CALL TO ORDER
Deb Schutte called the meeting to order at 1:00 p.m. and presided over the meeting. Introductions were made.

MEMBERS PRESENT
James Matsuda, Paul Romitti, Nancylee Ziese, Roger Williamson, Jerry Wickersham, Debra Schutte, Lisa Heddens, Linda Brown, Christina Trout, Diana Fritz Cates, Stan Berberich (representative for Mary Gilchrist), Carol Johnson (representative for Val Sheffield)

NONMEMBERS PRESENT
Mavis Rike, Catherine Evers, Pam DeBoer, Bradley McDowell, Stanley Grant, Karen Brewer, Tonya Diehn, Julie McMahon, Tammy O’Hollearn, Dawn Mouw, Sherry Smith

APPROVAL OF MINUTES
The minutes from April 2, 2004 meeting were presented for approval. Pam DeBoer indicated there were a few members from the Iowa City site who were not listed in the minutes: Judy Miller, Pam DeBoer, Bradley McDowell, and Marcy.

A motion was made by Jerry Wickersham and seconded by Nancylee Ziese to accept the minutes as corrected.

Lisa Heddens was introduced and welcomed as the newest member to the committee.

I. ADMINISTRATIVE RULES REVISIONS

The Chapter 4 “Birth Defects Institute” rules are being opened for renaming the Birth Defects Institute. Also, language has been added to the Newborn Screening Program, which discusses sharing of information and confidentiality. They will be easier to interpret. It lists the policies and procedures and the different responsibilities of the programs. The rules will be published in the administrative bulletin on June 9. The public hearing will be on June 29th.

Tonya reminded the group that during the April meeting, the members voted to change the name of the Iowa Birth Defects Registry to Iowa Congenital and Inherited Disorders Registry. The group agreed the title of the Registry should be changed to “Iowa Congenital and Inherited Disorders. Tonya met with the Kids Coalition group on Monday; they felt the new name is better than The Birth Defects Registry. The committee members offered language to make the rules clearer in regards to the Neuromuscular and Regional sections of the rules.

II. UPDATES
INMSP Update
Carol Johnson announced that Newborn Screening Program has a new metabolic geneticist, Sarah Copeland. It is hoped that Sarah will begin working with the program on August 1, 2004. Carol anticipates Sarah will be at the August 20th meeting.

NBS Retention Policy
Heather Adams, from the AG’s office, approved the recommendations to change the retention period of the blood spot cards. They will be stored at -70 degree temperature during the first year. The cards will be stored at room temperature for an additional 4 years and the program is permitted to use the specimens for research, granted the program has received parental consent. The lab cannot provide the information to other entities without having parental consent. A copy of this policy will be provided to all advisory members to be included in their BDAC books (they have a copy of these policies or are supposed to have a copy in their BDAC books. This would replace the old, right? You may want to consider adding more sections to the book. It is not very well organized or at least not organized how I would do it.)

Bylaws Subcommittee Volunteers
Tonya asked for volunteers to join the subcommittee. The following members volunteered: Nancylee Ziese, Christina Trout, Linda Brown, Jerry Wickersham. Tonya would like the group's first meeting to be face-to-face, sometime during May.

Genetic Implementation Grant Activities
Tammy went over the summary she had provided to the attendees. As of May 10th, the Parent Notification Program sent out 933 letters. The average time expected to complete each case is 5 months 11 days. She was able to speak to 421 parents on the phone. The response from the parents was very good. She received 137 response cards for requests for no further contact, which is an option not to receive a personal call. If she is unable to reach a parent, she does mail a notification stating she is making her final attempt to reach them. There were 81 address/phone changes and 91 incorrect physician or pediatricians notifications.

There have been 25 requests for families to be contacted with Spanish notification. Initially, the families receive the parent notification letter in English, but they have the option to request the information be translated into Spanish. She has been working with others from the department to ensure the Spanish families receive adequate information. She has also received one request for the information to be translated into Vietnamese. The Center for Genetics now has literature/brochures translated into Spanish and Vietnamese.

