REPORT OF THE
JOINT COMMISSION ON HEALTH CARE

Review of Newborn Screening in Virginia
(HJR 164, 2004)

TO THE GOVERNOR AND
THE GENERAL ASSEMBLY OF VIRGINIA

HOUSE DOCUMENT NO. 28

COMMONWEALTH OF VIRGINIA
RICHMOND
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COMMONWEALTH of VIRGINIA

Joint Commission on Health Care

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March 15, 2005

TO: The Honorable Mark R. Warner, Governor of Virginia
and Members of the General Assembly

The 2004 General Assembly, in House Joint Resolution 164, directed the Joint Commission on Health Care (JCHC) to collect information regarding the newborn screening programs, conducted in other states, as well as the costs and benefits of screening.

An executive summary of the requested information was submitted prior to the 2005 General Assembly Session. The executive summary and two JCHC presentations regarding newborn screening are enclosed for your consideration.

Respectfully submitted,

Harvey B. Morgan
Chairman

JOINT COMMISSION ON HEALTH CARE: 2004

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PREFACE

Knowledge of the devastating consequences of certain inborn metabolic disorders and the ability to screen for certain disorders have advanced significantly in recent years. House Joint Resolution 164, of the 2004 Session of the General Assembly, directed the Joint Commission on Health Care to review newborn screening, including the disorders screened in other states and the benefits and costs associated with screening.

Completion of the Joint Commission study coincided with the release of a study by the American College of Medical Genetics (ACMG) – *Newborn Screening: Toward a Uniform Screening Panel and System*. The Secretary of Health and Human Services is expected to issue a significant, new guideline based on the ACMG study that will encourage states to include at least 29 disorders in their newborn screening programs. (Currently, the federal recommendation is for newborns to be screened for PKU, congenital hypothyroidism, and sickle cell diseases.) The Joint Commission on Health Care introduced legislation in 2005 to require expansion of Virginia's newborn screening program to screen for the recommended disorders.

As Virginia considers this major expansion of its newborn screening program, the contributions of Delegate Kenneth R. Plum and Senator Patricia S. Ticer in support of newborn screening should be acknowledged. Delegate Plum introduced legislation over the last 22 years to ensure that Virginia's program adhered to the recommended practice for newborn screening. Moreover, House Joint Resolution 164, which requested this study of newborn screening programs, was introduced by Delegate Plum. Senator Ticer introduced legislation in 2001 and 2002 to require screening for additional disorders; including a disorder that required the Department of General Services to purchase tandem mass spectrometers. That purchase will facilitate the screening expansion recommended by this study.

On behalf of the Joint Commission on Health Care and its staff, I would like to thank the numerous individuals, agencies, and associations that assisted in the completion of this study including: Jana A. Monaco, American Academy of Pediatrics – Virginia Chapter, March of Dimes – Virginia Chapter, Pediatrix Screening, Virginia Association of Health Plans, Virginia Department of General Services, Virginia Department of Health, Virginia Genetics Advisory Committee, and Virginia Hospital & Healthcare Association.

Kim Snead
Executive Director

March 2005

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REVIEW OF NEWBORN SCREENING IN VIRGINIA

EXECUTIVE SUMMARY

Authority for Study

HJR 164 directed the Joint Commission on Health Care (JCHC) to review information regarding newborn screening (NBS) programs for metabolic disorders including the disorders screened in other states, and the benefits and the costs associated with screenings. The collected information and an executive summary are required to be submitted prior to the 2005 General Assembly Session for processing as a legislative document.

NBS Programs in Other States and Anticipated New Federal Guidelines

Newborn screening programs began in the 1960s, after the effects of phenylketonuria (PKU) were identified and a simple, inexpensive means of screening newborns for PKU was developed. Over the last 40 years, newborn screening programs have been established in each of the 50 states as additional disorders have been identified and corresponding screening procedures have been developed.

