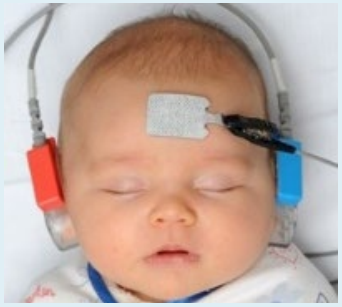
OVERVIEW OF MICHIGAN'S NEWBORN SCREENING PROGRAM

Angela Aldrich MSN, RN
Nurse Consultant, Newborn Screening Program

PURPOSE OF NEWBORN SCREENING (NBS)

- Early diagnosis
- Early treatment
- Reduce morbidity and mortality

THREE COMPONENTS OF NBS



Blood spot collection 24-36 hours

- Collected on filter paper

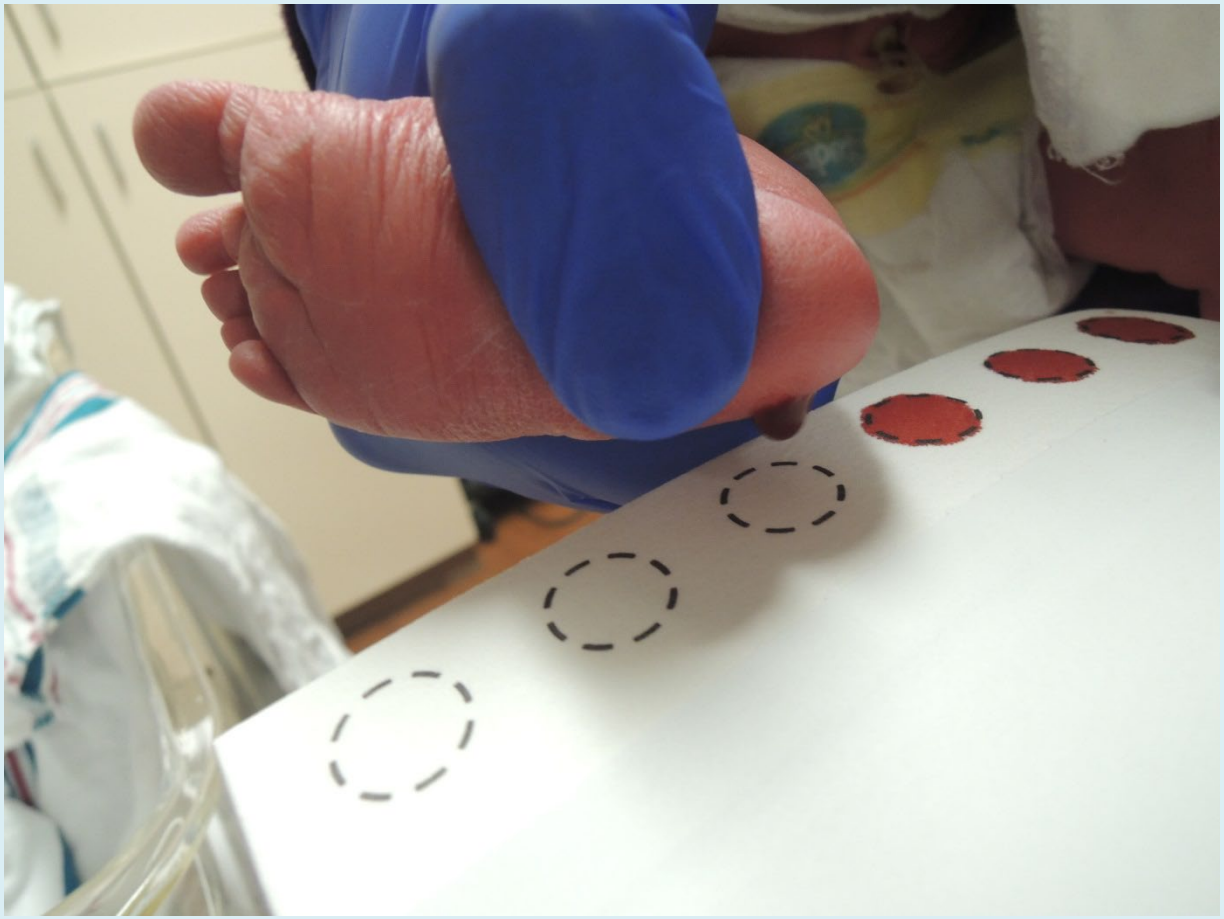
Hearing screening

- Prior to 1 month (usually included in 24-hour bundle)
- Point of care test

Critical congenital heart disease screening (CCHD)

- Pulse oximetry screening at approximately 24 hours
- Point of care test

Bloodspot Screening and Follow-up System



NBS in Michigan

- 1965: MI NBS Program began with phenylketonuria
- 2022: MI NBS panel includes over 50 disorders, plus hearing and critical congenital heart disease screening
  - Amino Acid Disorders
  - Organic Acid Disorders
  - Fatty Acid Oxidation Disorders
  - Endocrine Disorders
  - Hemoglobinopathies
  - Cystic Fibrosis
  - Primary Immune Deficiency Disorders
  - Lysosomal Storage Disorders
  - X-linked Adrenoleukodystrophy (X-ALD)
  - Spinal Muscular Atrophy (SMA)
  - Guanidinoacetate Methyltransferase (GAMT) deficiency

NBS Disorders

Michigan.gov/Newbornscreening

Disorders List

The Newborn Screening Laboratory screens all Michigan Infants for more than fifty disorders.

Amino Acid Disorders

1. Argininemia (ARG)
2. Argininosuccinic acidemia (ASA)
3. Citrullinemia Type I (CIT-I)
4. Citrullinemia Type II (CIT-II)
5. Homocystinuria (HCY)
6. Hypermethioninemia (MET)
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign hyperphenylalaninemia defect (H-PHE)
10. Biopterin cofactor biosysnthesis defect (BIOPT-BS)
11. Biopterin cofactor regeneration defect (BIOPT-REG)
12. Tyrosinemia Type I (TYR-1)
13. Tyrosinemia Type II (TYR-II)
14. Tyrosinemia Type III (TYR-III)

Fatty Acid Oxidation Disorders

15. Carnitine acylcarnitine translocase deficiency (CACT)
16. Carnitine palmitoyltransferase I deficiency (CPT-1A)
17. Carnitine palmitoyltransferase II deficiency (CPT-II)
18. Carnitine uptake defect (CUD)
19. Dienoyl-CoA reductase deficiency (DERED)
20. Glutaric acidemia type II (GA-2)
21. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
22. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
23. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
24. Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
25. Trifunctional protein deficiency (TFP)
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders

27. 2-Methyl-3-hydroxy butyric aciduria (2M3HBA)
28. 2-Methylbutyryrl-CoA dehydrogenase deficiency (2MBG)
29. 3-hydroxy 3-methylglutaric glutaric aciduria (HMG)
30. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
31. 3-Methylglutaconic aciduria (3MGA)
32. Beta-ketothiolase deficiency (BKT)
33. Glutaric acidemia type I (GA1)
34. Isovaleric acidemia (IVA)
35. Malonic Acidemia (MAL)
36. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
37. Methylmalonic aciduria with homocystinuria (Cbl C,D)
38. Methylmalonic acidemia methylmalonyl-CoA mutase (MUT)
39. Multiple carboxylase deficiency (MCD)
40. Propionic acidemia (PROP)

Hemoglobinopathies

41. S/Beta thalassemia
42. S/C disease
43. Sickle cell anemia
44. Variant hemoglobinopathies
45. Hemoglobin H disease

Endocrine Disorders

46. Congenital adrenal hyperplasia (CAH)
47. Congenital hypothyroidism (CH)

Lysosomal Storage Disorders

48. Glycogen Storage Disease Type II (Pompe)
49. Mucopolysaccharidosis Type I (MPS I)

Other Disorders

50. Biotinidase deficiency (BIOT)
51. Galactosemia (GALT)
52. Cystic fibrosis (CF)
53. Severe combined immunodeficiency (SCID)
54. T-cell related lymphocyte deficiencies
55. X-linked Adrenoleukodystrophy (X-ALD)
56. Spinal muscular atrophy (SMA)
57. Guanidinoacetate methyltransferase (GAMT) deficiency
58. Hearing
59. Critical Congenital Heart Disease (CCHD)

Updated September 2022

Newborn Screening (NBS)

- Mandated by Michigan law (MCL 333.5431)
- Funded by purchase of the newborn screening card
  - $144.50 first sample, $158.91 effective 1/1/23
  - $131.05 repeat sample, $144.15 effective 1/1/23
- Annual fee changes based on:
  - Changes in the Detroit Consumer Price Index
  - Addition of new disorders
- Fees entirely support:
  - Courier
  - Laboratory testing
  - Follow-up and coordinating centers

298 Babies Diagnosed through NBS in 2020

- Cystic fibrosis (26)
- Endocrine (130)
  - Congenital hypothyroidism (121)
  - Congenital adrenal hyperplasia (9)
- Hemoglobinopathies (68)
- Metabolic disorders (45)
- Primary immunodeficiencies (13)
- Lysosomal storage disorders (7)
- X-linked adrenoleukodystrophy (1)
- Spinal muscular atrophy (8)

