

**Virginia Genetics Advisory Committee  
Tuesday, April 28, 2009  
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**

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

- |  |   |
|--|---|
| <input checked="" type="checkbox"/> Willie Andrews, DCLS                 | <input type="checkbox"/> William Owen, EVMS/CHKD            |
| <input checked="" type="checkbox"/> Joann N. Bodurtha, VCU (VaGAC Chair) | <input type="checkbox"/> Arti Pandya, VCU, VEHDIP Adv Cmt   |
| <input type="checkbox"/> Nancy Bullock, VDH                              | <input type="checkbox"/> James Pearson, DCLS                |
| <input type="checkbox"/> Mary Ann Discenza, DMHMRSAS                     | <input type="checkbox"/> Ginny Proud, CHKD                  |
| <input checked="" type="checkbox"/> Laura Duncan, VCU                    | <input type="checkbox"/> Jene O. Radcliffe-Shipman, VDH     |
| <input type="checkbox"/> Mary Claire Ikenberry                           | <input checked="" type="checkbox"/> Charlie Stevenson, DCLS |
| <input type="checkbox"/> Anil R. Kumar, VCU                              | <input type="checkbox"/> Bill Wilson, UVA                   |
| <input checked="" type="checkbox"/> Sara Long, MOD                       | <input type="checkbox"/> Eileen Coffman, CHKD               |
| <input type="checkbox"/> Walter Nance, VEHDIP Adv Cmt                    |   |

**VaGAC and Subcommittee Staff:**

- |   |  |
|---|--|
| <input checked="" type="checkbox"/> Nancy Ford, Lead Staff - VaGAC & Steering Cmt | <input checked="" type="checkbox"/> Audrey Greene, Lead Staff - NB Subcmt            |
| <input checked="" type="checkbox"/> Tahnee Causey, Lead Staff - CP/PI Subcmt      | <input checked="" type="checkbox"/> Jennifer Macdonald, Staff - NB Subcmt            |
| <input type="checkbox"/> Heather Creswick, Lead Staff - SGP/PH Subcmt             | <input checked="" type="checkbox"/> Rafael Randolph, Staff - VaGAC                   |
|   | <input checked="" type="checkbox"/> Sharon Williams, Lead Staff - Contractors Subcmt |

**VaGAC Ad Hoc Members:**

- |  |  |
|--|--|
| <input type="checkbox"/> Joanne Boise, VDH     | <input type="checkbox"/> Gayle Jones       |
| <input type="checkbox"/> Joan Corder-Mabe, VDH | <input type="checkbox"/> David Suttle, VDH |

**Additional Subcommittee Members / Interested Parties:**

- |  |   |
|--|---|
| <input type="checkbox"/> Stephen Braddock, UVA                       | <input type="checkbox"/> Rees Lee, NMCP               |
| <input type="checkbox"/> Bonny Bukaveckas, VCU                       | <input checked="" type="checkbox"/> Phil Poston, DCLS |
| <input checked="" type="checkbox"/> Eileen Coffman, CHKD             | <input type="checkbox"/> Alice Schroeder, Parent      |
| <input checked="" type="checkbox"/> Sarah H. Elsea, VCU              | <input type="checkbox"/> Cliff Schroeder, Parent      |
| <input checked="" type="checkbox"/> Valerie Luther, Partnership, VCU | <input type="checkbox"/> Holly Tiller, VDH            |
| <input type="checkbox"/> Paula Miller, Parent                        | <input type="checkbox"/> Heather Trammell, Parent     |
| <input checked="" type="checkbox"/> Jana Monaco, Parent              | <input type="checkbox"/> Susan Ward, VHHA             |
| <input type="checkbox"/> Vicki Hardy-Murrell                         |   |

**Guests:** Edward H. Karotkin, MD, Professor of Pediatrics, Eastern Virginia Medical School; Wendy Golden, PhD, Professor, Department of Pathology, UVA (representing W. Wilson); Joe Spontarelli, CART Provider

**Recorders:** Rafael Randolph and Nancy Ford

1. Welcome: J. Bodurtha
  - a. Introductions: Completed.
  - b. Minutes of 10/7/08 Meeting: Approved as written.
  - c. Agenda: No changes.
  - d. Membership: Circulated for corrections.
  - e. Travel Reimbursement Reminder: See Rafael Randolph to receive a travel reimbursement form.

