Information on Virginia's Newborn Screening Dried Blood Spot Program

Objectives

- Review Newborn Screening and its history, nationally and in Virginia
- Review Virginia Code and Regulations relevant to the newborn screening dried blood spot program
- Identify common conditions that are found through the Newborn Screening Programs in Virginia
- Describe the newborn screening dried blood spot process in Virginia
- Discuss the hospital and nurse role in the newborn screening dried blood spot process

Baby's First To-Do List

1. Meet Parents
2. Sleep
3. Newborn Hearing Screening
4. Heart Disease Screening
5. Heel Stick
6. Sleep, Eat, & Sleep some more...

#3, #4 and #5 on your Baby's To-Do List are just as easy as the others!

Make Sure You Know Your Baby's Results!

VIRGINIA NEWBORN SCREENING SERVICES

For Genetic Screening:
Call: 804-864-7712

VDH VIRGINIA DEPARTMENT OF HEALTH

www.vdh.virginia.gov

Virginia VEHDI Early Hearing Detection and Intervention Program

For Newborn Hearing:
1-866-493-1090 or
TTY users dial 7-1-1 for Virginia Relay

www.facebook.com/VDHGenetics
www.facebook.com/vehdi

Mission of Newborn Screening

To prevent intellectual disability, permanent disability, or death through early identification and treatment of infants who are affected by certain heritable disorders and genetic diseases.

The Virginia Newborn Screening Program is a coordinated and comprehensive system consisting of education, dried-blood spot screening tests, follow up and referral, diagnosis, medical and dietary management, and treatment.

What is screened?

The Recommended Uniform Screening Panel aka RUSP

http://www.hrsa.gov/advisorycommittees/mchbadvisory/heritabledisorders/recommendedpanel/uniformscreeningpanel.pdf

- 34 core heritable disorders and genetic diseases are included in the HHS RUSP
  - These disorders may cause severe intellectual disability, illness, or death if not treated early in life.
  - If treated, infants may live relatively normal lives.
  - Results in savings in medical costs over time.

Statistics

- Over 4 million births per year in the U.S.
- ~1 in every 1500 births is affected by a condition screened for by NBS
- ~ Over 3,000 babies identified each year in the US with severe disorders
- ~ 100,000 births per year in Virginia
- ~ Since 1966, over 3,000 infants diagnosed with a disorder detected through NBS

History of NBS in Virginia

YEAR	DISORDER
1966	PKU
1984	Congenital hypothyroidism, Maple Syrup Urine Disease, Homocystinuria, Galactosemia
1986	Biotinidase Deficiency
1989	Hemoglobinopathies (eg. Sickle cell)
2000	Hearing
2002	Congenital adrenal hyperplasia
2004	MCAD
2006	CF & 18 other metabolic disorders
2014	CCHD
2015	SCID
2016	50 years of NBS in Virginia

50th National Anniversary of NBS 2013

Virginia Code

§ 32.1-65. Certain newborn screening required

http://lis.virginia.gov/lis.htm

- Every infant who is born in the Commonwealth shall be subjected to screening tests, consistent with, but not necessarily identical to the uniform screening panel
- Any infant whose parent or guardian objects thereto on the grounds that such tests conflict with his religious practices or tenets shall not be required to receive such screening tests
- The physician or certified nurse midwife in charge of the infant's care after delivery shall cause such tests to be performed. The screening tests shall be performed by the Division of Consolidated Laboratory Services or any other laboratory the Department of Health has contracted with to provide this service.

Virginia Code
§ 32.1-66. Commissioner to notify physicians; reports to Commissioner.

Whenever a newborn screening test result indicates suspicion of any condition pursuant to § 32.1-65, the Commissioner **shall notify forthwith the attending physician** and shall perform or provide for additional testing required to confirm or disprove the diagnosis. All physicians, certified nurse midwives, public health nurses, or any nurse receiving such test result, and administrators of hospitals in the Commonwealth, shall report the discovery of all cases of any condition for which newborn screening is conducted pursuant to § 32.1-65 to the Commissioner for infants and children up to two years of age.