Although knowledge has advanced about a variety of inborn, metabolic disorders and their devastating consequences if left unidentified and untreated, there is no uniformity in the screening conducted by each state. Currently, the number of disorders screened ranges from 3 to 54. This lack of uniformity is due in part to the absence of federal requirements related to NBS. Currently, the federal recommendation is for newborns to be screened for PKU, congenital hypothyroidism, and sickle cell diseases. However, it is anticipated that the Secretary of Health and Human Services (HHS) will issue a new recommendation based on the findings of a recently released study by the American College of Medical Genetics (ACMG) *Newborn Screening: Toward a Uniform Screening Panel and System*. The 3-year ACMG study to develop a uniform list of disorders for states to include in NBS screening programs was presented in September 2004 to an Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns or Children (established by the HHS Secretary). The ACMG study recommended screening for a core panel of 30 disorders with 25 additional disorders being report-only disorders. (Virginia screens for 12 of the 30 disorders as they are listed in the ACMG study.) It is expected that the HHS Secretary will recommend that states include a NBS panel of disorders that is different by only one or two disorders from the ACMG's initial study recommendation.

Reaction to the Anticipated Expansion of NBS Panels

On September 22, 2004, the March of Dimes revised its NBS recommendation from 9 disorders to the 30 disorders recommended in the ACMG study.

Expansion is supported by a number of Virginia-based associations including: VA Chapter of March of Dimes, VA Chapter of American Academy of Pediatrics, VA Association of Health Plans, VA Hospital & Healthcare Association (VHHA), VA Genetic Advisory Committee (VaGAC) and DMAS. VHHA indicated however the need for the societal benefits to be borne "fairly by all those who benefit." VaGAC, which is charged with recommending changes in the NBS program to the VA Board of Health suggests limiting the initial expansion to the disorders that can be tested on the tandem mass spectrometer to allow for further study on screening of cystic fibrosis (CF) and Glucose 6 Phosphate Deficiency (G6PD).

Expansion of the NBS Panel in Virginia

The Division of Consolidated Laboratory Services (DCLS) within the Department of General Services completes the NBS testing. DCLS has the equipment to complete testing for 28 of the 30 disorders included in the ACMG study (exceptions are CF and G6PD). A few additional staff with expertise in interpreting screening results will be needed. VDH (with DCLS) will need to educate and provide technical assistance to providers, expand databases, and enhance services for children who are medically indigent children or have certain disorders. VDH contracts with EVMS, UVA, and VCU to provide expert consultation, diagnostic testing, and treatment will need to be expanded.

Even if the decision is made to expand NBS as soon as possible, there will be a delay (likely to be until March 2006) before the expanded screening will be implemented. Virginia Newborn Screening Services and DCLS in cooperation with VaGAC will be adding information regarding expanded, supplemental screening options to their NBS pamphlet. The pamphlet is typically given to parents of newborns in hospitals. However, efforts are underway to provide the pamphlets to parents during the prenatal period. Moreover, information about expanded, supplemental screening options will be included on the VDH website.

OPTIONS AND PUBLIC COMMENTS

The following options were proposed and public comments received regarding those options. The Options that were approved by JCHC are shown in bold text.

Option I: Take no action.
No comments were received in support of Option I.

Option II: Introduce legislation to amend the *Code of Virginia*, Title 32.1 Chapter 2 to expand Virginia's panel for newborn screening to include the disorders recommended by the Secretary of Health and Human Services that may be tested on the tandem mass spectrometer (effective date March 2006).

Two comments were received specifically in support of Option II.
Albert B. Finch, MD, FAAP, Executive Medical Director of Children's Hospital of the King's Daughters commented in support of the Virginia Hospital and Healthcare Association (VHHA) position "with the qualification that possibly some of the additional tests may not meet the criteria" for required testing.
Dr. Finch was more supportive of testing that could be completed via the tandem mass spectrometer. Nancy Ford commented on behalf of VAGAC in stating that Virginia's "panel should be expanded to include those disorders currently listed as the 'uniform panel' in the ACMG study that are screened by tandem mass spectrometry" and that additional time should be given for further study regarding the inclusion of cystic fibrosis and G6PD and for the submission "funding options and proposed panel expansion dates."

Option III: Introduce legislation to amend the Code of Virginia, Title 32.1 Chapter 2 to expand Virginia's panel for newborn screening to include all of the disorders recommended by the Secretary of Health and Human Services (effective date March 2006).

