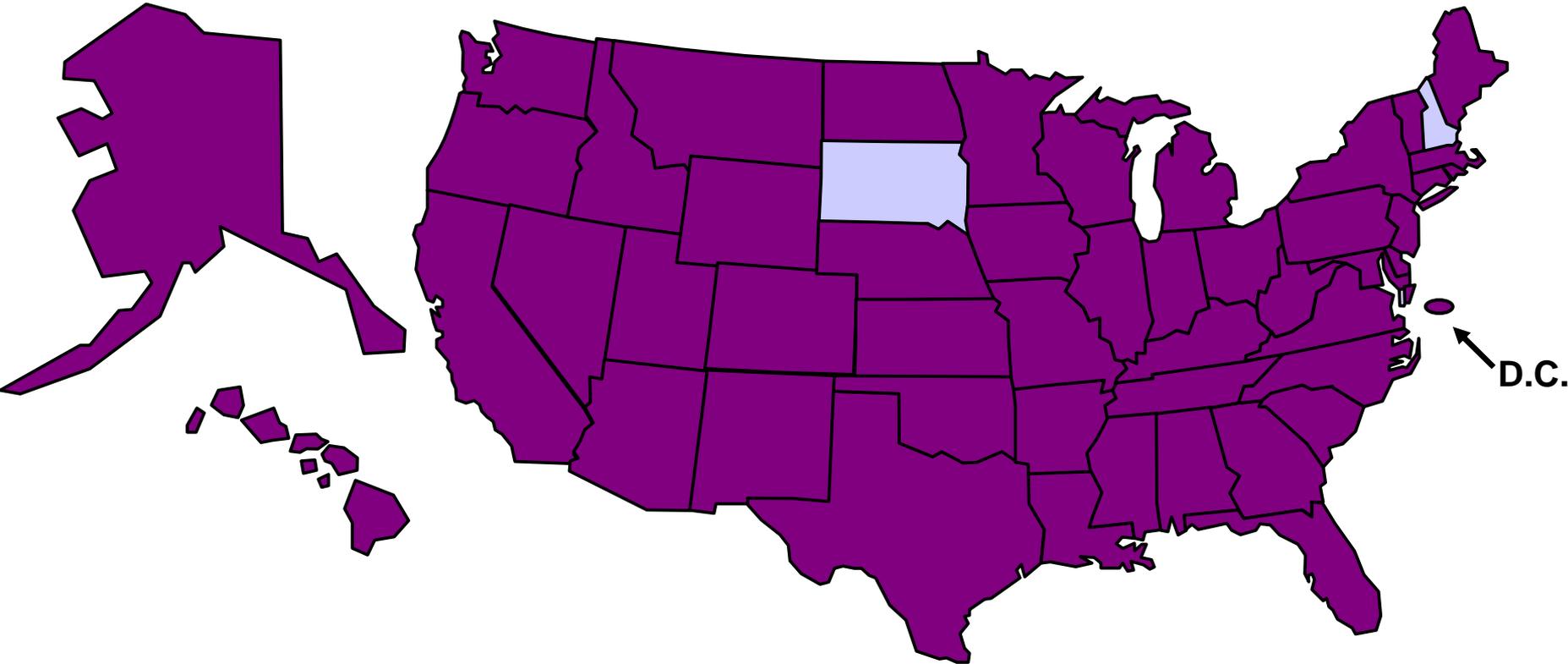
- Universal Newborn Hemoglobinopathy Screening Mandated
- Newborn Hemoglobinopathy Not Universally Mandated