Tammy has made 3 contact referrals on behalf of the families and has received 19 requests for additional information. She also received 14 requests for their children's diagnosis and 1 requested both contact referral information and the child's diagnosis. Typically, the referrals that have been made are for early intervention services, child health specialty clinics or requests to connect with parents who have the same
disorder. A couple of families she connected with the Parent Training Information Center. It has been a very positive outcome for the families that have been connected.

There were 25 cases excluded from the Registry, ten of those cases were brought to her attention because the parents or physicians stated the information was ineligible. Since that time, there have been modifications made to the manual that will help to decrease that number.

Tonya provided all advisory members with a copy of the proposal asking the committee to vote on using funding from the developmental funds to finance activities and services provided through the Parent Notification Project.

Tonya stated that the Center should be able to sustain the program within a year. Deb Schutte informed the group that if the developmental funds were not appropriated to the Parent Notification Project, the program would no longer exist.

Tammy mentioned that some states do not necessarily contact the parents to notify them that their child is on the registry, but do notify the parents regarding services available to them. The state of Colorado automatically makes a referral to their children with special health care needs program; however there are only certain diagnoses for which an automatic referral is made. Those public health agencies must then call and follow up with the families and provide them with any resource information that would be beneficial. Colorado receives the information through birth certificates, hospitals, physicians, etc. The Colorado turn around time for these cases is supposed to be 90 days, but usually takes longer. No other state performs the notification project on a large scale as Iowa. They may provide the service for specific birth defects, usually the more major defects requiring more medical attention.

Tonya thinks that CDC is getting close to funding Registry programs that connect families to early intervention services. The Iowa Birth Defects Registry does not qualify for this money at this time because it is considered a “Center” with a focus on research. In the future, CDC may incorporate some funding for centers, as well.

Motion was made to approve this request of $83,300 to be appropriated to the Parent Notification Project with the understanding that this funding is for a period of one year.

Motion was made by Nancylee Ziese.
Seconded by Paul Romitti

Discussion

Stan mentioned that if there were questions regarding the appropriateness of using the developmental funds for these activities, would one of the initiatives be clearly acceptable and one not be clearly acceptable. If one were not acceptable it would be pulled along with the other. Julie mentioned that she did not have information in front of her, but the information she reviewed yesterday, would support both
initiatives. She agreed they could be two distinct initiatives and stated they could be listed as such.

Motion approved

Stan mentioned there should be some constraints to the grants and they should be used for enhancement. He feels the funding would certainly benefit the CF and PN project, but the information should be verified to ensure the funding is appropriate. Tonya said she would be send an email to everyone, which consists of the guidelines for the developmental fund and will ask for those members who were present at the meeting to vote if they think it is appropriate to use developmental funds for these initiatives.

III. MATERNAL FIRST TRIMESTER AND INTEGRATED SCREENING QUESTIONS AND ANSWERS

Deb Schutte stated she had received an email from Julie McMahon stating she still had unanswered questions preventing her from acting on that proposal. She had requested the meeting be reconvened in order for her questions to be answered. The primary purpose of this agenda item was to provide additional information to the Department of Public Health in order for the department to make a decision regarding this proposal.

Today's meeting was an attempt to answer any remaining questions about the first trimester screening proposal. Tonya had requested committee members send questions regarding the proposal to her; those questions were forwarded on to Roger. Following an extensive discussion, the proposal to establish a pilot of the screening was approved by a role call vote. The next step of the process is for the proposal to be presented to the Department of Public Health.

What are the different screening options that will be offered by the program and what will the reporting options be?

Why is the integrated screening offered only to women of the maternal age of 35. Roger and Stanley Grant both responded that it is offered to any woman at any age. Women who are 35 years of age and above are informed they can forgo screening and proceed right to an amniocentesis test. Roger mentioned that no money from the Newborn Screening developmental fund. The proposed budget of $98,000 is money that is accumulated into the developmental fund for quadruple marker screening. That will be the only source for the funds to establish the mediums. For the first trimester screening, the program does require data by an ultrasound.

