NewSTEPs
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Short Term Follow Up Technical Assistance Webinar
March 2015

Presentations:

- State Profile Mississippi—Ashlyn Booker, MBA
- The Controversy of Mild Congenital Hypothyroidism—Caroline Nucup-Villaruz, MD
- Vermont Protocols for Follow-up of Mild Congenital Hypothyroidism—Cindy Ingham, RN
- Feasibility of Providing Long Term Follow-Up for Congenital Hypothyroidism—Ning Rosenthal, MD, PhD

Please direct all comments/questions pertaining to this presentation to Thalia Wood at Thalia.wood@aphl.org or 240-485-2701.

Careema Yusuf: Welcome, everybody. Again, this is the short-term follow-up webinar for this afternoon.

Carol, I'm going to turn it over to you.

Carol? Press "start," "7" to unmute.

Carol Johnson: Sorry.

Careema Yusuf: Hello?

Carol Johnson: Hi. This is Carol. Sorry, I don't know, my phone just kind of went dead there for a moment. I apologize, everyone.

My name is Carol Johnson. On behalf of John Thompson and I, as co-chairs of the APHL short-term follow-up committee, we'd like to welcome you to today's webinar. We're excited to have with us today Ashlyn Booker from the state of Mississippi to tell us a little bit about what happens in newborn screening in their state. Ashlyn is filling in for Phillis Hoggatt just in case you wondered where Phillis was.

Thank you, Ashlyn, for doing that.

Then, next, we have three speakers who are going to talk about congenital hypothyroidism. First, we have Caroline Nucup-Villaruz from the Washington Newborn Program, Cindy Ingham from Vermont and Ning Rosenthal, also talking about providing long-term follow-up for congenital hypothyroid patients in California and Hawaii for her project.

We are very, very, very happy to have these individuals join us today. Thank you very much. I will turn this over to Ashlyn to talk about what happens in Mississippi in the newborn screening program.

Ashlyn Booker: Hello, everybody. Can you hear me?

Carol Johnson: Yes, we can. Thank you, Ashlyn.

Ashlyn Booker: As Carol just mentioned, I am filling in for Phillis Hoggatt. She's, unfortunately, out with the flu today. I'm hoping that she is well very soon and join us.

In regards to Mississippi's newborn screening program, we do test for over-40-something genetic conditions and disorders. The conditions are identified for 2013. There were 5 metabolic disorders identified, 3 [inaudible 00:02:48] glutaric acidemia; [inaudible 00:02:52], there were 2; and then there was a classic PKU. There were 15 endocrine disorders, 61 hemoglobin disorders, 5 biotinidase deficiencies, 6 cystic fibrosis and 1 [SCD 00:03:17], which was a prenatal diagnosis. The other conditions, there 3 [inaudible 00:03:26], 4 of the [var 00:03:33] hemoglobin and then there were 2,261 hemoglobin traits identified.

We've got some exciting new developments for the newborn screening program. We were recently mandated to perform the CCHD screening, so that was added to our newborn screening panel in October of 2013. It was formally adopted and was approved by the board and it has been added, so we've been doing our due diligence in making sure that the hospitals can report that information to the health department so that we can follow up as necessar6y.

As of right now, our role is to just compile the data. We are working with our IT professionals here to make sure that we have a proficient method to transmit that information to the lab and, of course, from the lab to our department here in Mississippi. The agency has undergone some quality-improvement initiatives as of November of 2014. The state health officer sent out a directive that we are going to become an agency that is QI-driven. Newborn screening was also included in that, and so what we did was we formed a quality improvement group here.

Our first task was to address timeliness of newborn screening specimens to the lab. We did a preliminary survey of all of our birthing facilities throughout the state. We did receive a decent response rate. What we did was we gauge the questions around current practices and what types of things in terms of educational resources that they have available to them in the lab to ensure that they were collecting specimens properly and timely and to make sure that they were aware of the protocols in place for the courier service, which we shipped through UPS, to our lab in Pennsylvania.

The results of that survey pretty much revealed that a lot of our hospitals wanted some assistance in developing manuals and that they expressed the need to want to have some in-house educational materials. We have analyzed all of that data. We've sent out some targeted outreach through our district coordinators who are stationed throughout the state. We are waiting on our first quarter results to see if any of our timeliness quality measurements have improvement.

