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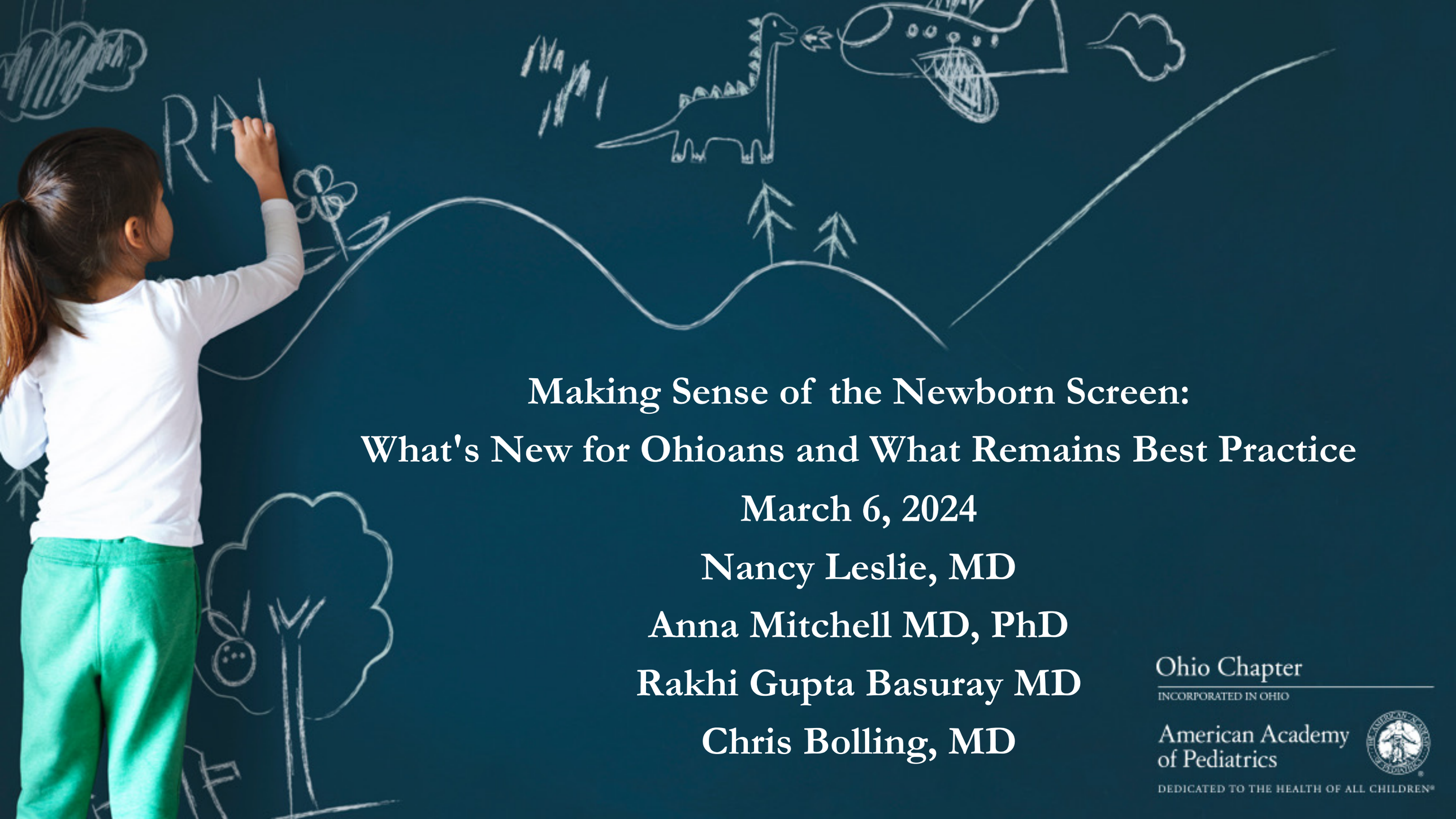

Making Sense of the Newborn Screen:
What's New for Ohioans and What Remains Best Practice

March 6, 2024

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CME Disclosure

Nancy Leslie, MD, Anna Mitchell MD, PhD, Rakhi Gupta Basuray, MD, Chris Bolling, MD,
faculty of this educational activity, have no relevant financial relationship(s) with ineligible companies to disclose.