Three comments were received specifically in support of Option III while three additional comments were received in support of expansion in general without specifying a preference for Option II or Option III.
Delegate Kenneth R. Plum, the Family Support Group, and Pediatrix Medical Group and Pediatrix Screening commented in support of Option III. Delegate Plum stated: "With the advances in technology and scientific research, it is possible to expand the current program in Virginia to include 30 disorders recommended by the United States Secretary of Health and Human Services. I would be honored to assist with the necessary statutory and budget amendments." The comment from Pediatrix Medical Group and Pediatrix Screening noted: "We respectfully request the Virginia Joint Commission on Health Care to consider public-private partnerships as a cost-effective means to deliver a comprehensive and quality newborn screening program."
Jana A. Monaco, a parent of a child who was affected severely by an undiagnosed metabolic disorder and Board member of the Organic Acidemia Association, VDA, and VHHA commented in support of expanding the NBS panel without specifying a preference between Options II or III. In supporting expansion via Options II or III, VDH wrote: "any action to expand the newborn screening program will require changes to *Code of Virginia* pertaining to treatment services, as well as additional fiscal resources. VDH requests the JCHC to propose amendments to § 32.1-67 that would enable VDH to adequately respond to the treatment needs of individuals served by the program. Currently, this section of the *Code of Virginia* does not provide VDH with the clearly defined ability to meet management and treatment needs under an expanded screening panel. VDH believes a better result can be achieved through regulatory action by the Board of Health, pursuant to legislative direction, rather than by attempting to include specific program rules and requirements in statute, as is currently the case. Modifications to existing regulations governing the Newborn Screening and Children

with Special Health Care Needs Programs, developed pursuant to the public participation requirements of the Administrative Process Act, would provide a more appropriate and responsive avenue to prescribe VDH's responsibility with regards to management and scope of treatment. VDH prefers this approach to enable the agency to react appropriately to anticipated increases in program participants as well as changes in the ever transforming genetics field."

Option IV: Introduce a budget amendment for as much as $1.15 million in general funds to fund ~~all or~~ a portion of the expanded screening for FY 2006. Include on the 2005 workplan of the Joint Commission, consideration of continued funding of the expanded program.

Three comments were received in support of Option IV – Albert B. Finch, MD, FAAP, for Children's Hospital of the King's Daughters, VDA, and VHHA. VDH noted: "Additional required resources for management and treatment needs resulting from newborn screening can be estimated. However these needs will likely change as the actual incidence of these rare conditions is realized in the Commonwealth. For example, certain conditions are much more prevalent among certain sub-populations. Most importantly, treatment costs vary significantly from screening costs. Since infants are screened, for the most part, only once, the costs associated with the screening itself are more finite and predictable. Medical management and treatment for diagnosed infants, however, will span many years with the cohort of those entering treatment growing every year. Since the management and treatment costs are projected to grow on an annual basis, VDH prefers that funding for follow up, management, and treatment be appropriated through a budget amendment using General Funds as proposed by Policy Option 4. This funding approach will provide VDH with more fiscal stability to support mandated services."

Option V: Introduce a budget amendment (language only) directing the Department of General Services to increase newborn screening user fees to fund ~~all or a~~ the testing portion of the expanded screening. (It is estimated that an increase from $32/filter to a range of $41 to $48 per filter would fund the screening-related activities ~~while an increase of an additional $4.50 or $6.00 per filter would fund screening, educational, and follow-up activities~~).

No comments were received in support of Option V. Three comments were received in opposition of Option V – Albert B. Finch, MD, FAAP for Children's Hospital of the King's Daughters, VDA, and VHHA. VHHA indicated that the benefits of NBS "saves society as a whole from expense by preventing the need for lifelong support of needlessly disabled citizens….and the burden of funding these programs must be borne fairly by all those who benefit and should not be imposed primarily on hospitals." VDH's letter stated: "If a user fee, as proposed in JCHC Option 5, is the sole source of funding for an expansion of Newborn Screening Services, then the ability to ensure that adequate resources are maintained for follow up, management, and treatment will be limited and funding shortfalls are likely to occur within several years."

Option VI: Introduce a joint resolution and accompanying budget amendment (language only) requesting the Board of Health to submit a plan for expanding Virginia's panel of newborn screening disorders.

No comments were received in support of Option VI.

REVIEW OF OPTIONS FOR VIRGINIA'S NEWBORN SCREENING PROGRAM

Joint Commission on Health Care

Kim Snead

May 4, 2004
Richmond, Virginia

Authority for the Study

- House Joint Resolution 164 (Delegate Plum) directed JCHC to review information regarding newborn screening programs for metabolic disorders including:
  - The disorders screened for in other states
  - The benefits of the screenings
  - Cost of screening programs.
- HJR 164 directs JCHC to submit, prior to the 2005 Session, an executive summary and the information collected about newborn screening to be processed as a legislative document.
- HJR 164 was adopted by the General Assembly.