I believe at the time that we initiated the QI project, specimens were reaching the lab at about 2.9 days. Of course, we strive for less than 3 days, which that number wasn't too bad, but since we started our QI project, the numbers for 2014 have gone down to about 2.5 days. We're really proud of our hospitals. They have been putting forth a great effort to ensure that they are shipping those specimens in a timely manner. This will be an ongoing QI initiative. We'll continue to monitor their timeliness.

Since then, we've engaged in part of 2 of our QI project, which will include a quality portion. Now, we're focusing on the quality of specimens because quite a significant amount of the specimens are returned to the lab or the lab is returning some of those results, indicating that some of the specimens are poor quality. We are trying to address that and make sure that the hospitals are aware of how to collect them and how to properly lay them and store them for shipment and things that will ultimately affect the quality of that particular specimen.

We're really excited about that. We're excited that we're able to reach out and do some targeted outreached based off of the data that we've already compiled through the survey and, of course, through the feedback that we're receiving on the onsite visits that are conducted quarterly. We'll be happy to update you guys throughout the process. We were one of the first QI teams that were formed with the agencies that we're very proud they were able to address our issues with newborn

screening and so that we can continue to uphold a standard for our newborn screening program.

Phillis is the nurse for the program. We're in the process of hiring another nurse who will be on board effective April 1st. There will be another nurse here along with her. I myself, I'm the director. I've been on board since about March of last year. It'll be a year that I've been here with the health department.

Phillis' information has been provided on the slide. I'm not sure if you guys would like for me to indicate that information, but you guys can always feel free to give us a call at the office if you have any questions specific to what we're doing here. We'll be happy to assist in any way that we can. Okay? Thank you.

Carol Johnson: Thank you, Ashlyn. That was very interesting. We're curious about what is on your survey.

I would encourage other states to contact Phillis if you have questions related to their survey.

Thank you for your presentation, Ashlyn.

Ashlyn Booker: Thank you.

Carol Johnson: Next, we have Caroline Nucup-Villaruz. She's going to talk to us about The Controversy on Mild Congenital Hypothyroidism, The Path We Took to Resolve the Dilemma in Washington Newborn Screening.

Caroline, please go ahead and share what you've learned with us.

Caroline Nucup-Villaruz: Hi. Good afternoon. Before anything else, I would like to thank the APHL and the short-term follow-up committee for giving me the opportunity to actually present today. This presentation was made possible with the input and invaluable contribution of our endocrine consultant, Dr. Patricia Fechner, who is also the coauthor of this paper.

Can we move to the next slide please?

The objectives of this presentation are as follows, to present the issues and dilemma of either confirming or ruling out the diagnosis of hypothyroidism based on the serum thyroid results, to give a definition of compensated or subclinical hypothyroidism, to determine the appropriate TSH threshold at a certain age that merits further monitoring

and follow-up and to share the consensus from the endocrinologist regarding appropriate follow-up of mild or compensated hypothyroidism.

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As we all, congenital hypothyroidism is the most prevalent disorder in any newborn screening program. I can say that globally and in the US, for Washington, it used to be about 1 per 3,600. This was way back in 1977 when we were still using total T4 until the time we switched to primary TSH in 2004. As you can see, the prevalence has significantly increased to 1 per 1,200. This may also be attributed to the fact that we are a 2-screen state were 30% of cases are detected on the second newborn screen or subsequent newborn screen. Aside from that, we have a significant size of Asian population here where Asian babies are at risk for mild congenital hypothyroidism.

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Compensated or subclinical hypothyroidism is defined as a serum TSH which is above the upper limit of the normal range, along with the fact that total T4 or free thyroxine level is normal. Certain questions come up wherein does lack of treatment result in mental retardation or does early treatment result in normal neurocognitive outcome.

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I think everyone is very familiar with the definition of permanent hypothyroidism and transient hypothyroidism. This is just a repetition. For interest of time, I'll just go fast in some slides.

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As we all know, there is a high TSH surge with the peak of 70 of 30 of life and there's an increase in the free thyroxine which is TSH dependent and could last for 1 to 2 months.

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This slide shows the different reference ranges of several diagnostic laboratories here in Washington State. This values are based off on serum and not the dried blood spot.

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As you can see, based on this table, a study by Kahler conducted in Argentina shows that 97.5% of TSH from normal newborn population do not exceed the level of 4.5.

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This is another study by [Lam 00:14:19], which was released in 2012, showing that, at 7 days to 1 year of age, the TSH remains to be below 6, if you can just focus on the last column there for babies at the ages of 7 days to 1 year.

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I just want to share with you the TSH cutoff that we have been using since 2012. So far, to our knowledge, we have not missed a confirmed case of a true congenital hypothyroidism.

