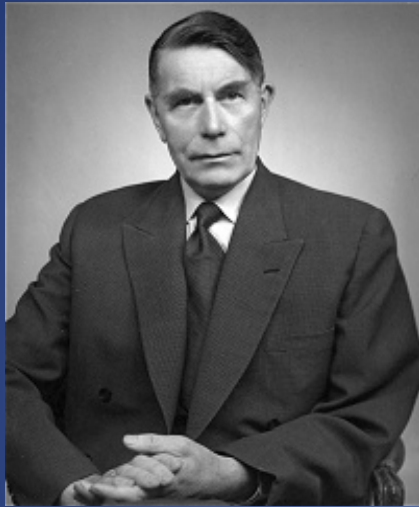




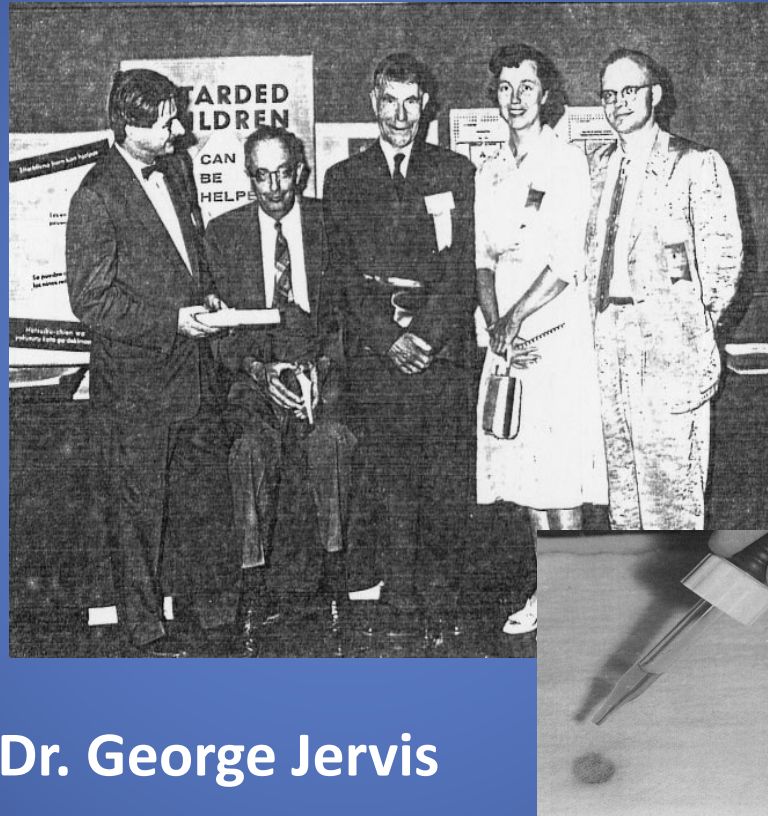
New York State's Contributions to the Field of Newborn Screening

Daniele Marisa-Thomas, BS; Heather Page, BS; Deborah Rodriguez, RN, MPH;
Joseph Orsini, PhD; Michele Caggana, ScD

