

**Virginia Genetics Advisory Committee**  
**Tuesday, April 17, 2007**  
**10:00 a.m. – 12:00 Noon**

**Division of Consolidated Laboratory Services**  
**Training Room T-23**  
**600 North 5th Street**  
**Richmond, VA 23219**  
**Telephone (804) 648-4480**

**MINUTES**

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**Present: VaGAC Members** (check=present)

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| <input checked="" type="checkbox"/> Willie C. Andrews, DCLS              | <input checked="" type="checkbox"/> Anil R. Kuma, VCU              |
| <input checked="" type="checkbox"/> Joann N. Bodurtha, VCU (VaGAC Chair) | <input checked="" type="checkbox"/> Sara Long, MOD                 |
| <input type="checkbox"/> Joanne Boise, VDH (Ad Hoc Member)               | <input type="checkbox"/> Walter Nance, VEHDIP Adv Cmt              |
| <input type="checkbox"/> Joan Corder-Mabe, VDH (Ad Hoc Member)           | <input checked="" type="checkbox"/> Kathy Odum, VDH                |
| <input checked="" type="checkbox"/> Nancy Ford, VDH (VaGAC Staff)        | <input checked="" type="checkbox"/> Arti Pandya, VCU               |
| <input type="checkbox"/> Virginia (“Ginny”) K. Proud, CHKD               | <input checked="" type="checkbox"/> James Pearson, DCLS            |
| <input checked="" type="checkbox"/> Jene O. Radcliffe-Shipman, VDH       | <input checked="" type="checkbox"/> Rafael Randolph, VDH           |
| <input checked="" type="checkbox"/> William (“Bill”) G. Wilson, UVA      | <input type="checkbox"/> Ramona Schaeffer, VDH                     |
| <input checked="" type="checkbox"/> Nancy Bullock, VDH                   | <input type="checkbox"/> Allison Schreiber, VCU                    |
| <input type="checkbox"/> Tia Campbell, VDOE                              | <input checked="" type="checkbox"/> Charlie Stevenson, DCLS        |
| <input type="checkbox"/> Mary Ann Discenza, DMHMRSAS                     | <input type="checkbox"/> David Suttle, VDH                         |
| <input checked="" type="checkbox"/> Laura Duncan, VCU                    | <input checked="" type="checkbox"/> Theresa Taylor, VDH            |
| <input checked="" type="checkbox"/> Audrey Greene, VDH                   | <input type="checkbox"/> Sharon Williams, VDH                      |
|  | <input checked="" type="checkbox"/> Melody Persinger Yeargin, CHKD |

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**Present: Others** (VaGAC Subcommittee members, guests)

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| <input checked="" type="checkbox"/> Stephen Braddock, UVA                | <input checked="" type="checkbox"/> Theresa Meadows, CHKD |
| <input checked="" type="checkbox"/> Kelcey Davis (for Charlie Stevenson) | <input checked="" type="checkbox"/> Jana Monaco, Parent   |
| <input checked="" type="checkbox"/> Karen Durst (for Mary Ann Discenza)  | <input checked="" type="checkbox"/> Phil Poston, DCLS     |
| <input checked="" type="checkbox"/> Sarah H. Elsea, VCU                  | <input checked="" type="checkbox"/> Susan Ward, VHHA      |
| <input checked="" type="checkbox"/> Valerie Luther, Parent               | <input checked="" type="checkbox"/> Thomas York, DCLS     |

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**Recorders:** Rafael Randolph, Nancy Ford

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**1. Welcome: J. Bodurtha**

- a. Introductions: Completed.
- b. Minutes of 10/3/06 Meeting: Approved as written.
- c. Agenda: Reviewed, no new items added.
- d. Membership (revised 4/17/2007): Circulated for corrections.