U.S. History of Hemoglobinopathy Screening By January 1, 1990



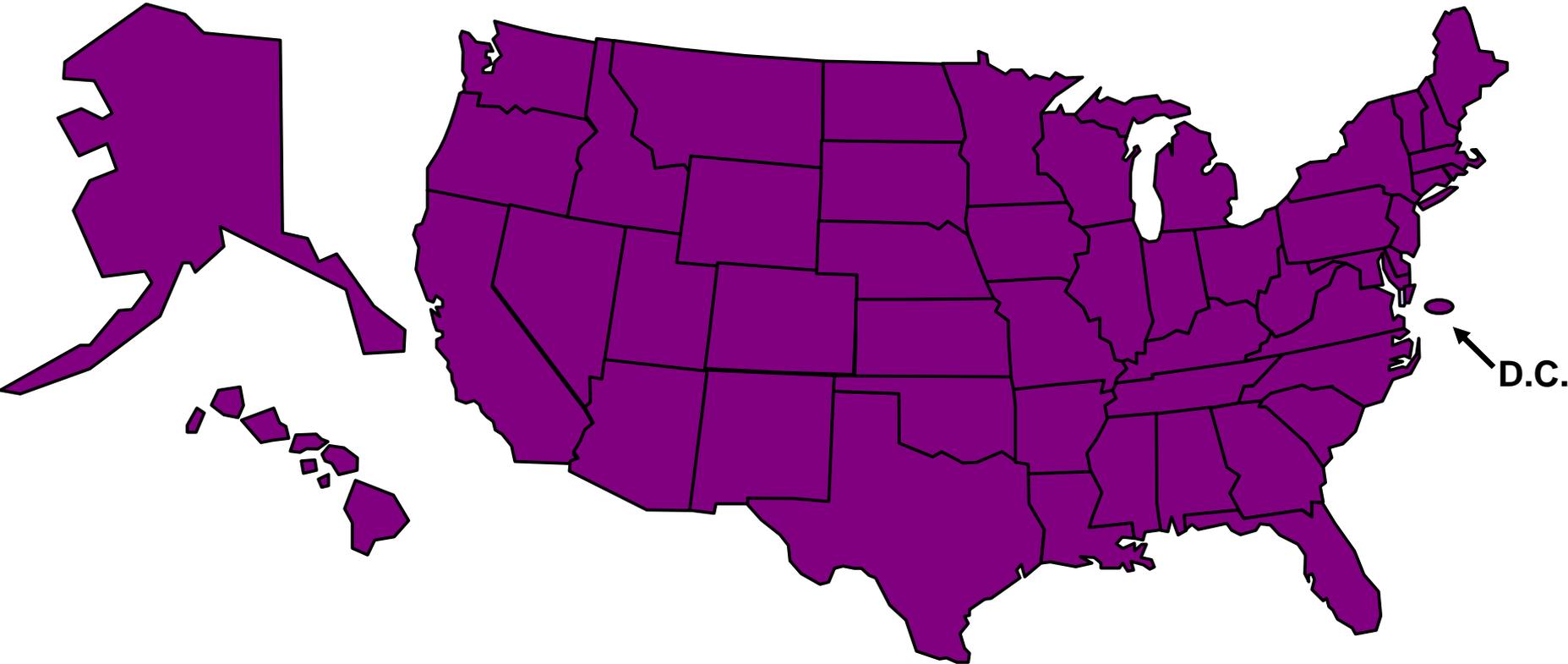
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U.S. History of Hemoglobinopathy Screening By January 1, 2000



- Universal Newborn Hemoglobinopathy Screening Mandated
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U.S. History of Hemoglobinopathy Screening By January 1, 2005



- Universal Newborn Hemoglobinopathy Screening Mandated
- Newborn Hemoglobinopathy Not Universally Mandated

**U.S. History of
Hemoglobinopathy Screening
By May 1, 2006
All 51 Programs**

Newborn Screening History

2000s

- 15th – 21st U.S. NBS Symposia
- Newborn hearing screening
- Public pressure to expand testing with MS/MS
- Emphasis on program integration (especially data)
- ACMG Report Recommending core screening panel in U.S.
- Privacy concerns – residual blood spot DNA, federal HIPAA rules (data sharing)
- Save Babies Act