Screening Detection Rates 2020

BLOOD SPOT

1:342

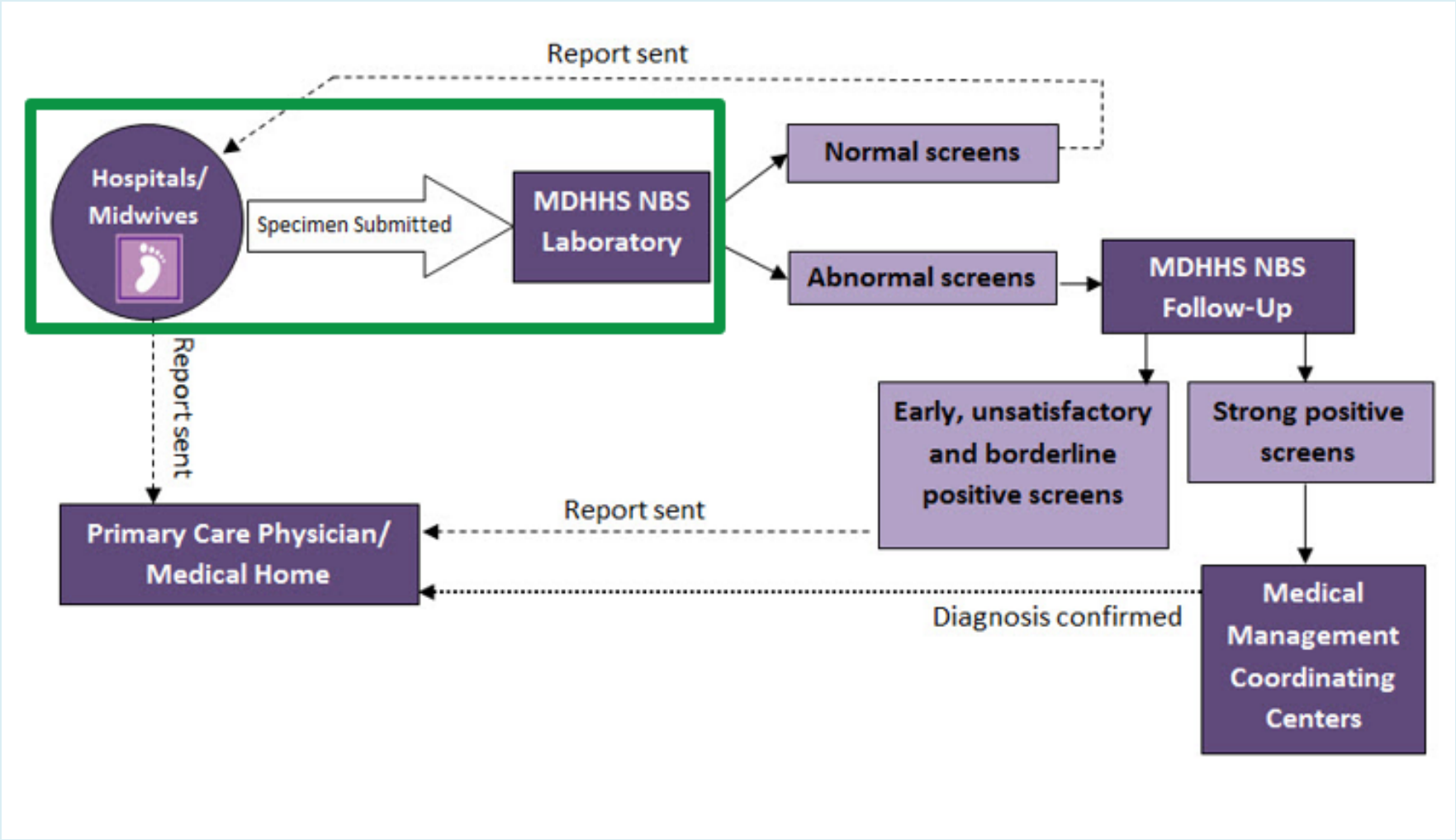
HEART

1:22,935

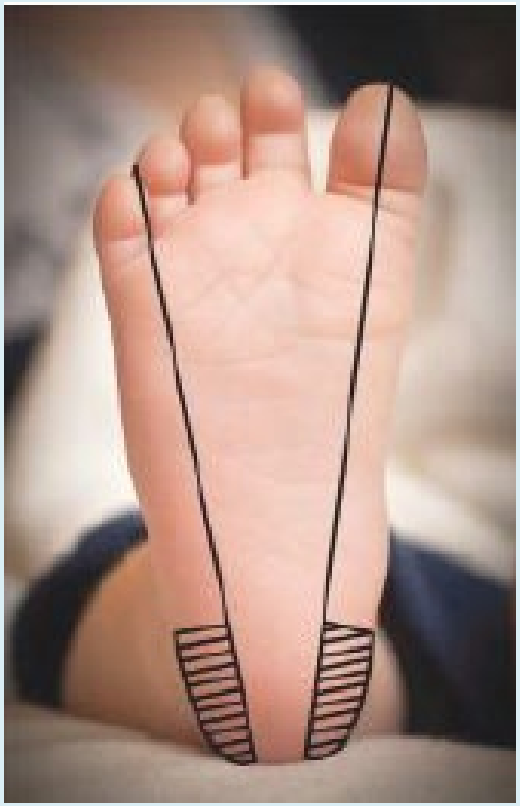
HEARING

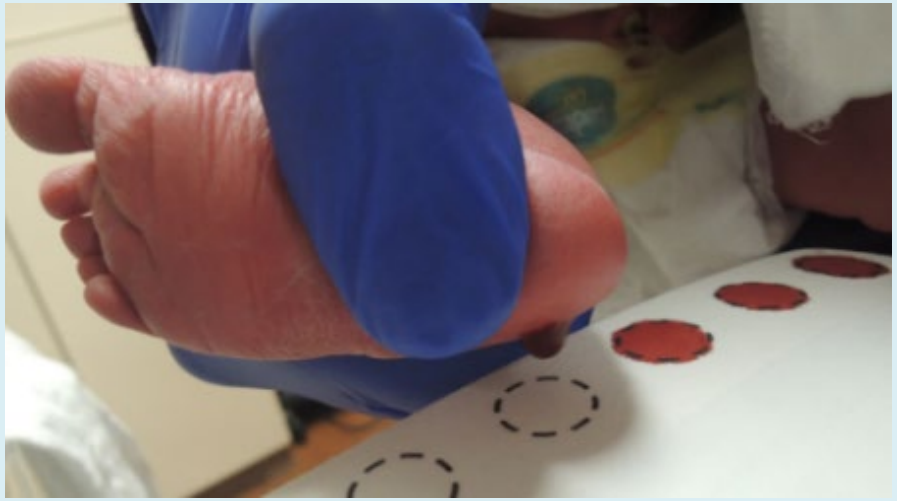
1:543

Michigan's Blood Spot Screening and Follow-Up System



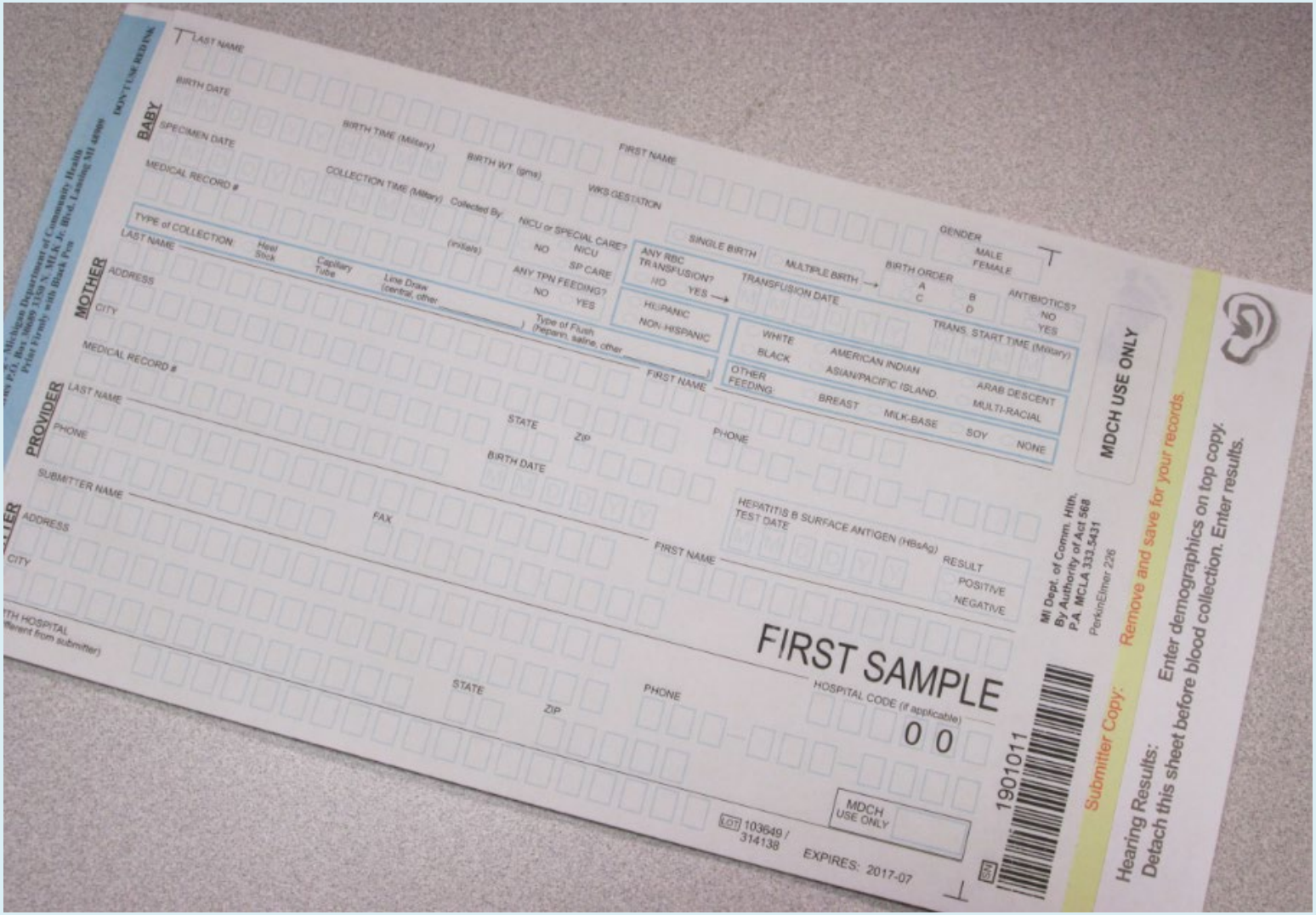
Specimen Collection





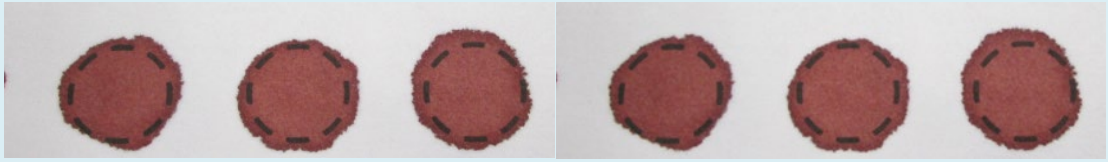
- Heel stick collection preferred
- Collected at 24-36 hours
- 5-6 blood spots on filter paper
- Done at hospital or home (midwife)