2. Public Comments: None
3. Review of VaGAC Infrastructure, if needed.
  - a. VaGAC Contractors Subcommittee: S. Williams proposed changing the VaGAC Contractors Subcommittee to an ad hoc group that would be convened by S. Williams on an “as needed” basis, such as after Newborn Screening Subcommittee meetings. The majority agreed. Plan:
    - N. Ford will make corrections on VaGAC Infrastructure document.
  - b. Membership: J. Bodurtha reminded members to continue to explore ways to increase diversity and expand membership on Subcommittees as appropriate
4. Subcommittees Reports
  - a. Newborn Screening Subcmt: W. Andrews
    - DCLS is very busy with the recent occurrence of novel influenza A (H1N1) virus (formerly referred to as swine-origin influenza virus). One of the reporting sites, Naval Medical Center Portsmouth, stopped sending in data subsequent to changing newborn screening laboratory contract from DCLS to PerkinElmer Inc. This brought up a concern about follow-up services and data control. There is a Newborn Screening Subcmt scheduled to take place immediately after today’s VaGAC meeting.
  - b. Birth Defects Prevention Subcmt: T. Causey
    - Has not convened the new subcommittee yet because they are waiting on carry forward funds from the CDC. They are planning a new folic acid campaign that focuses on multi-vitamin use for women of childbearing age. The new folic acid campaign will not concentrate on pregnant women but overall women’s health because data show that half of all pregnancies are unplanned. Some research has found that when women who are not planning pregnancies hear the word “pregnancy” connected with folic acid, they “tune out.”
    - Circulated birth defect prevention materials for comments.
  - c. State Genetics Plan and Public Health Subcmt: J. Bodurtha
    - Almost 10 years ago, the federal government released some grants for the development of a state genetic plan, but the leadership at VDH at the time chose not to participate. We have tried through smaller funding sources and efforts to develop a State Genetic Plan. Thanks went to Nancy Ford and Sharon Williams for their leadership in this effort. Thanks also were extended to the March of Dimes for supporting a statewide needs assessment. A draft state genetics plan, “Key Goals for Virginia State Genetics Plan for Public Health,” was distributed. Plan:
      - N. Ford will email the same draft plan that was distributed to the VaGAC distributed list for comments. Responses will be collated and sent to J. Bodurtha. Then, the subgroup that developed the draft plan will review the comments and develop a structure to finalize the plan via the VaGAC State Genetics Plan and Public Health Subcommittee.
  - d. VDH Contractors Subcommittee: S. Williams
    - In the process of revising the committee structure. See item 3a.