**2. Fetal Alcohol Spectrum Disorders: S. Braddock**

*PowerPoint Presentation:* “FAS/FASD Through the Ages.” Key points of presentation:

- Alcohol is the oldest and most widely used drug in the world.
- In 1991 FAS recognized as the leading known cause of mental retardation in the U.S.
- Alcohol-related birth defects are completely preventable.
- We don’t know what, if any, amount of alcohol is safe.
- Risk increases with how much you drink.
- 8,800 U.S. babies born with FAS annually.

- Health professionals should inquire about alcohol consumption by women of childbearing age.
- In the child's best interest for mother not to drink alcohol.
- Lifespan View of FASD – much of what we know is anecdotal, behavioral phenotype, IQ may not predict functional performance.
- Potential secondary disabilities: over 90% have mental health problems; 60% get into trouble with the law; 49% exhibit sexual misconduct; 60% are disruptive in school; 35% have problems with alcohol themselves; and 50% experience confinement.

3. **VaGAC Infrastructure: J. Bodurtha**

- a. Reviewed, no changes.
- b. Reminder: Subcommittee members can invite individuals to join their subcommittee without VaGAC approval.

4. **Subcommittees and Work Groups Reports**

a. **Newborn Screening Subcommittee: W. Andrews**

- *New Scientist*: Phil Poston, PhD, Lead Scientist, Newborn Screening, DCLS, introduced.
- *Meetings and Membership*: Subcommittee planning to convene more frequent meetings, probably quarterly, and expand its membership to include cystic fibrosis, endocrinology, and hematology experts.
- *International Data Collection Project*: DCLS is in process of determining if Virginia wants to participate in HRSA-funded grant conducted by Mayo Clinic for purpose of evaluating newborn dried-blood-spot screening cut-off values; currently, 33 states are participating.
- *Newborn Echocardiograms and Provider Education*: As follow up to a concerned citizen's request to Governor Kaine regarding birth defects—specifically that echocardiograms should be conducted on all newborns and there is a need for education of providers about birth defects, including hypoplastic left heart syndrome—the Subcommittee will confirm or not confirm recommendation that, given current information newborn, echocardiograms should not be conducted on all newborns and feasibility of increasing provider education about birth defects.
- *Molecular Testing for Hearing Loss*: This might be a future issue in Virginia. (i.e., that it be incorporated into core panel of conduction for which newborn screening is conducted). A request from W. Nance related to this issue was referred to the Subcommittee.
- *C0 Markers*: How to handle C0 Markers is a concern that is being addressed nationally. A request from V. Proud on this issue was referred to the Subcommittee.
- *Protocol*: Subcommittee will develop protocol for following up with requests from VaGAC Steering Committee to address questions or issues that have been referred by VaGAC to the Subcommittee.
- *NYMAC Focus Group*: There has been a request for family involvement in a NYMAC (New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services) focus group. J. Monaco will be the facilitator.
- *2007 Newborn Screening and Genetic Testing Symposium*: P. Poston will conduct a poster presentation, Challenges of Bringing Conformity Testing in Laboratories," at the May symposium entitled "Harmonization of Newborn Screening Systems."
- *Virginia Newborn Screening Services*: Of the 73 newborns that were diagnosed in the first year of the program's expansion, 30 were diagnosed with one of the new panel disorders.