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Now, having been the primary follow-up for congenital hypothyroidism since 2008, I have noticed a pattern of serum TSH among patients diagnosed with mild congenital hypothyroidism. Secondly, due to the challenges and dilemma that we have encountered as a newborn screening program, we have decided to present 1 year of data to the endocrinologist at the PEARL Conference last year in 2014. This data shows that 95 babies were referred to undergo thyroid function test, of which 56 or 60% were confirmed to have congenital hypothyroidism and 37 or 40% were ruled out.

This table also shows the range of the highest and lowest serum TSH value as well as the timeframe when the diagnosis was made and when treatment was initiated.

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The TSH values were plotted because I wanted to superimpose the true positive cases along with the false positive cases to drive our point to the endocrinologist.

Next slide please.

By magnifying and taking a closer look at these cases-

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There is obviously an overlap between the true positive cases and the false positive cases.

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As you can see, based on this 2012 data, it clearly exemplified the issues and dilemma that we have encountered as a newborn screening program. It was based on the fact that different state newborn screening programs have different methods. Some states use total T4, some states use primary TSH, some states do just a 1 screen versus states who do 2 screens. Also, different physicians and endocrinologists use different laboratories, and different laboratories different reference ranges.

The bottom line was hoping we would get a consensus from the endocrinologist regarding appropriate management of compensated hypothyroidism. That was the main objective of us presenting the 2012 data at the PEARL Conference on March 8, 2014 in Portland, Oregon.

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I just want to show that I've been trying to see what available studies or articles were out there and to get the fence from the endocrine specialists regarding their point of view on how to manage these cases. Based on this literature review, the majority of endocrinologists are very conservative in addressing and approaching this issue, wherein 6 authors favored treatment and follow-up, 2 will treat depending on confounding factors and only 2 favor no treatment.

The first and second journal articles served as the foundation or springboard for our current recommendations, which brings us to the ...

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... to the conclusions that endocrinologists differ in regards to management and opinion on how to manage congenital hypothyroidism based on our data. More endocrinologists favor monitoring, follow-up and treating mild congenital hypothyroidism based on literature review. Finally, we were glad that a consensus was reached during that PEARL Conference led by Dr. LaFranchi and Dr. Fechner that a serum TSH of 6.0 or higher at an age of 14 days or older will need a repeat serum thyroid studies or a referral to a pediatric endocrinologist. Thus, the TSH protocol and memo for our newborn screening program were modified based on the above consensus and recommendations.

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This slide was adopted from Dr. LaFranchi's article showing the algorithm in managing these mild cases of congenital hypothyroidism.

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It would be interesting to collaborate and conduct further studies on the following, to compare the outcome of short-term versus long-term follow-up, early versus late treatment, with or without treatment, benefits and adverse effects. It would also be helpful if we can get a consensus or clinical practice guidelines regarding these terms from the endocrinology section of the American Academy of Pediatrics.

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As always, I wish to thank again my colleagues here at the Washington State Newborn Screening, Dr. Fechner and the endocrinologists of Seattle Children's and Dr. LaFranchi for his guidance and leadership within the PEARL.

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Finally, it has been a pleasure meeting and conferencing with Cindy Ingham from Vermont Newborn Screening to coordinate our presentations today. She is the next speaker for this webinar. Thank you for your kind attention. I would be happy to answer questions later. Thanks.

Carol Johnson: Thank you, Caroline. That was very interesting. I was at the presentation at the meeting, but it's great to see this information again.

I forgot to mention that we're going to save our questions till the end of everybody's talk so that the speakers can stay on the line just in case somebody might have a question for you. Remember that you can also type your question in the webinar window to the far left. Also, we're going to move on to our next speaker that Caroline graciously gave me a great into to.

This is going to be Cindy Ingham from the Vermont Newborn Screening Program to talk about their protocols for mild congenital hypothyroidism.

Thank you, Cindy.

Cindy Ingham: Thank you very much, Carol. Can you hear me?

Carol Johnson: Yes, I still hear.

Cindy Ingham: Oh, great. Welcome to Vermont.

Next slide please.

This, I thought you'd enjoy seeing Church Street, which is just steps away from my office. That's the kind of winter we're having.

Next slide please.

Just to give you an overview of what Vermont is like, we're a fairly small state with a population of about 625,000 people. We have just under 6,000 births each year. You can see that it's going up slightly in 2014, which is great.

Next slide.

