

Birth Defects Advisory Committee Minutes

January 18, 2002

CALL TO ORDER:

Chairperson, Rebecca Boyd Parrott called the meeting to order at 1:00 p.m. She welcomed Dr. James Matsuda, the new American Academy of Pediatrics representative and Jeff Murray in his new role as ethics representative to the committee.

MEMBERS PRESENT:

Rebecca Boyd Parrott, M. Peggy Stokes, Representative Pat Murphy, Jeff Murray, James Matsuda, Debra Schutte for Janet Williams, Jeffrey Lobas, Val Sheffield, Cathy Evers, Michelle Hall, Jerry Wickersham and Joyce Kohl for Molly Guard

MEMBERS ABSENT:

David A McInnes, Senator Kitty Rehberg, Mary Gilchrist, Greg Garvin, Neil Mandsager, Roger Williamson, Linda Brown, Nancy Ziese, Julie Curry, Paul Romitti

OTHER ATTENDEES:

Julie McMahon, Tonya Diehn, Stan Berberich, Don Simmons, Jack Cameron, Eva Tsalikian, Robert Philibert

APPROVAL OF MINUTES:

Cathy Evers had two changes to the October minutes. The minutes should reflect that the Atlantic clinic has been discontinued not Carroll. The minutes should also reflect that the Regional Genetics Consultation Service has 4 part-time physicians. Jerry Wickersham approved the minutes with the stated amendments. Peggy Stokes seconded the motion.

I. IDPH UPDATE

A. Budget/ Administration Issues

Julie McMahon

1. Legislative session

- ◆ Will not move many new initiatives forward, as the focus will be on the budget.
- ◆ Legislation will address policy issues in January.
- ◆ The fiscal budget will be addressed after review conference in February.

2. Departmental changes

- ◆ Reduction in the number of divisions from six to five. The five divisions are:
 - ◆ The Division of Administration. Consolidation of Administrative and Regulatory Affairs and Executive Staff divisions. A few programs from the Administrative and Regulatory Affairs division will be going to other divisions, such as Radiological Health to the Division of Environmental Health.
 - ◆ The Division of Tobacco Control and Prevention (required by statute)
 - ◆ Division of Health Promotion
 - ◆ Division of Public Health Services (Dr. Gleason's suggestion for the Division of Family and Community Health)
 - ◆ Division of Environmental Health
- ◆ Renaming of some of the bureaus and restructuring of some of the programs.

- ◆ Eight to nine individuals have chosen early retirement. Their last day will be January 31 and their positions will not be replaced.
 - ◆ David Fries, the deputy director and director of the former Division of Administrative and Regulatory Affairs. Mark Schoeberl will assume many of David's responsibilities as the director of the new Administrative Affairs division.
 - ◆ Dr. Ronald Eckhoff, the Medical Director of the Division of Health Promotion. Dr. Eckhoff had also served at one time as the Interim Director and was quite knowledgeable about the Birth Defects Institute and its programs.
 - ◆ Other personal changes
 - Dr. Ed Schor has resigned as the Medical Director for the Division of Family and Community Health. Dr. Schor has accepted a position with the Commonwealth Fund in New York.
 - ◆ The health department will be looking for a candidate with medical and public health experience for a Medical Director of Health Care Services. This position will replace the positions held by Dr. Eckhoff and Dr. Schor.

3. **Governor Vilsack's State of the State Address**

- ◆ Transformation of government occurring to reduce some of the duplication, increase efficiency and better serve families and communities across the state.
- ◆ Signing an executive order to establish a "Children's Cabinet".
- ◆ Governor will release budget today.

4. **Metabolic Formula**

- ◆ Julie has met with Dr. Gleason and will meet with him again next week.
- ◆ She is hopeful that he will make a decision next week.

B. **Iowa Child and Family Household Health Survey**

Jeff Lobas

- ◆ First report from the survey distributed to attendees.
- ◆ Collaboration between Iowa Department of Public Health, Iowa Child Health Specialty Clinics and the University of Iowa Public Policy Center.
- ◆ Provides statewide data on health status, access to health care, insurance coverage, childcare, and social and family environment.

