Congenital and Inherited Disorders Advisory Committee
Minutes
January 29, 2016
12:00 p.m. to 2:00 p.m.
Conference Call

M i n u t e s

Members Present	Members Absent	Others Present
Sandra Daack-Hirsch	Stanton Berberich	Kimberly Noble Piper
Bobbi Buckner Bentz	Sarah Dricken	Carol Johnson
Hannah Bombei	Mary Larsen MoD	Elisa Seeger ALD
Lane Strathearn	Kate Small	Brenna Booth ALD
Lori Murphy-Stokes	Sarah Grotha	Kathy Mathews
Paul Romitti		Emily Phillips
Kelly Schulte		Lisa Neff-Letts
Stewart Boulis		Jennifer Marcy
George Wehby		
Val Sheffield		
Stacy Frelund		
Francis Degnin		
Dan Rowley		
Andrea Greiner		
	Representative Wessel Kroeschell	
	Senator Ragan	

Topics	Discussion/Action
Call to Order	▪ Buckner-Bentz called the meeting to order at 12:05 pm. ▪ Roll call attendance was taken. Quorum present
Approval of October 23, 2015 minutes	Vote on minutes from October 23, 2015 - approved.
Announcements	Newborn screening program and maternal prenatal screening program budgets have not changed from last year, so will not be presented at this meeting.
Approval of NBS Panel Management Policy	Policy #003 Management of the Iowa Newborn Screening Panel was presented for approval. Sheffield requested the description of the process to review nominated conditions be amended to state "The CIDAC Iowa Newborn Screening Panel Subcommittee will evaluate all presented and other available information about a nominated condition ~~using~~ considering the following criteria:" A motion was made and seconded to approve policy #003 as amended. Motion carried and policy #003 was approved as amended. When asked for input, parents Elisa Seeger and Brenna Booth stated they were pleased with the policy and appreciated that Iowa will consider conditions nominated by other than governmental entities.
Research Proposal: Assay Development for a Study of Genetic Prevalence in Myotonic Dystrophy	Dr. Nicholas Johnson presented his research proposal for "Assay Development for a Study of Genetic Prevalence in Myotonic Dystrophy (MDt). Phase 1 of this proposal requests 1500 de-identified residual newborn screening specimens to develop a laboratory testing assay to establish the feasibility of testing for Myotonic Dystrophy using dried blood spots (DBS). Discussion: Kathy Mathews (KM): in the assay development phase did you consider any alternative to using DBS? Nicholas Johnson (NJ): No. Part of the proposal is determining the

	feasibility of using DNA from DBS to detect MDt. Val Sheffield (VS): DBS are a scarce resource. What is the availability of DBS going forward in Phase 2? NJ: Phase 2 will require DBS from other states to have enough to examine the feasibility of testing on a population-wide basis. Bobbi Buckner Bentz (BB): Will other states provide additional DBS? NJ: They will work with the Virtual Repository of Dried Blood Spots Specimens (VRDBS) to obtain the larger amount of specimens. Paul Romitti (PR): How much of the blood spot is needed? NJ: 1/8th of a spot should be sufficient for the PCR assay. PR: Would you require positive controls from the 1500 specimens? NJ: That would be desirable, but we wouldn't know if any of the samples are from babies with MDt. George Wehby (GW): Would it be feasible to obtain residual DBS from newborn with MDt diagnosis? PR: Yes. We could correlate cases reported in the Registry (Iowa Registry for Congenital and Inherited Disorders) with their newborn screening specimens. After questions answered, Romitti moved to approve Johnson's research proposal, and providing case controls if able. Sheffield seconded. Roll call vote – Assay Development for a Study of Genetic Prevalence in Myotonic Dystrophy research proposal approved. Piper will draft a letter for the Chair's signature recommending approval of the research proposal and will provide it to Dr. Johnson.
Research Proposal Longitudinal pop-based study of gene-environment contributors to developmental delay	Dr. Lane Strathearn presented a proposed research project that will be submitted to NIH in February for grant funding. The proposal is "A longitudinal population-based study of gene-environment contributors to developmental disability using smart phone technology." Pregnant women throughout Iowa will use their smart phones to enroll in a longitudinal study, and provide data throughout their pregnancy and the first two post-natal years. They will also provide consent for their infant's residual newborn screening sample to be used for genetic testing. At age 2, children will be screened for developmental disorders such as autism and language delay. Genetic and environmental markers will then be identified from this data. Dr. Strathearn is requesting a letter of support for his application to NIH. Discussion: Kathy Mathews (KM): How are families recruited? Lane Strathearn (LS): The smartphone app will be made available to pregnant women through a promotional campaign. The app contains informed consent information. Paul Romitti (PR): If you are going to use e-consent (LS=yes), there is precedent for that in our programs. It will need to be a very focused proposal to obtain RERC approval. I can help you with that. George Wehby (GW): What about the epigenetics? LS: Due to the nature of epigenetics, there will be some limitations as to what can be done with the blood samples. GW: Is there an alternative to collecting DNA samples at 2 years of age? LS: The parent will be completing a developmental assessment of the child including a video they can upload, and the study will follow up with DNA testing if necessary. GW: Is the genetic information to identify the phenotype? LS: Yes, using polygenetic capabilities. This is cutting edge research. GW: Is self-reported assessment a reliable way to identify the

	phenotype? LS: Child Health Specialty Clinics will conduct comprehensive assessments as needed. Discussion was held as to what the CIDAC letter of approval for the premise of Strathearn's research proposal should say; members want to be clear they are not approving the research study, just the premise of the proposal. Agreed letter will state CIDAC approval for the premise of the proposal only, and that further review would require presentation of a final proposal with IRB approval. Sheffield moved to provide a letter supporting the premise of Strathearn's research proposal. Wehby second. Roll call vote: motion carried. Piper will draft a letter for the Chair's signature.
Healthy Iowans Priority Issues	Piper reviewed the request for members to identify congenital and inherited disorders-related priority health issues. Only one person provided suggestions for priority issues. Christina Trout suggested activities take place to assist individuals with an inherited disorder as they transition to adulthood; and have resources available to support their integration into the workforce or to assist with activities of daily living. Also need support for care givers. Sheffield stated they will address this at an upcoming staff meeting and will provide some issues for the CIDAC to consider. It was suggested that Piper provide the information from prior Healthy Iowans reports, and to resend the request to all members. Members are to have their suggestions to Piper by February 19.
Next meeting date and agenda	The next meeting will be April 22, 2016 in Grinnell. Program reports RERC presentation
Adjournment	Meeting adjourned at 1:50 pm.