

Genetic Diseases Program Advisory Committee Meeting

July 28, 2023, 2:00 p.m. – 3:30 p.m.

Location: Benson Tower, 1450 Poydras St., 20th floor, Room 2024

Zoom Link: <https://ldhophbfhgenetics.zoom.us/j/89076697817>

Minutes

I. **Roll Call**

- Cheryl Harris welcomed everyone to the meeting. 25 participants. Members present were Cheryl Harris, Dr. Hans Andersson, Dr. Joseph Bocchini, Michelle Duplantier and Tiffany Allemand. A quorum was not established.
- The Bureau of Family Health is working to update the statute to include more representation of other specialties.
- Self-introductions of all participants in meeting

II. **Review of the Committee Minutes from 4/28/23**

- The minutes from 4/28/23 were attached to the invitation prior to the meeting. Cheryl asked if there were any corrections, revisions, or questions.

III. **Changes within the Bureau of Family Health's Genetic Diseases Program**

- Statewide Nurse Consultant
 - Heiki Griffin introduces self to committee
- Genetic Diseases Program Strategy Manager
 - Rodney Goldsmith introduces self to committee
- Early Hearing Detection & Intervention (EHDI) Program
 - Cheryl shared org chart with explanation of new and ongoing roles within the Genetics program and in conjunction with EHDI Program

IV. **Laboratory Updates –Kelli Dugas, NBS Manager, OPH Lab**

- Status of In-House Testing for SMA, MPS I and Pompe
 - The lab is currently in the process of validation testing for MPS I and Pompe and hope to begin in-house testing at the end of this summer.

V. **Other Committee Updates – Rodney Goldsmith**

- Rare Diseases Advisory Committee
 - Introductory meeting held to establish future meeting dates and agendas
- Sickle Cell Commission – Upcoming August 8 Meeting
 - Filling vacancies
 - Event planning for September Sickle Cell Awareness Month

VI. **Open Discussion/Public Comment**

- Who will do follow up following Dr. Hollman’s retirement?
 - Region 9 neurology contract process begun and working on transition
 - Region 2 neurology clinic has transitioned to community providers successfully
- Who are positive X-ALD patients referred to?
 - We are currently not set up for this, working on this process
 - Due to complicated nature of the disease, it was suggested to begin discussion about the follow up care required for these patients. Inclusive of:
 - Endocrinology
 - Pediatric
 - Adult
 - Neurology
 - Pediatric
 - Adult
- Because we were not ready for referrals of positive X-ALD patients, we are looking into revising the Newborn Screening rules to reflect a halt on other facilities’ testing unless on the LA NBS Panel to ensure proper readiness
- Senate Bill 201 updates regarding virtual meetings/attendance
 - Cheryl/Heiki to follow up
- MPS I Data Share – Bonnie Brignac

VI. **Upcoming Meetings, Webinars, Workshops**

- HHS Advisory Committee on Heritable Disorders in Newborns & Children Meeting
 - August 10, 2023: 10:00 a.m. – 2:00 p.m. ET
 - August 11: 10:00 a.m. – 1:00 p.m. ET
- Registration Link: <https://www.achdncmeetings.org/registration/>

VII. **Upcoming Meeting Dates for 2023**

- October 27

VIII. **Announcements**

- Recognition of Joe Elsin
- We are a Unite Grant Recipient; 1 of 5 states
 - Goal is to strengthen partnership between NBS blood spot and Newborn Hearing Screening
 - Requires parent involvement; Tiffany Allemand to serve as our consumer representative

IX. **Adjournment**

- The meeting adjourned at 2:50 p.m.