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DEI Disclosure

I have considered the Ohio AAP Diversity, Equity, and Inclusion standards in preparing this presentation. I invite your feedback in promoting equity within this learning space. If you have suggestions, please email me at bolling.cf@gmail.com

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Objectives

- By the conclusion of this activity, participants will be able to:

1) Refer patients with an abnormal newborn blood spot screen to the appropriate follow-up clinic
2) Communicate effectively with the Ohio Department of Health regarding abnormal test results
3) Order appropriate testing for congenital CMV infection following an abnormal newborn hearing screen
4) Respond effectively to abnormal thyroid screening.
5) Manage and follow a patient with an abnormal newborn screening for Duchenne muscular dystrophy
6) Recall information about the three Newborn Screens
7) Describe how the Ohio Revised Code and the Ohio Administrative Codes address newborn screening

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Critical Congenital Heart Disease (CCHD)

- Screens for heart conditions that require intervention in the first year of life
- Implemented in Ohio in 2014
  - 99.9% pass rate
  - Sensitivity 50% - 76%
- Performed at >/= 24HOL (or shortly before discharge if less than 24 hours of age)
- Pass (both):
  - SpO2 >/= 95%
  - </=3% difference between pre-post ductal sites
- If does not Pass
  - Retest in 1 hour
  - ECHO if fails
- Immediate fail (no re-test) if SpO2 < 90% (in either right hand or foot)

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Original Wilson-Jungner NBS criteria

- The condition sought should be an important health problem
- There should be an accepted treatment for patients with recognized disease
- Facilities for diagnosis and treatment should be available
- There should a recognizable latent or early symptomatic phase
- There should be a suitable test or examination
- The test should be acceptable to the population
- The natural history of the condition should be understood
- There should be an agreed upon policy on whom to treat
- There should be an acceptable cost/ benefit
- There should be ongoing learning regarding case finding

Advisory Committee on Heritable Disorders in Newborns

The Committee advises the Secretary, U.S. Department of Health and Human Services on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and standards.

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ACHDNC process

- Is the condition serious?
- Can a case be defined?
- Analytic validity
- Clinical utility
- Are treatments available?
- Is there prospective data to support the process through population screening?

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What is the RUSP?

- Recommended Uniform Screening Panel-ultimate goal is predictable and uniform set of conditions in all states
- Initial set of conditions was vetted by a stakeholder survey commissioned by the American College of Medical Genetics, highly focused on MS/MS testing – around 1990
- Significant criticism of the process (was this actually expert opinion, doesn't fit current model of evidence-based decision making)
- New process includes nomination, evidence review, pragmatic capabilities, and sign off by Sec HHS

What's Included in the RUSP?

- Tests done on a dried blood spot
- Point of care tests: newborn hearing screening, pulse ox testing for critical congenital heart disease

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Numbers, Numbers (Dried Blood Spot Screening Data)

- ODH screened 129,344 specimens in 2022
- 1,622 repeat screens
- 190 rejected “unsats”
- 3,318 flags (excluding Hb trait)
- 129 confirmed cases
- 362 “other” carriers and conditions not on the newborn screen
- 1,706 open cases
- * Limitation for data is as of November 2023 and is subject to change

So What Happens When an NBS (Dried Blood Spot) Gets Flagged?

- State calls and faxes to PCP of record
- Often another designated entity (e.g., genetic center) gets a copy on the fax machine
- There's a process for critical values
- Following a flagged newborn screening result a baby may require: repeat screen, labs, urgent visit, sometimes Emergency referral
- Family is counseled, diagnostic labs ordered, plan developed
- Close the loop back to the NBS lab by faxing diagnosis and test results to the NBS Lab at 614-644-4648.

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Molecular Diagnostics are a Big Part of the Initial Evaluation

- Currently the only testing where molecular testing is the PRIMARY test is SCID and SMA (both done by qPCR)
- Second tier testing with sequencing (or another biochemical marker) is increasingly common (currently not done in Ohio)
- Currently there are no sequencing first disorders, but there are research efforts
- A panel for 39 common CF pathogenic variants is performed if the primary marker is elevated for CF.

What Has Been Added Recently?

- Krabbe disease (OH and KY 2016) NOT on RUSP yet
- Mucopolysaccharidosis I (KY 2016, OH began screening in 2017)
- Pompe (KY 2016, OH began screening in 2017)
- Spinal Muscular Atrophy (OH was in validation phase since 2018, did not officially launch until 2022)
- X-linked adrenoleukodystrophy (OH began high volume validation 02/2022, launch 2022)

And Newer Yet- But Not Yet Implemented

- Duchenne Muscular Dystophy (Ohio only, non RUSP)
- Mucopolysaccharidosis II
- GAMT – creatine synthesis disorder

How does Ohio address Newborn Screening?