A Disorder, Treatment and Diagnostic Test



Dr. Asbjorn Folling



Dr. George Jarvis

Dr. Willard Centerwall



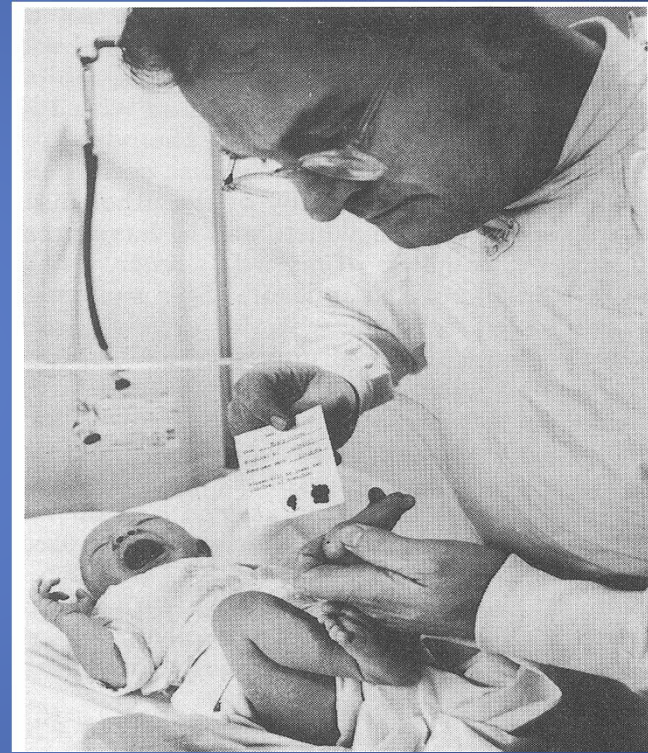
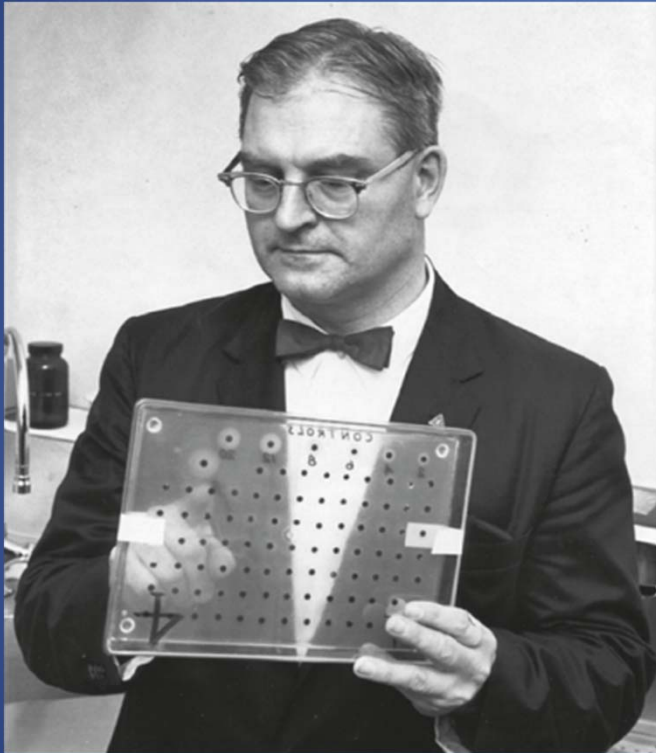
Dr. Horst Bickel

The World of PKU www.pkuworld.org

SA Centerwall, WR Centerwall (2000) The Discovery of Phenylketonuria: The Story of a Young Couple, Two Retarded Children, and a Scientist. Pediatrics 105: 89-103.

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NEW YORK STATE DEPARTMENT OF HEALTH

The Perfect Storm



Dr. Robert Guthrie

Pictures Courtesy of Dr. Kenneth Pass


Jamestown, New York

3/31/95
to Ken -
Fair winds,
Bob

Screening, I (1992) 5-15
© 1992 Elsevier Science Publishers B.V. All rights reserved 0925-6164/92/\$5.00

SCREEN 0005

The origin of newborn screening
Robert Guthrie



Robert Guthrie.