Specimen Transport

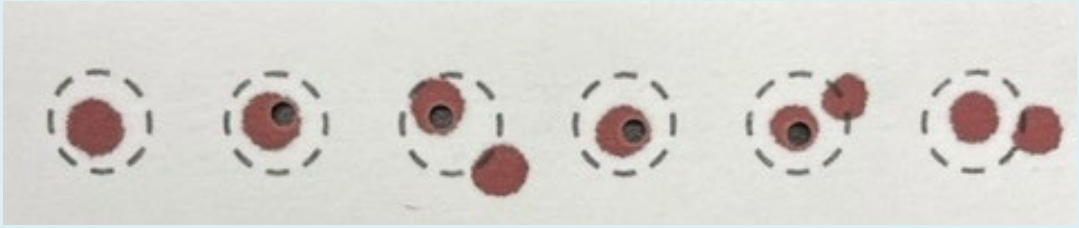


NBS Card Image

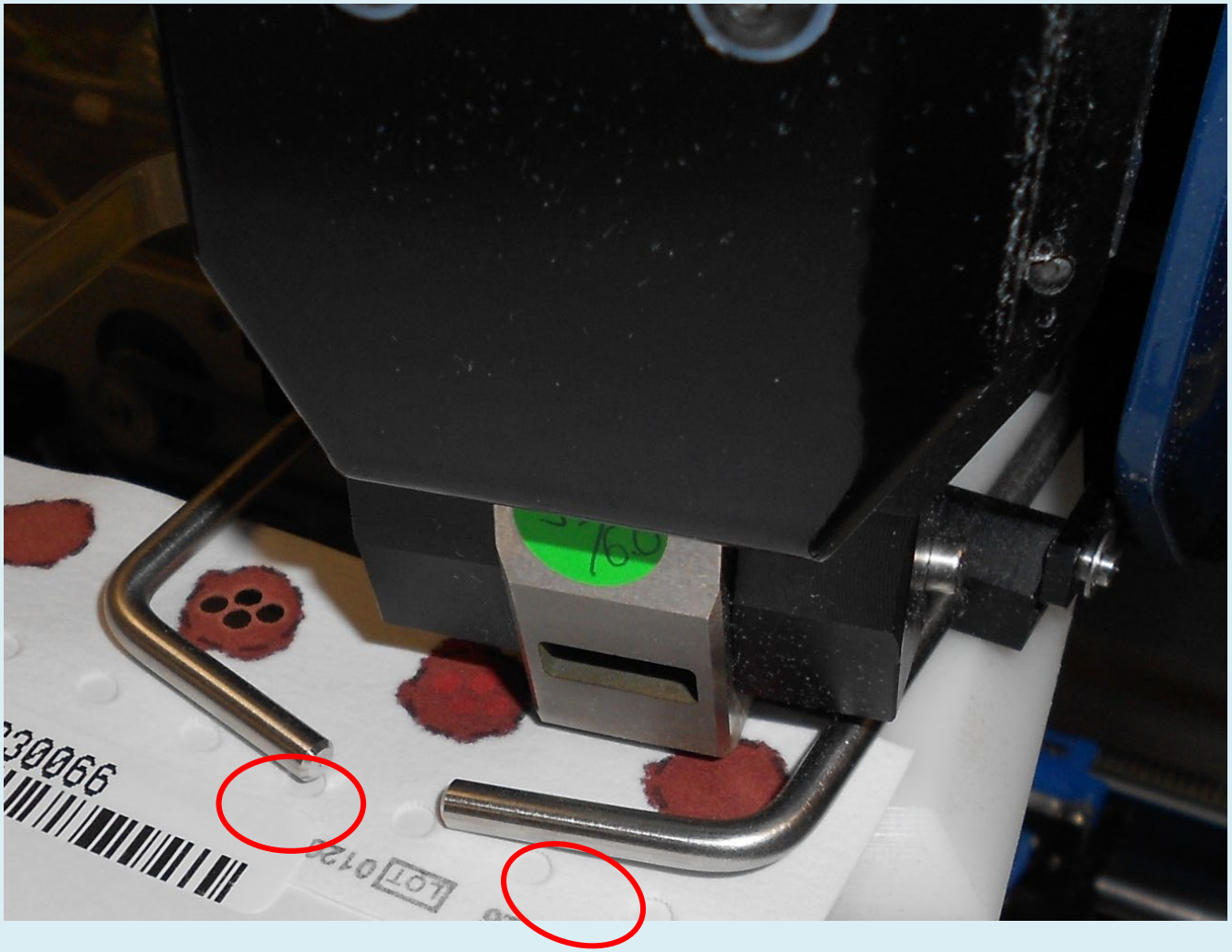
All specimens examined for quality



Satisfactory Specimen



Unsatisfactory Specimen

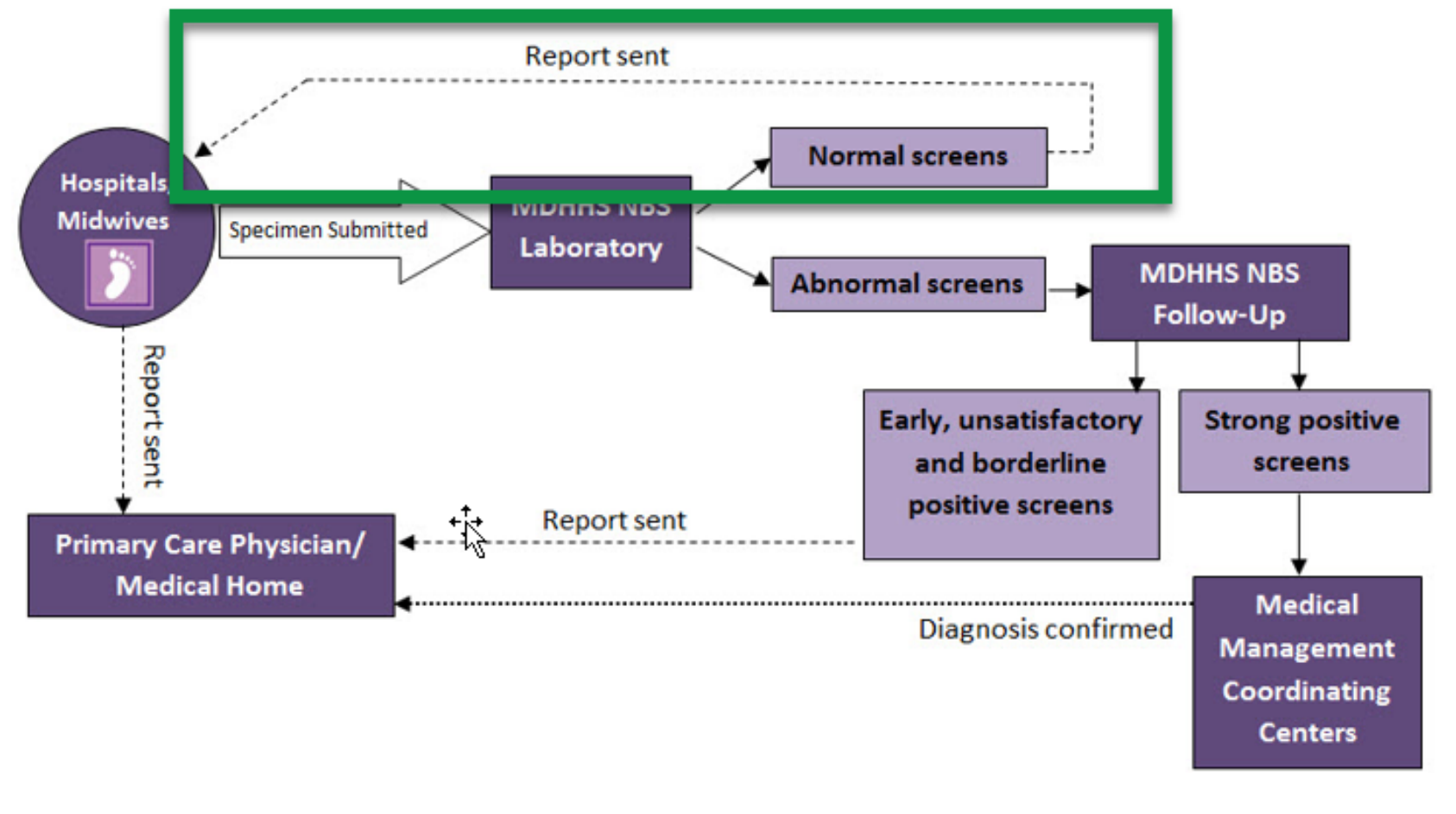


Blood Spots are punched. Need minimum of 11 punches for analysis.

Ready for Analysis

Next steps…normal results

Michigan's Blood Spot Screening and Follow-Up System



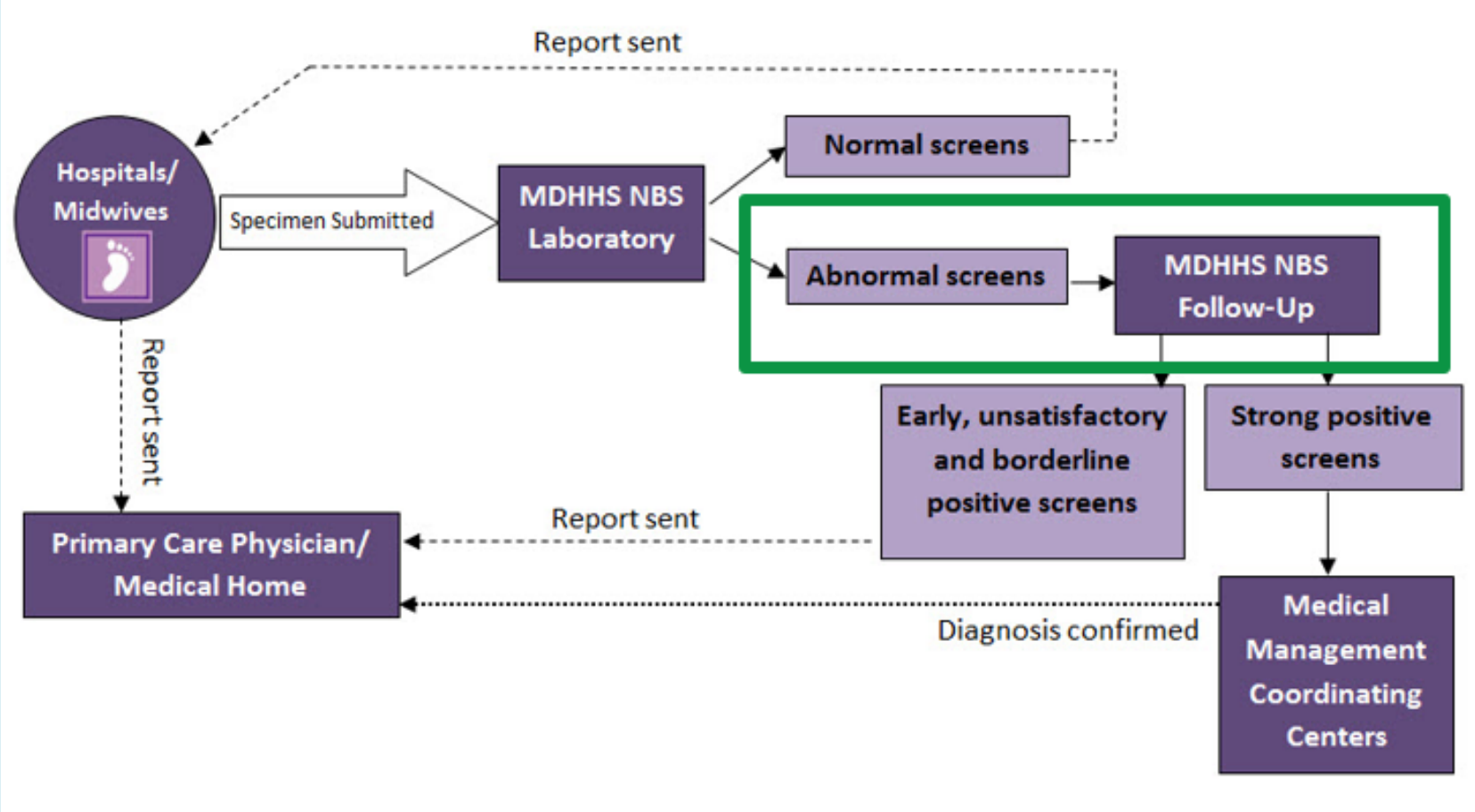
Testing is Completed

- Normal results are faxed or mailed from the lab to the submitter (hospital or midwife)

- NBS final reports are posted on Michigan Care Improvement Registry (MCIR) at ~ 2 weeks

