Congenital and Inherited Disorders Advisory Committee
Minutes
April 24, 2015
1:00 p.m. to 3:00 p.m.
Drake Community Library, Grinnell

M i n u t e s

Members Present	Members Absent	Others Present
Sandra Daack-Hirsch		Kimberly Noble Piper
Stewart Boulis	Sarah Dricken	Carol Johnson
Lori Murphy-Stokes	Andrea Greiner	Lisa Neff-Letts
Val Sheffield	Stacy Frelund	Travis Henry
Shannon Sullivan	Kari Atkinson	Ben Darbro
Bobbi Buckner Bentz	Michelle Gogerty	Tonya Millard
Mary Larew	Kelly Schulte	
Cathy Evers/Hannah Bombei	George Wehby	
Sarah Grotha*		
Kate Small		
Stanton Berberich		
Francis Degnin	Representative Wessel Kroeschell	
Paul Romitti	Senator Ragan	
Dan Rowley	*via phone	

Topics	Discussion/Action
Call to Order	▪ Degnin called the meeting to order at 1:05 pm. ▪ Roll call attendance was taken – quorum is present.
Approval of January 23, 2015 minutes	▪ Minutes approved by exception vote.
Announcements	Retirements – Kim Horton and Cathy Evers from the University of Iowa Division of Medical Genetics are both retiring at the end of May. There will be a retirement party May 29 at the Vine in Coralville. Everyone is invited. If interested in attending, please let Lisa Neff-Letts know. Hannah Bombei will be the new Coordinator of the Regional Genetics and Consultation Clinics upon Cathy's retirement. Dr. Bernat, a metabolic geneticist, will begin working at the Division in July. Dr. Bernat has an interest in lysosomal storage disorders. Subcommittee reports – Informed Consent –Daack-Hirsch reported that the subcommittee will meet in May to discuss options for informed consent. Daack-Hirsch and Piper have collected information about costs to store residual dried blood spots and costs for managing consent. Once a decision is made about the consent process, the issue of storage of the residual specimens and management of the consents goes back to the use of residual newborn screening specimens subcommittee. Residual Specimens – Romitti reported that this subcommittee is on hiatus pending the recommendations from the informed consent subcommittee. Adding Conditions – Piper – The subcommittee looking at the addition of lysosomal storage disorders is recommending this subcommittee be expanded and called the Subcommittee for management of the newborn screening panel. Jenny Marcy has

	agreed to chair that subcommittee. Additional members would be brought to the subcommittee on an ad hoc basis depending on the condition(s) being discussed. Members approved the recommendation for the new subcommittee on a voice vote.
Request for LOS for carving hemophilia and clotting disorders treatments out of Medicaid Managed Care	▪ Tonya Millard, Executive Director of Hemophilia Iowa, requested a LOS from CIDAC members to have hemophilia carved out of the Iowa Managed Care Act RFP and remain a fee for service model; if a carve out is not an option, then at the least that clotting factor be on a fee for service schedule. Letters from the National Hemophilia Foundation to the director of Iowa DHS was shared. Tonya also shared copies of informational sheets about Medicaid Managed Care and Hemophilia and a legislative fact sheet. ▪ Members request an opportunity to review the information Tonya provided before deciding on whether to provide a letter of support. Piper will disseminate the information, and members will have an opportunity to vote to provide a letter of support via email or SurveyMonkey. Tonya noted that there are over 100 Medicaid patients in IA, and that it averages $250,000 to treat one person with hemophilia per year.
Policy for release of data	▪ Members reviewed the draft of the policy for the release of CCID data, including newborn and maternal prenatal screening data. Edits were suggested. Piper will make the recommended edits and post it for approval via email or SurveyMonkey.
Research proposal	▪ Dr. Ben Darbro and Dr. Travis Henry presented a research proposal "Newborn screening and Genotype-Phenotype Associations in Immunodeficient Children with 22q11.2 Deletion. (Presentation and proposal attached.) 22q11.2 is the most common and significant SCID "false positive" – screen is not positive for SCID, but is significant for the secondary disorder of 22q11.2 deletion. This project would benefit newborns as it validates the screening process for targeting 22q11.2 deletion specifically. The project aims are to 1. Establish performance metrics for the detection of the 22q microdeletion by the TREC assay (current assay used for SCID screening). 2. Establish the correlation between TREC assay positive 22q deletion patients and the incidence of functional immune system dysfunction and an alternative measure of T-lymphocyte numbers. 3. Discover candidate modifier genes for T-lymphocyte depletion and immune dysfunction in children with 22q deletion. The researchers propose to use retrospective 22q samples from TREC assays already completed through the SCID pilot (half TREC + and half TREC -) linked with an already completed chart review to conduct a Proximity Ligation Assay (PLA) to determine those appropriate for Exome Sequencing. The development of this testing process may improve SCID screening; provide confirmation of immune defects and provide benefit to the Iowa Newborn Screening Program; and 22q deletion sequencing data may identify genes associated with impaired immune response and provide improved clinical intervention at an earlier time. ▪ Discussion: ▪ Dr. Paul Romitti – What is the project specifically asking this committee? - Darbro – Asking to associate data from previous TREC assays from SCID pilot with their database, and to use residual newborn screening specimens from + and – specimens to test PLA. ▪ CIDAC Members – Is this considered newborn screening new test development or is this considered research? Is informed consent required? Henry – U of I IRB has waived consent because this is

	using data already collected. Not sure of new test QA/QI vs. research. ▪ Piper – suggested PIs discuss proposal with Heather Adams, IDPH Assistant Attorney General, for opinion of new test development vs. research, prior to proceeding with presentation to RERC. ▪ Boulis – Is there potential to compare 22q deletion testing through non-invasive prenatal screening with specimens collected through this project? – Darbro – That would be beyond the scope of this specific proposal, but would be an interesting study. Boulis – Labs providing non-invasive prenatal testing have not demonstrated the efficacy of this screening. Would be beneficial to compare. Darbro – All these labs' work is proprietary, so it would be interesting to do a comparison study. ▪ Romitti moved that the premise of the research proposal be approved, pending Heather Adams' recommendation. Boulis seconded. Motion passed on voice vote with two abstentions.
CoIIN Timeliness in Newborn Screening Project	▪ Carol Johnson presented an overview of the state's timeliness in newborn screening Collaborative Improvement and Innovation Network project. Iowa has a goal of 95% of newborn screening collection forms being received by the State Hygienic Laboratory in Ankeny within 60 hours of birth. Birthing facilities will be provided with infographic reports on at least a quarterly basis that will describe their status in meeting the state goal. CoIIN team members will work with birthing facilities to lead a QI process specific to their organization's needs in order to meet the goal. CoIIN Team members are Stan Berberich, Team lead; Carol Johnson; Laura Malone from the Iowa Hospital Association; Kristen Ernsperger from Mercy Medical Center Des Moines; Ashley Comer from the State Hygienic Lab; Kim VonAhnson from UnityPoint Des Moines' Lab, and Kim Piper.
Next meeting date and agenda	The next meeting will be July 24 from 12:00 pm to 2:00 pm "ish" via conference call.
Adjournment	Meeting adjourned at 3:20 pm.