

# **Birth Defects Advisory Committee Minutes**

## **July 19, 2002**

### **CALL TO ORDER:**

Linda Brown called the meeting to order at 1:00 p.m. Linda presided over the meeting due to Chairperson, Rebecca Boyd Dublinske being absent. Introductions were made.

### **MEMBERS PRESENT:**

Peggy Stokes, Julie Curry, Linda Brown, Celeste Johnson, Jolene Johnson, Molly Guard, Paul Romitti, Nancy Ziese, Christina Trout, Jeff Lobas, Gregory Garvin

### **MEMBERS ABSENT:**

Rebecca Boyd Dublinske, James Matsuda, Debra Schutte, Roger Williamson, Michelle Hall, Neil Mandsager, Pat Murphy, Jeff Murray, Val Sheffield, Jerry Wickersham, Mary Gilchrist

### **OTHER ATTENDEES:**

Julie McMahon, Stan Berberich, Tonya Diehn, Stanley Grant, Tammy O'Hollearn, Karen Brewer, Amy Van Maanen, Judy Miller, Pam DeBoer, Don Simmons, Carol Johnson, Cathy Evers, Jack Cameron

### **APPROVAL OF MINUTES:**

The Committee indicated that the minutes from the April 19, 2002 meeting did not need any changes, but there was no motion to approve them. They will be on the October agenda to obtain approval.

## **I. COORDINATOR REPORT:**

**Tonya Diehn**

### **Status on Administrative Rules**

The notice of intended action has been sent to the code editor and will be published on August 7, 2002. Changes that were made to the rules included adding biotinidase as a routine screen in the metabolic screen, adding inhibin-A as a routine screen to the Expanded MSAPF screen, and adding a provision about metabolic formula funding. Comments can be made about the rules between now and August 27, 2002. There will be a public hearing on August 27, 2002. The Committee was encouraged to make comments regarding the rules as well as encouraging families to make comments. September 11, 2002 is the Board of Health meeting. Anyone can attend.

### **Birth Defects Institute Programs Overview**

Tonya provided an overview about the programs located within the Birth Defects Institute (BDI) including mission, services, funding and staff. A handout of the presentation was provided to the BDAC members by email at a previous date. Tonya has used this presentation to educate Dr. Gleason, Jane Colacecchi, Deputy Director, and other constituents about the BDI.

### **Other Items**

- Tonya will be participating in the Iowa Colon/Rectal Cancer Summit/Task Force on September 11, 2002 and asked if anyone on the committee would want to participate.

- Tonya has been working on a cooperative agreement for CDC for surveillance and long-term follow-up individuals identified with a disorder by tandem mass spectrometry. The deadline for submission is August 2, 2002.
- The Hemoglobinopathy Program submitted a grant to Maternal Child Health Bureau, HRSA to expand genetic counseling services for people with a hemoglobinopathy trait or disease.
- The Regional Genetic Consultation Service and Neuromuscular Program submitted their annual reports for review. There will be discussion on these two reports at the October meeting.
- The Registry was awarded a 5-year CDC grant for research and prevention to extend current surveillance programs. Paul Romitti indicated that the competition was very competitive this year. Only five of the existing seven centers received funding this year.
- Paul is working with Christina Trout of the neuromuscular program in writing a cooperative agreement for CDC for Duchenne and Becker muscular dystrophy.

## **II. STATE BUDGET/ADMINISTRATION ISSUES**

**Julie McMahon**

Julie explained how budget cuts had been made over the last two fiscal years. In FY01 there was over \$31 million in general funds supporting public health programs. At the start of FY03, the general fund portion of the department's budget is at \$24.2 million. At the beginning of FY02, a 9% reduction was made with another 5.3% cut was made during the FY02. An additional 9.32% reduction had to be made for FY03. Approximately 75% of IDPH programs are funded by federal or other (foundation, private grants) funds. The percentage of federal/other funding continues to rise. This is significant in that it is anticipated that federal cuts might need to be made in the future if revenues continue to fall nationwide.