Vermont is a single screen state and we contract with the New England Newborn Screening Program in the Boston area of Massachusetts for our laboratory services. T4 is the primary marker to screen for hypothyroidism and the TSH is done as a second-tier screen on a number of categories of infants, as you can see on the bottom left of the slide here.

We do collect data on the filter paper, asking if mothers have a history of thyroid disease and if they were on medication. We also ask hospitals or home birthing midwives to write in a comment if there is a known family member with hypothyroidism. This sort of information clues in the folks who receive the filter papers in the lab that TSH may be needed if the infants don't fit into one of the other categories.

At times, being a small state, we actually recognize names. If nothing is written in, but we can say, hey, wait a minute, I think that baby has a sibling with hypothyroidism, we can always call and request that a TSH be done.

The TSH is interpreted by the testing laboratory based on the infant's age in hours. As you can see on the screen, after 96 hours of life or 4 days, anything under 15, generates a report stating that the results are within range. These are, therefore, not flagged electronically as needing follow-up.

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The Vermont Newborn Screening Program enjoys very close working relationships with all of the consulting sub-specialists who, incidentally, are on our newborn screening advisory committee. In reviewing endocrine cases' recent statistics with the endocrinologists a couple of years ago, they recommended that we consider a more conservative approach in the follow-up of babies over a week old whose TSH was over 6.5 mostly because of the possibility of the subtle effects of persistent mild elevation in the TSH.

We started doing this in calendar year 2012 using the following procedures. First of all, the newborn screening program nurse reviews all of the filter paper results daily, looking for babies who are over a week old and whose TSH is over 6.5. When we identify one, if there's not a clear reason as to why that result is there, the nurse calls the primary care provider's office or the home birthing midwife to request a repeat.

Now, bearing in mind that the report that is sent to the PCP shows everything in range, when we first started doing this, some of the primary care providers preferred to discuss the rationale with the endocrinologist, and they were pleased to talk with the providers and explain it. This happens less frequently nowadays because it's become part of our routine. Depending upon the results and the situation of the individual baby, the PCP will either obtain a repeat filter paper or, perhaps, a serum T4 and TSH.

We are extremely lucky in Vermont that we're authorized to access both the New England Newborn Screening Program's database to obtain filter paper results as well as the electronic medical records for the medical center out of which all of the sub-specialists practice. This means that the nurse who's tracking them very closely can see laboratory results of repeat testing almost as soon as they're ready. When this happens, the nurse checks in with the endocrinologist for recommendations and relays these to the primary care provider.

Now, this does not take out the role of the primary care provider. They may or may not already be aware of the results and recommendations, but contacts from the newborn screening program minimizes the possibility of delays. This system if very effective in facilitating the earliest possible entry into care, which is actually the theme for the webinars that the short-term follow-up committee has been presenting.

Obviously, if the TSH was not just mildly elevated, the endocrinologist would already have seen the baby in the clinic, but, however, with mild elevations, they're more likely to opt for further testing at the local level to see if the TSH continues its downward trend before deciding whether or not the parents should bring the baby into the clinic for further workout and possible supplementation with thyroid hormones.

Then, finally, the newborn screening program monitors periodically to see if the babies are still on medication. I'll speak a little bit more about that in a minute.

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Now, this slide shows the impact of adding the monitoring of mild TSH elevations over a 2-year period. Because our numbers are so small, I chose an inclusive period of 5 years to show the difference. You can see, in 2013 and 2014, that, when we started this additional level of monitoring, the number of infants placed on medication was significantly higher than what it would have been using the standard algorithm.

In 2013, we treated, and are still treating, 8 infants, whereas, only 3 would have been flagged through the routine screening algorithm. In 2014, it was 12 versus 3. So far, in 2015, I think we're following 9 infants already, but don't quote me on that. These infants are not quite old enough yet to have been given a trial off medication. The long-term need for continued supplementation has not yet been measured, but we will do that. I do expect that, if we adopt the recommendations such as Caroline just mentioned that were decided upon at the PEARL Conference and start re-screening at 6 rather than 6.5, these numbers will increase.

I do want to point out that we have not yet tracked the numbers of babies whose mild elevations result without treatment. We do plan to do so starting with babies born in 2015 so that we can quantify the effects on staff time, which, certainly, hasn't changed.

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What's in the works? We continue to work very closely with Vermont's pediatric endocrinologists to refine our processes. We will continue to monitor to see if and when treatment is discontinued for babies with mild congenital hypothyroidism. We also do a retrospective review of infants who are seen in the endocrinology clinic to see if there were any

babies who may have been referred for treatment who might have been missed in the screening. I'm happy to say there have been none so far.