Virginia Regulations
Chapter 71. REGULATIONS GOVERNING VIRGINIA NEWBORN SCREENING SERVICES

- 12VAC5-71-70. Newborn dried-blood-spot screening specimen collection, specimen submission, and notification for hospital deliveries

- K. The healthcare provider in charge of the infant's care, on receiving notice of the infant's screened-abnormal result, shall:
  - 1. Cause the collection of a repeat newborn dried-blood-spot specimen for repeat or confirmatory testing as soon as possible but no later than two business days after notice;
  - 2. Cause the submission of the specimen; and
  - 3. Take immediate action, as instructed, when notified of a critically abnormal screening result.

http://lis.virginia.gov/lis.htm

12VAC5-71-90. Responsibilities of the chief executive officer.

- Shall assure that the hospital providing birthing services develops and implements policies and procedures to make certain that the following steps take place:
  - 1. Collection of newborn dried-blood-spot screening specimens shall occur after 24 hours of birth, and collection and submission of the specimens shall meet the standards required by the testing laboratory;
  - 2. Notification of the newborn's physician of record or designee shall occur **within one business day in the event that the infant is discharged** before the newborn dried-blood-spot screening specimen has been collected;
  - 3. Communication of the newborn dried-blood-spot screening test results to the **newborn's physician of record** or designee shall occur so that test results may become part of the infant's medical record on file with the physician;
  - 4. The newborn screening dried-blood-spot results and treatment shall be **recorded in the patient's medical record**, and retention of the information shall comply with applicable medical record retention requirements; and
  - 5. Training of staff on newborn dried-blood-spot screening specimen collection and submission and parental notification shall be implemented in a way that ensures an adequately trained and knowledgeable workforce is maintained for implementing specimen collection and submission and parental notification according to standards required by the testing laboratory and guidance from the department.

Virginia's Genetic Services

- Screening Programs
  - Dried Blood Spot
  - Hearing Loss
  - Critical Congenital Heart Disease
- Care Connection For Children
- VaCARES (birth defects registry)

DCLS
Dept of General Services
Commonwealth of Virginia

Currently 29 Recommended Disorders Screened at DCLS

- 4 Amino acid metabolism disorders
- 2 Urea cycle (amino acid metabolism) disorders
- 9 Organic acid metabolism disorders
- 5 Fatty acid oxidation disorders
- 3 Hemoglobin disorders
- 2 Endocrine disorders
- 1 Vitamin disorder
- 1 Galactose disorder
- 1 Pulmonary disorder
- 1 Immunodeficiency

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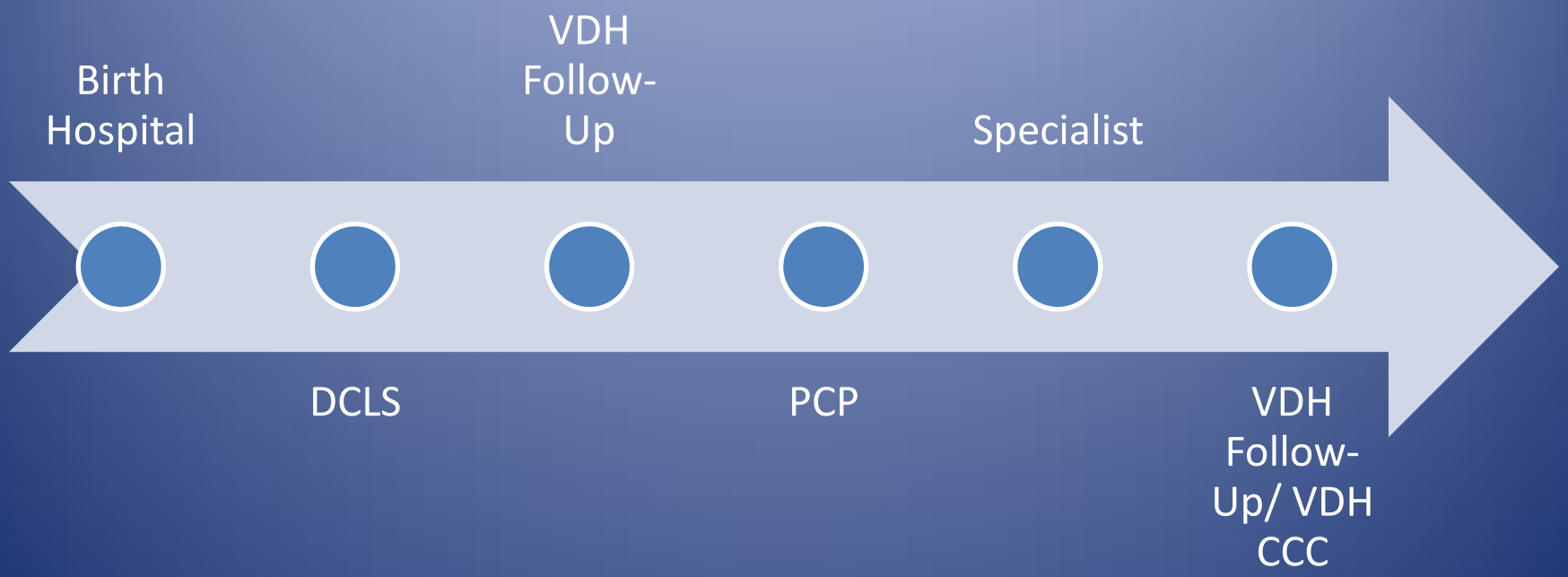
NBS Process


