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

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A Disorder, Treatment and Diagnostic Test

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The World of PKU www.pkuworld.org
The Perfect Storm

Dr. Robert Guthrie

Pictures Courtesy of Dr. Kenneth Pass
Jamestown, New York

• 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
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
**Simple Spot Check**

**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 without 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.

Information provided by The New York State Department of Health.
Neonatal Screening

Blood Specimen Collection and Handling Procedure

1. Equipment: sterile lancet with tip approximately 2.5 mm, sterile alcohol prep, sterile gauze pads, soft cloth, blood collection form, gloves.

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

3. Hatch area (**CIRCLES**) indicates safe areas for puncture site.

4. Warm site with soft cloth, consistent with room warm to 40°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 seep 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 3 through 7. Care of skin puncture site should be consistent with your institution's procedures.

9. Dry blood spots on a dry, clean, the 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.
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
NEW YORK BECOMES THE FIRST STATE TO OFFER AUTOMATED TELEPHONE REPORTING OF NEONATAL SCREENING PROGRAM RESULTS

1996  2012

5,378 Phone Calls  206,204 Phone Calls
1,285 Faxes  105,544 Faxes
Picture Courtesy of Dr. Kenneth Pass
Population Screening for Krabbe Disease

Times Union, Albany NY June 8, 2004

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.
Many variants have been detected

GALC gene with boxes representing the 17 exons

Krabbe Disease: August 7, 2006

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.
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 1973, 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
2014: Adrenoleukodystrophy

STATE OF NEW YORK

211
2013-2014 Regular Sessions
IN ASSEMBLY
(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:

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

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

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

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

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 Hornuth, Ken Pass, Harry Hannon, Joe Joseph, and Harvey Levy.