

**Indiana Genetics Advisory Committee
Meeting Minutes
April 13, 2010
1 – 2 pm EST
Teleconference**

ATTENDANCE:

Full Members				Ad-hoc Members	
Bader, Patricia	X	Heyen, Candy		Bowman, Bob	X
Bingle, Glenn		Hodgin, PT	X	Burrus, Connie	X
Cohen, Stephanie		Humbert, Larry		Dumm, Sue	X
Cook, Lola	X	Lesko, Barb	X	Eddy, Courtney	X
Cushman, Lisa	X	Olsen, Donna Gore		Ganser, Judith	X
Darroca, Roberto		Quaid, Kimberly	X	Hutsell, Gayla	
Escobar, Luis F.	X	Rautenberg, Fr. Joe	X	Mani, Lisa	
Eugster, Erica	X	Reed, Terry	X	Minniear, Kim	
Gill, Nicole	X	Stone, Kristyne		Silva, Ruwanthi	X
Hainline, Bryan	X	Weaver, David	X	Stone, Iris	X
Hendrix, Jon		Zunich, Janice	X	Warner, Dolly	
				Weber, Mary	X

Welcome – Dr. Zunich & Dr. Quaid

Minutes from October 2009 were approved without correction.

ISDH Update

- Courtney Eddy accepted the position of Follow-up Program Director for the new IN Newborn Screening Tracking & Education Program (INSTEP), effective January 1, 2010.
- Malorie Hensley, MS (genetic counselor), will be joining ISDH as the new Genomics Program Director on July 1, 2010.
- ISDH is working on developing policies/protocols related to proper storage of dried blood spot cards (DBS) from newborn screening. Currently, demographics are torn away from specimen and DBS are being catalogued & stored for 23 years in bankers’ boxes at the Clarian storage facility at room temperature without dessicant (no special storage considerations). Lots of debate related to proper storage, length of storage, and release for research is occurring at the national level; ISDH and NBS lab are waiting for additional information from Region 4 meeting in Lansing, as well as national NBS meeting in FL in May, to get more information.
 - Dr. Hainline stated that each individual laboratory (metabolic) can create its own storage guidelines re: time of storage (varies from 2 weeks to longer than 23 years). There have been issues related to viability of specimens.

Genetics Clinics’ Updates

Dr. Bader:

- Statistics show that patient numbers are continuing to increase; things are going well at new facility. Clinic has had large number of Amish patients with non-ketotic hyperglycinemia; recently, specific abnormality was found—may have implication on statewide screen. Dr. Bader has shared this information with Dr. Hainline.

- Amish are getting very active in wanting to start own clinic to perform follow-up for conditions (SCIDS, hemophilia, non-ketotic hyperglycinemia). Most wanting to organize are on northern border (limb-girdle muscular dystrophy more in southern part of area).
- Lots of dx/follow-up of FAS patients—these patients have many symptoms (including distinct form of ADHD). Kids are mostly on Medicaid; recently, have had patients who have been stabilized on ADHD medicine, but Medicaid suddenly requires a previous authorization that indicates the ADHD should be treated by a psychiatrist. Dr. Bader’s concern is that she is a medical geneticist and a pediatrician—pediatrics has its own guidelines for ADHD. Concerned that these patients would not be able to see a psychiatrist and/or Dr. Bader would not be able to prescribe medications for this ADHD.
 - Dr. Hainline recommended that Dr. Bader speak with Craig Erickson at IU or Medical Director of Medicaid for insight about where this new guideline came from.
 - Dr. Ganser has new address (policyconsideration@fssa.in.gov) – can send these types of questions there.

Dr. Escobar:

- 800 current patients; seeing average of 35 patients/day (75% genetics + 25% newborns)
- Going to be participating in clinical trials for Marfan patients beginning in July
- Completed clinical trial with Pfizer for Down syndrome; results were somewhat positive (2-year trial)
- Just joined largest database of hospitals that see children with birth defects; collecting data up to 10 years
- No satellite clinics at this time; will be opening clinic at exit 10 facility (on I-69 near Fishers)
- Looking for new genetic counselor—currently have 2 counselors; looking for a 3rd counselor

Dr. Hainline:

- Very busy; trying to expand lysosomal storage program that Dr. Wappner started
 - Have patients getting all available therapies for every lysosomal storage dz
 - Beginning to see younger patients (not easily dx—have been to pulmonary, GI, etc.)
 - Also seeing patients who previously received bone marrow/stem cell transplant have later-onset problems
 - Want to remind everyone that you may be getting flyers from every possible genetic testing/drug companies, trying to stir up interest in lysosomal storage dz and PKU

Dr. Weaver:

- Started adult (16 – 18 years old+) Marfan/connective tissue clinic at Methodist in October; associated with thoracic surgeon

Dr. Zunic:

- Pregnant lab tech delivering in 6 weeks
- Majority patients are out of office
- Interviewing GC for available position
- May start another cancer risk assessment clinic in 2010