Birth Hospital
DCLS
VDH Follow-Up
PCP
Specialist
VDH Follow-Up/ VDH CCC

How does Newborn Screening work?

- Dried blood spot submitted to the Division of Consolidated Laboratory by the birth hospital
- Letters sent to healthcare provider and submitting hospital
- Follow-up staff calls healthcare provider concerning results that require immediate attention
- Encourage consultation with a metabolic and genetic specialist and sickle cell specialists (UVA, VCU, Children's Specialty Group)
- Upon confirmation of diagnosis, refer to Care Connection for Children
- Support funding for medical and dietary management

Parts of the NBS system

- 1. ***Collection***
  - ***Newborn nursery/NICU/Birthing Centers/mid-wives***
- 2. Analysis
  - DCLS
  - Diagnostic labs
- 3. Follow-up
  - VDH
  - Pediatricians
  - Specialists
  - Long term and transition care

DBS Collection Form

1 2 4 2 8 0 0 1

12428001 SN

FILTER COPY

YYYY-MM

PLEASE PRINT

BABY'S NAME: LAST | FIRST | MEDICAL RECORD NUMBER | BIRTH DATE | BIRTH TIME (MILITARY) | SEX ( ) MALE ( ) FEMALE

BIRTH WEIGHT GRAMS / LBS | ETHNICITY/RACE 1 ( ) BLK. 2 ( ) WHT. 3 ( ) HISP. 4 ( ) ASIAN 5 ( ) AMER. INDIAN 6 ( ) OTHER | BIRTH 1 ( ) SINGLE 2 ( ) TWIN A 3 ( ) TWIN B 4 ( ) TRIPLET A 5 ( ) TRIPLET B 6 ( ) TRIPLET C 7 ( ) OTHER | STATUS 1 ( ) NORMAL 2 ( ) SICK | 3 ( ) PREMATURE Check (✓) Only if <37 weeks | 4 ON ANTIBIOTICS ( ) Y ( ) N TYPE | 5 TRANSFUSED ( ) Y ( ) N DATE

DATE OF FIRST FEEDING | TYPE: 1 ( ) Breast Only 2 ( ) Cow's Milk Formula 3 ( ) TPN 4 ( ) Soy Formula 5 ( ) Breast + Formula 6 ( ) Other 7 ( ) NPO | DATE OF COLLECTION | TIME OF COLLECTION (MILITARY) | AGE AT COLLECTION 1 ( ) ≤ 24 HRS. 2 ( ) > 24 HRS. | SPECIMEN TYPE 1 ( ) INITIAL SPEC. 2 ( ) REPEAT SPEC.

MOTHER'S NAME: LAST | FIRST | AGE | MOTHER'S SSN | MOTHER'S TELEPHONE NUMBER

MOTHER'S ADDRESS | CITY | STATE | ZIP CODE (+4 CODE) | COUNTY OF RESIDENCE

PLACE OF BIRTH 1 ( ) HOSPITAL BIRTH 2 ( ) HOME BIRTH 3 ( ) OTHER | BIRTH HOSPITAL CODE | BIRTH HOSPITAL NAME | BIRTH HOSPITAL ADDRESS | CITY | STATE | ZIP CODE (+4 CODE)

DOCTOR CODE NUMBER | TELEPHONE NUMBER | (SUBMITTER) RESULT TO CODE | TELEPHONE NUMBER | FORM COMPLETED BY | FOR LAB USE: UNSAT CODE

BABY'S DOCTOR'S NAME | (SUBMITTER) RESULT TO NAME | SPECIMEN COLLECTED BY | Date / Int.