**Newborn Hearing Screening
(OAE)**

HRSA Continues to Meet the Challenge

1998 – Funds AAP to Develop National Newborn Screening Blueprint

1999 – Initiates National Newborn Screening and Genetics Resource Center (National Focal Point for NBS Information)

2000 – AAP Blueprint published

2000 – Establishes Regional Collaboratives for Genetic Services and National Coordinating Center (Aid in NBS Long-term Follow-up)

2002 – Funds ACMG to Develop National NBS Policy Guidance Including 'Core' Panel of Tests

Jana Monaco, BS

Parent and Newborn Screening Advocate

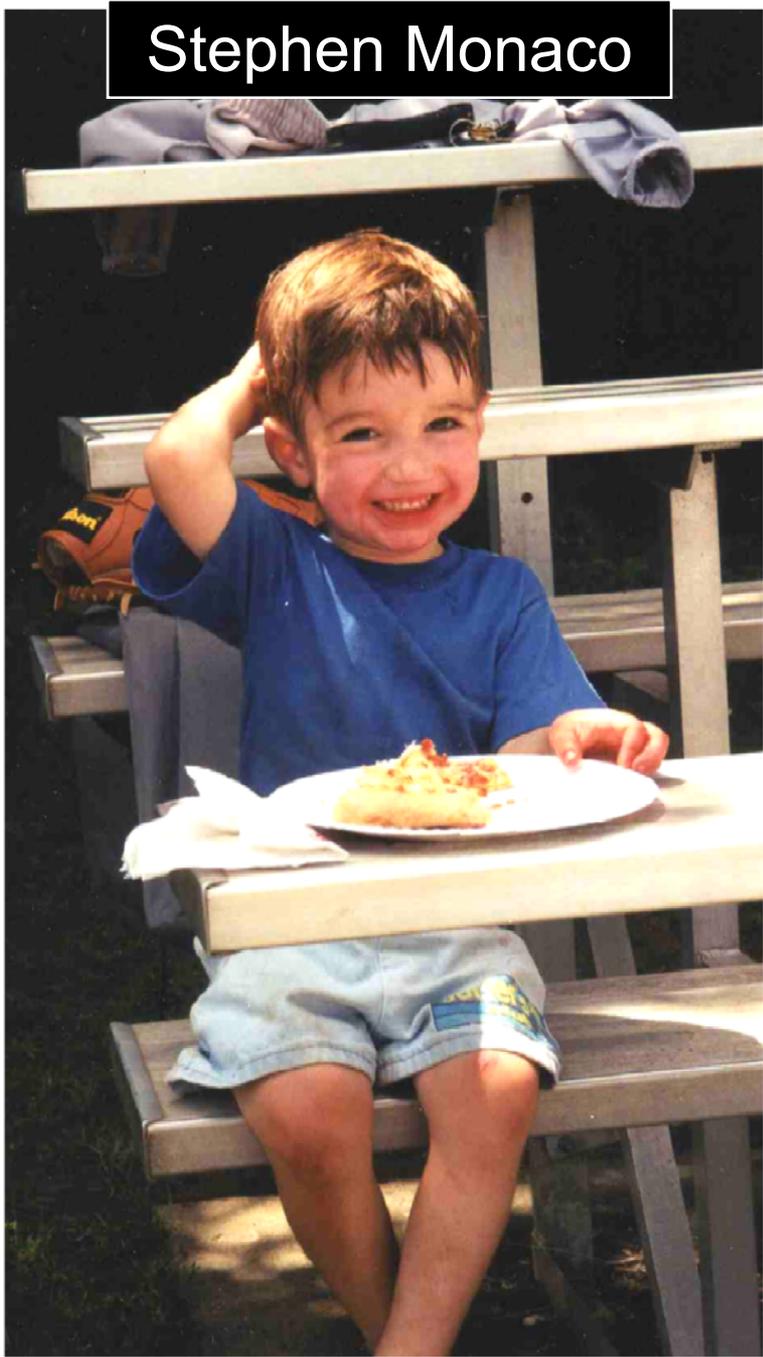
Member, Secretary's Advisory Committee on
Heritable Disorders in Newborns and Children