C. **Birth Defects Institute Activities**

Tonya Diehn

- ◆ **Personnel.** The Community Health Consultant and Program Planner positions for the Genetics Implementation Grant have been filled and today is their first day of work. Tammy O'Hollearn is the Community Health Consultant. Tammy was IDPH liaison to the Early ACCESS program prior to accepting this position. She will be working closely with the Iowa Birth Defects Registry on the notification project. Amy Van Maanen, the Program Planner also comes from within IDPH. She was previously the director of licensing for the Board of Medical Examiners. Amy will be working on the newborn metabolic screening aspects of the grant.
- ◆ **Site Visit.** The All Kids Counts Program of the Center for Innovation in Health Information Systems will be performing a site visit of the genetics implementation grant activities on February 28th. The site visit will focus on data integration.
- ◆ **Proclamation Signing.** There will be a proclamation signing for Birth Defects Prevention Month at the capital on January 25th. Everyone is welcome to attend.
- ◆ **Pilot Study Proposal Updates.** Dr. Gleason approved the biotinidase deficiency pilot study on November 6. The Cystic Fibrosis carrier screening pilot study was approved on December 5.

- ◆ **Teratogen Grant.** The March of Dimes' investment opportunity program (MIOP) grant proposal for a statewide teratogen information service was not awarded. The main reason that the project wasn't funded was because sustainability within the Iowa Department of Public Health could not be demonstrated. There is an opportunity to resubmit a revised proposal in the next funding cycle.
- ◆ **Retention Policy Subcommittee.** Tonya hopes to organize this subcommittee in the near future.
- ◆ **Expanded MSAFP Program letter.** Health care professionals in family practice and obstetrics/gynecology were sent a letter discussing the Inhibin A pilot study, prenatal screening for Smith-Lemli-Opitz Syndrome and the maternal serum screening policy in Iowa. The letter was distributed to attendees.

II. RESEARCH STUDY PROPOSAL

“The Role Of HOPA/TRAP230 Polymorphism In Congenital Hypothyroidism”

A. Presentation

Eva Tsalikian, Robert Philibert

- ◆ Handout of power-point presentation distributed.
- ◆ Overall goal of study defined.
- ◆ Neonatal screening for, clinical presentation and incidence of congenital hypothyroidism.
- ◆ Thyroid Response Associated Proteins and HOPA gene defined. HOPA/TRAP230 polymorphisms associated with behavioral illness and hypothyroidism in adults.
- ◆ Purpose of the study is to determine the frequency of HOPA defects in the Iowa newborn population and correlate the presence of those defects to TSH and T4 levels.
- ◆ Protocol for use of newborn blood spot cards in the project described.

B. Discussion

- ◆ **T4 levels** will be done on all samples sent to study. Abnormal T4 levels would be reported back to lab with the expectation of doing follow-up and notifying physicians of results. This part of the protocol doesn't meet the definition of anonymized specimen. Michelle Hall indicated that if a researcher were using newborn bloodspot cards to do a study with potential clinical significance, as a parent, she would want to know. Informed consent discussed.
- ◆ **Study correlates.** Jeff Murray expressed that he sees that the study is looking at two correlations: HOPA with T4 levels and HOPA with clinical significance. He stressed the need to decouple the two aspects of the study until the clinical significance of mild/borderline hypothyroidism in newborns is known. Marcia Willing indicated that the study could cause undue parental anxiety. Recommendation to send a letter to physicians about the study.
- ◆ **No Internal Review Board approval yet.** There needs to be justification in the IRB about how samples would be followed up, if they were unanonymized. Discussion that again this doesn't meet bloodspot card protocol. Dr. Tsalikian and Dr. Philibert indicated that they would take the T4 follow-up and notification out of the proposal.
- ◆ **Randomization.** The study needs randomized specimens. Sample number, randomization of sex and birthweight were discussed.

C. Birth Defects Advisory Committee Decision.

- ◆ Jeff Murray motioned to approve the proposal with the following amendment. The laboratory would provide the researchers completely anonymized blood spot cards with sex, birthweight and TSH level information.
- ◆ Jeff Lobas seconded the motion. Committee members approved the motion unanimously.
- ◆ Proposal changes must be submitted to the BDAC chairperson or Tonya Diehn before a letter of support for the project can be written.