Will a specimen without ultrasound information be accepted?

If the lab doesn't have an ultrasound follow-up with the doctor would the lab results be incomplete? Stan Berberich stated that if something as important as an interpretation of a result were missing, they would need to obtain that information. Stanley Grant mentioned the results are interpreted between the lab
and the Newborn Screening Program. A repeat of the screening is not repeated in order to prevent a false-positive reading.

**What is the detection rate and false positive rate for PAPP-A and beta-HcG only screening?**

When you look at screening in general, if a woman wants to be screened only by first trimester parameters, the detection rate for Downs Syndrome and a false-positive rate can be very comparable to the current quadruple marker screening. Currently, the quadruple marker screening has a detection rate of 85% that is a very high false-positive detection rate. Roger reported that the neural translucency screening greatly decreases the false-positive rate. Testing will not be available everywhere when this is first implemented, however sonographers across the state will be trained so that this is more readily available in their local communities. Roger does not feel that training will take long. Eventually, women across the state will have access to a very good first trimester screening.

Linda stated that one of the documents Roger had provided mentioned that neural translucency is not considered by all to be a standard of care. She asked him about the accuracy of the screening. Roger stated that the neural translucency screening does require a lot of skill. A formal learning course would train a sonographer to perform the test with accuracy. That is why only individuals who are trained to perform the neural translucency will be able to provide an accurate screening. Currently, Iowa City has 5 or 6 sonographers who have been trained, and Des Moines has 3. Linda stated that the document reported that there is no consensus on the definition of increased neural translucency. She also mentioned the statement was dated as far back as 1999 and asked if the consensus has changed. Roger answered it has changed. There is a pretty good standard now.

**Will there be a mass education campaign for this screening?**

Roger stated the program plans to make efforts for mass education for the screening.

Tonya asked if a doctor's office had a large group of patients who were interested in having the screen, would their program be able to send a sonographer to that doctor's office until more sonographers are trained? Stanley Grant said that they do not have the staff to provide that kind of service. Roger stated that the program does provide outreach for the eastern part of the state because there is a high need. They do not provide outreach to other parts of the state because the service is available in those areas. Stanley stated that they currently provide outreach to Dubuque, Waterloo, Davenport, Cedar Rapids, and Burlington.

Stanley Grant mentioned it would be nice to have CEU's for the sonographers. She also mentioned that the sonographers have expressed an interest in meeting once per month for continuing education. The sonographers would like to have some kind of presentation. She asked them if they had monthly meetings and would they like to have someone come to their office to speak during those
meetings. That option presented a favorable response and Stanley thinks it would be beneficial, however they would not be able receive CEU’s. The program is willing to present for those people who organize and provide CEU’s. She also mentioned the sonographers are interested in the education of cystic fibrosis, first trimester screening, and maternal serum screening.

**What insurance companies will pay for this screening?**

Linda asked Roger if his answer covered Medicaid? Roger stated that Medicaid would not cover the screening until it becomes standard of care. Roger stated when private insurance companies began to pay for quadruple or triple marker screening, Medicaid and Medicare began paying for those procedures shortly thereafter. Linda asked how it is determined when the screening becomes standard of care. Roger stated that when the insurance companies realize the cost savings from the screening, it quickly becomes a standard of care.

**Will the test be available for those who cannot pay?**

Linda asked what the uninsured patients will do during the interim before the screening becomes standard of care and how they will pay for care. Roger said that it shouldn't take long for the test to become standard of care. First trimester screening is being either implemented or piloted all over the country. The program will conduct the pilot project, which will pay for those patients that want to receive their first trimester screenings. By the time the pilot is complete, Roger hopes that the screening will be standard of care.