Next steps…Abnormal results

Michigan's Blood Spot Screening and Follow-Up System



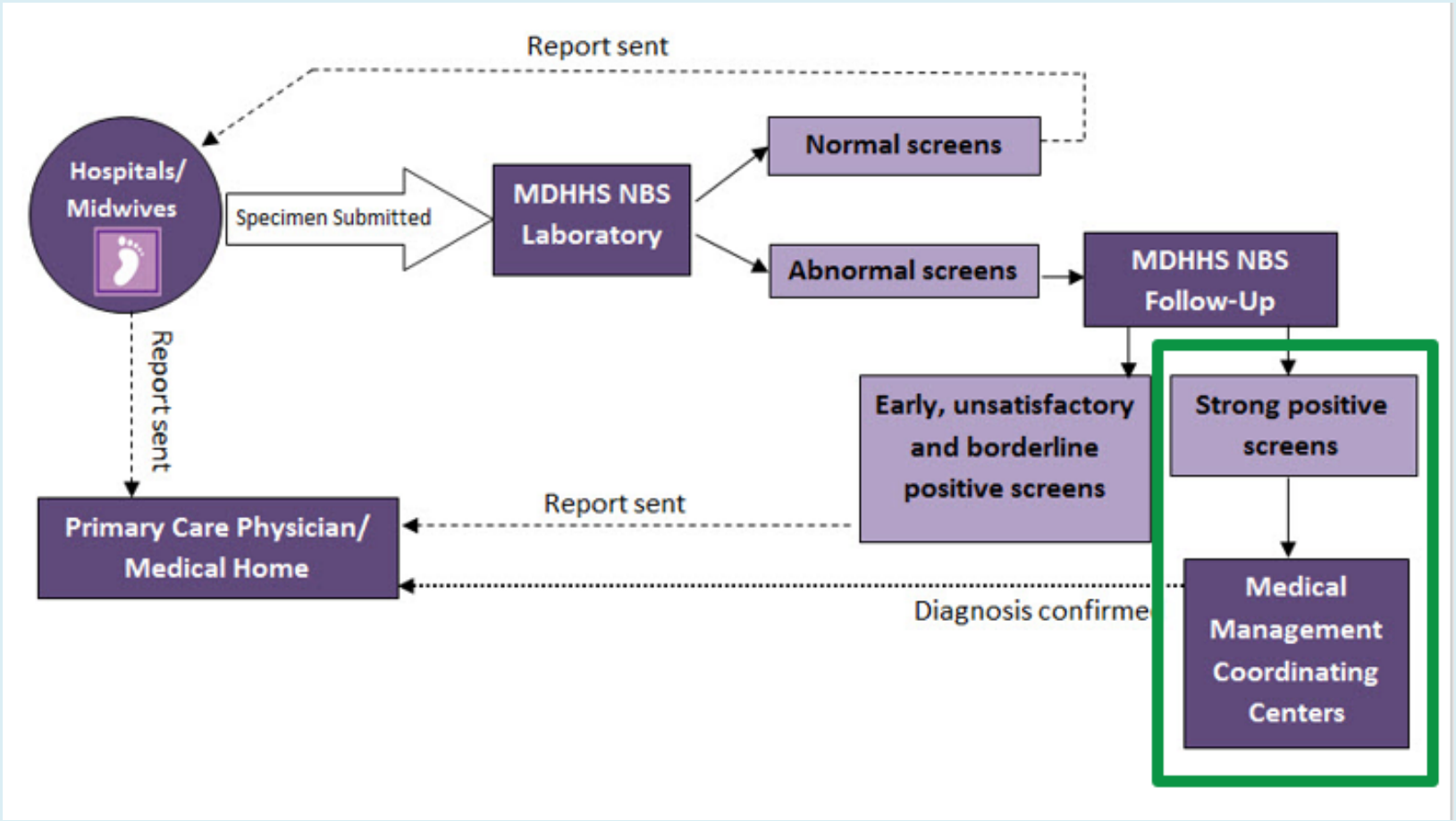
Abnormal Results

Borderline, inconclusive, or unsatisfactory results:

- Reported to primary care provider (PCP) to arrange with family for repeat screen
  - Birth hospital lab collects repeat NBS
  - "Don't worry, but don't wait."
  - ***Importance of parent selecting PCP prior to delivery***
- Reported to hospital, if infant is in NICU or SCN, so repeat screen can be collected prior to discharge.

Next steps…strong positive results

Michigan's Blood Spot Screening and Follow-Up System



NBS Results – Strong Positive

1

Fax report to PCP

Hospital notified, if infant has not been discharged

2

Fax report to follow-up coordinating center; assure referral received

3

Follow-up coordinating center assumes follow-up of strong positive referral

Follow-up Coordinating Centers

- Newborn Screening and Coordinating Program for Cystic Fibrosis (Michigan Medicine)
- Newborn Screening Endocrine Follow-up Program (Michigan Medicine)
- Sickle Cell Disease Association of America, Michigan Chapter
- Children's Hospital of Michigan Metabolic Clinic
- Children's Hospital of Michigan Coordinating Center for Primary Immunodeficiencies
- Pompe and Mucopolysaccharidosis, MPS I (Michigan Medicine and Children's Hospital of Michigan)
- Newborn Screening X-Linked Adrenoleukodystrophy Follow-up Program (Michigan Medicine)

Children's Special Health Care Services

- Diagnostic evaluation for confirmatory testing for NBS disorders
  - Family income not a factor
  - No payment agreement needed
  - Helps when families have high deductibles
  - Transportation assistance
- Coordinated through local health department

Pulse Oximetry Screening for Critical Congenital Heart Disease (CCHD)

Critical Congenital Heart Disease (CCHD)

- Occurs when heart or major blood vessels do not form properly
- Many different types of heart defects (mild to severe)
- “Critical” heart defects need urgent treatment; often requiring surgery shortly after birth
- Some diagnosed prenatally through ultrasound
- Pulse oximetry screening can sometimes detect a CCHD before oxygen levels in the blood become too low

Pulse Oximetry Screening for CCHD

- Mandated screening began April 2014
- Pulse oximetry screen
  - Point of care screen done at 24 hours of life
  - If pulse oximetry is low, infant **may** have a CCHD.
  - Additional testing required to confirm
- Screening does not identify all infants with CCHD
  - Clinical assessment important
  - Symptomatic infants should be evaluated asap

Home Visitors
Making a Difference

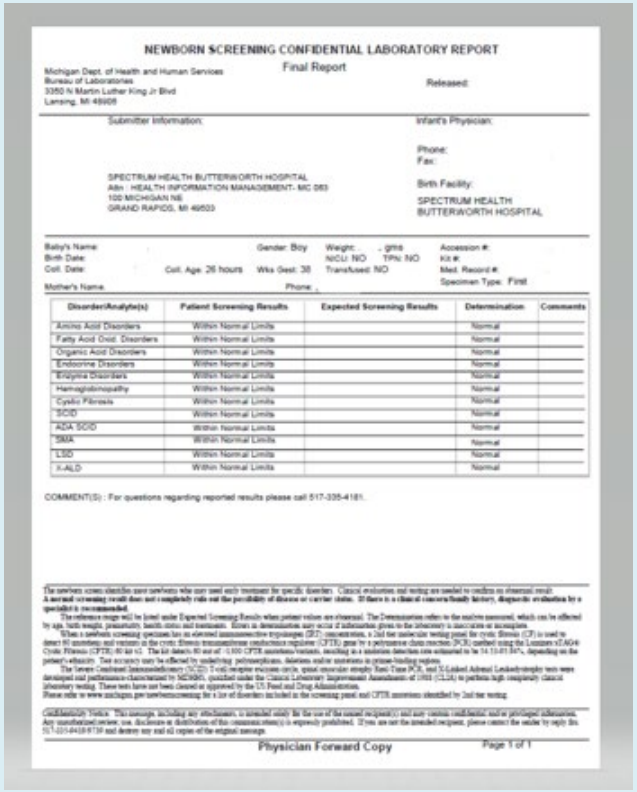
Prenatal Period

Anticipatory Guidance is Key

- Discuss NBS and its importance before expected delivery
- Selection of PCP prior to delivery is critical. Remind to give PCP name to hospital staff at time of delivery.

- Abnormal results are reported right away for follow-up action. Ensure contact information is up-to-date.
- Encourage family to discuss questions or concerns with their medical provider.
- Provide NBS brochures. For more information, provide NBS Program contact details:
  - P: 517-335-4181
  - Email: Newbornscreening@michigan.gov
  - Website: Michigan.gov/Newbornscreening

Review NBS Results Status



NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48909

Released:

Submitter Information:

SPECTRUM HEALTH BUTTERWORTH HOSPITAL
Attn: HEALTH INFORMATION MANAGEMENT- MC 093
100 MICHIGAN NE
GRAND RAPIDS, MI 49503

Infant's Physician:

Phone:
Fax:

Birth Facility:

SPECTRUM HEALTH
BUTTERWORTH HOSPITAL

Baby's Name:
Birth Date:
Coll. Date:
Coll. Age: 26 hours
Gender: Boy
Wks Gest: 38
Weight: . gms
NICU: NO
TPN: NO
Trans/fused: NO
Accession #:
Kit #:
Med. Record #:
Specimen Type: First

Mother's Name:
Phone: ,

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within Normal Limits		Normal	
Fatty Acid Oxid. Disorders	Within Normal Limits		Normal	
Organic Acid Disorders	Within Normal Limits		Normal	
Endocrine Disorders	Within Normal Limits		Normal	
Enzyme Disorders	Within Normal Limits		Normal	
Hemoglobinopathy	Within Normal Limits		Normal	
Cystic Fibrosis	Within Normal Limits		Normal	
SCID	Within Normal Limits		Normal	
ADA SCID	Within Normal Limits		Normal	
SMA	Within Normal Limits		Normal	
LSD	Within Normal Limits		Normal	
X-ALD	Within Normal Limits		Normal	

COMMENT(S) : For questions regarding reported results please call 517-335-4181.

Physician Forward Copy

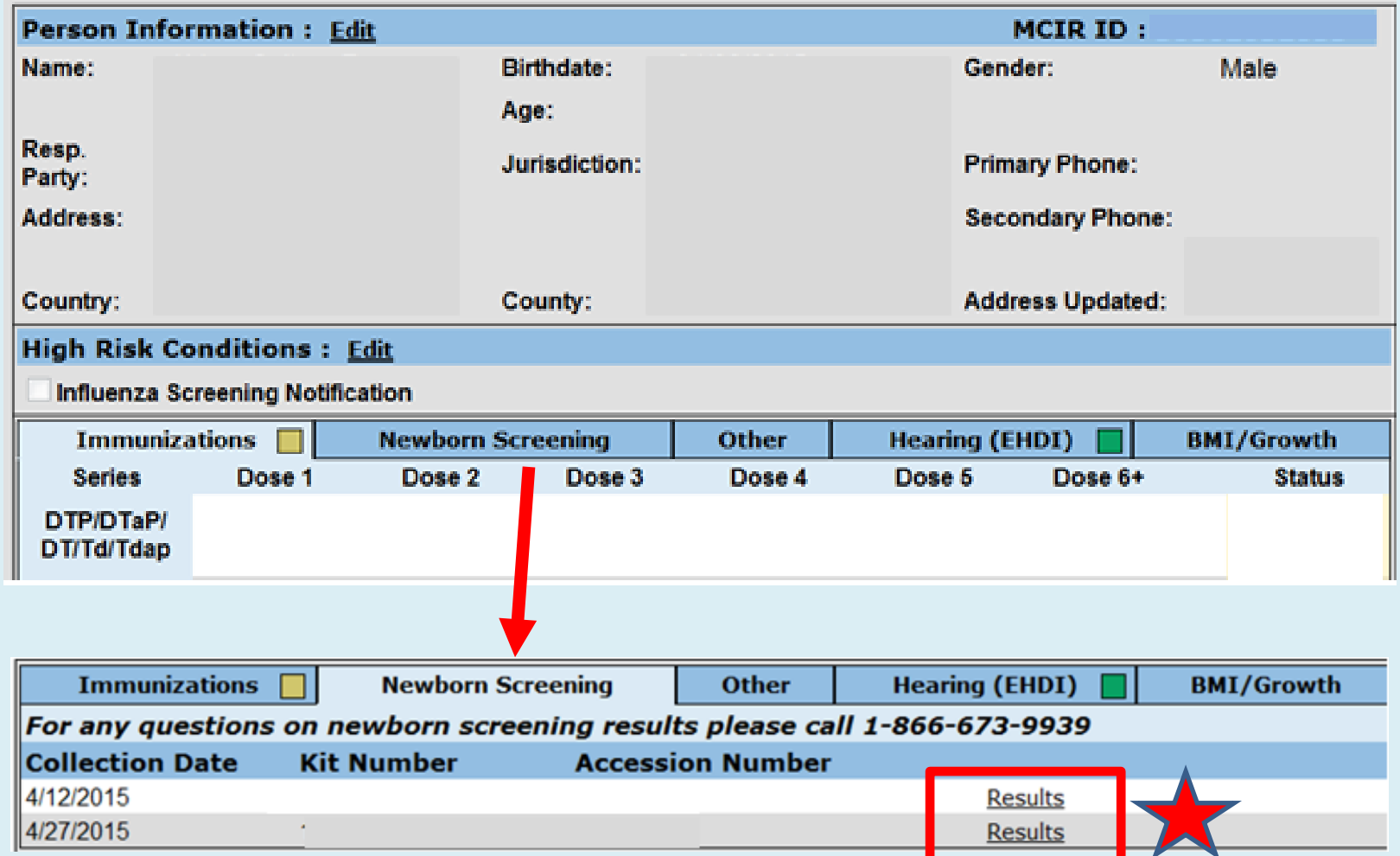
Page 1 of 1

Once infant is enrolled in MIHP, check MCIR to review NBS results.