Discussion of NBS for SCIDS

- NBS has been around for 40+ years, but standardization has been difficult (many factors—e.g., personnel, equipment, program’s interpretation of available resources of conditions, expense of new methodologies, state resources, advocacy of family/health care professionals/state legislators)
 - Many disparities among screening services available between states
 - IN performed expanded MS/MS NBS screening for children born in KY
- 1999: Task force indicated that greater uniformity would benefit families, etc.
 - HRSA/ACMG asked to develop guidelines for state screening program & to recommend uniform panel of conditions for each state should include in NBS (“target conditions”)
 - Originally 84 conditions; looked at incidence, analytical characteristics, treatment options, testing options (acute & chronic forms), etc. – developed scoring system that would evaluate strengths/weakness of conditions and then ranked conditions on list
- Handouts:
 - Summary of different conditions that were evaluated & how they were evaluated
 - Table 1, Figure 1 – of 84 conditions, scores ≥ 1200 were “high”

- Lower scores didn't have available treatment or poor understanding of natural history of dz, etc.
 - Scored and categorized into "core," "secondary," and "not appropriate"
- SCIDS initially had score of 1047 ("secondary") – was not deemed appropriate
- Wisconsin presented poster in 2008; had been screening for SCIDS for 2 months (~ 18,000 screens) – see handout
 - No positive cases were ever reported on their poster
 - 2008: Screened 71,000 infants; 17 had abnormal assays; 11 had whole blood samples to test T-cells; evaluated by immunologist
 - Have been screening for 2 years; data submitted to NNSGRC indicates they have zero confirmed cases (incidence of SCIDS varies)
 - Low-birth-weight and very-low-birth-weight infants were large contributors to WI's high false-positive rates
 - WI –only state that mandates SCIDS NBS
- 2002: Duke published results of 19-year study where they performed stem cell transplants on patients
 - 21 patients diagnosed after birth & received stem cell transplant; 20 were alive and leading normal lives (oldest – 19 years old)
 - Survival rate for infants diagnosed later was 73% (dx after 6 months of age)
- Official recommendation made to Secretary of Health & Human Services that all newborns should be screened for SCIDS; Secretary must respond in 180 days
 - With support of ACMG, SCIDS could be 1st condition added to core panel since 2005
- Testing is DNA-based (high through-put, low hands-on)
 - Using WI's repeat data, expect to perform 111,500 DNA assays for SCIDS (much higher than currently being done for *CFTR* analysis)
 - Currently performing DNA testing in small space within neighboring lab; will need to expand and spend money to add SCIDS
 - Remember that education and follow-up are essential to providing complete NBS service
 - Positive recommendation from HHS Secretary will put pressure on IN to add SCIDS to NBS
- Dr. Hainline: Other enzyme/immunological conditions (e.g., ADA deficiency) will be left off NBS panel; very similar to SCIDS – how can they be distinguished in newborn period?
- Fr. Joe: Looking at Executive Summary, didn't see anything about feedback to parents or offers of genetic counseling, etc.—shouldn't parents be given access to information/resources?
 - Barb: Report was trying to put together standardized way to evaluate/compare screening for conditions; don't know how large a part access to care played in this report
 - Bob: Bringing up issue that has also been brought up at federal level—e.g., what the general public's perception/knowledge of NBS is at this point. Trying to determine what information will be helpful, how this information can be distributed in cost-effective manner, etc.

Medicaid and Reimbursement for Services

- Dr. Zurich requested an update from Dr. Ganser about the status of the white paper submitted re: reimbursement for first-trimester screening. This information was re-submitted in October/November 2009 to Dr. Ted Grisell so that Dr. Grisell could pass it along to Dr. Tony Pelizio.

Discussion of Future Topics

- Dr. Escobar would like to discuss NBS for channelopathies at the October 2010 meeting.
- **ALL COMMITTEE MEMBERS**: Please e-mail Dr. Zurich (jjunich@iun.edu) your name, contact information, clinics, and specialties/areas of interest by April 30th. This information will be compiled into a list of contact information that will be offered to OMPP.
- The October 2010 meeting will bring elections of a new chair or co-chairs, as well as nominations and/or re-elections for members whose IGAC tenure will expire after the 10/10 meeting. **Members whose terms will expire in 10/10 are:**
 - **Patricia Bader, MD**
 - **Glenn Bingle, MD, PhD**
 - **Lisa Cushman, PhD, CGC**
 - **Barb Lesko, MT**
 - **Kimberly Quaid, PhD**

- **Fr. Joe Rautenberg**
- **Janice Zunich, MD**
- An updated IGAC membership directory will be e-mailed out with the minutes from the April meeting. The membership directory has been updated with term expiration dates for full members.

At this point, the Committee moved to adjourn the meeting. This motion was approved without dissent.

Next Meeting Date: Tuesday, October 19th, 2010, 1 – 3 pm, ISDH