Christina asked what insurance companies including Medicaid pay for now? Do they currently pay for the quadruple screen that is offered at this time? Stan answered yes. Christina stated that the current billing method is for the quadruple screen is to bill by each individual marker. Worse case scenario is that if Medicaid does not pay for the screening, the only marker that would not be paid for is PAPP-A. The standard of care is already established for the other markers. Tonya reported that Medicaid has a policy that specimens that go through Medicaid or HMOs, have to go to the University of Iowa Hygienic Laboratory. She stated she has received calls that providers have attempted to send specimens to other places and they do not receive reimbursement.

**The charge for the integrated test is $22.00; but the current screen is going to be $91.00. How can it be cheaper to report two screenings?**

Tonya asked if the quad screen is $86, what is the $22 for. Roger stated it is the integrated testing for the PAPP-A test. He said if someone opts to have only the first trimester screening, the best screen would include neural, beta HcG, and the PAPP-A. If the patient selects the integrated test, the first trimester screening will only include the PAPP-A and the neural. They do not use the HcG in both the first and second trimester. Tonya clarified the integrated testing would include PAPP-A in the first trimester, along neural translucency if it's available. The second trimester screening would include HcG, Esterol, AFP, and Inhibin A.
What does it mean to integrate markers?

Roger was asked to discuss why it is not appropriate to perform the first trimester screening and provide the results to the patient and then allow them to decide if they want to do the second trimester screening. Stanley stated that if you perform a first trimester screening and then you perform a second screen on top of that, the chances for false positive increases. That would place the patient at greater risk to have a more evasive procedure that they may not need if they would have went with both screens. It was also asked, when you do the integrated markers, are both results integrated into one risk assessment. Stan stated that if the patient selects first trimester screening only, and doesn't like the results, the next step would be a diagnostic test instead of another screening.

Roger mentioned that if a patient opts for the first trimester screening only, she would receive a fairly accurate screening. If the patient later decides to have the integrated test, she will not receive a very good test because the PAPP-A screening will not have been performed.

What criteria are used to determine when a type of screening is a public health issue?

Julie answered that any time we are providing additional information to a population or individuals in order for them to make informed choices. This screening would fall in with access to best care. She sees this as a public health initiative, but it comes from making new information available to a population.

What criteria are used to determine when a type of screening is a public health issue?

It was mentioned there is a lot of information made available to the public; however, the Iowa Department of Public Health is not the one who provides the community with all of that information. Julie responded that the Public Health Department does not provide all of the information to everyone, but instead works through all the various partnerships with local and private providers to provide what is believed to be legitimate and scientific based information. What she likes about the pilot program is the fact it is a pilot and the information will be available to all patients and will become a standard of care. Those individuals who are on Medicaid and who are of low income will have this information available to them and have the option to participate. The department's goal is to try as much as possible through partnerships to provide new information and new tests available to all Iowans.

IV. IOWA NEONATAL METABOLIC SCREENING PROGRAM ANNUAL REPORT

Stan provided a copy of the annual report to the committee. Stan mentioned that Dari Sharizi and himself have put together some tools to provide easy access to reports. The report consisted of the highlights from the laboratory's perspective of issues that occurred over the course of the year and a summary of the screening
that was performed by the laboratory. It also consisted of a break down of various disorders screened. He mentioned that in December of 2004, the hygienic laboratory staff from the Des Moines site will be moving to a new building in Ankeny. Their office is currently housed in the Wallace building. There is a lot of support and features that will now be available to them. At the end of last year, Dari and Stan began putting together tools that could be accessed via the facility's web site. Facilities will be query and pull up reports for submitted specimens. They will also be able to attain routine reports opposed to the laboratory mailing them via ground mail. The reports will also give information about the quality of the specimens submitted by the facilities and breaks down to nine specific conditions that cause a specimen to be rejected. This will also allow them to put in place quality improvement initiatives.

Pam DeBoer touched on a few of the highlights of the report for 2003. They had 10 detected diseases that were found through newborn screening. For each child that had been detected, she contacted their local physician to validate the tests. They had 379 traits detected through newborn screening. The program recommended genetic counseling for those families.

Each summer the program sponsors a summer camp, both Pam DeBoer, Dr. Lowe, and Myrl attended. Many fun activities are available to the children, such as rock climbing, swimming, B-B gun shooting, canoeing, archery, etc.