NBS results are posted on Michigan Care Improvement Registry (MCIR)

- Click on “Newborn Screening Tab”
- Click on “Results” (opens PDF)

MCIR View


Person Information : Edit
MCIR ID :
Name:
Birthdate:
Gender: Male
Age:
Resp. Party:
Jurisdiction:
Primary Phone:
Address:
Secondary Phone:
Country:
County:
Address Updated:
High Risk Conditions : Edit
Influenza Screening Notification
Immunizations
Newborn Screening
Other
Hearing (EHDI)
BMI/Growth
Series
Dose 1
Dose 2
Dose 3
Dose 4
Dose 5
Dose 6+
Status
DTP/DTaP/ DT/Td/Tdap
Immunizations
Newborn Screening
Other
Hearing (EHDI)
BMI/Growth
For any questions on newborn screening results please call 1-866-673-9939
Collection Date
Kit Number
Accession Number
4/12/2015
Results
4/27/2015
Results

NBS Results **Normal**

- No follow-up action needed.
- <u>Remember this is a screen</u>. Normal screening results does not rule out all disorders.

NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48906

Released: 11/7/22 [redacted]

Submitter Information:

SPECTRUM HEALTH BUTTERWORTH HOSPITAL
[redacted]

Infant's Physician: [redacted]
Phone:
Fax: [redacted]

Birth Facility:
SPECTRUM HEALTH
BUTTERWORTH HOSPITAL

Baby's Name: [redacted]
Birth Date:
Coll. Date:
Coll. Age: 24 hours

Gender: Boy
Wks Gest: 40

Weight: [redacted]
NICU: NO TPN: NO
Transfused: NO

Accession #:
Kit #:
Med. Record #: [redacted]
Specimen Type: First

Mother's Name: [redacted]
Phone: [redacted]

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within Normal Limits		Normal	
Fatty Acid Oxid. Disorders	Within Normal Limits		Normal	
Organic Acid Disorders	Within Normal Limits		Normal	
Endocrine Disorders	Within Normal Limits		Normal	
Enzyme Disorders	Within Normal Limits		Normal	
Hemoglobinopathy	Within Normal Limits		Normal	
Cystic Fibrosis	Within Normal Limits		Normal	
SCID	Within Normal Limits		Normal	
ADA SCID	Within Normal Limits		Normal	
SMA	Within Normal Limits		Normal	
LSD	Within Normal Limits		Normal	
X-ALD	Within Normal Limits		Normal	

COMMENT(S) : For questions regarding reported results please call 517-335-4181.

The newborn screen identifies most newborns who may need early treatment for specific disorders. Clinical evaluation and testing are needed to confirm an abnormal result. **A normal screening result does not completely rule out the possibility of disease or carrier status. If there is a clinical concern/family history, diagnostic evaluation by a specialist is recommended.**

The reference range will be listed under Expected Screening Results when patient values are abnormal. The Determination refers to the analyte measured, which can be affected by age, birth weight, prematurity, health status and treatments. Errors in determination may occur if information given to the laboratory is inaccurate or incomplete.

When a newborn screening specimen has an elevated immunoreactive trypsinogen (IRT) concentration, a 2nd tier molecular testing panel for cystic fibrosis (CF) is used to detect 60 mutations and variants in the cystic fibrosis transmembrane conductance regulator (CFTR) gene by a polymerase chain reaction (PCR) method using the Luminex xTAG® Cystic Fibrosis (CFTR) 60 kit v2. The kit detects 60 out of >1300 CFTR mutations/variants, resulting in a mutation detection rate estimated to be 54.53-95.94%, depending on the patient's ethnicity. Test accuracy may be affected by underlying polymorphisms, deletions and/or mutations in primer-binding regions.

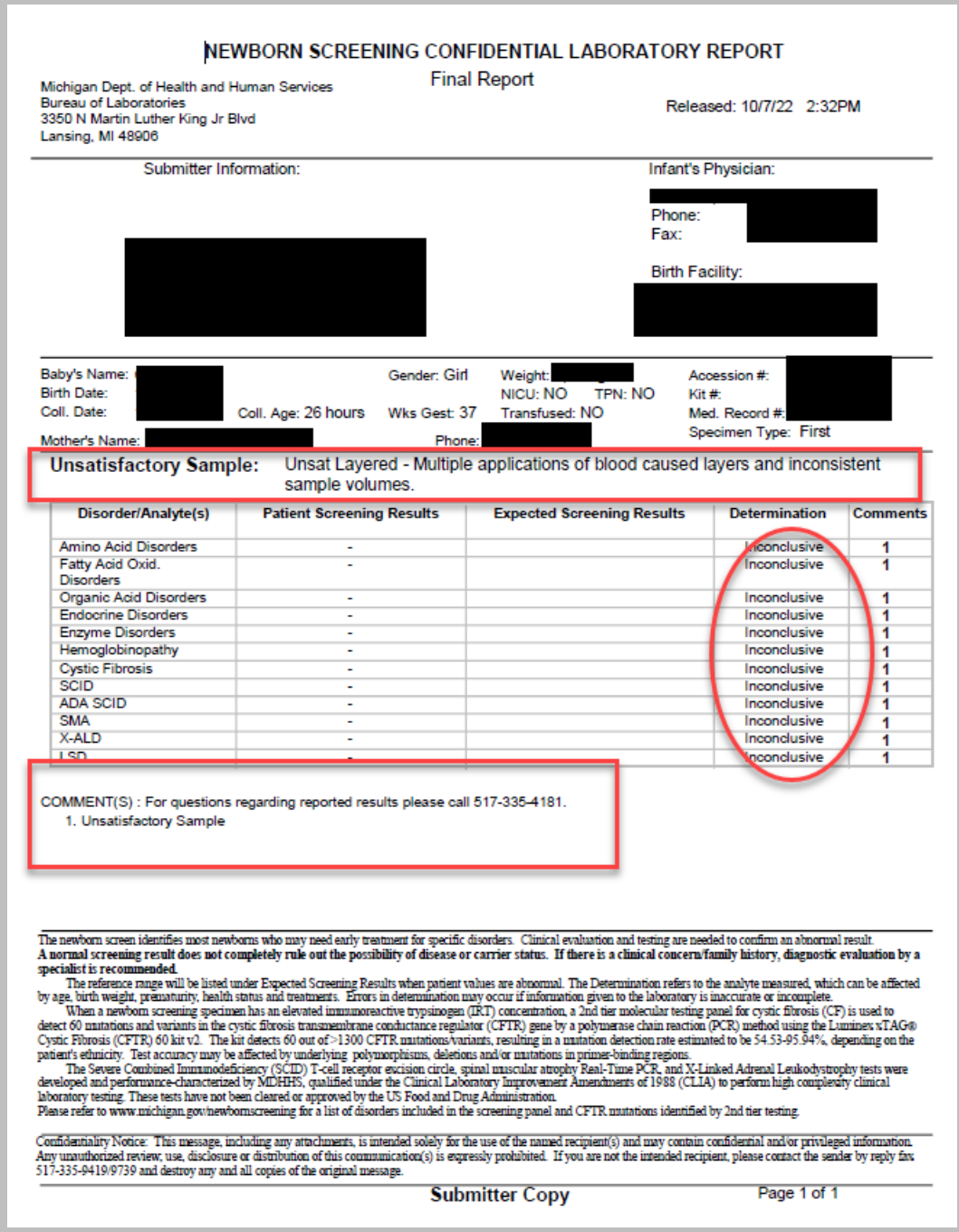
The Severe Combined Immunodeficiency (SCID) T-cell receptor excision circle, spinal muscular atrophy Real-Time PCR, and X-Linked Adrenal Leukodystrophy tests were developed and performance-characterized by MDHHS, qualified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to perform high complexity clinical laboratory testing. These tests have not been cleared or approved by the US Food and Drug Administration.

Please refer to www.michigan.gov/newbornscreening for a list of disorders included in the screening panel and CFTR mutations identified by 2nd tier testing.

Submitter Copy

NBS Results **Unsatisfactory Specimen**

- Result will be inconclusive
- Comment section will show unsatisfactory sample
- Repeat often necessary
- Instruct parent to call PCP for further instruction



NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48906

Released: 10/7/22 2:32PM

Submitter Information: [redacted]

Infant's Physician: [redacted]
Phone: [redacted]
Fax:

Birth Facility: [redacted]

Baby's Name: [redacted]
Birth Date:
Coll. Date: Coll. Age: 26 hours
Gender: Girl
Wks Gest: 37
Weight: [redacted]
NICU: NO TPN: NO
Transfused: NO
Accession #: [redacted]
Kit #:
Med. Record #:
Specimen Type: First
Mother's Name: [redacted]
Phone: [redacted]

Unsatisfactory Sample: Unsat Layered - Multiple applications of blood caused layers and inconsistent sample volumes.

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	-		Inconclusive	1
Fatty Acid Oxid. Disorders	-		Inconclusive	1
Organic Acid Disorders	-		Inconclusive	1
Endocrine Disorders	-		Inconclusive	1
Enzyme Disorders	-		Inconclusive	1
Hemoglobinopathy	-		Inconclusive	1
Cystic Fibrosis	-		Inconclusive	1
SCID	-		Inconclusive	1
ADA SCID	-		Inconclusive	1
SMA	-		Inconclusive	1
X-ALD	-		Inconclusive	1
LSD	-		Inconclusive	1

COMMENT(S) : For questions regarding reported results please call 517-335-4181.
1. Unsatisfactory Sample

The newborn screen identifies most newborns who may need early treatment for specific disorders. Clinical evaluation and testing are needed to confirm an abnormal result. **A normal screening result does not completely rule out the possibility of disease or carrier status. If there is a clinical concern/family history, diagnostic evaluation by a specialist is recommended.**

The reference range will be listed under Expected Screening Results when patient values are abnormal. The Determination refers to the analyte measured, which can be affected by age, birth weight, prematurity, health status and treatments. Errors in determination may occur if information given to the laboratory is inaccurate or incomplete.

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Please refer to www.michigan.gov/newbornscreening for a list of disorders included in the screening panel and CFTR mutations identified by 2nd tier testing.

Submitter Copy

NBS Results
Early Specimen

- Specimen was collected prior to 24 hours
- Comments inform that repeat screen is needed
- Instruct parent to call PCP for further instruction

NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48906

Released: 11/7/22 9:43 AM

Submitter Information:

[redacted]

Infant's Physician:

[redacted]
Phone:
Fax:

Birth Facility:

PROMEDICA MONROE REGIONAL HOSPITAL

Baby's Name: [redacted]
Birth Date:
Coll. Date:
Coll. Age: 5 hours
Gender: Boy
Wks Gest: 40
Weight:
NICU: YES TPN: NO
Transfused: NO
Accession #:
Kit #:
Med. Record #: [redacted]
Specimen Type: First
Mother's Name: [redacted] A
Phone: [redacted]

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within Normal Limits		Normal	
Fatty Acid Oxid. Disorders	Within Normal Limits		Normal	
Organic Acid Disorders	Within Normal Limits		Normal	
Endocrine Disorders				
TSH	-	* Varies with Age	**Inconclusive**	1
Other	Within Normal Limits		Normal	
Enzyme Disorders	Within Normal Limits		Normal	
Hemoglobinopathy	Within Normal Limits		Normal	
Cystic Fibrosis	Within Normal Limits		Normal	
SCID	Within Normal Limits		Normal	
ADA SCID	Within Normal Limits		Normal	
SMA	Within Normal Limits		Normal	
X-ALD	Within Normal Limits		Normal	
LSD	Within Normal Limits		Normal	

* TSH Age Categories: < 24 hrs is undefined; 24 hrs - 36 hrs < 27; 37 hrs - 167 hrs < 21; 168 hrs - 743 hrs < 11; > 744 hrs < 9

COMMENT(S) : For questions regarding reported results please call 517-335-4181.