- b. **Genetics and Public Health Subcommittee: J. Bodurtha**
    - *March of Dimes*: As MOD further develops an advocacy and education plan, will need help from VaGAC.
    - *State Genetics Education Plan Work Group*: This year's activities include developing modules for health care providers on family health history.
    - *Family Health History Month*: Governor Kaine has proclaimed November 2007 as "Family Health History Month" in the Commonwealth of Virginia. Leadership Metro Richmond is developing a family health history campaign and has involved VDH, via its Virginia Congenital Anomaly Tracking and Prevention Improvement Project (VaCATPIP), in campaign activities.
    - *Future Issues*: Might involve privacy and discrimination.
  - c. **Community Partnerships Subcommittee, Parent Involvement Work Group: J. Monaco**
    - *Meetings*: Work Group will not have face-to-face meetings; instead, work group will meet via conference calls as needed, which has worked well in the past.
  - d. **VDH Contractors Subcommittee: N Ford for S. Williams**
    - *Site Visits*: VDH conducted site visits recently; the visits were part of a recent VDH Genetics Needs Assessment, which included surveying contractors (EVMS, UVA, VCU, and GIVF).
5. **Updates: Agencies, Organizations, Grants**
- a. **VDH: PSGS Grants VaCATPIP II, VaCHISIP, EHDI: N. Ford**
    - *Virginia Congenital Anomaly Tracking and Prevention Improvement Grant (VaCATPIP II)*: Year-3 funding for this five-year CDC-funded birth defects surveillance and evaluation grant, which supports the VaGAC, was awarded. The award is for budget period March 1, 2007 – February 28, 2008.
    - *Virginia Child Health Information System Integration Project (VaCHISIP)*: The year-3 continuation application for this three-year CDC-funded early hearing detection and intervention grant, which funds the redesign of the Virginia Infant Screening and Infant Tracking System (VISITS II), was submitted. The application is for the budget period July 1, 2007 – June 30, 2008.
    - *Universal Newborn Hearing Screening Grant*: The year-3 continuation application for this three year HRSA-funded grant will be submitted in May. The application is for the grant period September 1, 2007 – August 31, 2008. (Post meeting note: Using funds from the year-2 award, the Virginia Early Hearing Detection and Intervention Program collaborated with partners to design and implement the Virginia Guide-By-Your-Side program, which is designed to meet the needs of parents of newly-diagnosed children with hearing loss.)
  - b. **VDH: Virginia Sickle Cell Awareness Program: J. Radcliff-Shipman**
    - *Funding for Follow Up*: For the first time since 1995, the General Assembly has increased the allocation of funds for follow up of children with sickle cell disease.
    - *Funding for Sickle Cell Projects*: There will be an increase in allocation of funds via Request for Proposal (RFP) process to community-based sickle cell project.
    - *Hemoglobinopathies*: Using data from the VASCAP Access Tracking Data Base, it has been determined that from March 1, 2006, through April 16, 2007, 91 hemoglobinopathies have been identified and 74 hemoglobinopathies have been confirmed. The trait follow-up program sent out nearly 2000 letters; the return rate was only 7 percent.
    - *Sickle Cell Conference*: A flier for the upcoming conference entitled "Sickle Cell Disease: Facing Challenges and Improving Outcomes," on April 27, 2007, at the Crowne Plaza Richmond—West, was distributed.