5. Updates: Agencies, Organizations, Grants

a. VDH Cooperative Agreements: N. Ford

- Virginia Congenital Anomalies Tracking and Prevention Improvement Project II (VaCATPIP II): The last year of this 5-year cooperative agreement, which is funded by the Centers for Disease Control and Prevention (CDC), started March 1, 2009. Funds are used to partner with VCU to provide genetic counseling services focusing on the quality component of the birth defects registry, new case ascertainment, and a birth defects prevention campaign. N. Ford attended a CDC birth defects surveillance and prevention conference last February where it was announced that instead of having two grant groups (like it is now), CDC plans to combine the groups into one and send out a competitive application for all states to apply. There will still be as many awards as if you combined the two. The focus will be on evaluation and quality.
- Virginia Child Health Information Systems Integration Project I (VaCHISIP I): This 3-year cooperative agreement, which was funded by CDC, ended June 30, 2008. Funds were used to redesign the Virginia Infant Screening and Tracking System (VISITS) by the VDH Office of Information Management (OIM), which continues to work on the redesign through a no-cost contract extension. It is anticipated that VISITS II will be launched statewide soon after the new electronic birth certificate (EBC) system is implemented statewide, as VISITS II will be integrated with the EBC. OIM plans to launch the EBC system on a phase-in basis beginning with Vital Records staff during May 31, 2009; thereafter, the EBC will be phased in statewide with full implementation completed by October 2009. Currently, VDH is in the process of de-duplicating thousands of records that will be uploaded into VISITS II.
- Virginia Child Health Information Systems Integration Project II (VaCHISIP II): This 3-year cooperative agreement, which is funded by CDC, focuses on newborn hearing screening follow up and quality. G. Jones is the VaCHISIP II Project Director. Funds are being used to support two new wage positions: Follow-up Analyst, which has been filled, and Data Management Coordinator, which is expected to be filled in May 11, 2009.

b. VDH: Virginia Sickle Cell Awareness Program: J. Shipman

- No report.

c. VDH: Children with Special Health Care Needs Program, including Care Connection for Children (CCC) and Child Development Clinics: J. Bodurtha for N. Bullock

- The Crater Child Development Clinic has closed, and there is a new one in Richmond in the 3600 building. They are currently in the process of conducting a needs assessment about the child development clinics throughout the state. They are looking at autism and other diagnosis through the child development clinics.

d. DCLS: W. Andrews

- Dr. Pearson is busy with the H1N1 flu (swine flu). DCLS will soon be able to perform confirmation testing; DCLS is receiving samples that will be tested for final approval by CDC to perform confirmatory tests. Currently, DCLS performs

presumptive testing and then sends the specimens to CDC for confirmatory testing. They are in constant contact with the epidemiologists at VDH and CDC.

- DCLS has a new hire in Newborn Screening, Dr. Tom Hickey. In addition to technical responsibilities, the position includes quality assurance coordinator responsibilities.
- DCLS is in the second round of procurement processes to bring in new instrumentation for newborn screening.

e. March of Dimes: S. Long

- The March of Dimes is pushing very strongly to reduce the number of preterm births. Last year was their 5-year point for the prematurity campaign. Last November (prematurity awareness month), the national organization published its 2008 Premature Birth Report Cards, which showed that Virginia received a “D” grade along with the nation. The March of Dimes is working with hospitals and physicians on teaching messages related to c-sections and induced labor. A 9-page booklet entitled “Why the Last Weeks of Pregnancy Count,” which tells women why it is important not to schedule an induction or c-section for non-medical reasons before 39 weeks of pregnancy, is available from the March of Dimes Web site: <http://www.marchofdimes.com/catalog/product.aspx?productid=5141&categoryid=&productcode=37-2209-07>

The March of Dimes advocated for legislation banning smoking in Virginia's bars and restaurants. The bill was passed by the 2009 General Assembly and recently signed by Governor Kaine. The legislation goes into effect December 1, 2009.

f. DMHMRSAS - Part C: M.A. Discenza,

- No report.

g. Dept. of Education: N. for T. Campbell

- There was some miscommunication about the DOE Chronic Health Conditions survey. Some VaGAC members sent in comments about the DOE survey as requested, and Nancy sent them to DOE. After speaking to T. Campbell, it was determined that they actually were not looking to do a big overhaul to the survey. T. Campbell thanked us for our input and said she would consider it for future references.

h. New York-Mid-Atlantic Consortium for Genetic and Newborn Screening Services (NYMAC): J. Bodurtha, S. Williams, L. Duncan

- J. Bodurtha: Unfortunately, J. Bodurtha cannot attend the May meeting. There have been a number of workgroups and the one J. Bodurtha has been concentrating on is the medical home and the professional education workgroup. NYMAC is working to incorporate the medical home model around newborn screening diagnosis. VCU submitted a small grant to see about infusing the medical home model into pediatric medical home teaching and for those conditions that are shared between endocrine and genetics through our pediatric group practice and private practice. Follow-up grant was not funded.