Objective data are really critical when it comes to establishing consensus about the screening algorithm with the specialists, including those out of state who may be used to a different system. I think that information gleamed from webinars like the one we're having right now have a lot of practical value for those of us in the newborn screening program management, and we can draw on other states' experiences to modify our own programs. When you're dealing with small numbers the way we are here in Vermont, it can be less convincing to make a case. When you add in the experiences of the states around you, it is, I think, more convincing.

Finally, we would like to have an explanatory fact sheet for primary care providers to share with families when mild, but persistent elevations of the TSH particularly when the newborn screening report is considered within range because that can be confusing. If any of you actually have a brochure or a fact sheet that you share with parents, perhaps, that's something you can forward to tell you to attach to these minutes because that would be great if we could not reinvent the wheel.

May I have the next slide please?

It's time for me to get back to work. This is just down the street. I want to thank you for your attention. It's time to move on to our next speaker, speaker Dr. Rosenthal. Thanks.

Carol Johnson: Thank you, Cindy. That was great. We're sorry for your weather. I know many states from the west coast to the east cost have had crazy weather this year.

Cindy Ingham: Absolutely. Anyone who's listening in from Boston can say amen to that.

Carol Johnson: Exactly. Exactly.

Our next speaker is Ning Rosenthal. Dr. Rosenthal is going to talk to us about providing long-term follow-up for congenital hypothyroidism patients in California and Hawaii.

Dr. Rosenthal, please go ahead and start your presentation.

Careema Yusuf: Please press "star," "7" to unmute your phone.

Ning Rosenthal: Hello?

Careema Yusuf: Hi. Yes, we can hear you.

Ning Rosenthal: Oh, you can hear me now?

Careema Yusuf: Yes.

Ning Rosenthal: Okay. Okay. Good afternoon, everyone. This is Ning Rosenthal. I'm with the California Genetic Disease Screening Program. [inaudible 00:33:48] a little bit, my presentation is not about the diagnostics and algorithm of [PH 00:33:55]. It's rather focusing on the findings from a provider-based survey we conducted last year on the feasibility of providing long-term follow-up for congenital hypothyroidism patients at primary care settings in California and Hawaii.

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First, some background information, during the past decade, there has been an increase in national attention on long-term follow-up of children diagnosed during the newborn screening [inaudible 00:34:27]. In 2013, [inaudible 00:34:31] released a grant [inaudible 00:34:32] proposals to integrate newborn screening and long-term follow-up to these primary care practices.

When [Lisa 00:34:39] and I reviewed the RFP, we didn't think we can apply because it didn't really match with the California long-term follow-up model. In California, we contract with specialty care centers to collect yearly long-term follow-up data for our patients. We started collecting data on MS/MS disorders in July of 2007, and then, in November 2011, we started collecting long-term follow-up data for endocrine and hemoglobin disorders. Collecting data from primary care practices really does not match with what we were doing.

Then, later that year, we heard from the chief of our program that there may be a possibility of cutting long-term follow-up funding in our program. We figured, oh, gosh, if they cut, we will start from PH, so we figured it does not hurt us to look for alternative routes to collect long-term follow-up data.

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We applied for the grant, and then we decided to focus on the primary congenital hypothyroidism, known as the PCH, because it's the most

common disorder with screening in our program. We used the primary TSH screen method. Our birth incidence is about 1 in 2,000 live births. Most children with PCH need long-term treatment. Delayed or inappropriate treatment may cause severe neurocognitive disability.

Compared to many other complicated disorders we screen such as metabolic disorders, PCH is easy to manage, but little is known about the role of PCH or PCPs in PCH management.

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Before we applied and received the grant, we requested our sister state, Hawaii, to participate in the study with us. We targeted primary care doctors in both states and patients with PCH.

Nest please.

The study objectives were to assist the willingness and the capability of PCPs to provide long-term care for patients with PCH and their need for a PCH-related continuing medical education, to use the the current case management patterns in clinical outcomes to assess the PCPs willingness to obtain informed consent and provide data to the PCH project long-term follow-up database, to investigate the practicality of providing real-time long-term follow-up data that PCPs and to identify barriers incurred and to improve the PCPs' knowledge on PCH and increase their capability of providing long-term follow-up for patients with PCH.

