Virginia Genetics Advisory Committee  
Tuesday, April 20, 2010  
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):  
☑ Willie Andrews, DCLS  
☑ Joann N. Bodurtha, VCU (VaGAC Chair)  
☑ Nancy Bullock, VDH  
☑ Mary Ann Discenza, VDBHDS  
☑ Laura Duncan, VCU  
☑ Mary Claire Ikenberry  
☑ Anil R. Kumar, VCU  
☑ Sara Long, MOD  
☑ Walter Nance, VEHDIP Adv Cmt  
☑ William Owen, EVMS/CHKD  
☐ Arti Pandya, VCU, VEHDIP Adv Cmt  
☐ James Pearson, DCLS  
☐ Ginny Proud, CHKD  
☐ Charlie Stevenson, DCLS  
☐ Theresa Taylor, VDH  
☐ Bill Wilson, UVA  
☐ Eileen Coffman, CHKD  

VaGAC and Subcommittee Staff:  
☑ Nancy Ford, Lead Staff - VaGAC & Steering Cmt  
☑ Audrey Greene, Lead Staff - NB Subcmt  
☑ Jennifer Macdonald, Staff - NB Subcmt  
☐ Rafael Randolph, Staff - VaGAC  
☐ Sharon Williams, Lead Staff - Contractors Subcmt  

VaGAC Ad Hoc Members:  
☐ Joanne Boise, VDH  
☐ Joan Corder-Mabe, VDH  
☐ Ruth Frierson  
☐ David Suttle, VDH  

Additional Subcommittee Members / Interested Parties:  
☐ Stephen Braddock, UVA  
☐ Bonny Bukaveckas, VCU  
☐ Tahnee Causey, VCU  
☐ Sarah H. Elsea, VCU  
☐ Valerie Luther, Partnership, VCU  
☐ Paula Miller, Parent  
☐ Jana Monaco, Parent (via conf call)  
☐ Vicki Hardy-Murrell  
☐ Jane Juusola, VCU  
☐ Rees Lee, NMCP  
☐ Phil Poston, DCLS  
☐ Alice Schroeder, Parent  
☐ Cliff Schroeder, Parent  
☐ Holly Tiller, VDH  
☐ Heather Trammell, Parent  
☐ Susan Ward, VHHA  
☐ Rachel Arculin, Parent  

Guests:  
Joe Spontarellio, CART Services; Dr. Tom Hickey, DCLS; Megan Farley, Genetic Counselor  
Student  

Recorders:  
Rafael Randolph and Nancy Ford  

1. Welcome: J. Bodurtha  
   a. Introductions: Completed.  
   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: N. Ford  
There were no public comments.

3. Review of VaGAC Infrastructure, if needed.  
There were no changes.

4. Subcommittees and Groups Reports  
a. Newborn Screening Subcm – W. Andrews  
Reminder that the Newborn Screening Subcm will meet after today’s VaGAC meeting.  
Lab purchased a new mass spectrometer; the instrument is not yet in production as it is  
being validated. Subcommittee continued to discuss reorganization within VDH’s  
newborn bloodspot and hearing screening programs; concerns were raised about the  
ability to meet workload for newborn screening bloodspot testing, especially in the event  
that new diagnoses are added to the panel. DCLS continues to submit data to the Region  
4 Collaborative project regarding diagnosed cases and associated levels. At the last  
Subcommittee meeting, Dr. Phil Poston gave a presentation and solicited the help of the  
metabolic specialists to determine if additional or different markers are needed to identify  
the disorders.

b. Birth Defects Prevention Subcm – N. Ford  
On hold until the new Preconception Health Coordinator position is filled; this is a new  
position within VDH Division of Women’s and Infants’ Health (DWIH).

c. State Genetics Plan and Public Health Subcm – J. Bodurtha  
As reported in the last meeting, the plan has been place on hold due to other priorities  
within the VDH Division of Child and Adolescent Health (DCAH). (Note: Following the  
VaGAC meeting, the VaGAC/State Genetics Plan and Public Health Subcm Chair  
(J. Bodurtha) and Subcm staff (S. Williams and N. Ford) made a decision to disband the  
Subcm and reconvene it in the future if indicated. if needed.)

d. VDH Contractors Ad Hoc Work Group – N. Ford  
As reported in the last meeting, management of the genetics and metabolic treatment  
centers contracts has been transferred from S. Williams to N. Ford. Members are  
working on developing a FY 11 Annual Report and revising the quarterly report form via  
conference calls and emails. The next conference call is scheduled for April 28 at 2 p.m.  
In process of renewing the three genetic and metabolic treatment contracts (EVMS,  
UVA, and VCU) and the prenatal genetics contract (Genetics and IVF).