Ohio Revised Code Section 3701.501

(A)(1) Except as provided in division (A)(2) of this section, all newborn children shall be screened for the presence of the genetic, endocrine, and metabolic disorders specified in rules adopted pursuant to this section.

(2) Division (A)(1) of this section does not apply in any of the following circumstances:

(a) If the parents of the child object to the screening on the grounds that it conflicts with their religious tenets and practices;

(b) With respect to the screening for Krabbe disease described in division (C)(1)(b) of this section, if the parents of the child communicate their decision to forgo the screening;

(c) If appropriate laboratory equipment is not available.

How Does Ohio Address Newborn Screening?

Ohio Administrative Code Chapter 3701-55

Advisory Panel of 14 members selected by and reporting to the Director of ODH

(a) The disorder's incidence, mortality, and morbidity;

(b) Whether the disorder causes disability if diagnosis, treatment, and early intervention are delayed;

(c) The potential for successful treatment of the disorder;

(d) The expected benefits to children and society in relation to the risks and costs associated with screening for the disorder;

(e) Whether a screening for the disorder can be conducted without taking an additional blood sample or specimen;

(f) Whether the secretary of the United States department of health and human services has included the disorder in the federal recommended uniform screening panel.

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Case 1: Duchenne Muscular Dystrophy

Baby Li is a 30-week preterm newborn male delivered at Metro General in Cleveland. He has an inconclusive DMD screen reported from his newborn blood spot test.

- Inconclusive screen for muscular dystrophy, what's next?
- What kind of resource challenges do you anticipate in your community?
- What can AAP and ODH do for you?

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Case 2: CMV

Baby Carson is a female infant with a low birth weight born at 38 weeks in Zanesville. She failed her newborn hearing screening in the hospital twice.

- CMV saliva/urine testing be done in the first 21 days after birth
- CMV can also develop late-onset hearing loss, so stay vigilant!
- Diagnostic hearing evaluations for all who do not pass newborn hearing screening
- The first year of life critical for language and communication
- Diagnostic Hearing Evaluations by pediatric audiologist by three months of age

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Case 3: Thyroid

Baby Djokic is a 40-week LGA baby born in Toledo following an uneventful pregnancy and delivery. He was discharged at 26 hours of life and his newborn blood spot was drawn prior to discharge . His newborn screen showed a TSH of 37mU/L.

- What is your clinic process when this is received?
- What do resources look like for you?
- What are your biggest challenges with thyroid screening?

Case 4: Standard of Care/ Family Considerations

A new mother in your practice informs you that her family recently moved to Dayton from California. They have a newborn and the clinic there told her something like "the baby's screening is fine, but you should be concerned about ALD for future pregnancies when you get settled in Ohio". She did not remember the name adrenoleukodystrophy being used.

- Anything in particular we need to know about ALD?
- What is your role as a pediatric provider?
- What are the challenges with having information like this on people who are not your patients?

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Case 5: Resolving Cases with ODH

Landon Childers is a six-month old male infant in your practice in Ironton. He is in the care of his father with significant help from his maternal grandmother because his mother passed away in a car accident when he was two months old. He had a positive blood spot for Krabbe Disease, but you had not seen him since that report came back at one week.

- What is the best course of action now?
- What role can ODH and OH-AAP play to help you?
- What do you think are the best ways to help prevent incidents like this?