III. REGIONAL GENETICS CONSULTATION SERVICE

Semi-Annual Report

Cathy Evers

A. Personnel

- ◆ No changes in RGSC program staff since 2001 annual report.
- ◆ The Department of Pediatrics has made an offer to physician from Jordan. They are waiting to hear his decision.
- ◆ They are interviewing for the counselor position.

B. Clinic Activities

- ◆ Decreased the number of clinics and Spencer and Ottumwa (heard some disgruntling from Ottumwa).
- ◆ No clinics held in Decorah and Atlantic.
- ◆ Referrals to Familial Cancer Counseling Program increasing, had a cancer risk-counseling clinic in Dubuque.
- ◆ No show rate continues to be very low at 5.9%. Staff talks with individuals and families three to four times before appointment.

C. Program Activities

- ◆ Continued participation in quarterly Medicaid administrative claiming time studies.
- ◆ The Genetics Division received grants from the March of Dimes and the Children's Miracle Network to assist with the 1-800 number, interpretative services, and patient educational materials.
- ◆ The program continues to do a lot of educational outreach to a variety of audiences.

D. Discussion

- ◆ Jeff Lobas asked how the undergoing changes in the hemoglobinopathy program affect RGSC. Also how does the program connect with families and refer to other clinics? Cathy indicated that the hemoglobinopathy program sends a letter to physicians about genetic counseling and RGSC when a newborn is identified to be a carrier. Counselors regularly refer patients and families to clinics such as CHSC and local providers.
- ◆ Jeff Murray asked if the RGSC could retain the budget they are not using because of reduced staff and clinics and use it to add clinics in the next year when the program has the staff. Cathy indicated that the patient/family caseload not really impacted by the reduction in clinics. They have 16 clinics/year in Des Moines and could do more because they have many new referrals. More new patients in every clinic site. They are increasing the time span in which they see return patients and utilizing more local physician follow-up. There is also better and more effective use of the counselors during the clinics.
- ◆ Julie McMahon talked about working with the cancer programs at IDPH. Jeff Lobas suggested Early ACCESS as a resource.

IV. IOWA NEONATAL METABOLIC SCREENING PROGRAM

Annual Report

Stan Berberich

A. Pilot Study Activities

- ◆ Formal proposals for Expanded MS/MS and biotinidase deficiency pilot studies written. Timeline reviewed for BDAC presentations and IDPH approval of proposals.
- ◆ Attendees provided with a handout describing the symptoms and incidence of the disorders on the current newborn screening panel, in pilot studies, or currently under consideration.
- ◆ The expanded MS/MS pilot study began in October 1, 2001.
 - ◆ 112 specimens have been first screen abnormal. Second blood spot was then tested.
 - ◆ 26 cases were referred on for follow-up and further testing.
 - ◆ They are following two cases to determine significance.
 - ◆ They are maintaining a follow-up report.

- ◆ Don Simmons presented at a national workshop for tandem mass spectrometry. Iowa is entwined with other state NBS programs doing expanded MS/MS screening and is able to utilize their research. The programs are working to form a network of laboratories. CDC quality control materials for MS/MS should be available within the year.

B. Program Reports

Stan Berberich

- ◆ The Newborn Screening Data Collection 2000 report compiled by Marcia Valbracht for the National Newborn Screening and Genetics Resource Center was distributed.
 - ◆ 433 specimens or 1.09% of specimens received were unacceptable for analysis
 - ◆ Discussed the number of specimens collected and the infant's age at time of screen
 - ◆ Report lists screening results for each disorder and the number of infants confirmed with disorder, clinically significant variant, or variant not clinically significant.
- ◆ Iowa Reported & Confirmed Positive Report for January 1-December 31, 2001 distributed.
 - ◆ For hypothyroidism, 8 of the cases initially reported as borderline were later confirmed to have the disorder, one confirmed case was initially reported as normal. Discussion about the missed case and the reasons occurred.
 - ◆ Dr. Tsalikian indicated that in the last six months they have had more borderline TSH levels. Stan discussed that the laboratory had seen an upward shift of the mean TSH level. The shift may possibly attributable to one lot number of reagents.
 - ◆ There was discussion if SIDS cases are followed up to see if they have had metabolic screening. The medical examiner's office contacts the laboratory with SIDS case of concern. Don Simmons shared that there is a new disorder on the scene called carbonyl phosphatase. The marker for the disorder is glycine.
 - ◆ Highlights of the expanded MS/MS follow-up report were shared.
 - ◆ INMSP Facilities Summary Report distributed.