Each department was given a dollar amount of reductions that they had to achieve. Reductions in operational costs, reduction in personnel through vacancy factors, furloughs or layoffs and program reductions were implemented to achieve the reductions. The department's executive team met and prioritized all programs based on the functions of the programs and the benefits to the health status of Iowans. They developed a proposal for achieving the reductions. This was sent to the Department of Management and to the governor's office for review. The final budget represents this review and recommended changes.

Julie discussed specific budget reductions to the Genetics programs. Several University of Iowa programs in the Department of Pediatrics under contract with IDPH, including Neuromuscular and Regional Genetics Consultation Service, received reductions. The U of I Pediatric department chair will have the option of making a decision on how much each of these programs' budgets will be reduced or choosing an across-the-board budget of 9.32% reduction to each of the programs. A 9.32% reduction made to the Neuromuscular and Regional Genetics Consultation Service would be \$10,775 and \$53,017 respectively.

The department is also looking at consolidating their public health service contracts that are not research-based or statewide. One contract would cover all services provided by an agency where now each service within an agency receives an individual contract. A commission was developed out of the Omnibus II bill to review all programs with state monies within the next year to determine if there is a need for that program. Julie also indicated that the outlook for the upcoming years is grim and little relief is in site.

### **III. EXPANDED MSAFP PROGRAM**

#### **Annual Report**

**Stanley Grant**

Stanley provided a handout to the committee. From July 1, 2000 to June 30, 2001 there were 16,124 women that chose to have the expanded MSAFP screen done. One case of open spina bifida or anencephaly was detected per 21 women with persistent MSAFP elevations. One in 13 patient with elevated MSAFP values carried a fetus with a defined fetal anomaly. One chromosome abnormality or serious abnormality was detected for every 24 amniocentesis procedures performed. The statistics are a year behind because they wait until after birth to obtain final information. The Iowa Birth Defects Registry is now working with the program to determine the birth and pregnancy outcomes of screened pregnancies.

The Iowa Expanded MSAFP/ Quad screen will be started routinely within the month hopefully. New pamphlets have been developed and are ready. The Expanded MSAFP Program is also participating in a multi-center study with Smith-Lemli-Opitz screening.

#### **Iowa Expanded MSAFP/Quad Screen Status**

**Stan Berberich**

Stan reported that the lab was having significant problems with starting the quad screen due to software problems. The company has been out to the lab trying to pin point the problem. They ended up setting up the lab with an older version of the program and so far it has been working well.

#### **CF Carrier Screening Program**

**Karen Brewer**

The pilot study started in mid-June. Several centers are participating in the pilot study throughout Iowa including Des Moines Perinatal Center, six offices in Davenport, and the IVF, OB and Fetal Diagnosis programs at the University of Iowa. The program was designed from the ACOG model. Karen was unsure how long the pilot would run, they would like to have 60 to 100 samples for the pilot.

The program will be doing sequential couple screening. The program will require samples from both partners to perform the testing. Samples will be obtained from both the mother and father, the laboratory will then screen the mother first, if the mother has a positive result, then the father will be tested. If the mother does not carry one of the mutations tested for, the father will not be screened. Results will be reported for the individuals and the pregnancy as a unit. If there is a positive between both parents then they will contact the physician and recommend genetic counseling for themselves and possibly other family members. The cost of the screen is \$197.00 and includes screening both parents, if applicable. CF carrier screening is voluntary. The uptake rate for this screening test is uncertain.

### **IV. IOWA NEONATAL METABOLIC SCREENING PROGRAM**

#### **Status of Biotinidase Deficiency Screening**

**Stan Berberich**

On July 1, routine screening for biotinidase deficiency began. Stan reported that 15,000 screens have been done to date. Ten have been reported as abnormal to follow-up staff. One has been confirmed and it appears that another will be confirmed this week. Three normal babies have had false positives. The treatment for this disorder is a vitamin. Judy Miller indicated that they will be using a new confirmatory laboratory in Baltimore due their previous confirmatory laboratory closing. She also indicated that most of the infants were false positive results were very sick. There is a reported association between hepatic failure and reduced biotinidase

activity. Tonya indicated that Iowa Department of Public Health will be doing a press release about the biotinidase deficiency screening. There was an article about the screening and the disorder in the July IDPH Focus newsletter.