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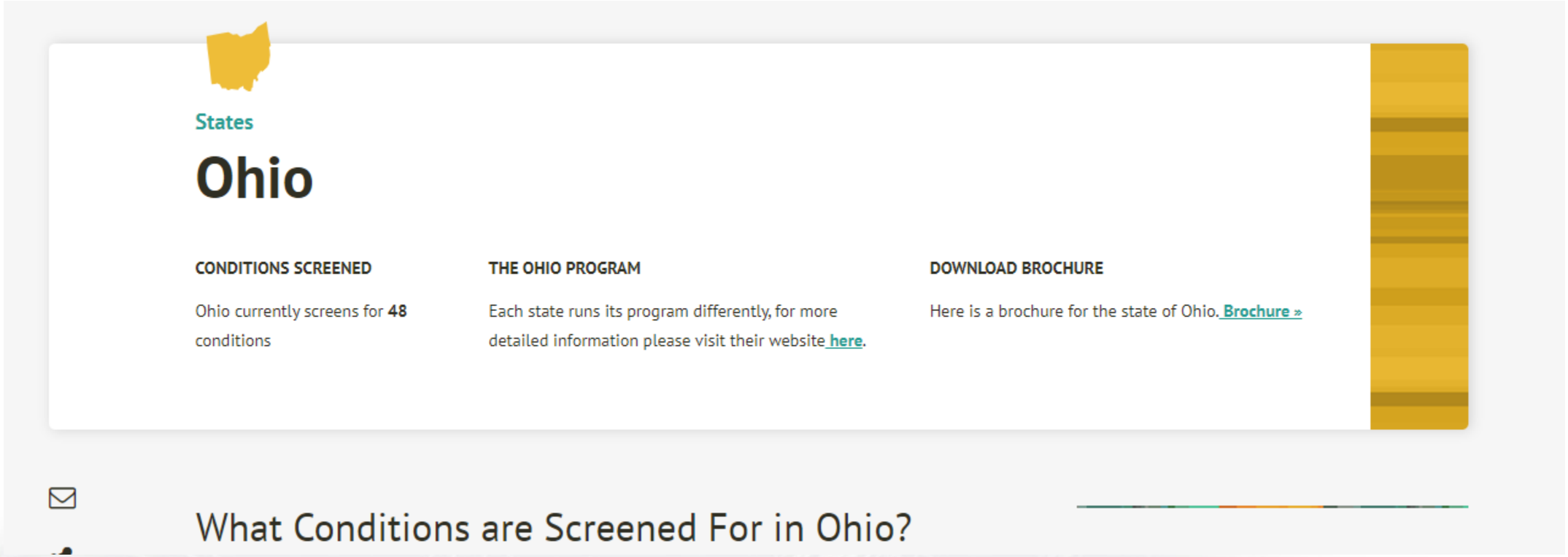
How Do I Find Out What is Screened for in my State?
Babysfirsttest.org



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How Can I Get a Copy of a Baby's Dried Blood Spot Results?

NEWBORN SCREENING NEWSLETTER DECEMBER 15, 2014

Introducing eReports

Attention Pediatric Practice Managers. Tired of waiting for Newborn Screening results? Sign up now for 24/7 web access to Newborn Screening Reports!

The Newborn Screening Laboratory is proud to announce a new service allowing designated persons at your practice to access and print newborn screening reports. To participate in eReports a legally responsible, authorized user will be required to sign and date the eReports Security Agreement consent form sent with this letter. Other information required includes the name of your practice, a valid e-mail address for the authorized user, a username, and strong password. Return the signed Security Agreement by fax to 614-644-4648. You will be notified by email when your access is granted. Please be sure to retain a copy of the Security Agreement for your records.

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Advocacy Opportunities

- Advisory councils in many states need a voice from primary care
- Informatics integration between EHR, NBS system, birth certificate system: is every baby screened, does every primary care provider have access to the results
- Process improvement in birth hospitals: NBS labs have timelines for TAT in critical conditions, but they need to get samples on time
- Long term follow up opportunities

ODH Lets Us Download a Supplemental Data Form

Ohio Department of Health - Newborn Screening

Results shown as follows: Result (Reference Range)