1. Early specimen; submit another sample to Newborn Screening Laboratory; confirmatory serum hypothyroid testing is not recommended at this time.

The newborn screen identifies most newborns who may need early treatment for specific disorders. Clinical evaluation and testing are needed to confirm an abnormal result. **A normal screening result does not completely rule out the possibility of disease or carrier status. If there is a clinical concern/family history, diagnostic evaluation by a specialist is recommended.**

The reference range will be listed under Expected Screening Results when patient values are abnormal. The Determination refers to the analyte measured, which can be affected by age, birth weight, prematurity, health status and treatments. Errors in determination may occur if information given to the laboratory is inaccurate or incomplete.

When a newborn screening specimen has an elevated immunoreactive trypsinogen (IRT) concentration, a 2nd tier molecular testing panel for cystic fibrosis (CF) is used to detect 60 mutations and variants in the cystic fibrosis transmembrane conductance regulator (CFTR) gene by a polymerase chain reaction (PCR) method using the Luminex xTAG® Cystic Fibrosis (CFTR) 60 kit v2. The kit detects 60 out of >1300 CFTR mutations/variants, resulting in a mutation detection rate estimated to be 54.53-95.94%, depending on the patient's ethnicity. Test accuracy may be affected by underlying polymorphisms, deletions and/or mutations in primer-binding regions.

The Severe Combined Immunodeficiency (SCID) T-cell receptor excision circle, spinal muscular atrophy Real-Time PCR, and X-Linked Adrenal Leukodystrophy tests were developed and performance-characterized by MDHHS, qualified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) to perform high complexity clinical laboratory testing. These tests have not been cleared or approved by the US Food and Drug Administration.

Please refer to www.michigan.gov/newbornscreening for a list of disorders included in the screening panel and CFTR mutations identified by 2nd tier testing.

Submitter Copy

NBS Results **Borderline**

- Report will vary – based on disorder
- Repeat NBS is needed
- If parent has not heard from provider about next steps, instruct to call PCP **ASAP** for further instruction

NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48906

Released: 6/10/21 7:35 AM

Submitter Information:

[redacted]

Infant's Physician:

[redacted]

Phone:
Fax:

Birth Facility:

[redacted]

Baby's Name: [redacted]
Birth Date:
Coll. Date: Coll. Age: 24 hours

Gender: Boy
Wks Gest: 38

Weight: [redacted]
NICU: NO TPN: NO
Transfused: NO

Accession #: [redacted]
Kit #:
Med. Record #:
Specimen Type: First

Mother's Name: [redacted] Phone: [redacted]

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within Normal Limits		Normal	
Fatty Acid Oxid. Disorders	Within Normal Limits		Normal	
Organic Acid Disorders	Within Normal Limits		Normal	
[illegible]	[illegible]		[illegible]	
Enzyme Disorders				
BIO	**Partial Activity**	**Normal Activity**	**Abnormal**	**1**
Other	Within Normal Limits		Normal	
Hemoglobinopathy	Within Normal Limits		Normal	
Cystic Fibrosis	Within Normal Limits		Normal	
SCID	Within Normal Limits		Normal	
ADA SCID	Within Normal Limits		Normal	
SMA	Within Normal Limits		Normal	
LSD	Within Normal Limits		Normal	
X-ALD	Within Normal Limits		Normal	

COMMENT(S) : For questions regarding reported results please call 517-335-4181.
1. Biotinidase Deficiency Borderline Positive

The newborn screen identifies most newborns who may need early treatment for specific disorders. Clinical evaluation and testing are needed to confirm an abnormal result. **A normal screening result does not completely rule out the possibility of disease or carrier status. If there is a clinical concern/family history, diagnostic evaluation by a specialist is recommended.**

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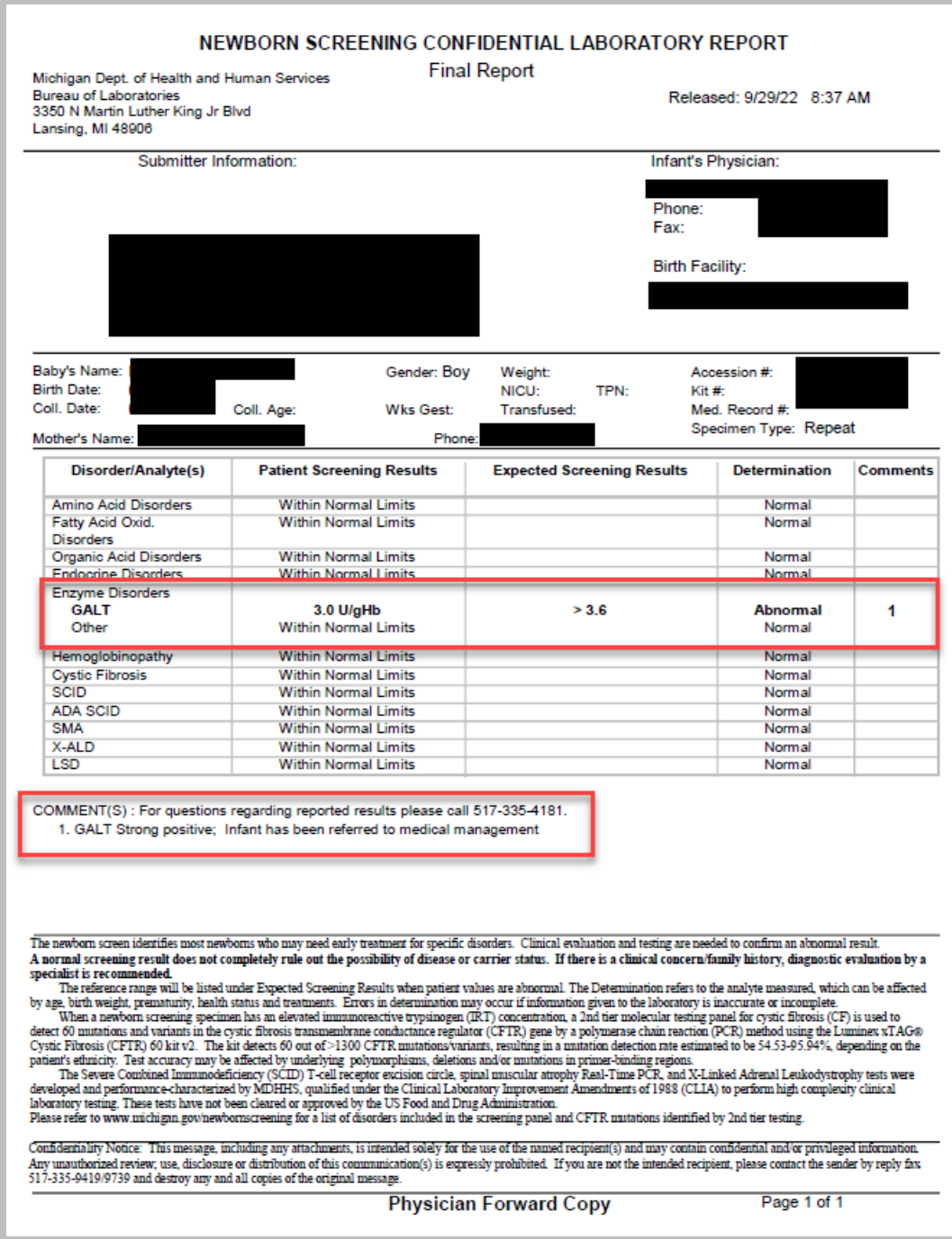
Please refer to www.michigan.gov/newbornscreening for a list of disorders included in the screening panel and CFTR mutations identified by 2nd tier testing.

Submitter Copy

NBS Results **Abnormal**

- Report will vary – based on disorder
- Infant referred to Coordinating Center for diagnostic testing
- If parent has not heard from provider about next steps, instruct to call PCP **ASAP** for further instruction



NEWBORN SCREENING CONFIDENTIAL LABORATORY REPORT

Final Report

Michigan Dept. of Health and Human Services
Bureau of Laboratories
3350 N Martin Luther King Jr Blvd
Lansing, MI 48906

Released: 9/29/22 8:37 AM

Submitter Information: [redacted]

Infant's Physician: [redacted]
Phone: [redacted]
Fax:

Birth Facility: [redacted]

Baby's Name: [redacted] Gender: Boy Weight: Accession #: [redacted]
Birth Date: NICU: TPN: Kit #:
Coll. Date: Coll. Age: Wks Gest: Transfused: Med. Record #:
Mother's Name: [redacted] Phone: [redacted] Specimen Type: Repeat

Disorder/Analyte(s)	Patient Screening Results	Expected Screening Results	Determination	Comments
Amino Acid Disorders	Within Normal Limits		Normal	
Fatty Acid Oxid. Disorders	Within Normal Limits		Normal	
Organic Acid Disorders	Within Normal Limits		Normal	
Endocrine Disorders	Within Normal Limits		Normal	
Enzyme Disorders				
GALT	**3.0 U/gHb**	**> 3.6**	**Abnormal**	**1**
Other	Within Normal Limits		Normal	
Hemoglobinopathy	Within Normal Limits		Normal	
Cystic Fibrosis	Within Normal Limits		Normal	
SCID	Within Normal Limits		Normal	
ADA SCID	Within Normal Limits		Normal	
SMA	Within Normal Limits		Normal	
X-ALD	Within Normal Limits		Normal	
LSD	Within Normal Limits		Normal	

COMMENT(S) : For questions regarding reported results please call 517-335-4181.
1. GALT Strong positive; Infant has been referred to medical management

The newborn screen identifies most newborns who may need early treatment for specific disorders. Clinical evaluation and testing are needed to confirm an abnormal result.
A normal screening result does not completely rule out the possibility of disease or carrier status. If there is a clinical concern/family history, diagnostic evaluation by a specialist is recommended.

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Please refer to www.michigan.gov/newbornscreening for a list of disorders included in the screening panel and CFTR mutations identified by 2nd tier testing.

Confidentiality Notice: This message, including any attachments, is intended solely for the use of the named recipient(s) and may contain confidential and/or privileged information. Any unauthorized review, use, disclosure or distribution of this communication(s) is expressly prohibited. If you are not the intended recipient, please contact the sender by reply fax 517-335-9419/9739 and destroy any and all copies of the original message.

Physician Forward Copy

Page 1 of 1

No results in MCIR

Ask if baby received NBS

- If **yes**, check MCIR again later as they may not be uploaded yet
  - Note: Results not posted to MCIR if in foster care or released for adoption

- If **no**:
  - Engage in conversation to see why infant was not screened
    - If NBS was missed, tell family to contact PCP right away
  - Provide educational resources about NBS
  - Share personal family stories about NBS
    - GAMT Deficiency Story: The Power of Newborn Screening (https://youtu.be/sw0zZRq6kZ0)
    - Hunter's Hope – Hope through Newborn Screening (https://youtu.be/lATzs_LqIYg)
    - Baebies Maggie's Story – Importance of Newborn Screening - MPSI (https://youtu.be/cAjJYCjWe6o)
  - Encourage family to discuss concerns with PCP
  - Remind family that they can still receive NBS for their infant

Contact Information

- Angela Aldrich MSN, RN, Nurse Consultant
  - aldricha1@Michigan.gov
  - 517-335-1966

- Websites:
  - www.Michigan.gov/newbornscreening
  - www.Michigan.gov/biotrust
  - www.Michigan.gov/cchd
  - www.Michigan.gov/ehdi

Newborn Screening

Parent Education

Is your client ready for their baby's first test?