5. Updates: Agencies, Organizations, and Grants  
a. VDH: Organizational Changes, Grants – N. Ford  
(1) VDH and DCAH Organization: Dr. Suttle, Director of the Office of Family and  
Health Services (OFHS), is retiring at the end of the month; Dr. David Trump, who is  
currently the health district for Peninsula Health District, will be the Acting OFHS  
Director. An updated DCAH organizational chart was distributed and reviewed.

(2) VISITS II: The new Virginia Infant Screening and Infant Tracking System  
(VaCATPIP II) was released statewide on April 12. VISITS II is a component of the  
Virginia Vital Events and Screening Tracking System (VVESTS), which also  
includes the new Electronic Birth Certificate. Eight statewide trainings were held for  
hospital users. Training materials are available online at  
(3) CDC Birth Defects Grant: The Virginia Congenital Anomalies Tracking and Prevention Improvement Project (VaCATPIP II) ended February 28, 2010, and the VaCATPIP II final report, which is due May 31, is being completed. The VDH application to CDC to fund VaCATPIP III was recommended for approval but did not rank high enough to be funded. If awarded, the grant would have funded a 1500-hour genetic counselor or similarly skilled staff position and birth defects prevention activities. A thank you was extended to everyone who wrote a letter of support. Birth defects prevention responsibilities have been transferred to the to-be-hired DWIH Preconception Health Coordinator. VaCARES quality improvement responsibilities have been transferred to the recently hired DCAH Surveillance and Evaluation Coordinator.

(4) VEHDIP: The vacant Virginia Early Hearing Detection and Intervention Program (VEHDIIP) Manager position was filled by Ruth Frierson, who had been serving as the Acting VEHDI Program Manager and Follow-up Coordinator. VEHDIP continues to support the Virginia Hearing Aid Loan Bank and Guide By Your Side family-to-family support program. Virginia has had over 200 matches for family-to-family support. VEHDIP also continues to manage two cooperative agreements (CDC and HRSA) that support the part-time (wage) Data Manager Coordinator, Follow-up Analyst, and Quality Assurance Coordinator positions; a full-time Follow-up Specialist position; and six VEHDIP learning collaborative sites.

b. VDH: Virginia Sickle Cell Awareness Program – T. Taylor
   Jean Radcliffe Shipman has retired. Virginia Sickle Cell Awareness Program responsibilities will be under the to-be-hired Preconception Health Coordinator.

c. VDH: CSHCN Program, including CCC and CDCs – N. Bullock
   No report. (Children continue to be referred from Virginia Newborn Screening Program to Care Connection for Children.)

d. VDH: DWIH – K. Moline
   No report.

e. Division of Consolidated Laboratory Services –W. Andrews
   (1) National Laboratory Week: Reminder that it is National Medical Laboratory Professionals Week, which provides the profession with a unique opportunity to increase public understanding of and appreciation for, clinical laboratory personnel.
   (2) Virginia State Furlough Day: The Newborn Screening Lab will be operational during furlough day, May 28.
   (3) DCLS-VDH Conference Calls: About one year ago, the newborn screening follow-up and laboratory team started having monthly conference calls; these calls continue and have been very beneficial in terms of staying connected and addressing issues as they occur.
   (4) CDC Epidemiology and Laboratory Capacity (ELC) Grant: Funds are available for developing a standardized data exchange between DCLS and hospitals. VDH Department of Epidemiology would need to sponsor the grant on behalf of DCLS. While the primary focus is not newborn screening, it could be a component of the exchange. This is the first funding made available to public health labs for electronic data exchange. So far, letters of support have came in from UVA, VCU, and Sentara hospitals.

f. March of Dimes – S. Long
(1) March for Babies: The Richmond March for Babies fundraiser walk is planned for May 2 at Monroe Park.

(2) Prematurity Campaign: The March of Dimes Prematurity Campaign is in its seventh year. The national rates for preterm births did drop a little in 2008 and 2009. The campaign has two goals: (1) to raise public awareness of the problems of prematurity and (2) to decrease the rate of preterm birth in the U.S. (15 percent reduction in rate). As part of a national effort to save babies from premature birth, the March of Dimes has designated November as Prematurity Awareness Month. The third annual Premature Birth Report Card will be released around November 18, 2010, which has been designated Prematurity Awareness Day. It was noted that Dr. Jerome Strauss, dean of the VCU School of Medicine, identified a gene variant related to increased risk of premature rupture of the fetal members, a common cause of premature birth; the gene variant is much more common among African-American women (2006).

g. VDBHDS: Part C – M. A. Discenza
No report.