17OHP	15.6967 ng/mL S (< 35 ng/mL)	**ALA**	319.29 µmol/L (No Range)
AP-GALT	8.1226 U/gHb (> 2.0 U/gHb)	**ARG**	6.415 µmol/L (<110 µmol/L)
BIO	149.87 ERU (> 17 MRU)	**C0**	27.525 µmol/L (> 6 µmol/L)
C10	0.23 µmol/L (No Range)	**C10:1**	0.075 µmol/L (No Range)
C10:2	0.07 µmol/L (No Range)	**C12**	0.725 µmol/L (No Range)
C12:1	0.525 µmol/L (No Range)	**C14**	1.025 µmol/L (No Range)
C14:1	1.035 µmol/L (< 0.8 µmol/L)	**C14:2**	0.11 µmol/L (No Range)
C14-OH	0.06 µmol/L (No Range)	**C16**	5.84 µmol/L (< 8.5 µmol/L)
C16:1	0.585 µmol/L (No Range)	**C16:1-OH**	0.095 µmol/L (No Range)
C16-OH	0.065 µmol/L (<0.09 µmol/L)	**C18**	1.6 µmol/L (No Range)
C18:1	2.525 µmol/L (No Range)	**C18:1-OH**	0.045 µmol/L (No Range)
C18:2	0.24 µmol/L (No Range)	**C18-OH**	0.03 µmol/L (No Range)
C2	31.58 µmol/L (No Range)	**C3**	2.155 µmol/L (< 5.0 µmol/L)
C3/C2	0.0682624 (< 0.3 µmol/L)	**C3DC/C4OH**	0.19 µmol/L (No Range)
C4	0.195 µmol/L (No Range)	**C5**	0.12 µmol/L (< 1.0 µmol/L)
C5/C2	0.00379783 (No Range)	**C5:1**	0.01 µmol/L (No Range)
C5DC	0.22 µmol/L (< 0.8 µmol/L)	**C5-OH**	0.225 µmol/L (< 0.8 µmol/L)
C6	0.075 µmol/L (No Range)	**C6DC**	0.185 µmol/L (No Range)
C8	0.13 µmol/L (< 0.6 µmol/L)	**C8/C10**	0.565714 (No Range)
C8:1	0.095 µmol/L (No Range)	**CIT**	14.815 µmol/L (< 75 umol/L)
GAA	6.08 (> 1.10 µmol/Lhr)	**GALC**	6.53 (> 0.65 µmol/L/hr)
GLY	460.025 µmol/L (No Range)	**Hemoglobin (HPLC)**	1 (FA)
IDUA	8.43 (> 0.65 µmol/Lhr)	**IRT**	38.906 ng/mL (< 96 Percentile)
LEU	131.82 µmol/L (< 400 µmol/L)	**LEU/VAL**	1.52223 (No Range)
LSD Disorder	()	**MET**	39 µmol/L (< 100 µmol/L)
ORN	67.8 µmol/L (No Range)	**PHE**	90.795 µmol/L (< 160 µmol/L)
PHE/TYR	0.824751 (No Range)	**PRO**	228.59 µmol/L (No Range)

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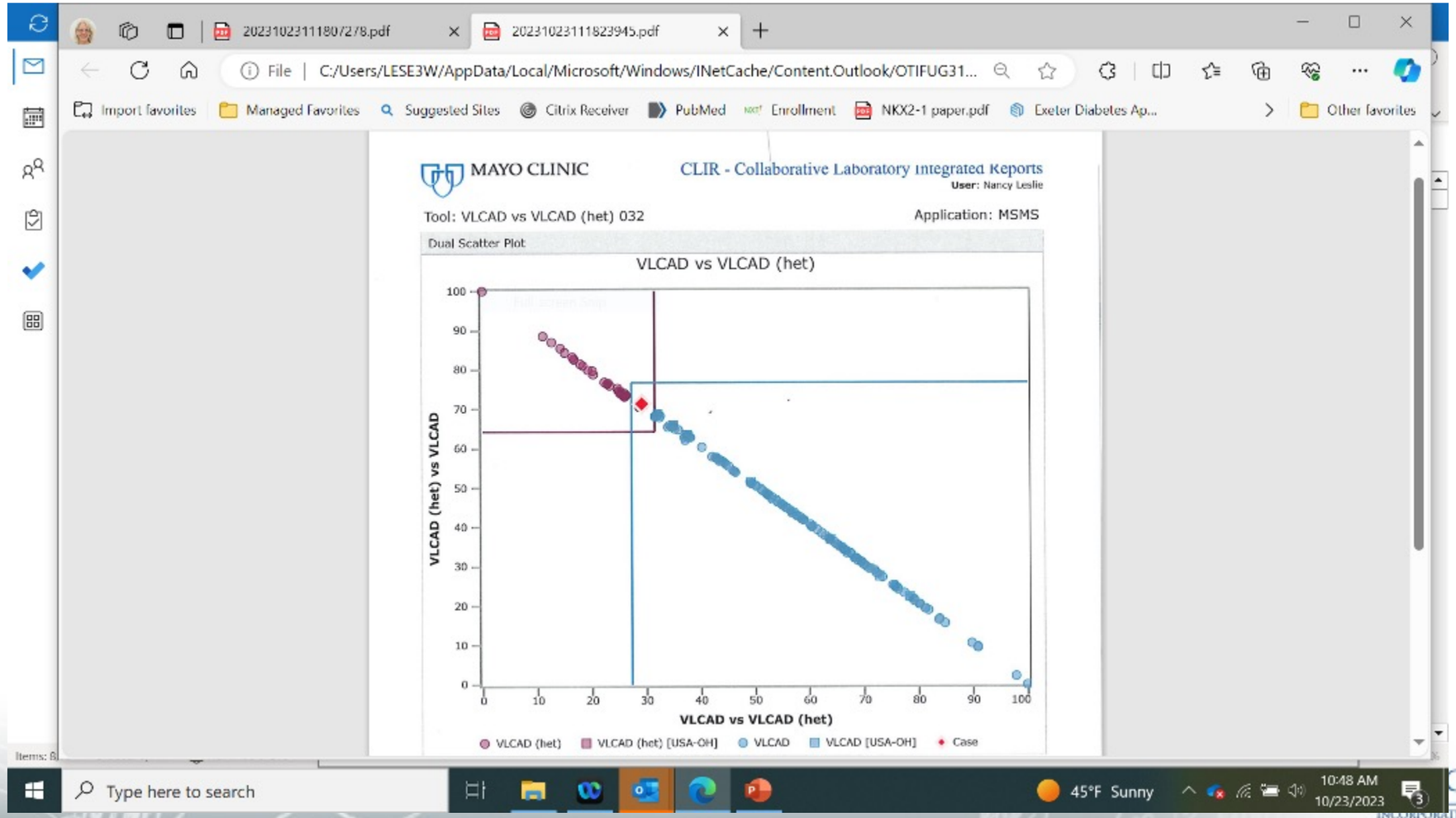