Advocacy Liaison,
Organic Acidemia Association



Organic Acidemia
Association
www.oaanews.org

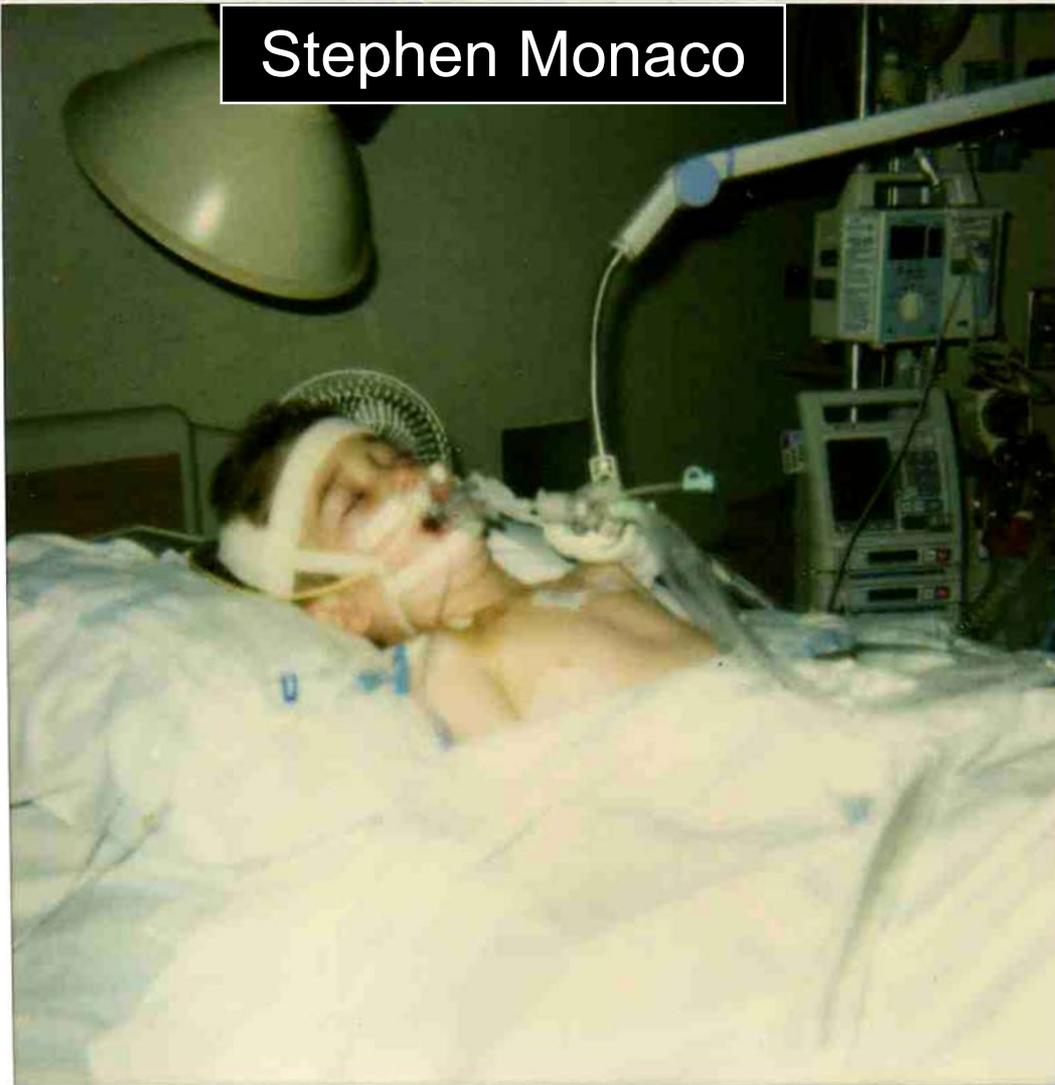
Stephen Monaco



Caroline Monaco



Stephen Monaco



June 1, 2001

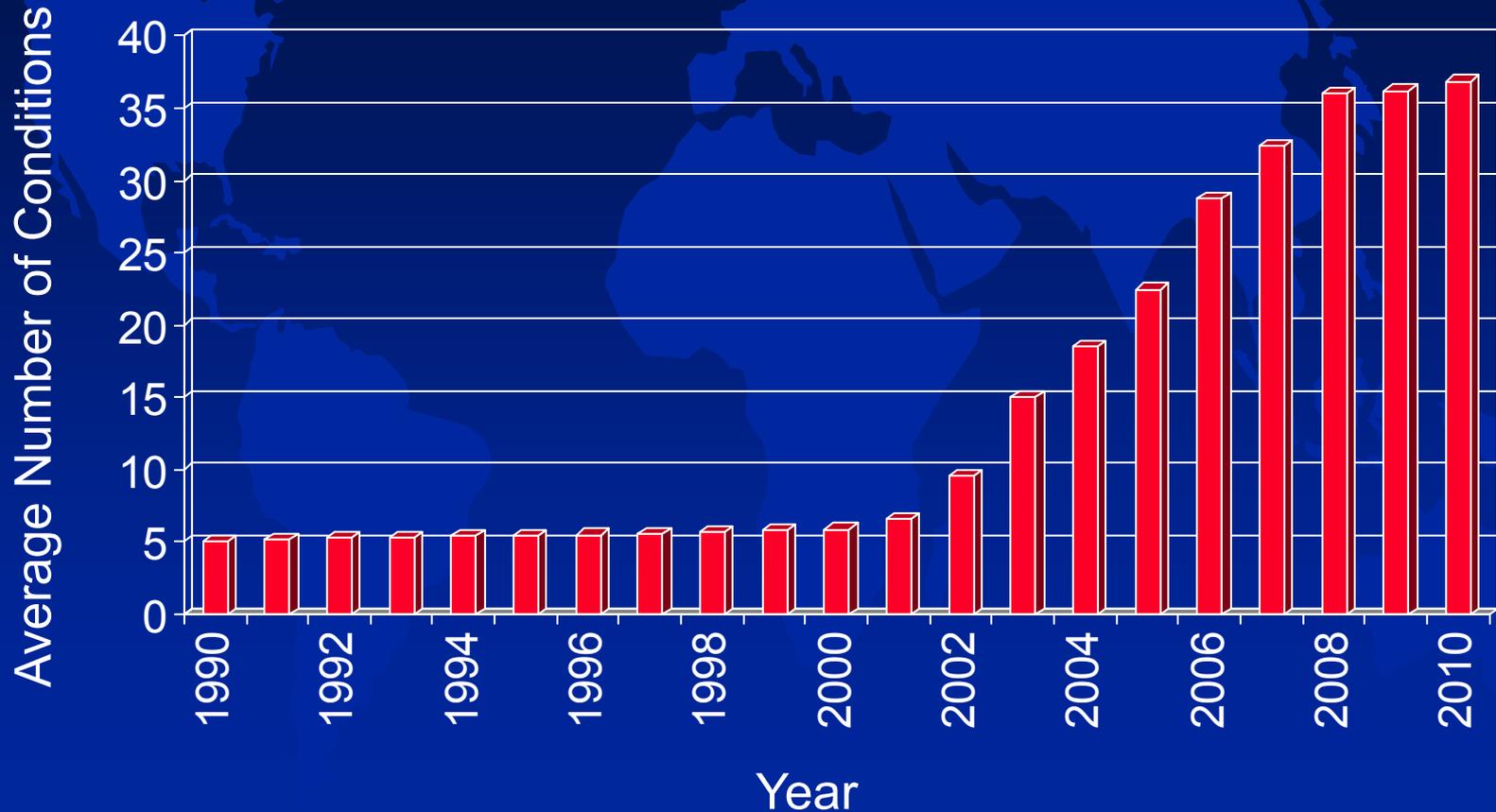


Monaco Family

HRSA Continues to Meet the Challenge

- 2003 – Organizes Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC)
- 2006 – ACMG Report approved by SACHDC and HRSA supplement to develop educational action sheets (ACT Sheets) for primary care physicians
- 2009 – Funds National Newborn Screening Clearinghouse as part of the Save Babies Through Newborn Screening Act
- 2009 – Assumes leadership role in improving newborn screening information technology