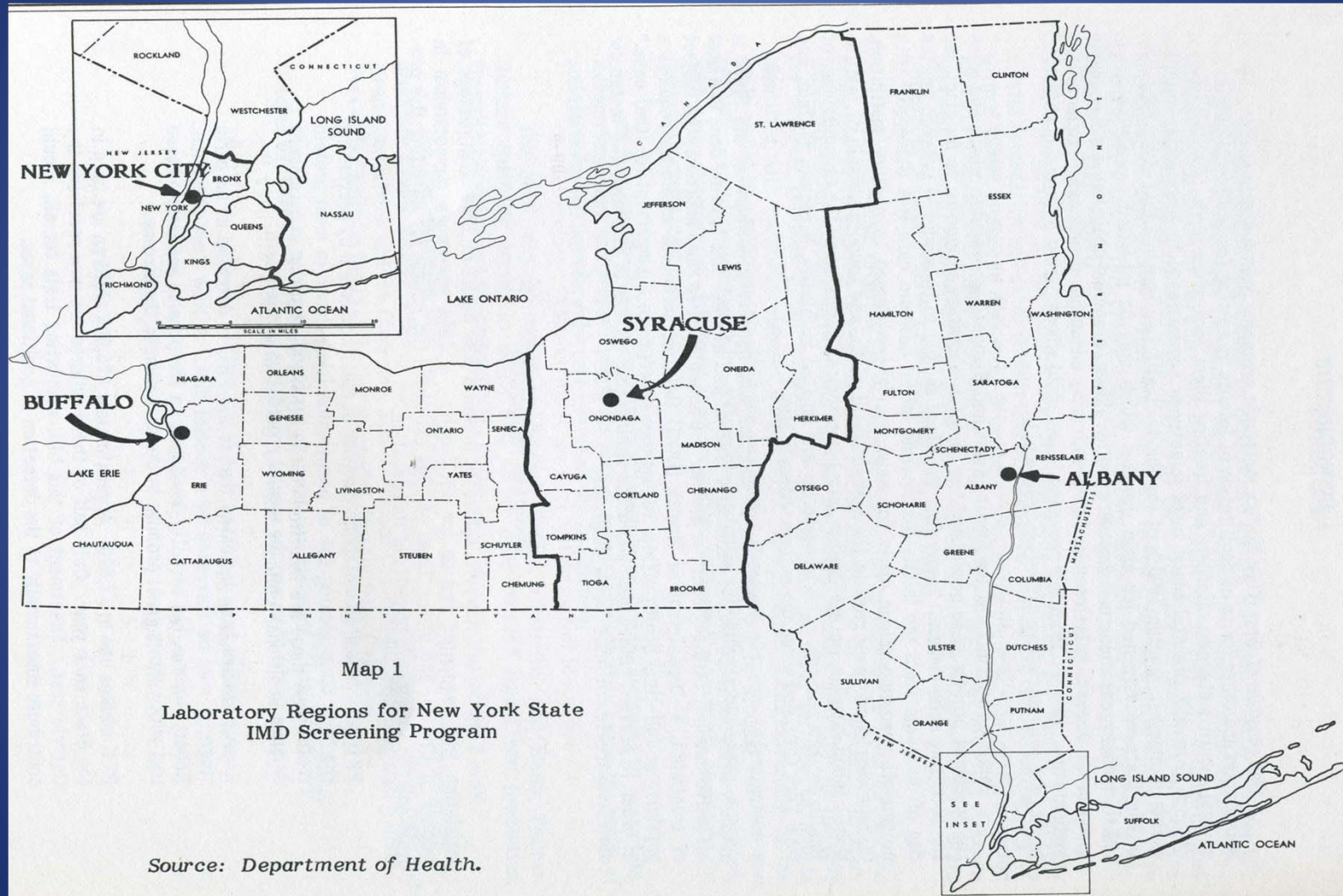
It began with our second child, John. He is mentally retarded. John stimulated me to go into research aimed at preventing mental retardation and developmental disabilities.

In 1957 I had been in cancer research for 12 years. Because of Johnny, my wife Margaret and I had become very active in the local Buffalo Chapter of the New York State Association for Retarded Children. As Vice-President of the Chapter, I was responsible for the program at the monthly meeting. For one of these programs

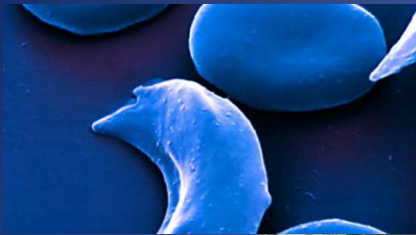
- Fall 1961 talk for The Association for Retarded Children
- Began to receive newborn filter-paper specimens
- “Thus, screening had its start in Jamestown, New York in 1961”

Picture Courtesy of Dr. Kenneth Pass

1965 Public Health Law 2500a



Sickle Cell Disease Screening



- First universal testing for SCD in the U.S. is implemented on April 1, 1975
- Until 1978, only NYS screened infants for sickle cell disease

Any Guesses?