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Best Overall Web Site

- www.babysfirsttest.org

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Coming Soon!

Our PHP education program continues with informative topics!

- **Updates to Diabetes 1 and 2 (Thurs, March 28 @ 1pm)**
- **Anxiety in the "Tween" Aged Child (Wed, May 15 @ Noon)**

Register Here:

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2024 Spring Meeting!

- Friday, April 19, 2024, at the Dublin Integrated Education Center

  (12:00 – 3:30)
- Topics Include:
  - Solutions for ADHD and Behavior Problems in the Young Child (4-6)
  - Helping You Make the Case: The Pediatricians Role in Lead Prevention, Working with Your System and Resources for Families

Register Here!

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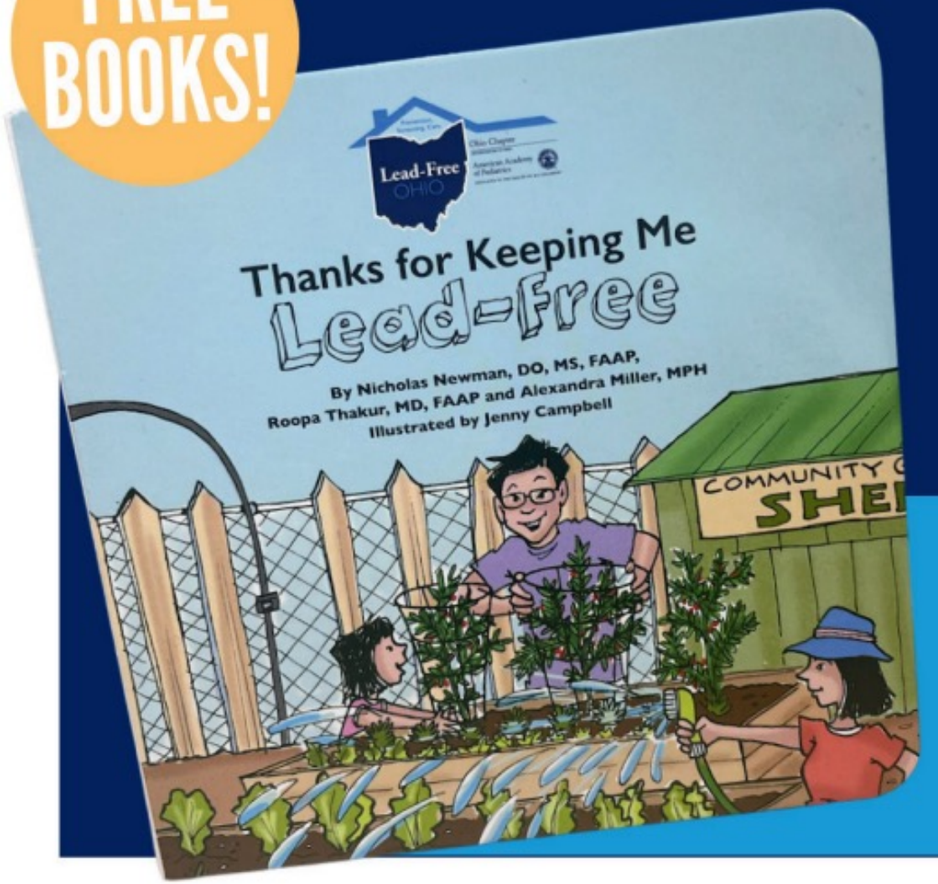
Lead Poisoning
PREVENTION RESOURCE