What all parents need to know about newborn screening.

Prior to Delivery

- Encourage parents to pick a primary care provider for their newborn and tell them to be ready to give the name and phone number of the provider to the hospital after delivery.

During Hospital Stay

- Make sure to tell parents what they can expect at the hospital. Explain each step of the NBS process: the blood spot and both point of care screens.
- Encourage parents to ask hospital staff questions about their child's NBS.
- Make parents aware of the options they have with the blood spots after NBS is completed.
- Tell parents to ask hospital staff for their baby's results for the two screens done at the hospital (hearing and pulse oximetry).

After Discharge

- Parents can ask their baby's primary care provider for the results of the newborn blood screen. The results should be ready before their baby is one week old.

How to explain the three parts of a newborn screen to a parent

Heel Stick

A small blood sample is taken from your baby's heel and placed on a newborn screening card. This card is delivered by courier to the State of Michigan Laboratory for analysis. If an out-of-range result is detected, your baby's healthcare provider will be notified.

Hearing Test

One or two tests may be used to screen for hearing loss in your baby: an otoacoustic emissions or an auditory brainstem response. Both tests are simple, safe and can be done while your baby is asleep!

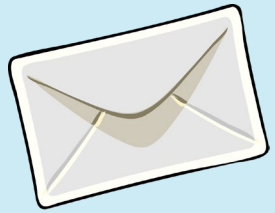
Pulse Oximetry

Pulse oximetry is a test that measures the amount of oxygen in your baby's blood and can detect some heart problems called critical congenital heart disease (CCHD).

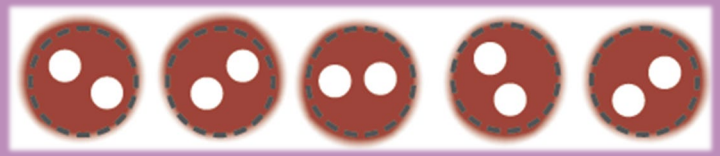
You do not need to request that a newborn screen be taken!

What happens next?

What parents need to know



At the lab, blood spots are punched into smaller circles for each of the tests. Your baby is tested for more than 50 health problems.



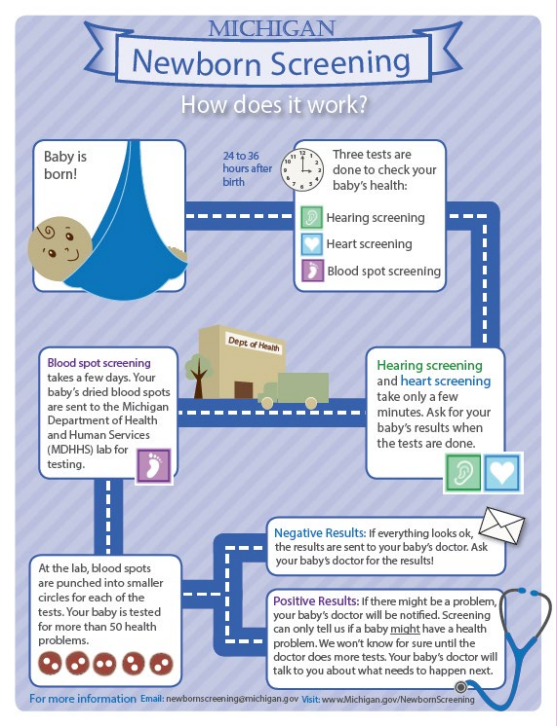
Negative results: If everything looks okay, the results are sent to your baby's doctor. Ask your baby's doctor for the results.

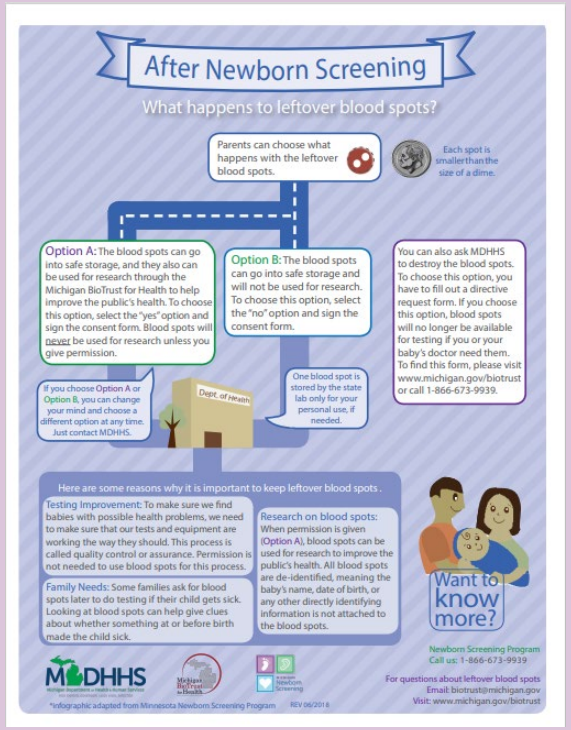
Positive results: If there might be a problem, your baby's doctor will be notified. Screening can only tell us if a baby might have a health problem. We won't know for sure until the doctor does more tests. Your baby's doctor will talk to you about what needs to happen next.

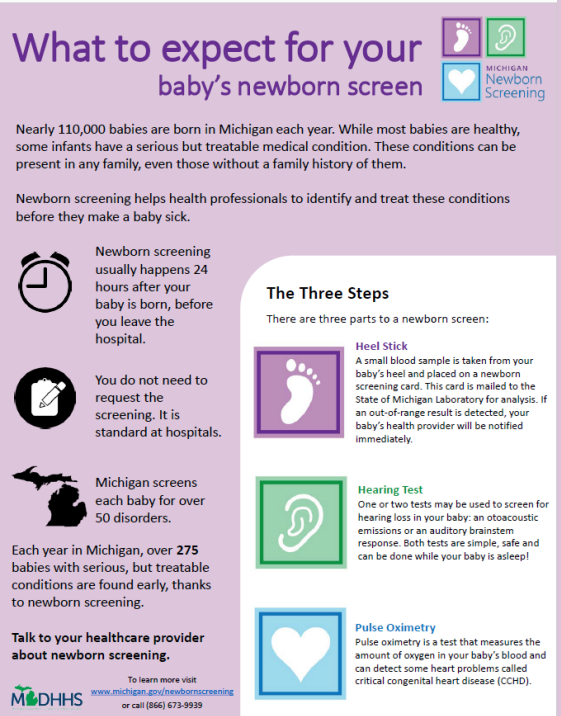
Talk to your healthcare provider about newborn screening.

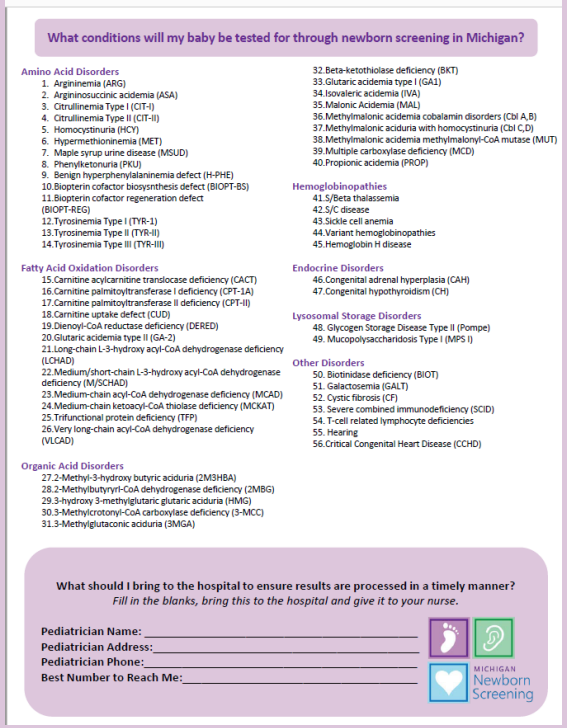
All of our educational resources are FREE

- Please place all orders for educational materials through NBSO
- Website: michigan.gov/NBSorders
- Phone number: (517) 335-1400
- Email: MDHHS-NBSOrders@michigan.gov



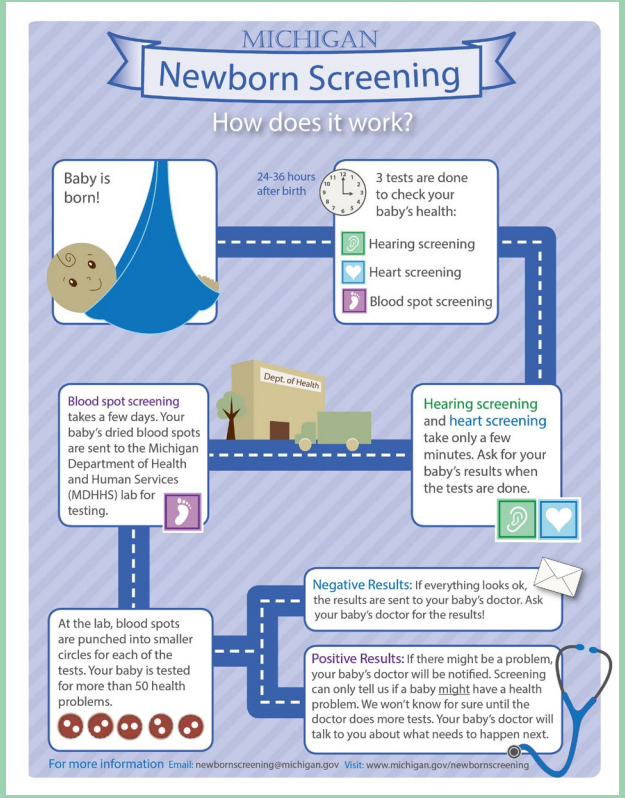


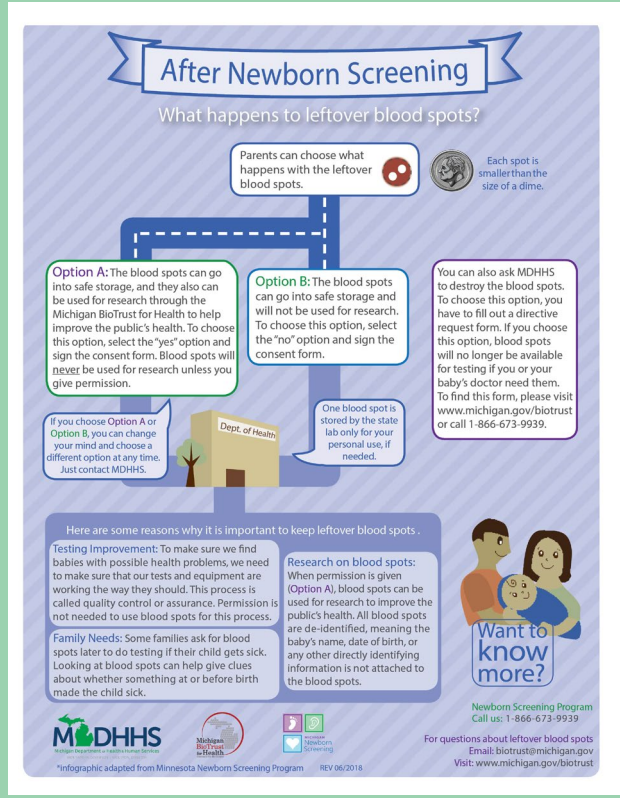




Infographics

NBS and BioTrust Road Map





- ✓ Can be ordered through NBS Online Ordering Module in English, Spanish, and Arabic
- ✓ Visual summary of newborn screening on the front and the parents' options for their child's blood after the screen is complete on the back
- ✓ Easy to follow road map that helps parents understand exactly what is happening at each step of the NBS process
- ✓ Great for new or expecting parents

What to Expect

What to expect for your baby's newborn screen

MICHIGAN Newborn Screening

Nearly 110,000 babies are born in Michigan each year. While most babies are healthy, some infants have a serious but treatable medical condition. These conditions can be present in any family, even those without a family history of them.