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The study method included 4 key components. The first one is a provider-based professional survey that addresses objectives 1, 2, 3. This survey has been completed by June of last year. We did a survey among 823 PCPs who are listed as contact doctor for who have at least 1 patient with PCH during 2009 and in 2014, by the time we conducted the survey. This presentation is really about the findings on this survey.

The second component is a 3-year long-term follow-up data collection which is just this objective 4. This part of the study is during the progress. We just completed the first year of data collection. My study manager is checking the completeness of each questionnaire we receive.

The third component is a post-follow-up survey which also addresses objective number 4 to be implemented after the long-term follow-up

data collection is over. The last component is continuing medical education about PCH.

In a the later slides, you will see that we identified a knowledge gap on PCH management among PCPs. Based on those finding, we created a continuing medical education course targeting PCPs in both states. We just conducted a CME in Hawaii on February 19th. Our next CME event will be in the UCSF Benioff Children's Hospital Oakland on March 17th.

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This flowchart shows the response rate. Because there is no significant difference between the 2 states on any of the questions we assessed, so I'm reporting the data collated together for both states. The total response rate was 31.1%, which is pretty good considering the type of survey we conducted in the lens of surveys. The doctors had to go back to their medical charts to get data to do this survey. This is incomparable to other provider surveys conducted in the states.

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This table shows the characteristics of the responding PCPs, about half were male. The majority of doctors were either white or Asian. When we asked what percentage of their patients were of Hispanic origin, about half said it's 0 to 29% of their patients were Hispanic and about a third had patient population of over 50% of their patients were Hispanic origin, which means that they're probably a third of the population.

The majority of doctors were either private practice or group practice. In terms of their medical specialty, as we expected, that was our target as well, the majority has been more generalist, general pediatrics and about 80% were in family practice. The median years in medical practice was 18 years with a range of 2 to 43 years.

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This next slide is about the case management patterns. About 78% of PCPs reportedly caring for one or more patients with PCH. When we asked who usually manages the patient's PCH condition, 18% said mainly the PCPs, but the endocrinologists are involved. [63% 00:42:05] said the patients were mainly managed by endocrinologists, but the PCPs were involved as well, and then 19% were solely by endocrinologists. Note that no patient was solely managed by a PCP.

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Over 90% of the PCPs reported a willingness to provide long-term care to new patients with PCH, which we were very happy to see. In terms of perceived barriers for providing long-term care for patients with PCH, the number one barrier is their need for guidance or support from endocrinologists and number 2 is not familiar with PCH treatment guidelines.

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About 94% of PCPs reported knowing an endocrinologist with whom they could consult for PCH. When we asked whether they encountered any difficulties with coordinating care with specialists, to our delight, 2/3, they didn't encounter any difficulty and about 30% said some endocrinologists were hard to reach, difficult to make a referral. Then there were a couple of doctors who were located in the rural area that could not find an endocrinologist to work with in their area.

Next please.

In terms of knowledge on PCH management, when we asked how familiar they were with the PCH recommended treatment guidelines, only 4% said they were very familiar. 2/3 said they were somewhat familiar, and about 28% said they were not familiar at all. Then, when we asked if such a question [inaudible 00:43:55] recommended [inaudible 00:43:57] follow-up, less than 50% of the respondents practically answered all 3 questions.

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In addition, when we asked how familiar they were with the [inaudible 00:44:11] diagnostic indications, only 2.5% said they were very familiar, and 28% somewhat familiar, and over 50% were not familiar at all. Consistently, only 23% know at what age to consider trying the patient off the [inaudible 00:44:33] treatment [inaudible 00:44:34] PCH. There an obvious knowledge gap there.

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When they assessed the continuing medical education needs, about 84% of PCPs reported that they are likely or very likely to participate in CME on PCH long-term follow-up if it's available. In terms of the preferred formats for CMEs, 61% preferred webinars, 34% preferred [inaudible

00:45:05], and about 30% preferred in-person classes. Based on these finding, we are using [inaudible 00:45:17] in-person classes as well as webinars for disseminating our CME materials.

Next please.

When assessing their capability of obtaining informed consent, within California, we don't really write informed consents when we [inaudible 00:45:38] follow-up data [inaudible 00:45:41], but, nationally, like new staff, like the [CRNs 00:45:44] they request to get informed consent from the patients, so [inaudible 00:45:52] assess whether these doctors are willing to obtain informed consent and whether it's difficult to do.

Luckily, 76% of PCPs reported willingness to obtain informed consent and only 5% said they're not willing to do so. When we asked of their difficulty in obtaining informed consent from patients, only 4% said very difficult, a third said difficult, but doable, over 40% they're not difficult at all, and 21% did not know what it is.