- c. **VDH: Children with Special Health Care Needs Program: N. Bullock**
- *Care Connection for Children:* The section of the *Code of Virginia* that expanded Virginia newborn screening effective March 1, 2006, includes a requirement that all children under 21 years of age be referred to Care Connection for Children, which is a statewide network of six centers of excellence for children with special health care needs. The centers provide leadership in the enhancement of specialty medical services; care coordination; medical insurance benefits evaluation and coordination; management of the CSHCN Pool of Funds, which is a small pool of funds is available for families 300% below the federal poverty level; information and referral to children with special health care needs (CSHCN) resources; family-to-family support; and training and consultation with community providers on CSHCN issues.
  - *Child Development Clinics:* The Child Development Services Program is a specialized program for children and adolescents suspected of having developmental and behavioral disorders such as developmental delays, developmental disorders associated with sensory or physical disabilities, disorders of attention and hyperactivity, learning problems, emotional and behavioral concerns, mental retardation, or a combination of these problems. A professional team consisting of a pediatrician, nurse, social worker, educational consultant, and psychologist provides services. Core services include diagnostic assessment and care planning, 6-12 months of follow-up care coordination and referral. Consultations are available from other pediatric specialists as needed. There are ten child development clinics throughout the state (Danville, Fredericksburg, Gate City, Harrisonburg, Lynchburg, Newport News, Norfolk, Petersburg, Roanoke, Winchester). About 3,000 children are evaluated annually.
- d. **Division of Consolidated Laboratory Services: J. Pearson, W. Andrews**
- *Clinical Laboratory Improvement Amendments (CLIA):* DCLS recently completed its successful CLIA inspection.
  - *DCLS Information System:* Additional laboratories continue to be incorporated.
  - *Public Health Laboratory Interoperability Project (PHLIP):* DCLS was asked to be one of six state public health laboratories to participate in PHLIP, a project of the Association of Public Health Laboratories. PHLIP's overarching objective is to foster collaboration in the areas of information technology and laboratory science with the ultimate goal of developing and piloting viable IT architecture options and tools for the exchange of electronic laboratory data at all levels of public health laboratories, including HL7 message specification and standard nomenclature.
  - *National Medical Laboratory Professionals Week: April 22-28, 2007.*
  - *DCLS Tour:* Those who have not toured DCLS are invited to do so.
- e. **March of Dimes: S. Long**
- *Preconception and Prenatal Genetic Screening Pocket Facts:* This updated fact card, which describes prenatal genetic screening tests and included ethnic-specific risks for several common genetic conditions, was circulated. The fact card is available from MOD (contact Sara Long, [slong@marchofdimes.com](mailto:slong@marchofdimes.com)).
- f. **Part C: K. Durst for M. A. Discenza**
- *Infant & Toddler Connection of Virginia:* Provides early intervention supports and services to infants and toddlers from birth through the age of 2 who are not developing as expected or who have a medical condition that can delay normal

development. There are 40 central points of entry throughout Virginia. About 9,000 children were served in the past year.

- *Early Intervention Awareness Month*: Governor Kaine has proclaimed May 2007 as “Early Intervention Awareness Month” in the Commonwealth of Virginia.

g. **Virginia Department of Education**: No report.

6. **Updates: Virginia Genetic and Metabolic Centers**

a. **EVMS: M. Persinger Yeargin for V. Proud**

- *Newborn Screening*: Continue to be busy with expanded newborn screening activities
- *Virginia PKU Walk-A-Thon*: Takes place on Saturday, May 19, 2007, in Virginia Beach and will feature low protein cooking demonstrations, TV interviews, and information on PKU research.

b. **UVA: B. Wilson**

- *Newborn Screening*: Continue to be busy with expanded newborn screening activities; there are 3 NICUs in UVA’s catchment area.
- *Biopterin Expanded Access Program*: At the American College of Medical Genetics meeting, Biomarin announced that they were conducting an expanded access program for their product sapropterin (a synthetic form of biopterin). They are in the process of applying for FDA approval for this medication, which has been shown to improve phenylalanine tolerance in some patients with PKU due to phenylalanine hydroxylase deficiency. The number of patients for whom the medication will be available as part of the expanded access program is limited, and each institution will need to get IRB approval to participate.

c. **VCU: J. Bodurtha**

- Genetic counseling program received full accreditation.
- W. Nance retired but is back in the laboratory.
- Waiting to hear about fellowship accreditation (post meeting update: received 5-year full accreditation).
- Becoming more involved with enzyme therapy.
- Expanded newborn screening activities have been quite a challenge, including developing materials in other languages.
- Developing relationships with catchment area NICUs, including Northern Virginia.

7. **New Business**

J. Monaco was appointed by the U.S. Secretary of Health and Human Services to the Secretary’s Advisory Committee on Heritable Disorders and Genetic Diseases in Newborns and Children. The first meeting that she attended as an appointed member was December 18-19, 2006. Information about the Committee, including past minutes, is available online at <http://mchb.hrsa.gov/programs/genetics/committee/>.

8. **Next Meeting:**

- a. Date: Tuesday, October 16, 2007 (secondary date is October 2, 2007)
- b. Time: 10 a.m. – 12 noon
- c. Location: Division of Consolidated Laboratories
- d. Recommended Speaker: Phil Postum, DCLS.

9. **Adjourn**

Meeting adjourned at 12 noon.