Hint: 1988

NEONATAL SCREENING BLOOD SPECIMEN COLLECTION AND HANDLING PROCEDURE

1 • Equipment - sterile lancet with tip less than 2.5 mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.
WEAR GLOVES FOR PROCEDURE.

2 • Complete ALL information.
• Do not contaminate filter paper circles by touching or spillage before or after blood collection.
• Keep "SUBMITTER COPY".

3 • Indicate safe areas for puncture site.

4 • Warm site with soft cloth saturated with warm water up to 42° C, for three to five minutes.

5 • Choose site with alcohol prep.
• Wipe dry with sterile gauze pad.

6 • Puncture heel.
• Wipe away first blood drop with sterile gauze pad.
• Allow another LARGE blood drop to form.

7 • Lightly touch filter paper to LARGE blood drop.
• Allow blood to SOAK THROUGH AND COMPLETELY FILL CIRCLES with SINGLE application to LARGE blood drop.
• To enhance blood flow, VERY GENTLE intermittent pressure may be applied to area surrounding puncture site.
• Apply blood to one side of filter paper, only.

8 • Fill remaining circles in same manner as step 7, with successive blood drops.
• If blood flow is diminished, repeat step 3 through 7.
• Care of skin puncture site should be consistent with your institution's procedures.

9 • Dry blood spots on a flat, non-absorbent surface for a minimum of four hours.

10 • Mail completed form to testing laboratory within 24 hours of collection.

Simple Spot Check

Valid Specimen



Allow a sufficient quantity of blood to soak through to completely fill the preprinted circle on the filter paper. Fill all required circles with blood. Do not layer successive drops of blood or apply blood more than once in the same collection circle. Avoid touching or smearing spots.

Invalid Specimens:



1. Specimen quantity insufficient for testing.



2. Specimen appears scratched or abraded.



3. Specimen not dry before mailing.



4. Specimen appears supersaturated.



5. Specimen appears diluted, discolored or contaminated.



6. Specimen exhibits serum rings.



7. Specimen appears clotted or layered.



8. No blood.

Possible Causes:

- Removing filter paper before blood has completely filled circle or before blood has soaked through to second side.
- Applying blood to filter paper with a capillary tube.
- Touching filter paper before or after blood specimen collection with gloved or ungloved hands, hand lotion, etc.
- Allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood specimen collection.
- Applying blood with a capillary tube or other device.
- Mailing specimen before drying for a minimum of four hours.
- Applying excess blood to filter paper, usually with a device.
- Applying blood to both sides of filter paper.
- Squeezing or "milking" of area surrounding the puncture site.
- Allowing filter paper to come in contact with gloved or ungloved hands or substances such as alcohol, formula, antiseptic solutions, water, hand lotion or powder, etc., either before or after blood specimen collection.
- Exposing blood spots to direct heat.
- Not wiping alcohol from puncture site before making skin puncture.
- Allowing filter paper to come in contact with alcohol, hand lotion, etc.
- Squeezing area surrounding puncture site excessively.
- Drying specimen improperly.
- Applying blood to filter paper with a capillary tube.
- Touching the same circle on filter paper to blood drop several times.
- Filling circle on both sides of filter paper.
- Failure to obtain blood specimen.

Schleicher & Schuell knows this information to be true only when using S&S 903™ Specimen Collection Paper.

Schleicher & Schuell
P.O. Box 2012, Keene, New Hampshire 03431 • 800/437-7003 • FAX: 603/357-3627

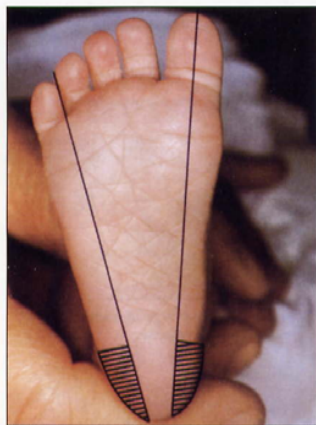
Information provided by The
New York State Department of Health.