Easily accessible resource for parents & kids

FREE BOOKS!



ORDER FREE BOOKS FOR YOUR PATIENTS TODAY!

Receive up to 120 books!
**Books come in quantities of 60*

"This board book is a great conversation starter with families on the topic of lead prevention. Any time I can hand out a book at a visit, I am happy!"
-Matthew Tien, MD, FAAP, Beachwood Health Center

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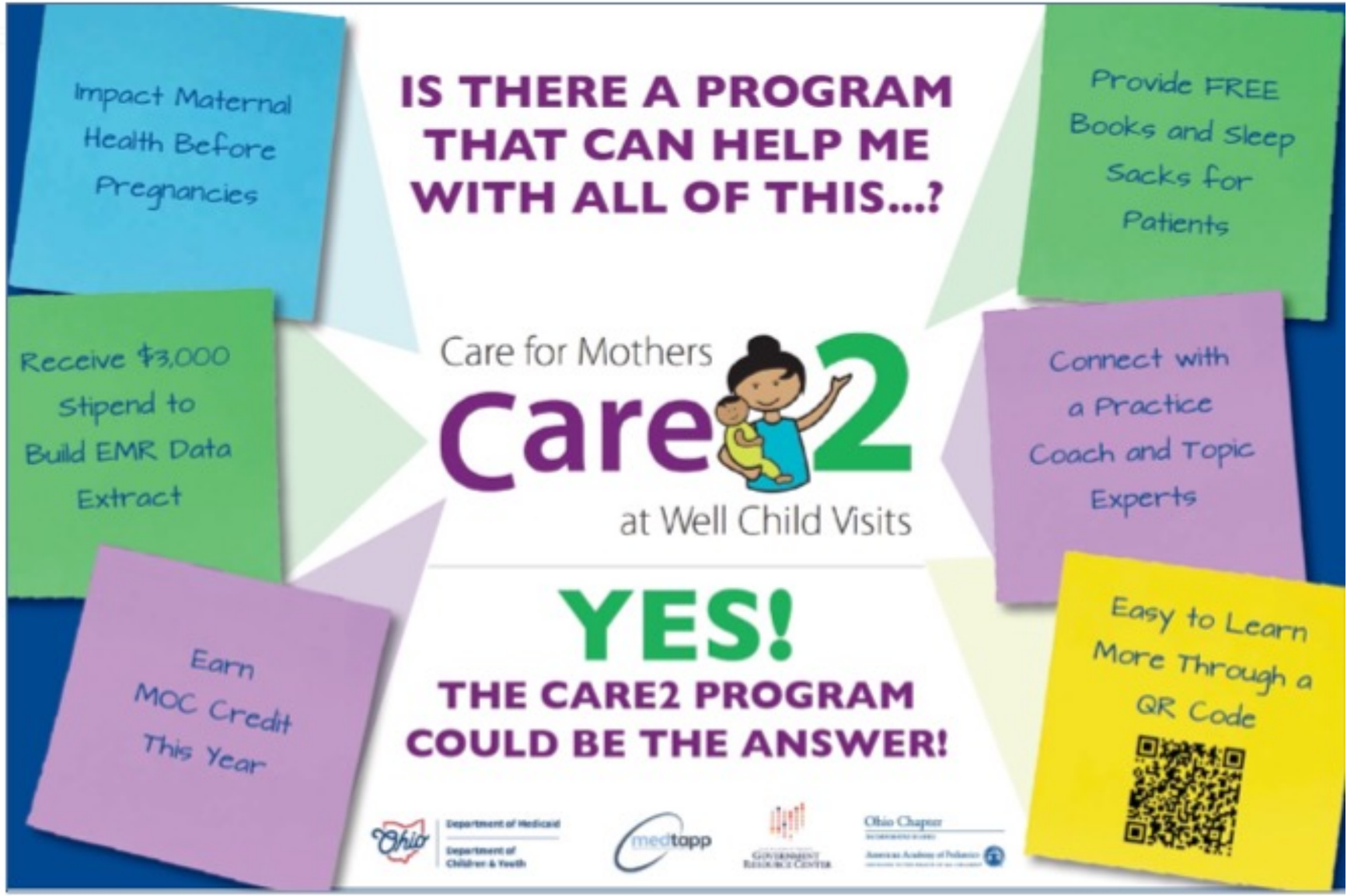
Care2 Quality Improvement Program – Wave 2 Enrolling Now!