- S. Williams: The professional education committee met yesterday. They are finalizing the work about the emergency medical cards for diagnosed patients. The concern is that if a child with one of these disorders goes to an emergency room, the staff might not know how to manage the child.
  - L. Duncan: A NYMAC committee is conducting a survey on reimbursement coverage for metabolic formulas. UVA is probably participating on this project. L. Duncan participates on the transition workgroup for NYMAC, and they are pulling together a plan for funding educational programs for adolescents with PKU.
- i. Virginia Association of Genetic Counselors (VaAGC): T. Causey
- VaAGC last met in October 2008, at which time they elected officers. They have been meeting via teleconference monthly, and they have completed by-laws and a redesigned the Web site (<http://www.vaagc.org>). The biggest push right now is the upcoming state meeting, traditionally called the Genetic Professionals Meeting but now called the Virginia Genetics Education Conference. They will be meeting in Williamsburg on June 1, 2009. The conference is being sponsored by LabCorp, and registration is open for anybody interested in attending. If you would like to attend and are not a genetic counselor just email T. Causey ([tahnee.causey@vdh.virginia.gov](mailto:tahnee.causey@vdh.virginia.gov)). There will be speakers on various topics including Operation Smile (a new update in facial surgery). The VaAGC Licensure Committee will have their first conference call today. VaAGC is looking at applying for a grant next year.
- j. HRSA Advisory Committee on Heritable Disorders and Genetic Diseases: J. Monaco
- The last two Committee meetings were convened on November 24, 2008, and February 26-27, 2009.

The November meeting included a preliminary report on the candidate nomination *Severe Combined Immunodeficiency Disease* (SCID) by the Evidence Review Workgroup and a report on the candidate nomination *Spinal Muscle Atrophy* (SMA) by the Internal Review Work Group. The Internal Review Work Group recommended no evidence review for SMA at this time and further recommended a nominator to conduct prospective pilot studies in one or more traditional public health laboratories in order to show reproducibility of the preliminary findings. The recommendation was unanimously approved by the HRSA Committee. The November meeting minutes are available online at <http://www.hrsa.gov/heritabledisorderscommittee/meetings/2008november/default.htm>

The February meeting included a final draft report on the candidate nomination of *Severe Combined Immunodeficiency* (SCID) by the Evidence Review committee. The Committee recommended not adding SCID to the core panel now and recommended additional studies. The recommendation was approved by HRSA Committee. The meeting also included a discussion on progress in developing the Newborn Screening Translational Research Network (NBSTRN), which involves building a research infrastructure that is large and potentially national in size and scope to facilitate the introduction of new screening tests and treatments in a research

mode to gain knowledge as quickly as possible so that no potential data on rare disorders would be lost.. The February meeting minutes are available online at <http://www.hrsa.gov/heritabledisorderscommittee/meetings/2009february/default.htm>

The next meeting, which will take place via teleconference, is scheduled for May 12, 2009. The agenda includes a preliminary report on the candidate nomination *Krabbe Disease* by the Evidence Review Workgroup.