DOCTOR'S ADDRESS | SUBMITTER'S ADDRESS

CITY | CITY

STATE | ZIP CODE (+4 CODE) | STATE | ZIP CODE (+4 CODE)

Commonwealth of Virginia
Department of General Services
Newborn Screening Laboratory
600 N. 5th St.
Richmond, VA 23219
Telephone: (866) 378-7730
DGS-22-174 (Rev. 11/02)

Use By YYYY-MM

LOT W051 6261507 REF 1063782 Rev. 0

LOT W051 6261507 Whatman 903®

VDH VIRGINIA DEPARTMENT OF HEALTH
Protecting You and Your Environment

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:

Possible Causes:

1. Specimen quantity insufficient for testing.

- 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.

2. Specimen appears scratched or abraded.

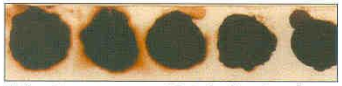
- Applying blood with a capillary tube or other device.

3. Specimen not dry before mailing.

- Mailing specimen before drying for a minimum of four hours.

4. Specimen appears supersaturated.

- Applying excess blood to filter paper, usually with a device.
- Applying blood to both sides of filter paper.

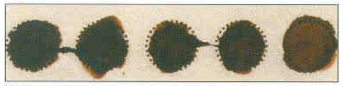


5. Specimen appears diluted, discolored or contaminated.

- 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.

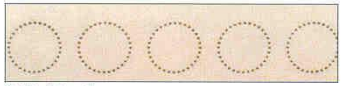
6. Specimen exhibits serum rings.

- 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.



7. Specimen appears clotted or layered.

- Touching the same circle on filter paper to blood drop several times.
- Filling circle on both sides of filter paper.



8. No blood.

- 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.

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Complications in Follow-Up

- False positive results in baby that were secondary to mother's metabolic status or prematurity.
- Language and cultural barriers to learning in family.
- Non-compliance due to perceived lack of disease since no obvious symptoms.
- Lack of insurance and medical care.
- ***Lack of knowledge of what newborn screening is.***
  - Confidentiality and retention of blood spots

The Diagnosis Comes In…Now What?

- Team reviews documentation and closes case.
  - If more info is needed we can communicate with PCP and/or specialists that PCP referred child to
- Automatic referral is made to Care Connection for Children (CCC)
  - Parents may/may not accept case management services through CCC

Care Connection for Children

- Statewide network of centers of excellence for children with special health care needs (CSHCN)
- Provide
  - leadership in the enhancement of specialty medical services
  - care coordination
  - medical insurance benefits evaluation and coordination
  - management of the CSHCN Pool of Funds
  - information and referral to CSHCN resources
  - family-to-family support
  - training and consultation with community providers on CSHCN issues.

Service Areas
For
Information, Referrals, & Care Coordination for Children and Youth with Special Health Care Needs

Care Connection Case Manager can help the child and family to:

- Find a pediatrician or family practice doctor
- Find specialty medical care
- Coordinate specialty care services
- Learn about their health insurance choices
- Become familiar with the health insurance that they have
- Explore health care payment options for services not covered by their health insurance
- Find information about and refer to community resources
- Talk to schools, doctors, & others
- Plan for the child's future
- Solve problems when they come up
- Find someone to listen when they do not know where to turn
- Connect with other families experiencing similar situations

Northern Virginia
Falls Church, Va.
703-531-3131
Toll Free: 1-866-222-0372

Blue Ridge
Charlottesville, Va.
434-924- 0222
Toll Free: 1-866-596-9367

Central Virginia
Richmond, Va.
804-827-1795
Toll Free: 1-866-737-5965

Southwest Virginia
Bristol, Va
276-645-4900
Toll Free: 1-800-704-1285

Roanoke Area
Roanoke, Va.
540-985-8470
Toll Free: 1-866-906-2999

Hampton Roads
Norfolk, Va.
757-668-7132
Toll Free: 1-800-864-8903

October 2010

VACares

- The development of VaCARES was mandated in 1985 and amended in 1986 and 2006 in the Code of Virginia, § 32.1-69.1. According to this mandate, hospitals are required to report cases of birth defects to VaCARES. The relevant information can be reported and reviewed without the prior written consent of the patient or family. The mandate also allows for review and evaluation of the system and activities.