Presentation Outline

- ***Newborn Screening in the United States***
- **Newborn Screening in Virginia**
- **Next Steps**

Joint Commission on Health Care

History of Newborn Screening Programs in the United States

- Newborn screening programs began in the 1960s, after Dr. Robert Guthrie developed both a screening test for phenylketonuria (PKU) and a means of preserving blood samples on filter paper to allow for an inexpensive, simple means of screening a large number of newborns.
- PKU was only the first of a number of metabolic and genetic disorders that have been recognized in the last 40 years to be capable of causing serious health problems, mental retardation, and in some instances death.

Joint Commission on Health Care

Early Developments in Newborn Screening in the United States

1970s
- 43 states have statutes requiring screening of newborns
- Centers for Disease Control and Prevention (CDC) establishes the Newborn Screening Quality Assurance Program (NSQAP)
- Federal legislation is passed to support screening for certain genetic diseases such as sickle cell diseases

1980s
- 34 states receive federal funding to improve NS programs
- Council of Regional Networks for Genetic Services (CORN) established and publishes guidelines for 5-part system – screening, follow-up, diagnosis, treatment/management, evaluation (of NS)
- 12 states allow in statute for charges of fees for screening

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Newborn Screening

- Today, newborn screening is "recognized internationally as an essential, preventive public health program for early identification of disorders in newborns that can effect their long term health." (National Newborn Screening & Genetics Resource Center Website)
- Some form of newborn screening is conducted in each of the 50 states. It should be understood, however, that screening simply indicates the presence of metabolic or genetic markers, screening is not a foolproof diagnosis of the presence of a disorder
  - False positives and false negatives are generated. In part this is because states conservatively set the parameters for "normal results" in order to minimize false negatives.
  - As technology advances, the number of false positives and false negatives will be reduced.
- More than 4 million newborns are screened each year in the United States.

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Newborn Screening Programs Vary by State as There Are Few Federal Requirements

- There is no federal requirement for states to screen newborns for metabolic or genetic disorders
  - *Newborn Screening: Characteristics of State Programs*, a study published by GAO reported: "With the exception of federal recommendations that newborns be screened for PKU, congenital hypothyroidism, and sickle cell diseases, there are no federal guidelines on the set of disorders that should be included in state screening programs."
  - "All laboratories that perform testing for state newborn screening programs voluntarily participate in CDC's Newborn Screening Quality Assurance Program (NSQAP). This enables them to meet the federal regulatory requirement under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to have a process for verifying the accuracy of tests they perform."
- GAO also reported "in deciding which disorders to include in their programs, states generally consider similar criteria, such as how often the disorder occurs in the population, whether an effective screening test exists, and whether the disorder is treatable."

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GAO Found that States Varied Significantly in Screening Required for Newborns (Dec. 2002)

# of Disorders	States
3	3 (MT, SD, WV)
4	8 (AR. CA, KS, KY, ND, OK, UT, WA)
5	11 (AL, DE, FL, ID, LA, MN, MS, MO, NE, TN, TX)
6	8 (AK, IA, NV, NH, NM, PA, SC, WY)
7	4 (CO, HI, MI, VT)
8	4 (AZ, CT, GA, **VA**)
9	4 (IN, ME, MD, RI)
10	2 (MA, NY)
12, 14	OH & NJ respectively
21, 27	WI & IL respectively
32, 33	NC & OR respectively

Source: *Newborn Screening* GAO-03-449 (March 2003)

Joint Commission on Health Care

In 2002, States Reported Significantly Different Screening Fees and Per-Infant Expenditures to GAO

- Newborn screening fees ranged from **"no fee" in 7 states** (GA, KS, NY, PA, SD, WV, WY) **to a high of $60 in California** (which had required screening for four disorders but conducted a pilot program that screened for up to 28 disorders)
  - — Virginia's fee for screening for 8 disorders was $27.
    (Virginia's current fee for 9 disorders is $32.)
- Reported average expenditures for each infant screened ranged from **$14.75 in NC to $61.28 in Delaware** (for the 45 states that reported an estimate)
  - — Virginia's estimated per infant expenditures were reported to average $30.89 (as compared with the $27 fee).

Source: *Newborn Screening* GAO-03-449 (March 2003)

Five States Have Significantly Increased the Number of Screened Disorders Since the GAO Report

- According to the **Save Babies Through Screening Foundation**, 5 states have significantly increased the number of disorders included in their newborn screening programs since 2002
  - — Alaska, Indiana, Nevada, and North Dakota screen for 30 or more disorders
  - — Mississippi screens for 55 disorders.