Average Number of Newborn Screening Conditions Required in US Programs 1990-2010



What Next?



MS/MS – Better quality; more uses
– second tier procedures

Beyond MS/MS New techniques;
increased efficiency

Why Does Everything in the Universe Rotate?

Pheromones: Profoundly Mysterious

Dust Devils

VOL. 24, NO. 7

Discover

JULY 2003

SCIENCE, TECHNOLOGY, AND MEDICINE

Now the
Genetic
Testing
Really
Begins

It Starts With
a Single
Drop
of **Blood**
Taken From
Each Newborn

And Ends When
Scientists Predict
Everyone's
Physical
and Mental
Future

Human red blood cells. Magnification: 19,600x



Examples of Candidate Conditions for Expansion of Uniform Panel (in alphabetical order)

➤ α-thalassemia

➤ CMV

➤ DMD

➤ G6PD

➤ Fabry disease

➤ Fragile-X

➤ HIV

➤ Krabbe disease

➤ Pompe disease

➤ SCID (Adopted)

➤ SMA

➤ Toxoplasmosis

➤ Wilson disease

➤ Many (?) others.....

Design and Characterization of a Multisource Hand-Held Tandem Mass Spectrometer

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Received for review June 23, 2008. Accepted August 19, 2008.

Abstract:

A wireless-controlled miniature rectilinear ion trap mass spectrometer system, total weight with batteries 5.0 kg, consuming less than 35 W of power, and having dimensions of 22 cm in length by 12 cm in width by 18 cm in height, is characterized. The design and construction of the mass spectrometer including mass analyzer, vacuum system, electronics system, and data acquisition and processing systems, is detailed. The mass spectrometer is compatible with various types of ionization sources including a glow discharge electron impact ionization source used in the internal ionization mode, and various atmospheric pressure ionization sources, including electrospray ionization, atmospheric pressure chemical ionization, and desorption electrospray ionization, which are employed for external, atmospheric pressure ionization. These external sources are coupled to the miniature mass spectrometer via a capillary interface that is operated in a discontinuous fashion (discontinuous atmospheric pressure interface) to maximize ion transport. The performance of the mass spectrometer for large and small molecules is characterized. Limits of detection in the parts-per-billion range were obtained for selected compounds examined using both the internal ionization and external ionization modes. Tandem mass spectrometry and fast in situ analysis capabilities are also demonstrated using a variety of compounds and ionization sources. Protein molecules are analyzed as the multiply protonated molecules with mass/charge ratios up to 1500 Da/charge.

Anal. Chem., 80 (19), 7198–7205, 2008. 10.1021/ac801275x

A dark blue world map is centered in the background of the slide. The text is overlaid on the map.

International Cooperation and Collaboration



Conference on Strengthening Newborn Screening in the Middle East and North Africa
Morocco, Marrakech November 13-15, 2006



**1st Conference of Asia Pacific Regional Newborn Screening Network for Developing Programs
Cebu, Philippines – April 5 -7, 2008**

Summary

- ◆ Newborn Screening is an essential and productive public health prevention activity.
- ◆ HRSA has played a MAJOR role in developing and sustaining newborn screening – both dried bloodspot and hearing screening.
- ◆ Expansion of newborn screening is inevitable and the SACHDNC will play a major role in shaping national policy.
- ◆ Equal access for all newborns will require continued HRSA foresight and support.

Newborn Screening Improves and Saves Lives