VaCARES

The Virginia Congenital Anomalies Reporting and Education System (VaCARES) is a registry of children under age two with birth defects.

The mission of VaCARES is to:

- Collect data to evaluate possible causes of birth defects.
- Improve the diagnosis and treatment of birth defects.
- Establish a mechanism for informing families of children with birth defects and their physicians about available health resources.

Looking forward to the future

- Review of recently recommended disorders to Virginia's Panel
  - Pompe, MPS-1, X-ALD
- NBS education training modules
  - Medical practitioners---offers CEUs
  - Early Hearing Detection Module coming soon

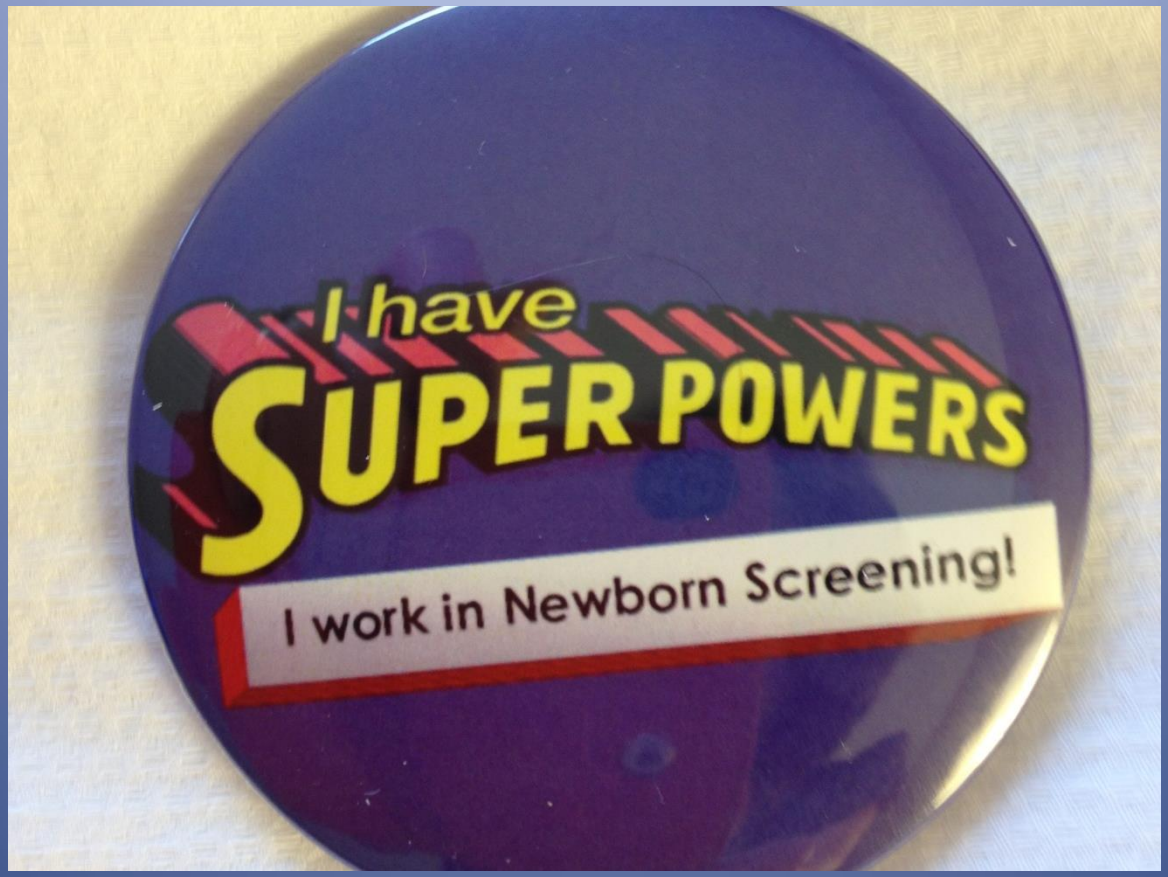
References

- Virginia Department of Health Newborn Screening website:
  http://www.vahealth.org

- Virginia Department of Health Newborn Screening website:
  http:/www.vdh.virginia.gov/ofhs/childandfamily/childhealth/gns/

- CDC website:
  http://www.cdc.gov/nbslabbulletin/bulletin.html

- Baby's First Test website
  http://www.babysfirsttest.org/



Thank you!