Recommendations for Newborn Screening

- The **March of Dimes** recommends screening for the 9 disorders that Virginia now includes in its program.
- The **Save Babies Through Screening Foundation** recommends screening for the 55 disorders that Mississippi screens.

Presentation Outline

- ❑ **Newborn Screening in the United States**
- ■ ***Newborn Screening in Virginia***
- ❑ **Next Steps**

Code of Virginia, Title 32.1, Chapter 2 Delineates Requirements for Screening of Newborns in Virginia

- Except for an "infant whose parent or guardian objects..on the grounds that such [newborn screening] test conflicts with his religious practices or tenets"
  - Every infant born in Virginia is required to be screened for eight specified metabolic disorders
  - Every infant born in Virginia "determined at risk" for sickle cell diseases is required to be screened for those diseases.
    - However, since, it is simpler and "safer" to screen all infants that is the practice in Virginia
  - Beginning July 1, 2004, the physician or certified nurse midwife who is responsible for the newborn's care after rather than during delivery will be responsible for ensuring the newborn screening test is performed. (HB 1133 – 2004)

Source: *Code of Virginia* §32.1-65.

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History of Legislative Requirement for Newborn Screening in Virginia

1963 Phenylketonuria (PKU)

1978 Congenital Hypothyroidism (CH)

1984 Maple Syrup Urine Disease (MSUD)
Homocystinuria (HCU)
Galactosemia (GAL)

1986 Biotinidase Deficiency

1989 Sickle Cell Diseases (SCD)

2002 Congenital Adrenal Hyperplasia (CAH)

2004 Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)

Joint Commission on Health Care

Code of Virginia, Title 32.1, Chapter 2 Delineates Requirements for Screening of Newborns in Virginia

- The State Health Commissioner is responsible for administration of the newborn screening program in Virginia.
- The Board of Health is responsible for establishing procedures for:
  - Making recommendations regarding the treatment of the 9 specific disorders
  - Providing for "treatment for infants in medically indigent families"
  - Providing parents/guardians of infants identified as having PKU with "special food products …[and] with such funds as are appropriated…reimbursement from the Department for the cost of such special low protein modified foods in an amount not to exceed $2,000 per diagnosed person per year."

Source: *Code of Virginia* §32.1-67.

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Screening of Newborns in Virginia

- In order to screen newborns for metabolic disorders, blood is taken from the heel of the newborn and collected on special filter paper which must be purchased by the hospital, birthing center, or health care provider.
- Time intervals, established for when blood should be drawn to ensure accurate readings, are included in the *Virginia Administrative Code*, Title 12
  - For full-term infants with an attended birth, blood samples should be taken just prior to discharge but not before the infant is 24 hours old or after the infant is 3 days old.
  - For pre-term infants (gestation period of less than 38 weeks) with an attended birth, blood samples should be taken at 7 days of age or at discharge whichever occurs first
  - For unattended births, "the first attending health care provider shall cause the initial newborn screening tests to be performed" at that time. If testing is performed before the infant is 24 hours old, testing should be repeated before the infant is 14 days old.

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Blood Sample Analysis Is Completed by the Division of Consolidated Laboratory Services (DCLS) Within the Department of General Services

- DCLS reported for Calendar Year 2003:
  - 100,000 initial screens
  - 12,000 repeat screens
  - Screening was completed for Virginia hospitals, birthing centers , health care providers and 7,465 screenings for 11 military bases

Disorder	Abnormal Screens	Diagnosed Cases
PKU	**18**	**7**
Congenital Hypothyroidism CH	**1,646**	**24**
Maple Syrup Urine Disease MSUD	**19**	**1**
Homocystinuria HCU	**25**	**0**
Galactosemia GAL	**295**	**3**
Biotinidase Deficiency	**30**	**3**
Sickle Cell Diseases SCD	**5,489** (carriers too**)**	**85** (disease only)
Congenital Adrenal Hyperplasia CAH	**853**	**6**

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Cost of Newborn Screening in Virginia

- From the inception of the newborn screening program in 1963 until 1992, program costs were funded by State general funds
- Beginning in 1992, the filter paper kits had to be purchased from DCLS
  - July 1992, testing charge was set at $16/sample (7 disorders)
  - January 2002, charge was increased to $27/sample (8 disorders)
  - March 2004, charge was increased to $32/sample (9 disorders)
- DCLS' newborn screening (NBS) laboratory services are completely supported by user fees through the $32/sample charge which funds :
  - NBS laboratory costs (personnel, fixed assets, materials and supplies, and contractual services including courier services)
  - VDH educational efforts regarding the screening program
  - Some of VDH's costs related to follow-up activities.
- DCLS budget for FY 2003 was $3.1 million
  - $383,000 (12.4% of the budget) was allocated to VDH to partially fund follow-up services.