C. Cystic Fibrosis Ad-Hoc Subcommittee

Tonya Diehn

- ◆ An update was provided. Judy Miller, chair of the subcommittee was unable to attend today because she is at home with a sick child. The subcommittee has been meeting for quite some time. Members attending today include Stan Berberich, Michelle Hall, and Marcia Willing. The members have done a thorough review of the laboratory, clinical, educational and follow-up aspects. However, the committee continues to struggle over pieces like cost, process, counseling, carrier detection, and educational components. A Pro and cons table has been developed by the subcommittee but a consensus as to whether CF screening should be offered has not. Michelle Hall commented she and other members have gone back and forth whether there should be screening.
- ◆ Marcia Willing expressed that the subcommittee continues to struggle with the impact of newborn CF screening on the current Iowa Neonatal Metabolic Screening Program and the Regional Genetics Consultation Service. RGSC doesn't have the staffing to meet the potential counseling cases. Follow-up plan would need to be in place before laboratory components. Discussion occurred at length.
- ◆ The primary care physician would be an important component of CF newborn screening, Jeff Lobas indicated. Not all patients identified as carrier will necessarily need or want to have genetic counseling through RGSC, their primary care physician may feel comfortable in providing this information to the family. Dr. Matsuda shared that in his group practice, they discuss hemoglobin traits with patients. The program should focus on the children who are identified with Cystic Fibrosis; the intent of the program is not to identify carriers.

- ◆ Stan suggested talking with the Wisconsin program to see how they dealt with the some of the issues being discussed. Attendees indicated that the committee could ask for two-part proposal that clearly defines the follow-up plan and the laboratory component.
- ◆ Debra Schutte discussed that nurse and nurse practitioners need to be included in provider education and with follow-up.

V. IOWA BIRTH DEFECTS REGISTRY PROGRESS REPORT

Developmental Fund Use

Jeff Murray

A. Program Overview

- ◆ Progress report distributed to attendees.
- ◆ Until last year, the registry shared field representatives with the Iowa Cancer Registry. These individuals were responsible for abstracting all cases of cancer and birth defects statewide. Individuals specifically trained in cancer abstraction.
 - ◆ Not timely, poor quality of birth defects abstraction
 - ◆ Cancer abstraction needs increased
 - ◆ The State Health Registry could no longer finance birth defects abstraction.
- ◆ Funded primarily by CDC. Also receives 50,000 from the board of reagents. This funding does not support abstracting activities.

B. INMSP and MSAFP Developmental Funds

- ◆ Asked for supplemental funding last year
- ◆ Received one year of support with the possible renewal of an additional year to fund field services coordinator and five abstractors.

C. Abstraction Progress

- ◆ Abstracting from births in 2000 and forward
- ◆ Currently have a six-month period between birth and abstracting. Goal is 4 months.
- ◆ Since 7/01 have abstracted 1,429 records.
- ◆ 62 hospitals completed through 6/01
- ◆ Also re-reviewing records of pre-2000 to verify patient ascertainment completeness.
- ◆ Goals for January through June include completing abstraction in all current hospitals through 12/01 and expanding out into hospitals in adjacent states.

D. Discussion

- ◆ Jeff Lobas asked about the ability to monitor census tracts and outbreaks. Information important for program planning. Jeff Murray talked about Lake Rathbun, GIS studies, and Des Moines infant mortality study. Dr. Paul Romitti has a personal interest in cluster studies.
- ◆ Val Sheffield stated that while supporting the abstractors was a unique use of the developmental funds, he feels that it was a good use of the funds and that the money has been well used.

VI. ADJOURNMENT

The motion for adjournment was by Jerry Wickersham and seconded by Jeff Murray.

VII. NEXT MEETING INFORMATION

The next meeting will be held on Friday April 19, 2002 from 1:00 PM to 4:00 PM at the Wells Fargo Bank-Sullivan Room in Grinnell, Iowa.