1 Equipment: sterile lancet with tip approximately 2.0 mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.



2 Complete ALL information. Do not contaminate filter paper circles by allowing the circles to come in contact with spillage or by touching before or after blood collection. Keep "SUBMITTER COPY" if applicable.



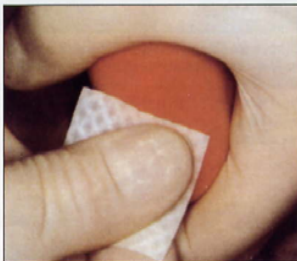
3 Hatched area (▨▨▨▨▨▨▨▨▨▨) indicates safe areas for puncture site.

Neonatal Screening

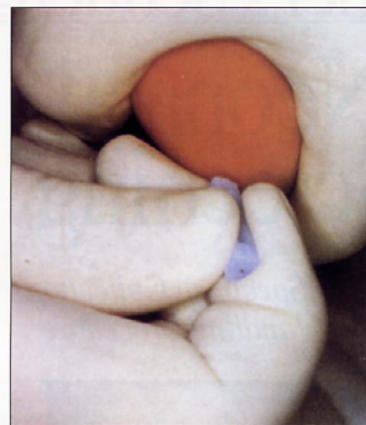
Blood Specimen Collection and Handling Procedure



4 Warm site with soft cloth, moistened with warm water up to 41°C, for three to five minutes.



5 Cleanse site with alcohol prep. Wipe DRY with sterile gauze pad.



6 Puncture heel. Wipe away first blood drop with sterile gauze pad. Allow another LARGE blood drop to form.



7 Lightly touch filter paper to LARGE blood drop. Allow blood to soak through and completely fill circle with SINGLE application to LARGE blood drop. (To enhance blood flow, VERY GENTLE intermittent pressure may be applied to area surrounding puncture site). Apply blood to one side of filter paper only.



8 Fill remaining circles in the same manner as step 7, with successive blood drops. If blood flow is diminished, repeat steps 5 through 7. Care of skin puncture site should be consistent with your institution's procedures.



9 Dry blood spots on a dry, clean, flat non-absorbent surface for a minimum of four hours.



10 Mail completed form to testing laboratory within 24 hours of collection.

Information provided by The New York State Department of Health.

Schleicher & Schuell Inc. • 10 Optical Avenue • Keene N.H. 03431 USA • Tel. (603) 352-3810 • Fax (603) 355-6524 • Internet: <http://www.s-and-s.com> • e-mail: solutions@s-and-s.com
 Schleicher & Schuell GmbH • P.O. Box 4, D-37582 Dassel • Germany • Tel. 49-5561-791-0 • Fax 49-5561-791536 • Internet: <http://www.s-und-s.de> • e-mail: filtration@s-und-s.com
 Other Subsidiaries of Schleicher & Schuell include the Netherlands, Switzerland, France, Belgium, Hungary, Italy and Great Britain

HIV Screening: February 1, 1997

NY became the only state to require mandatory Newborn Screening for HIV-1 and notification of mothers



"The opposition was formidable"



78% drop in the number of infected babies in the first 6 years

1997 10.9% infection rate

2002 2.4% infection rate



1996

5,378 Phone Calls
1,285 Faxes

2012

206,204 Phone Calls
105,544 Faxes

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NEW YORK POST

LATE CITY FINAL

WEDNESDAY, JANUARY 20, 1993 / Sunny, low 40s today; clear, 30 tonight / Details, Page 2

40¢ in New York City 50¢ elsewhere

TOT TAX



GOV. CUOMO
Eyes new revenue.

Gov wants \$17 for every infant born in New York

Details: Page 5

Picture Courtesy of Dr. Kenneth Pass

Population Screening for Krabbe Disease

Times Union, Albany NY June 8, 2004

SPORTS

Alb

PEOPLE

Kelly seeks newborn screening

Associated Press

BUFFALO— Hall of Fame quarterback **Jim Kelly** is taking his ailing son **Hunter's** message to Washington.