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Understanding the Operation of Tandem Mass Spectrometers (MS/MS)

- MS/MS are complex analytical instruments that "can be thought of as two mass spectrometers in series connected by a chamber that can break" a blood sample down for sorting and weighing
  - — MS/MS sorts through hundreds of compounds in blood to sort out the molecules of interest by "electronically" weighing molecules; every molecule has a unique weight or mass and MS/MS can simultaneously check for a number of different types of molecules within a single analysis that takes minutes (60,000,000,000,000,000,000,000 water molecules with a mass of 18 for each molecule would fit in a tablespoon)
  - — MS/MS determines not only the presence of molecules but also the "amount" that is present in the blood sample
  - — MS/MS is very precise in its measurements, producing fewer false positives than other available screening instruments.

Source: *A Layperson's Guide to Tandem Mass Spectrometry and Newborn Screening,* Donald H. Chace, Ph.D., M.S.F.S.

Purchase of the Tandem Mass Spectrometers

- DCLS purchased three tandem mass spectrometers in 2004 (total cost of $850,000), MS/MS were needed to test for MCADD
  - — Analysis of 3 disorders (PKU, MSUD and HCU) were moved to the MS/MS.
  - — Two scientists and 1 computer support technician were hired to support the MCADD testing bringing the total number of staff for the laboratory and the computer entry section to 25.

Presentation Outline

- ❑ **Newborn Screening in the United States**
- ❑ **Newborn Screening in Virginia**
- ■ ***Next Steps***

Completion of the Review of Newborn Screening

- Complete review (to include working with a subgroup of the Virginia Genetics Advisory Committee) to evaluate the 55 metabolic and genetic disorders identified by the **Save Babies Through Screening Foundation** on such factors as:
  - — Are the effects of the disorder understood?
  - — Can the disorder be prevented, treated, or the effects minimized if detected early and action is taken?
  - — Is testing for the disorder reliable, practical, and what will it cost?
  - — Can the testing be completed by the MS/MS?
  - — Who is most likely to bear any increase in the cost of screening (parents, insurers, health care providers)?
  - — What are the options for screening for additional disorders and what are the associated costs?
- Develop Options for improving Virginia's newborn screening program for JCHC consideration.

FINAL REVIEW OF OPTIONS FOR VIRGINIA'S NEWBORN SCREENING PROGRAM

Joint Commission on Health Care

Kim Snead

October 26, 2004
Richmond, Virginia

Authority for the Study

- House Joint Resolution 164 (Delegate Plum) directed JCHC to review information regarding newborn screening programs for metabolic disorders including:
  - The disorders screened for in other states
  - The benefits of the screenings
  - Cost of screening programs.
- HJR 164 directs JCHC to submit, prior to the 2005 Session, an executive summary and the information collected about newborn screening to be processed as a legislative document.
- HJR 164 was adopted by the General Assembly.

History of Newborn Screening Programs in the United States

- Newborn screening programs began in the 1960s, after the effects of phenylketonuria (PKU) were identified and a method was developed to preserve blood samples on filter paper to allow for an inexpensive, simple means of screening a large number of newborns.
- Since the 1960s, a number of metabolic and genetic disorders have been identified as being capable of causing serious health problems, mental retardation, and in some instances death.

Newborn Screening in the U.S.

- Today, some form of newborn screening is conducted in each of the 50 states
  - The number of disorders screened ranges from 3 to 54
    - Virginia screens for 9 distinct disorders (and hearing loss)
  - More than 4 million newborns are screened each year in the United States
    - Virginia analyzes approximately 100,000 initial screens per year.