- June 2024 – May 2025
- Designed for settings that include primary care for **BOTH** children and adult caregivers
- MOC credit for ABFM and ABP
- **$3,000 data stipend**
- **FREE** books and sleep sacks
- Topics in the project include:
  - Tobacco/vaping cessation
  - Family planning
  - Safe sleep environments
  - Maternal depression
- **FOR MORE INFORMATION:**
  - **Email:** Hayley Southworth - hsouthworth@ohioaap.org
  - **Click Here**
  - **QR Code on this slide**



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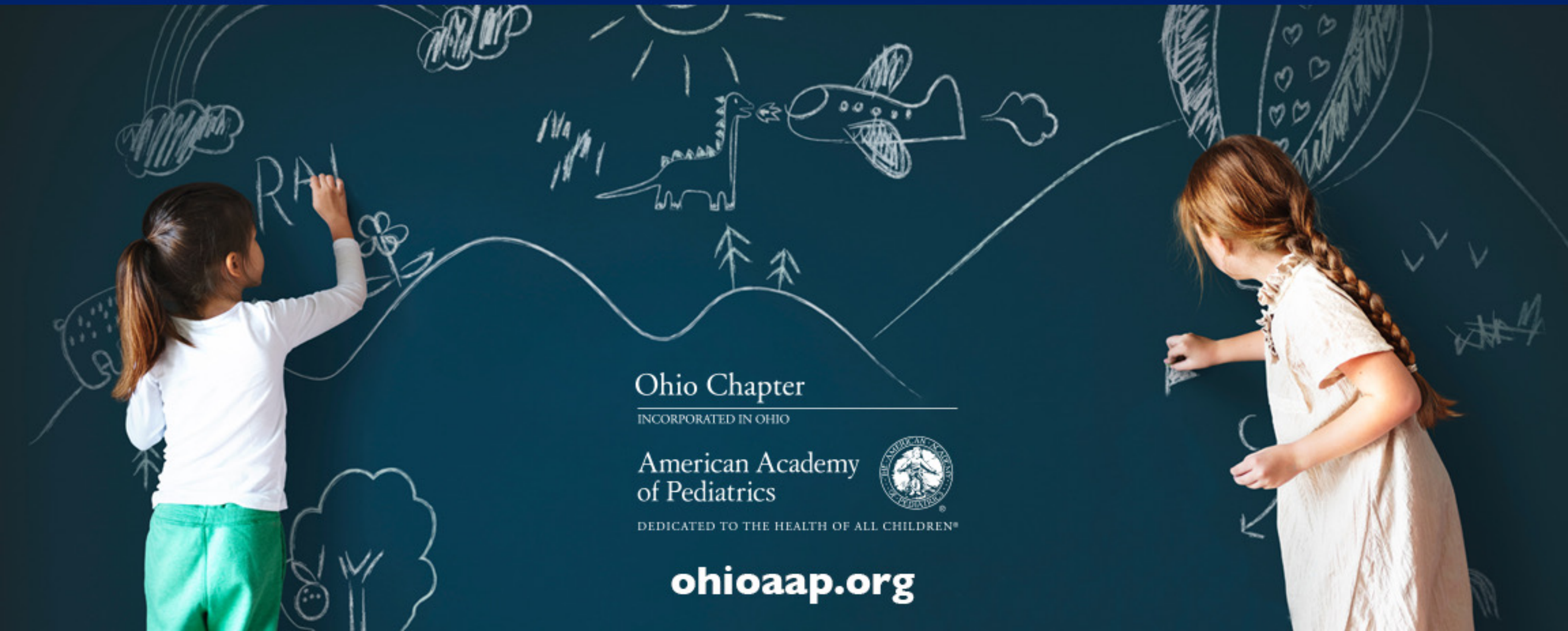
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