h. NYMAC – J. Bodurtha
The New York-Mid-Atlantic Consortium (NYMAC) for Genetic and Newborn Screening Services recently went through a process of review of grants. They included transitions particularly for children with genetic conditions, emergency preparedness, laboratory technical issues, and how to continue to improve communication medical home management issues. If you have interest in any of these areas, some small funds may be available. For more information, contact Katharine Harris, NYMAC Project Manager, phone: (518) 474-7148, e-mail kbh02@health.state.ny.us.

i. Virginia Association of Genetic Counselors – T. Causey
The second round of elections was conducted on November 2, 2009. The Fourth Annual Virginia Genetics Education Conference will be held on May 25, 2010, in Falls Church, Virginia. Additional information is available from the VaAGC web site http://www.vaagc.org/. Continue to work on licensure.

j. HRSA Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) – J. Monaco
(1) Last Meeting: The 20th ACHDNC meeting was held January 21-22, 2010. The minutes are available online at http://www.hrsa.gov/heritabledisorderscommittee/meetings/. Meeting highlights:
(a) The Internal Nomination Review and Prioritization Workgroup recommended to the Advisory Committee that the nominations of neonatal hyperbilirubinemia and critical congenital heart disease be forwarded for an evidence review by the Evidence Review Workgroup.
(b) The Advisory Committee recommended adding severe combined immunodeficiency disorder (SCID) to the uniform newborn screening panel and recommended adding other T-cell lymphocyte deficiencies to the list of secondary targets as a comprehensive entity. (Post meeting note: On May 21, 2010, Kathleen Sebelius, Secretary of Health and Services, announced the addition of SCID to the core panel of 29 genetic disorders as part of her recommendation to adopt the national Recommended Uniform Screening Panel. SCID is the first nominated condition to be added to the core panel of disorders.)
(c) The Newborn Screening Saves Lives Act of 2008 called for the Health Resources and Services Administration (HRSA) to establish and maintain a clearinghouse of current materials, resources, research, and data related to newborn screening. In late 2009, HRSA awarded a five-year, $3.75 million cooperative agreement to the Genetic Alliance to develop such a clearinghouse.

(d) The second draft of the Advisory Committee white paper on national policies regarding the retention and use of dried blood spots after newborn screening was presented for the Advisory Committee’s review. The final white paper will be published.

(e) The report from the Sickle Cell Disease Association of America’s (SCDAA) Workshop on Carrier Screening was discussed, including the National Athletic Trainers’ Association consensus statement promoting screening for sickle cell trait (SCT). It was decided that Dr. Howell (Committee Chairperson) will contact the Secretary’s Advisory Committee on Genetics, Health, and Society (SACGHS) about possibly collaborating on some work in developing recommendations pertaining to ethical, legal, and social issues related to carrier screening.

(f) It was noted that the Evidence Review Workgroup had revised its report on Krabbe disease and was planning to submit it to Genetics in Medicine for publication. (Note: During the September 2009 meeting, the Advisory Committee recommended not adding Krabbe disease to the recommended newborn screening panel at the present time.)

(2) Next Meeting: The 21st ACHDNC meeting will be held May 13 -14, 2010.

6. Updates: Virginia Genetics and Metabolic Centers
   a. EVMS – G. Proud and W. Owen
      Recruited fourth genetic counselor and he/she will start in June. A new dean is coming; the current dean is moving to be the head of research at EVMS. Sickle cell program recently hired a new social worker.
   b. UVA – B. Wilson
      Genetic counselor fellow Lindsey Paule started in early April.
   c. VCU – L. Duncan, J. Bodurtha
      Subsequent to the end of the VDH VaCATPIP II grant, Tahnee Causey joined the VCU genetic counseling team. Various student projects are on-going. Had an excellent recruitment year in which 25 students were interviewed. There is a particular interest in this year in the genetic counselor PhD combined program. They currently have two PhD genetic counseling trainees.

7. New Business
   a. Virginia Newborn Screening Program – S. Williams
      Distributed 2008 Virginia newborn screening data, which are a component of the VDH MCHB Title V Block Grant application: Number and Percentage of Newborns and Others Screened, Cases Confirmed, and Treated.
   b. No other new business reported.

8. Next Meetings
a. VaGAC Meeting:
   Date: Tuesday, October 5, 2010
   Time: 10 a.m. - 12 p.m.
   Place: DCLS
b. VaGAC Steering Cmt Conference Call:
   Date: Tuesday, September 14, 2010
   Time: 10 a.m.
   Contact Information: N. Ford will send.

9. Adjournment
   Meeting adjourned at 12 p.m.