6. Updates: Virginia Genetics and Metabolic Centers

a. EVMS: E. Karotkin

- No report.
- Dr. Karotkin interest in genetics was sparked by a visit to India as a speaker. While there he had the chance to meet someone that was involved in some new test for genetic screening. It involved GCNMS and a computer algorithm that allowed the analysis of the anolites and the biomedical markers. It also uses urine instead of blood and it claims to screen for over 100 metabolic diseases.

b. UVA: B. Wilson

- No report.

c. VCU: J. Bodurtha, L. Duncan

- L. Duncan: VCU has completed eight Kuvan<sup>®</sup> (sapropterin hydrochloride) trials. Kuvan is a new prescription medication that may help lower phenylalanine (Phe) blood levels more than the use of diet alone for people with PKU. Most of the people in their trails have not responded, but a few have. They have one PKU patient who has successfully given birth before and has decided to become pregnant again. VCU is having some concerns with the lost of the child development centers across the state. They relocated the Fredericksburg clinic to the health department there. They had been using the Peninsula Child Development Center for their satellite clinic there, but they are finding it difficult to find a space to work in.
- J. Bodurtha: They are proud of their genetic counseling students, who are in the process of going through oral exams. They have added some PhD genetic counseling students also. Dr. Walter Nance, whose mentor was Dr. Oliver Smithies, co-recipient of the 2007 Nobel Prize in physiology or medicine, still remains in his lab today. On April 24, 2009, VCU opened its second Medical Science building on Broad St. (125,000 sq. ft.). VCU officials have announced that 42-year-old Michael Rao, president of Central Michigan University, will succeed Eugene P. Trani on July 1, 2009, as the school's new president.

7. New Business

a. J. Bodurtha: Telehealth

- Telehealth is the delivery of health-related services and information via telecommunications technologies. Telehealth delivery could be as simple as two health professionals discussing a case over the telephone or as sophisticated as using videoconferencing between providers at facilities in two countries, or even as complex as robotic technology.

- UVA has a very active telehealth program which has been quite successful in its limited arena.
  - EVMS usually tries to get the patient into the hospital to see the baby by various means, but they really like the idea of putting a camera in the nursery. There may be an issue with the families that could benefit from this in that they probably do not have computers at home. A suggestion to fix this problem was to give them a loaner computer or to obtain low-tech computer similar to the ones that are given out to kids in Africa.
  - T. Causey provided telehealth services in Savannah, Georgia, both through paternal field medicine and genetic counseling services. This was part of a state grant for populations under served and people that were three hours away from maternal medical services in Georgia. They contracted with a local hospital where the patients could go instead of traveling three hours to the hospital. From a genetic counselor point of view, sometimes it was difficult to have a good relationship with the patient where they were uncomfortable with being on TV.
  - Dr. Hickey stopped by. He worked with Dr. Anderson out of Tulane who has been doing telehealth medicine with great effectiveness.
  - N. Ford has seen this done with school based clinics. She was at a conference where a presentation on telehealth in Kansas was conducted and the community seemed to really embrace this technology. The kids liked because they were on TV.
  - S. Williams stated that this may become more prevalent as we move to national healthcare because of reimbursement issues.
  - J. Bodurtha suggested PerkinElmer, Inc. might be able to support telehealth for criticals.
- b. L. Duncan: PKU
- Ryan's PKU walk-a-thon is Saturday, May 9, 2009. It will be in Virginia Beach this year.
  - National PKU Alliance, a fairly new organization, has selected two states to look at in terms of the distribution of metabolic formulas: Virginia and Georgia.
- c. S. Williams: Newborn Screening Report
- Copies of the summary table "Virginia Newborn Screening Diagnosed Cases 10/1/08 through 3/31/08" were distributed. In addition, copies of the report "Virginia Newborn Screening Services: Protection Newborns for 40 Years, 1966 – 2006," were distributed, and additional reports were made available for members to take back to their organizations. Because of VDH budget restrictions, there is as hold on printing future reports.
- d. Jana Monaco: Formula Legislation
- Had a question about a date for the formula legislation. S. Williams responded that the bill died in committee.

*DRAFT*

8. Next Meetings
  - a. VaGAC Meeting:
    - Date: Wednesday, October 28, 2009
    - Time: 10 AM – 12 PM
    - Location: Division of Consolidated Laboratory Services
  - b. VaGAC Steering Committee Meeting: To be announced.
9. Adjournment: Meeting adjourned at 11:55a.m.