Newborn Screening Programs Vary by State as There Are Few Federal Requirements

- There is no federal requirement for states to screen newborns for metabolic or genetic disorders
  - *Newborn Screening: Characteristics of State Programs*, a study published by GAO reported: "With the exception of federal recommendations that newborns be screened for PKU, congenital hypothyroidism, and sickle cell diseases, there are no federal guidelines on the set of disorders that should be included in state screening programs."
- However, it is expected that the Secretary of Health and Human Services will issue a new recommendation based on the findings of a recently released study by the American College of Medical Genetics (ACMG) – Michael S. Watson, Ph.D. primary author.

Developments at the Federal Level Regarding Recommendations for Newborn Screening

- The purpose of the 3-year ACMG study was to develop a "uniform panel of newborn screening conditions" to recommend for use by all states
  - The study was requested by the Maternal and Child Health Bureau of the Health Resources and Services Administration (HRSA)
  - The study findings were presented in September to an Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns or Children established by the Secretary of Health and Human Services.
- The ACMG study recommended a core panel of 30 disorders that are specifically screened for which will produce another 25 "report-only" disorders in the process
  - Virginia currently screens for 12 of the 30 disorders as they are listed in the ACMG study (11 inborn errors of body chemistry and hearing loss).

Developments at the Federal Level Regarding Recommendations for Newborn Screening

- The disorders recommended for screening reflect the ACMG study participants' judgment that the disorders in the core panel meet the criteria for scoring disorders for inclusion:

Incidence of conditions	Identifiable at birth
Burden of disease	Availability of test
Test characteristics	Availability of treatment
Cost of treatment	Efficacy of treatment
Benefits to individual	Benefits to family and society
Mortality prevention	Diagnostic confirmation
Acute management	Simplicity of therapy

Source: Watson, M.S., *Standardization of Outcomes and Guidelines for State Newborn Screening Programs.*

Developments at the Federal Level Regarding Recommendations for Newborn Screening

- The Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns or Children is expected to recommend a panel of disorders for use by the Secretary of Health and Human Services as a guideline for states in structuring their newborn screening programs
  - The recommended panel is expected to be different by only one or two disorders from the ACMG study recommendations according to discussions taking place at the federal level.

The March of Dimes Has Revised its Recommendation on Newborn Screening

- On September 22 2004, the March of Dimes revised its recommendation on newborn screening from 9 disorders to the 30 disorders recommended in the ACMG study.

"We will urge every state to screen every baby for at least the 30 disorders listed in the ACMG report. These 30 disorders meet our inclusion criteria, and include all of the nine metabolic tests plus hearing screening contained in our previous policy. We will urge states to provide test results for an additional 25 'reportable' conditions named in the ACMG report for which there are reliable tests but not yet documented treatments."

Source: March of Dimes Statement, September 22, 2004.

Considerations in Implementing the Recommended Panel in Virginia

Basic Outline of VA's System for Newborn Screening

Screening of blood samples submitted by hospitals, physicians, nurse midwives, etc.	Div. of Consolidated Laboratory Services (DCLS) of DGS
Monitoring and initial follow-up on abnormal screens reported by DCLS	VDH staff
Providing expert consultation, diagnostic testing as available, treatment as appropriate, genetic counseling, etc.	VDH contracts with EVMS, UVA, and VCU for services of physicians and nutritionists
Providing services to medically indigent children and children who have certain types of disorders as well as monitoring to ensure resolution of all abnormal screens.	VDH staff

Considerations in Implementing the Recommended Panel in Virginia

- DCLS has the equipment to complete testing for 28 of the 30 disorders (shown as appendix to these slides on teal paper) included in ACMG study; exceptions being cystic fibrosis (CF) and glucose-6 phosphate dehydrogenase deficiency (G6PD).
  - A few additional staff with expertise in interpreting screening results will be needed.
- VDH in consultation with DCLS will need to educate and provide additional technical assistance to hospitals, physicians, and other health care providers; expand databases that track screening and follow-up results; and enhance services provided for medically indigent children and children with certain types of disorders. These activities will require a few additional VDH staff.
- Contracts with EVMS, UVA, and VCU will need to be expanded.

Reactions to Expansion of Recommended Panel in Virginia

- Expansion of the number of screened disorders is supported with some caveats
  - Virginia Chapter of March of Dimes
  - Virginia Chapter of the American Academy of Pediatrics
  - Virginia Association of Health Plans
  - Virginia Department of Medical Assistance Services
  - Virginia Hospital & Healthcare Association
    - Benefits of screening are societal benefits; "screening for treatable conditions is an important function that can save babies from a lifetime of preventable impairment, saves their families from the associated burden, and saves society as a whole from expense by preventing the need for lifelong support of needlessly disabled citizens. However, those benefits are truly societal benefits and the burden of funding these programs must be borne fairly by all those who benefit."
  - Virginia Genetic Advisory Committee (VaGAC)
    - Suggests expansion to include disorders that can be tested on MS/MS but to allow for further study on screening of CF and G6PD
    - Requests "the opportunity to submit additional recommendations in the near future regarding funding options and proposed panel expansion dates."

