

Webinar Zoom Chat

14:03:42 From Amy Gaviglio to Everyone:

Welcome everyone!! Please put any questions or comments in the chat box throughout the presentations. We will save all questions/comments for Q&A at the end!

14:04:55 From Cindy Ingham to Everyone:

So nice to see you all! Greetings from snowy Vermont.

14:07:08 From APHL to Everyone:

Greetings!

14:07:33 From Lani Culley to Everyone:

Hello from Washington!

14:09:56 From Cindy Ingham to Everyone:

I'd love to have a copy of the decision tree model.

14:11:57 From Amy Gaviglio to Everyone:

Will ask Michael if they are able to share, Cindy!

14:12:23 From John Thompson to Everyone:

@ Cindy - please provide your email in the chat and we can send it to you (hi there!).

14:12:28 From Lani Culley to Everyone:

Hi Cindy and Amy, I'm sure Michael will be happy to share :)

14:13:24 From Cindy Ingham to Everyone:

Thanks John, and hi to you too! My email address is director@negg.org

14:14:22 From Jeremy Penn to Everyone:

It is probably a mistake to only count value of life saved in terms of mortality. Reducing disability and morbidity also has value (and can be computed in \$\$) and should be incorporated as part of a cost-benefit model.

14:15:20 From John Thompson to Everyone:

@ Jeremy - survival with permanent disability was indeed included in our analysis.

14:15:49 From Jeremy Penn to Everyone:

Computed in terms of DALYs produced by screening?

14:16:06 From Jeremy Penn to Everyone:

Sorry, QALYs, not DALYs

14:21:08 From Kristy Karasinski to Everyone:

Is WA planning to just use low cit as the marker?

14:26:13 From John Thompson to Everyone:

@ Jeremy - not QALYs. Our model was simple in that we applied a value of annual cost of severe disabilities (expert opinion from Scott Grosse, CDC) for those babies expected to have long-term disability. We're happy to share with you and talk offline if you want.

14:27:24 From John Thompson to Everyone:

@ Kristy - low citrulline will be the primary marker. We will be working through the algorithm and learning from the programs already doing this, including the great information being shared by Patrice right now! Stay tuned... We expect to go live in the Summer/Fall of 2023.

14:27:54 From Carol Johnson to Everyone:

Which programs are already screening for OTC?

14:28:31 From Susan Berry to Everyone:

quite a few list themselves as doing this on NewSteps

14:28:53 From Carol Johnson to Everyone:

OK - thank you. I haven't looked for this information for this disorder.

14:29:08 From Amy Gaviglio to Everyone:

<https://www.newsteps.org/resources/data-visualizations/newborn-screening-status-all-disorders>

14:29:57 From Amy Gaviglio to Everyone:

I think it depends a bit on how you define "screening for" - actively setting cut-offs versus may be picking up a low citrulline as you look for elevated citrulline as well

14:30:08 From APHL to Everyone:

@Carol- there are 8 states screening for OTC

14:32:00 From Susan Berry to Everyone:

depends on how you count 😊

14:33:53 From John Thompson to Everyone:

@ Carol - we reached out to California, the New England program and Puerto Rico. They were all helpful in sharing information with us as we prepared for the policy process.

14:36:55 From Guisou Zarbalian to Everyone:

<https://www.newsteps.org/data-resources/reports/screened-conditions-report> for OTC, see the "other" tab within the screened conditions report

14:37:21 From Guisou Zarbalian to Everyone:

it is listed under the amino acid disorders

14:38:49 From Amy Brower to Everyone:

<https://nbstrn.org/tools/nbs-cr/ornithine-transcarbamylase-otc-deficiency>

14:47:53 From Amy Gaviglio to Everyone:

Hi all! Just a reminder to put any comments/questions here in the chat box, and we will get to them shortly after Dr. Berry's talk!

14:48:14 From Amy Brower to Everyone:

<https://nbstrn.org/tools/nbs-vr>

14:51:13 From Jeremy Penn to Everyone:

In GA, (assuming the rate of OTCD is 1 in 158,000), the chance of having 0 babies with OTCD out of 138,000 screens is 42%.

14:52:16 From Patricia Hall to Everyone:

most published incidences of OTC are considerably higher (~1:20,000 seems common), but there is a wide range.

14:53:02 From Amy Gaviglio to Everyone:

Agreed that the published birth prevalences are all over the place 1 in 158,000 actually is quite low compared to the published literature interesting enough.

14:54:42 From Amy Brower to Everyone:

<https://nbstrn.org/tools/nbs-vr?tab=estimate-of-cases-per-year>

14:58:05 From Cindy Ingham to Everyone:

I'm wondering if the concept of Whole Genome Sequencing of newborns (rather than DBS identification of disorders) came up in any of the discussions the UCD Consortium has had?

15:03:21 From Mei Baker to Everyone:

Dr. Berry, could you verify that the same genotype in a family can have different phenotypes in OCT?

15:05:22 From Amy Gaviglio to Everyone:

Sue - can you remind me - I believe this case also, sadly, had a family hx of childhood deaths as well, correct?

15:05:26 From Cindy Ingham to Everyone:

That answers my question about sequencing, Thanks, Sue.

15:08:48 From Melissa Parisi to Everyone:

There needs to be a high index of suspicion in these neonates with unclear, non-specific symptoms--poor feeding is common and doing NH3 testing in all of these babies would be difficult.

15:10:14 From Mei Baker to Everyone:

Dr. Berry, well said about TIMING, bot testing and treatment!

15:12:53 From Melissa Parisi to Everyone:

If this condition is as common as stated, why are we not finding these babies--are they all dying in the neonatal period without detailed workup??

15:13:33 From Amy Gaviglio to Everyone:

I have the same question Melissa - will ask!

15:13:33 From Susan Berry to Everyone:

condition is overall likely more frequent if you include all forms, not just neonatal presentation

15:13:59 From Susan Berry to Everyone:

some die undiagnosed at all ages!

15:20:05 From Jeremy Penn to Everyone:

Really hard to know the prevalence without screening :)

15:21:07 From Amy Gaviglio to Everyone:

True point Jeremy!

15:24:59 From Cindy Ingham to Everyone:

Amy, could you share the contact info for the GC at Valley Children's Hospital? I'd love to know more about this targeted AP screening panel.

15:27:49 From Sarah Bradley to Everyone:

Thank you to APHL, the speakers, and the moderators - this webinar has been very helpful.

15:27:52 From Amy Brower to Everyone:

Very helpful session!

15:28:13 From Cindy Ingham to Everyone:

This was fabulous. Thanks so much.

15:28:45 From Lani Culley to Everyone:



Wonderful presentation everyone! So grateful for all of you :)