Kelly is scheduled to speak before lawmakers today in support of universal newborn screening, something which could have helped his son, who suffers from the deadly Krabbe Disease.

Kelly will speak before a committee that advises Health and Human Services Secretary **Tommy Thompson**, and will also meet with Sen. **Charles**

Schumer, D-N.Y.

Krabbe is an inherited degenerative disorder that hinders development of the fatty sheath, known as myelin, that protects the brain's nerve fibers. Those born with the disease can be treated with a cord blood transplant.

Hunter Kelly, 7, was not diagnosed until he was four-months-old, too late for the transplant surgery to be successful.

Currently, Hawaii tests for 48 diseases, New York for 12 and California and Alabama for four.



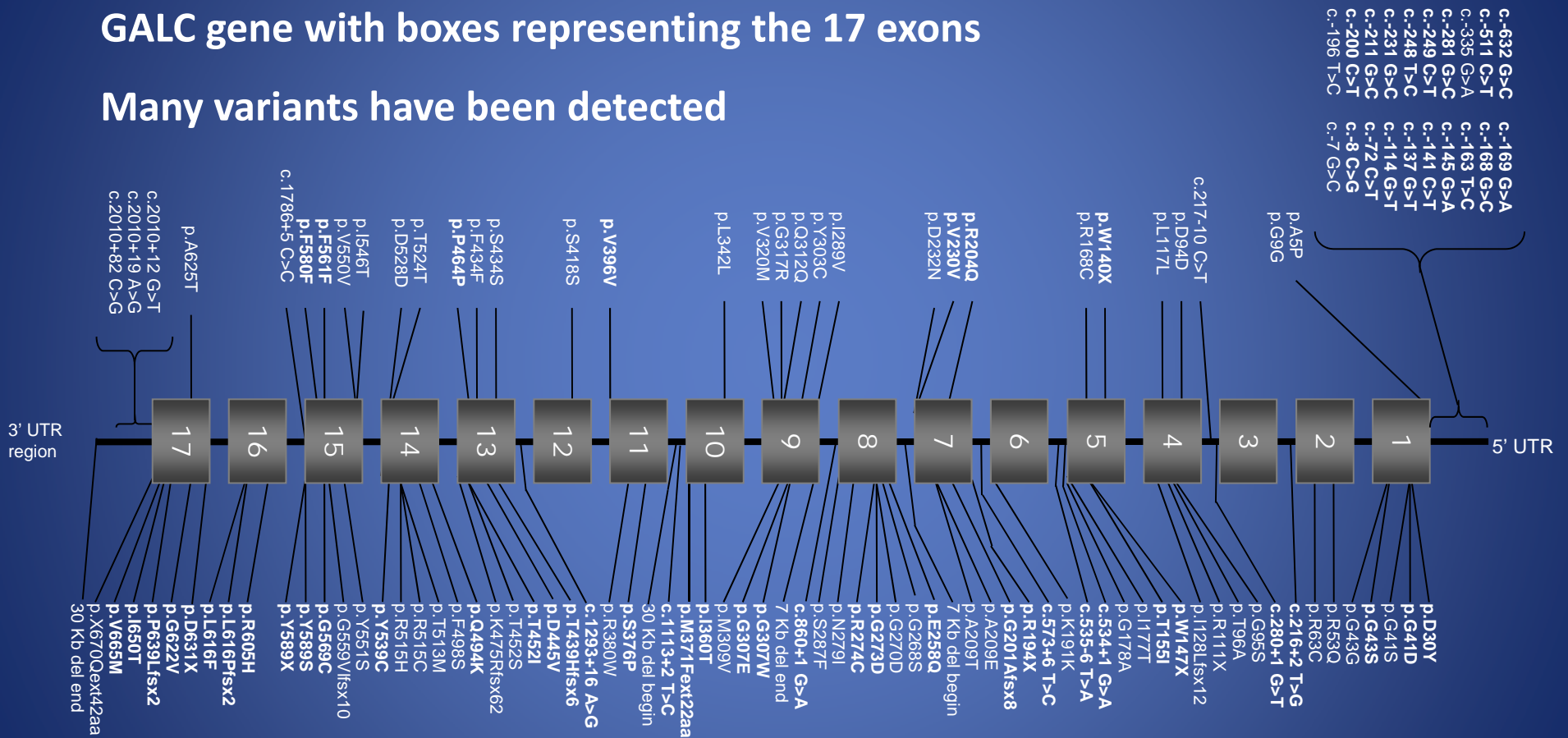
Delta Sky Magazine, "Legacy of Hope" by Brion O'Connor (who obtained picture courtesy of Hunter's Hope Foundation)