The program provides six outreach clinics each year: two in Waterloo, two in Davenport, and two in Iowa City. Dr. Lowell presents several different lectures throughout the state Iowa the year to the residents, staff at the university, family practice office and other groups consisting of physicians and people throughout Iowa. Currently, there are nine cycle sigations that are on chronic transfusion or exchange transfusion; they are performed at their local physician or at the university.

Pam also discussed that last week proved to be quite exciting. A newborn was diagnosed with galactosemia. The baby was very sick and a screen was performed. The program processed the screening within one day of receiving the specimen and was able to report the findings, which most likely saved the baby's life. Kim Keppler mentioned the mother had been breast-feeding the baby. After the diagnosis was made, they supplemented the baby's nutrients with soy formula.

Mavis highlighted the endocrine follow-up care. She works with 3 endocrinologists. They follow-up with on babies who are detected with CAH disorders throughout the state. She discussed hyperthyroid disease and how it requires special treatment.

They are looking at the cut-offs for the TSAs on the newborn screening. The number of confirmed cases was 26 for FY2003. Half of these babies have normal or borderline screens and are not detected with the disorder until later. There is not a lot of literature that supports the cut-off age for screening.
Dawn provided the group with an update regarding the new data system used to track babies who show up having missed the newborn screen. The new data system was released last week, which will work closely with the new web based birth certificate system when it is implemented. The system will electronically match vital records with lab records to ensure all babies are screened.

During the past year, Dawn has collaborated with the Child Death Review Team. Dawn is sent a list of babies who have died in order to determine if the cases need to be reviewed further.

Dawn mentioned she attended the Newborn Screening meeting in Atlanta, Georgia at the beginning of May. The meeting allowed her to meet with the follow-up staff from surrounding states. They discussed the possibility of collaborating more in order to track babies who have relocated to another state. Tonya mentioned that Dawn has been working very diligently on the next edition of "Heel Stick News," which will be issued in July.

Tammy reported that the FA Council has worked together to create folic acid web pages which will appear within the genetic website. Tammy finalized the project with the IT staff within the Department of Public Health and the pages should be up and available within the next week. Within the genetics website, a new feature has been added; family stories.

V. COORDINATOR REPORT

Tonya thanked Tammy for all of her hard and heartfelt work with the Parent Notification Project. Tammy in turn thanked Kim Keppler and the rest of the Iowa Birth Defects Registry staff for all their support and contributions to the project.

Tonya told the group that she is beginning a new chapter in her life. She will be going back to school to pursue a career as a physicians assistant. In the past, she had worked as a genetic counselor for six years and would like to work with patients once again. She asked the group to consider having her as a member on the Birth Defects Advisory Committee.

Julie mentioned that Tonya has shown fantastic leadership abilities. She feels it will be challenging to fill Tonya's vacant position. Tonya has worked hard to get recognition for the Center for Genetics within the Iowa Department of Public Health, as well as in the community. Julie stated that what she has seen thus far in terms of progress within the genetics program in the state of Iowa has a lot to do with the BDAC members, but as much to do with Tonya. There have been many fiscal challenges that Tonya was able to work through while in her position with the Iowa Department of Public Health. Julie wished Tonya the best and said she knows all of the leadership and skills Tonya has portrayed and demonstrated for the genetics program will be well used in the physician assistant world.
Although it will be difficult to fill Tonya’s shoes, the coordinator position will hopefully be posted next week. When the position is posted, Julie will notify the members of the committee in order to obtain their assistance in recruiting someone to fill the position.

Tonya mentioned that Dawn would continue working very closely with the Newborn Screening Program. Tonya stated that Dawn has a lot of abilities and desires to provide education as part of a team; therefore she will take a stronger role in that area as the program continues to move forward.

Meeting adjourned at 4:05

The next Birth Defects Advisory Committee meeting will be held on August 20 at the Wells Fargo bank in Grinnell.

Minutes respectfully completed by Sherry Smith