Congenital and Inherited Disorders Advisory Committee
Minutes
April 22, 2016
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
Bobbi Buckner Bentz		Carol Johnson
Hannah Bombei		Myrl Holida
Stanton Berberich		John Bernat
Lori Murphy-Stokes	Sarah Grotha	Travis Henry
Paul Romitti	Lane Strathearn/Tom Scholz	Judy Miller
Kelly Schulte	Stacy Frelund	Lisa Neff-Letts
Stewart Boulis		Jennifer Marcy
Andrea Greiner		
Val Sheffield		
Dan Rowley		
George Wehby		
Francis Degnin		
	Representative Wessel Kroeschell	
	Senator Ragan	

Topics	Discussion/Action
Call to Order	▪ Daack Hirsch called the meeting to order at 1:15 pm. ▪ Roll call attendance was taken. Quorum present
Approval of January 29, 2016 minutes	Vote on minutes from January 29, 2016 - approved.
Pompe disease addition to NBS panel	• Purpose of CIDAC vote discussed. Per policy: CIDAC will vote whether to recommend the subcommittee develop a proposal to CIDAC as to whether Pompe pilot screening should be considered. • Explained that the subcommittee vote on whether to add Pompe to the Iowa NBS panel was 5 for, 5 against, and 2 undecided. • Marcy presented the background of Pompe disease. See attached presentation. • Bernat discussed the treatments for Pompe. Questions discussed. • Berberich presented information about the circumstance of adding conditions to a mandatory screening panel based on limited knowledge of the early identification and treatment benefits. Questions discussed. Harm/benefits of early identification and consideration of late onset variant. Treatment (enzyme replacement therapy available last 10 years) improves quality of life. Life expectancy approx. to 20s. No treatment = fatal condition. Berberich – are we doing research to determine the course of disease with treatment through mandatory screening? Sheffield – essentially translational research. Daack-Hirsch – different types of research/clinic studies other than randomized clinical trials that conduct research within the clinical setting i.e., implementation science . Berberich – when

	there are issues that are unanswered, i.e., when should enzyme replacement be started – pre symptomatic? Also don't understand the cost benefit/ratio to patient. Rowley – what is the penetrance? Bernat diagnosis based on • Wehby – agrees with Stan's position. • Stan clarifies his position that due to the uncertainty, we should not put it under a mandated program, but work with other states to develop larger studies to address newborn screening for these conditions to an environment where patients can be informed of the research and elect participation. • Bernat – this is do we think that newborn screening for infantile Pompe disease is beneficial enough to justify the "incidental finding" of late onset • Stan – as a public health program, we need to consider the greatest benefit for the greatest numbers • Rowley – the key issue is "mandated" By mandating screening, we are requiring families to take on the burden of addressing the unknown. If research – families have the option to be involved in the studies to their child's benefit. • Sheffield – is Pompe the best of the LSDs to address? Bernat – Pompe has the larger body of evidence • In other states Pompe was legislatively mandated. • Daack-Hirsch – the NBS program could lead the way of testing in a program that provides a "safe" environment to "pilot" test Pompe NBS. Not necessarily a dichotomy research vs clinical screening. • Stan – investigate proceeding with screening, but with informed consent. How? Bigger hazard to NBS if we use current mandated screening (without their consent) to discover Pompe course. • Miller – long term follow up shows that families always are motivated by what could possibly happen once given a diagnosis • Daack-Hirsch – what is the question? Subcommittee to continue and expand review of conditions that come with "ambiguity" and give updates to the CIDAC. Explore use of newborn screening framework to research new conditions, i.e., structured pilots that obtain informed consent for participation. • We don't have the ability to provide enough information to parents about the course and treatments of the disease for them to make an informed decision to participate in this mandatory screening program. Consensus of CIDAC Members: a Subcommittee should be established to: • Continue and expand review of conditions that come with "ambiguity" and give updates to the CIDAC. • Explore use of newborn screening framework to research new conditions, i.e., structured pilots that obtain informed consent for participation. • Follow and report on the experiences of the States where Pompe NBS has been mandated.
Informed Consent and Use of Residual NBS Specimens for Research	Daack-Hirsch reviewed the information presented from the Informed consent subcommittee. Distinction of retaining residual NBS for INSP QI/QI programming vs research. Degnin - Motion to accept the subcommittee's proposal to not pursue the storage and consenting for use of residual NBS

	specimens for research. And not restrict research requests for specimens stored. Rowley second. Motion carried with 7 yes, 1 no, 1 abstain. The retention subcommittee will convene to determine length of time specimens will be stored for newborn screening program activities.
Healthy Iowans Priority Issues	Piper stated that CIDAC members prioritized the issue of transition from child/adolescent to adult genetic health services for the Healthy Iowans Health Improvement Plan. We need to develop action steps to address and performance measures for this activity. Piper will work with "Got Transition?" website and the Heartland Regional Genetics Collaborative transition planning work group to explore options.
Next meeting date and agenda	The next meeting will be July 22, 2016 via conference call. Maternal Prenatal Screening Program Program reports RERC presentation
Adjournment	Meeting adjourned at 3:50 pm.