Options

- Take no action.
- Amend the *Code of Virginia*, Title 32.1 Chapter 2 to expand Virginia's panel for newborn screening to include the disorders recommended by the Secretary of Health and Human Services that may be tested on the tandem mass spectrometer (effective date of March 2006).
- Amend the *Code of Virginia*, Title 32.1 Chapter 2 to expand Virginia's panel for newborn screening to include all of the disorders recommended by the Secretary of Health and Human Services (effective date of March 2006).
- Introduce a budget amendment to fund all or a portion of the expanded screening through a general fund appropriation rather than through an increase in user fees. General estimate for the entire increase $2.3 million per year.
- Introduce a budget amendment (language only) directing the Department of General Services to increase newborn screening user fees to fund all or a portion of the expanded screening. (General estimate of increase: from $32/filter to $50/filter for testing and educational activities or $55/filter for testing, educational, and follow-up activities.)
- Introduce a joint resolution and accompanying budget amendment (language only) requesting the Board of Health to submit a plan for expanding Virginia's panel of newborn screening disorders.

Joint Commission on Health Care

APPENDIX A

ENROLLED

HOUSE JOINT RESOLUTION NO. 164

Directing the Joint Commission on Health Care to collect information concerning infant screening program for metabolic disorders. Report.

Agreed to by the House of Delegates, February 17, 2004
Agreed to by the Senate, March 9, 2004

WHEREAS, metabolic disorders involve defects produced by inactive genes that prevent the body from making enzymes necessary to break down certain amino acids or fats; and

WHEREAS, metabolic disorders are rare, but the consequences of these disorders if undetected or untreated are usually severe, often resulting in neurological impairment, mental retardation, and even death; and

WHEREAS, these harmful effects can often be reduced or even avoided when such disorders are detected in infants and the appropriate dietary or other treatment is prescribed; and

WHEREAS, § 32.1-65 of the Code of Virginia currently provides that "In order to prevent mental retardation, permanent disability or death, every infant who is born in this Commonwealth shall be subjected to a screening test for biotinidase deficiency, phenylketonuria, hypothyroidism, homocystinuria, galactosemia, congenital adrenal hyperplasia, and Maple Syrup Urine Disease, and each infant determined at risk shall be subject to a screening test for sickle cell diseases"; and

WHEREAS, the State Board of Health is required to recommend procedures for treating these disorders, and is required to provide such treatment for infants in medically indigent families; and

WHEREAS, many metabolic disorders may still go undetected and untreated because current screening requirements are too limited or current screening procedures are not utilizing available, improved technologies; and

WHEREAS, new technologies such as tandem mass spectrometry can improve diagnoses and expand infant screening to 20 or more metabolic disorders; now, therefore, be it

RESOLVED by the House of Delegates, the Senate concurring, That the Joint Commission on Health Care be directed to collect information concerning infant screening program for metabolic disorders.

In collecting the information, the Joint Commission on Health Care shall compile a list of the (i) types of metabolic disorders for which infants are screened in other states, including a summary of the benefits of such screening; and (ii) the costs of such screening programs.

Technical assistance shall be provided to the Commission by the State Department of Health and the Department of Mental Health, Mental Retardation and Substance Abuse Services. All agencies of the Commonwealth shall provide assistance to the Commission in collecting the information, upon request.

The Joint Commission on Health Care shall submit to the Division of Legislative Automated Systems an executive summary and the information collected on infant screening programs for metabolic disorders no later than the first day of the 2005 Regular Session of the General Assembly. The executive summary and information shall be submitted as provided in the procedures of the Division of Legislative Automated Systems for the processing of legislative documents and reports and shall be posted on the General Assembly's website.

JOINT COMMISSION ON HEALTH CARE

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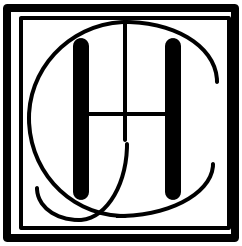

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