Stan also shared information about meeting with the Nebraska laboratory. Nebraska recently passed legislation that required all their labs to combine and that there be one single laboratory by July 2003. The legislation also required that they start testing for MCADD now and they do not have a mechanism for that. Stan presented information to them how Iowa could assist them until they are able to test for MCADD. Stan was very pleased in the way the meeting went and thought this opened up the door for some good opportunities.

### **Expanded Tandem Mass Spectrometry (MS/MS) Panel Pilot Study**

**Don Simmons**

Don provided an update report on the MS/MS pilot study and distributed a handout to attendees. The study has been underway since October 2001 with approximately 22,000 babies screened for MS/MS detectable disorders through April 2002. 80 newborns required follow-up, representing 0.4% of the screened population. One infant was confirmed for medium chain acyl Co-A dehydrogenase deficiency (MCADD), one was identified as a possible carrier for 3-methylcrotonyl Co-A carboxylase deficiency (3-MCC) and a mother was identified as positive for 3-MCC. Four infants are still in follow-up. A second tandem mass spectrometer has been ordered and shall be delivered in the next month.

Don also explained that they have been doing a comparison study using the MS/MS and the current screening method for PKU and achieving good results. The MS/MS method detected 8 screen positives, which were all, confirmed by repeat testing, resulting in one PKU found that was missed by the fluorometric assay.

Judy Miller shared a story about a Bosnian Family that recently had a baby in Iowa with a presumptive positive screen for PKU. When learning of the results, the parents indicated that one of their other sons was handicapped. The baby was confirmed to have PKU and the handicapped son was brought into clinic. He had profound mental retardation and has subsequently been diagnosed with PKU. Both boys were put on formula. Judy also shared another story about a baby confirmed to have PKU whose three-year old mentally retarded sibling born in Mexico was diagnosed when the family was seen in clinic. The three-year old was also put on the special diet and they have seen positive developmental changes.

## **V. GENETICS IMPLEMENTATION GRANT**

### **Matching Activities**

**Amy Van Maanen**

Amy previously reported that she had been matching birth records against lab information trying to identify infants that did not receive the metabolic screen. She had been doing this manually but recently a database was developed that will match the records for her. She has been able to match March through June electronically and has February left to match manually. From July to December, 13 infants were identified not to have received newborn metabolic screening. Letters were sent to the physicians of these babies, six of those missed have been brought in for screening, one signed a waiver, two patients have transferred to another physician, one letter was returned undeliverable, and one has been a no show at their last two appointments. We have received seven waivers from March to June 2002.

Amy shared information about placing the collection cards and brochures in the home birth packets and suggested that maybe we should consider putting the waivers in the packets also. This would at least show us that people are making a conscious decision not to have the screen where we might be thinking they were missed. Amy and Tonya have been visiting hospitals with the newborn hearing-screening program to discuss their programs and the addition of newborn metabolic and hearing screening questions to the birth certificate worksheet revisions.

The Iowa Neonatal Metabolic Screening Program practitioner manual is complete and will be on the Center for Genetics website. The website is close to completion, upon completion the committee will be notified.

### **Parental Notification System Activities**

**Tammy O'Hollearn**

Tammy provided handouts for the advisory members regarding the notification system for the Birth Defects Registry (BDR) and a summary of a survey mailed to physicians/nurse practitioners. Tammy reviewed her progress to date on the development of a statewide notification system for the BDR and talked briefly about next steps. Tammy also reviewed the survey and highlighted some areas of specific concern as she moves forward with the development of the notification system. One striking finding of the survey was that physicians/nurse practitioners lack awareness or knowledge in regards to the Iowa Birth Defects Registry and its activities.

### **VI. WRAP-UP**

The next meeting will be on October 18, 2002. Dr. Garvin suggested that we evaluate road construction activities and offer alternatives for the meeting if travel is hindered.

### **VII. ADJOURNMENT**

The meeting was adjourned at 3:55pm.

Submitted by Amy VanMaanen.