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Krabbe Disease: August 7, 2006

GALC gene with boxes representing the 17 exons

Many variants have been detected



Courtesy of Matthew Nichols, NYS DOH Newborn Screening

Population Screening for Krabbe Disease

An Age-old Debate?

- “...undertaken at a time when many questions remained unanswered concerning the detection and treatment of the disease”¹
- “An unexpected consequence of the screening programs was that many more infants were identified with elevated ... levels than would have been predicted on the basis of previous statistical data... A minor upheaval in the concepts...was caused by the discovery of relatively large numbers of these variants...These variant patients...are an enigma and the cause of great confusion.”²
- “It is suggested that more information on the validity of this test should be made available *before*state health departments embark on this mass testing program.”³

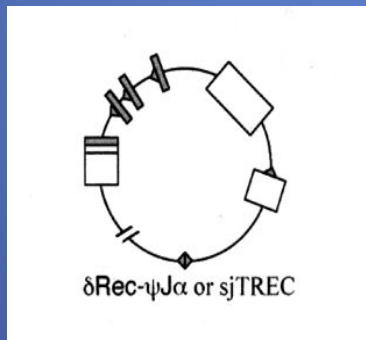
1 National Research Council, Committee for the Study of Inborn Errors of Metabolism (1975) Screening for Phenylketonuria. Genetic Screening : Program, Principals and Research, p. 92. National Academy of Sciences

2 Blaskovics ME and Nelson TL. Phenylketonuria and Its Variations A Review of Recent Developments. Western Journal of Medicine, 1975, 115; p. 44.

3 Wright SW. Mass screening for phenylketonuria. The Journal of Pediatrics, 1962, Vol. 61, No. 4, 651-652.

2010: SCID

New York State is the only state currently testing newborns for adenosine deaminase deficiency and histidinemia. After testing more than 3 million newborns since 1975, the blood constituent, adenosine deaminase, has been found to be a poor "marker" for severe combined immuno-deficiency (SCID). The Legislature intended to enhance the ability to identify children with SCID by amending PHL section 2500-a in 1974.



4th state to add to SCID to the panel, with funding from the NICHD and the Jeffery Modell Foundation

Quantification of T cell receptor rearrangement excision circles to estimate thymic function: an important new tool for endocrine-immune physiology. Geenen V et al. *Journal of Endocrinology* 2003 Mar; 176(3):305-311.

2014: Adrenoleukodystrophy

S T A T E O F N E W Y O R K

211

2013-2014 Regular Sessions

I N A S S E M B L Y

(PREFILED)

January 9, 2013

Introduced by M. of A. BRENNAN -- read once and referred to the Committee on Health

AN ACT to amend the public health law, in relation to requiring adrenoleukodystrophy screening of newborns

THE PEOPLE OF THE STATE OF NEW YORK, REPRESENTED IN SENATE AND ASSEMBLY, DO ENACT AS FOLLOWS:

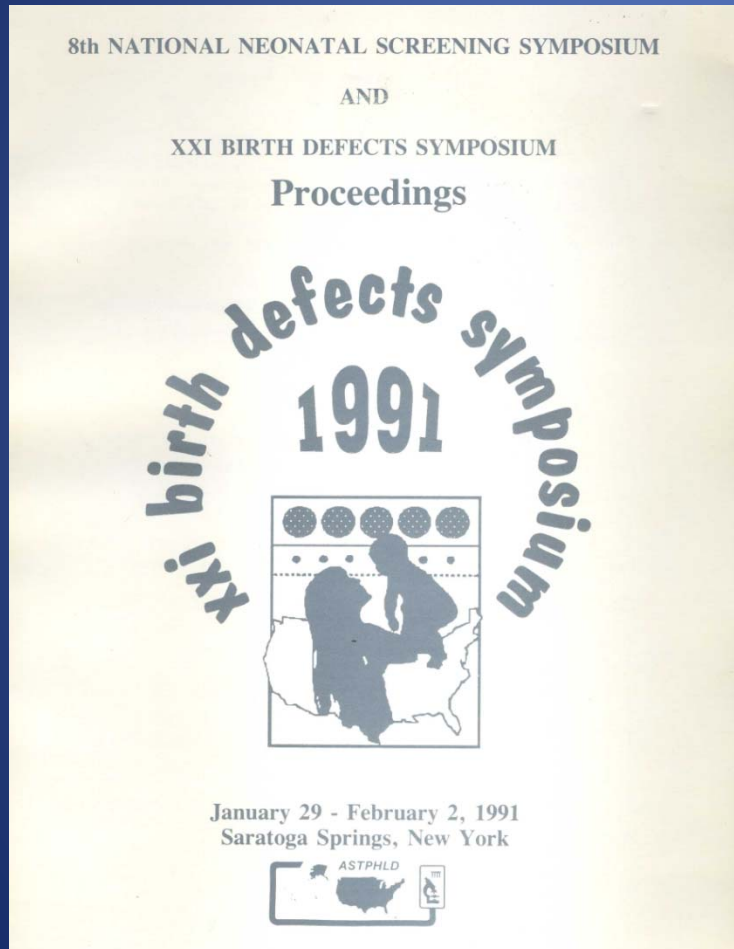
1 Section 1. This act shall be known and may be cited as "Aidan's Law".

2 S 2. Subdivision (a) of section 2500-a of the public health law, as
3 amended by chapter 863 of the laws of 1986, is amended to read as
4 follows:

5 (a) It shall be the duty of the administrative officer or other person
6 in charge of each institution caring for infants twenty-eight days or
7 less of age and the person required in pursuance of the provisions of
8 section forty-one hundred thirty of this chapter to register the birth
9 of a child, to cause to have administered to every such infant or child
10 in its or his care a test for phenylketonuria, homozygous sickle cell
11 disease, hypothyroidism, branched-chain ketonuria, galactosemia, homo-
12 cystinuria, ADRENOLEUKODYSTROPHY and such other diseases and conditions
13 as may from time to time be designated by the commissioner in accordance
14 with rules or regulations prescribed by the commissioner. Testing, the
15 recording of the results of such tests, tracking, follow-up reviews and
16 educational activities shall be performed at such times and in such
17 manner as may be prescribed by the commissioner. The commissioner shall
18 promulgate regulations setting forth the manner in which information
19 describing the purposes of the requirements of this section shall be
20 disseminated to parents or a guardian of the infant tested.

21 S 3. This act shall take effect on the one hundred eightieth day after
22 it shall have become a law; provided, however, that effective immediately,
23 the addition, amendment and/or repeal of any rule or regulation
24 necessary for the implementation of this act on its effective date are
25 authorized and directed to be made and completed on or before such
26 effective date.

Yesterday and Today



7

ASTPHLD Newsletter

8th Neonatal Screening Symposium Attracts 300 to Saratoga Springs

— Kenneth A. Pass, Ph.D.
Organizing Committee Chairman

An outstanding feature of each Neonatal Symposium is the exchange of ideas.



From left to right: Bob Guthrie, Bob Phillips, Lindsay Hofman, Rudy Hormuth, Ken Pass, Harry Hannon, Joe Joseph, and Harvey Levy.

Wadsworth Center
NEW YORK STATE DEPARTMENT OF HEALTH

Acknowledgements



Dr. Michele Caggana



**Heather Page
Daniele-Marisa Thomas**

Anita Girard, RN



Dr. Kenneth Pass

Dr. Joseph Orsini