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When we asked whether they were willing to provide long-term follow-up data, over 2/3 said that, yes, they were willing to provide long-term follow-up data, and 18.7% said maybe. We're really glad to see those results. In terms of the format of data collection, a third preferred Web-based forms, a third preferred paper forms, and 33% had no preference.

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In terms of compensation required to provide long-term follow-up data, about a quarter of doctors were really generous [inaudible 00:47:15]. Put together, nearly 90% of doctors were happy with [inaudible 00:47:23] $200 per patient per year or less to provide this long-term follow-up data. This is much lower than what it entailed for specialty care centers to collect data.

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When asked for reasons for not willing to provide long-term follow-up data, among the 27 participants indicated they're now willing to provide data, a third said they don't have enough time, and a third said they don't provide long-term care for patients with PCH, and nobody said it's not important to collect long-term follow-up data for PCH.

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In conclusion, the majority of PCPs are willing to provide long-term care for patients with PCH, and there's a lack of knowledge about up-to-date PCH-related treatment and management standards. Support from endocrinologist is needed for successful management of cases, and most PCPs are willing to obtain informed consent from patients and provide long-term follow-up data with a reasonable compensation.

Next please.

I want to say thanks to the following colleagues for their contribution to this study. I also want to thank the short-term follow-up committee for giving me this opportunity to present our findings. I'm happy to answer any questions. Especially, I want to ask Ashlyn to give my regards to Phillis because I'm citing her [inaudible 00:48:53] as well here.

Ashlyn Booker: I sure will.

Ning Rosenthal: Thank you.

Carol Johnson: Sorry you're not feeling well, but that was a great presentation. Again, like Caroline's presentation, I really enjoyed hearing this again.

We wanted to make sure that those of you who are unable to attend the newborn screening meeting in Anaheim this past October had the chance to hear these presentations because we felt they were very important. Go ahead and ask your questions.

To unmute, it's "star," "7," or, again, online, you can type your question in from there.

Careema, I'll turn the question-and-answer session over to you, all right?

Careema Yusuf: Yes. Thanks, Carol.

Carol Johnson: Thank you.

Careema Yusuf: While we're waiting for folks to unmute and type, I do have a question for Ashlyn. We'd like to know how is CCHD data being reported to the Mississippi Department of Health?

Ashlyn Booker: I'm sorry. What kind of data?

Careema Yusuf: The CCHD data that you've just been mandated to screen for.

Ashlyn Booker: Okay. Great. We've got our new blood spot card and we had to do some slight modifications to the actual filter paper to collect the new information. As of right now, our lab is receiving the information as it's being reported on the card, and they are able to transmit that data to us. As of right now, we have hospital discharge data that we're able to utilize and bump the data up against it just to ensure that we don't have any missing babies, and then we're also in the process of trying to develop a delayed screening report form, which is to be completed for those babies who did not get the screening done prior to that newborn screening result coming out to the lab.

We have encouraged the hospitals not to delay sending in the actual specimens waiting on the point of service test. We're going to shoot that form out, the delayed reporting form, to them as well and we'll have to manually update our database with the information provided on that form.

As of right now, we are collecting the data. We have a genetics advisory committee meeting coming up this spring where we've got a new chairperson that was newly elected. We have some more discussions that need to be had in terms of what we'll do with the data and the necessary follow-up that will be needed and all of that good stuff. We have that info in it. We've got the forms. We have an effective way of retrieving the data. We just have to kind of finalize what exactly we're going to do with the data once we do have it.

Careema Yusuf: Great. Thank you. I do have another question. This time, it's for Caroline in California.

For the mild hypothyroidism cases, does your newborn screening program continue to follow those cases in short-term follow-up or do these cases get closed out?

Caroline Nucup-Villaruz: Is that question for us here in Washington State? I'm sorry.

Careema Yusuf: Sorry. It is in Washington, yes.

Caroline Nucup-Villaruz: Oh, okay. The question is "do we follow up on those confirmed cases" or the false positive cases? Could you clarify?

Careema Yusuf: Sure. It says, "For the mild hypothyroidism cases."

Carol, would you like to elaborate on your question?

Carol Johnson: Sure. Yes, that was my question.

In your presentation, you talked about how you did surveillance on those cases and that, sometimes, that process seemed like that could go on through maybe a somewhat significant period of time. Do you keep that case open in your short-term follow-up processes or does that get turned over to what I would consider to be a long-term follow-up staff?

Caroline Nucup-Villaruz: Okay. Thanks. Thanks for clarifying. That's a great question. That's the space within the short-term follow-up. As soon as the final diagnosis is made, whether the case is confirmed or ruled out, then that's the time we close the case. As you can see on that slide, it's our 2012 data, for some cases, it may take a while. It may take even more than a year before I close the case, until I hear back either from the endocrinologist or the primary care provider, if they have either started treatment or they would just reassure the family that the patient does not have a congenital hypothyroidism based on the trend of the TSH if it continues to go down.

Carol Johnson: Okay, that does answer my question. Thank you very much.

Caroline Nucup-Villaruz: Thank you.

Careema Yusuf: I do have a question for Cindy.

Cindy Ingham: Yes.

Careema Yusuf: Does Vermont have age-related cutoff for TSH?

Cindy Ingham: In the slide that was presented, you can see that, through our laboratory testing, it's over 96 hours under 15, but we're using 6.5 at the moment for older babies. Does that answer the question?

Careema Yusuf: I think so, and there is a follow-up question. It's asking, "What are the cutoffs for the drive for the filter paper on the initial screen and then on the routine repeat screen?"

Cindy Ingham: Oh, okay. If you look at that slide, that information is right there, for babies who are less than 24 hours, between 24 and 96 hours, and then over 96 hours. Something that can be a confounder in this is that we need to keep in mind that filter papers are for the purpose of screening and not diagnosis. I think, sometimes, you'll see filter papers submitted

on much older kids just because it's sometimes easier than drawing a serum level. In interpreting those results, you have to take that into account and see how old that child is.

Sometimes, I'll get them in on an 8-year old, monitoring this baby, but it's easier to poke his finger. There's some education necessary there, too.

Careema Yusuf: Thank you.

Cindy Ingham: Yep.

Careema Yusuf: Another question for Dr. Rosenthal.

Could you please comment on collecting long-term data versus treatment and management? How do the pediatric endocrinologists view this process?

Ning Rosenthal: How do the pediatric endocrinologists view it or how do the primary care doctors view it?

Careema Yusuf: It's specifically the endocrinologists, the pediatric endocrinologists.

Debby: Yeah. This is Debby [inaudible 00:56:50].

Careema Yusuf: Oh, there you go. Hi.

Debby: Yeah. That was my question. When you were on your slides, it varied between collecting the long-term data and managing this patient. Your slide said that the majority of the pediatric endocrinologists preferred, I mean, the pediatric primary care docs, preferred to have the pediatric endocrinologist involved in the care of the child. It seemed to be flipping back and forth between collecting data and treating, and I was wondering how endocrinologists felt about the primary care docs, not just collecting the data, but, if you're going to management, how they felt about that particular aspect of your project.

Ning Rosenthal: Great. Thank you for the question. We have an advisory committee for our project which is made of 4 renowned pediatric endocrinologists, 2 Californians, 1 from Stanford, 1 from [inaudible 00:57:52] Children's Hospital of Los Angeles and [inaudible 00:58:01]. During the whole process, like this project, it was actually motivated by these endocrinologists because they felt like, in their daily work, it creates a lot of burden to follow these patients. They are happy to serve as

consultants, but they really think that the main follow-up responsibilities can be shifted to primary care doctors.

The study in a way identified there's a knowledge gap. These doctors are willing to do the job, but they need some help. What we are strongly recommending in our state and also in Hawaii as well, we strongly recommend a collaborative model, like the PCP to work closely with an endocrinologist to manage these patients in long-term.

Currently, some of the PCPs, they just like, after they referred patients to the endocrinologists, they just let them go. They don't get involved anymore. That was the pattern that we are starting to recommend in our state.

Debby: As a follow-up, if there is some mechanism in place to compensate the endocrinologist for serving as the consultants to the PCP?

Ning Rosenthal: Not really. The endocrinologists in our committee, they just provide the regular consultations for the PCPs in their area for free, pretty much.

Careema Yusuf: Great. Thank you so much, everybody. Thank you to the speakers for some really great presentations. Unfortunately, we're going to have to stop the webinar as we have gone over time. If you do have any additional questions, please feel free to contact [Thalia 01:00:19]. Her contact information is on the slide that is up now.

Thank you very much, and have a great rest of the afternoon.

Carol Johnson: Thank you, everyone.

Ashlyn Booker: Thank you.

Caroline Nucup-Villaruz: Thank you.

Cindy Ingham: Thank you.