Newborn screening helps health professionals to identify and treat these conditions before they make a baby sick.

Newborn screening usually happens 24 hours after your baby is born, before you leave the hospital.

You do not need to request the screening. It is standard at hospitals.

Michigan screens each baby for over 50 disorders.

Each year in Michigan, over **275** babies with serious, but treatable conditions are found early, thanks to newborn screening.

Talk to your healthcare provider about newborn screening.

MDHHS

To learn more visit www.michigan.gov/newbornscreening or call (866) 673-9939

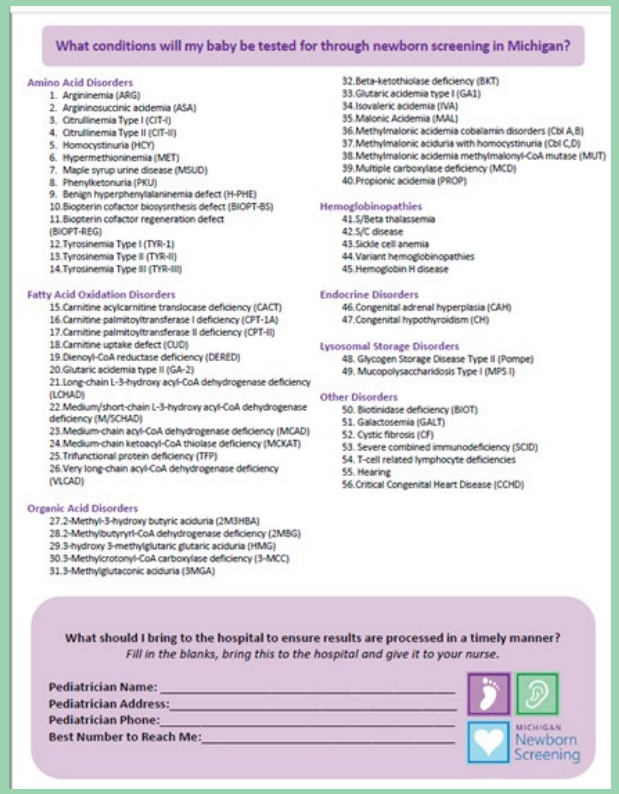
The Three Steps

There are three parts to a newborn screen:

Heel Stick
A small blood sample is taken from your baby's heel and placed on a newborn screening card. This card is mailed to the State of Michigan Laboratory for analysis. If an out-of-range result is detected, your baby's health provider will be notified immediately.

Hearing Test
One or two tests may be used to screen for hearing loss in your baby: an otoacoustic emissions or an auditory brainstem response. Both tests are simple, safe and can be done while your baby is asleep!

Pulse Oximetry
Pulse oximetry is a test that measures the amount of oxygen in your baby's blood and can detect some heart problems called critical congenital heart disease (CCHD).



What conditions will my baby be tested for through newborn screening in Michigan?

Amino Acid Disorders
1. Argininemia (ARG)
2. Argininosuccinic acidemia (ASA)
3. Citrullinemia Type I (CIT-I)
4. Citrullinemia Type II (CIT-II)
5. Homocystinuria (HCY)
6. Hypermethioninemia (MET)
7. Maple syrup urine disease (MSUD)
8. Phenylketonuria (PKU)
9. Benign hyperphenylalaninemia defect (H-PHE)
10. Biopterin cofactor biosysnthesis defect (BIOPT-BS)
11. Biopterin cofactor regeneration defect (BIOPT-REG)
12. Tyrosinemia Type I (TYR-1)
13. Tyrosinemia Type II (TYR-II)
14. Tyrosinemia Type III (TYR-III)

Fatty Acid Oxidation Disorders
15. Carnitine acylcarnitine translocase deficiency (CACT)
16. Carnitine palmitoyltransferase I deficiency (CPT-1A)
17. Carnitine palmitoyltransferase II deficiency (CPT-II)
18. Carnitine uptake defect (CUD)
19. Dienoyl-CoA reductase deficiency (DERED)
20. Glutaric acidemia type II (GA-2)
21. Long-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (LCHAD)
22. Medium/short-chain L-3-hydroxy acyl-CoA dehydrogenase deficiency (M/SCHAD)
23. Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
24. Medium-chain ketoacyl-CoA thiolase deficiency (MCKAT)
25. Trifunctional protein deficiency (TFP)
26. Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Disorders
27. 2-Methyl-3-hydroxy butyric aciduria (2M3HBA)
28. 2-Methylbutyryrl-CoA dehydrogenase deficiency (2MBG)
29. 3-hydroxy 3-methylglutaric glutaric aciduria (HMG)
30. 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC)
31. 3-Methylglutaconic aciduria (3MGA)
32. Beta-ketothiolase deficiency (BKT)
33. Glutaric acidemia type I (GA1)
34. Isovaleric acidemia (IVA)
35. Malonic Acidemia (MAL)
36. Methylmalonic acidemia cobalamin disorders (Cbl A,B)
37. Methylmalonic aciduria with homocystinuria (Cbl C,D)
38. Methylmalonic acidemia methylmalonyl-CoA mutase (MUT)
39. Multiple carboxylase deficiency (MCD)
40. Propionic acidemia (PROP)

Hemoglobinopathies
41. S/Beta thalassemia
42. S/C disease
43. Sickle cell anemia
44. Variant hemoglobinopathies
45. Hemoglobin H disease

Endocrine Disorders
46. Congenital adrenal hyperplasia (CAH)
47. Congenital hypothyroidism (CH)

Lysosomal Storage Disorders
48. Glycogen Storage Disease Type II (Pompe)
49. Mucopolysaccharidosis Type I (MPS I)

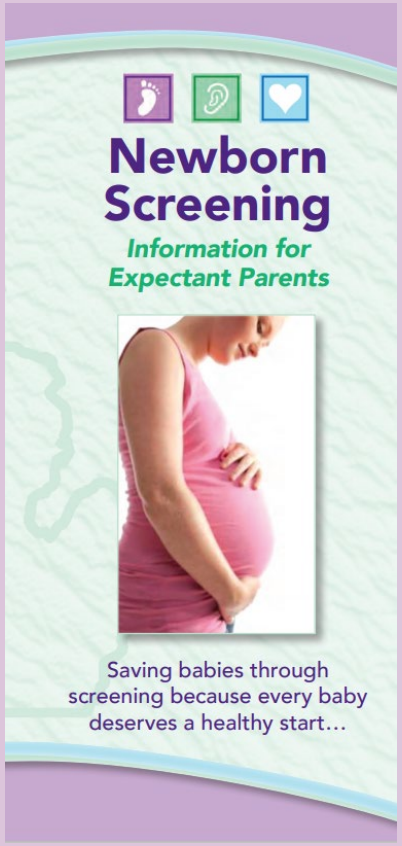
Other Disorders
50. Biotinidase deficiency (BIOT)
51. Galactosemia (GALT)
52. Cystic fibrosis (CF)
53. Severe combined immunodeficiency (SCID)
54. T-cell related lymphocyte deficiencies
55. Hearing
56. Critical Congenital Heart Disease (CCHD)

What should I bring to the hospital to ensure results are processed in a timely manner?
Fill in the blanks, bring this to the hospital and give it to your nurse.

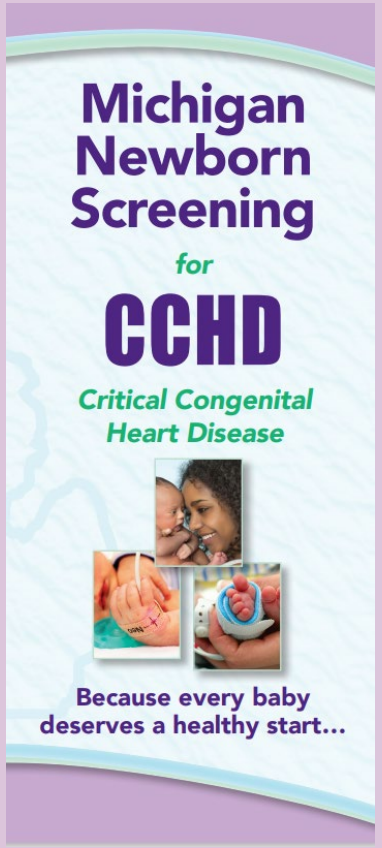
Pediatrician Name: _____
Pediatrician Address: _____
Pediatrician Phone: _____
Best Number to Reach Me: _____

MICHIGAN Newborn Screening

- ✓ Can be ordered through NBS Online Ordering Module in English
- ✓ Greater detail of the 3 parts to the newborn screen: blood spot, heart screening, and hearing screening
- ✓ Lets the parents know exactly what disorders are included on the Michigan NBS panel
- ✓ Has an area where the parents can write down PCP contact information, so they are able to easily share this with the birthing attendant
- ✓ Great for new or expecting parents


Newborn Screening
Information for Expectant Parents
Saving babies through screening because every baby deserves a healthy start…


Michigan Newborn Screening
Saving babies since 1965
Learn about blood spot screening…


Michigan Newborn Screening
for
CCHD
Critical Congenital Heart Disease
Because every baby deserves a healthy start…


After Newborn Screening
Your Baby's Blood Spots
Michigan BioTrust for Health
research for the future
Learn More About the Facts and Choices You Need to Understand

Brochures

Michigan Newborn Screening Saving Babies

Is there anything else I need to do?

ASK Hospital staff or your midwife if newborn screening was done.

BE SURE The hospital or midwife and your baby's healthcare provider have the right phone number and address to reach you.

CHECK With your baby's healthcare provider or midwife about the NBS results.

FOLLOW Directions from your baby's healthcare provider if more tests or medical appointments are needed.

Saving babies since 1965

Would you like to learn more?

Please talk to your baby's healthcare provider or contact us by:

Telephone:
1-866-673-9939 (toll-free)

Fax:
517-335-9419

Email
newbornscreening@michigan.gov

P.O. Box 30195
Lansing, MI 48909

www.michigan.gov/newbornscreening

MDHHS is an equal opportunity employer, services and program provider.

9/17

Michigan Newborn Screening

Saving babies since 1965

Learn about blood spot screening...

Newborn Screening (NBS)?

ogram that screens all babies ours of age for rare but serious

arly. As part of newborn your baby is checked for hearing gns of critical congenital heart few drops of blood are also taken from your baby's heel to fill spots on a filter paper card. The card is sent to the State Newborn Screening Laboratory where blood spots are tested for over 50 different disorders that benefit from early treatment. This pamphlet describes newborn blood spot screening.

My baby seems really healthy. Is NBS still needed?

YES! Whether your baby is born in a hospital, non-hospital setting or at home, screening should be done. Most babies with these disorders seem healthy at birth but can become very sick in a short time. If not treated early, serious health problems, severe developmental delay and even death can occur. NBS is the best way to find nearly all babies with these disorders as early as possible.

What happens if screening suggests a health problem?

The NBS Follow-up Program will alert your baby's healthcare provider. You will get a call about what to do next, but it does not always mean your baby will have a problem. Additional testing may be needed.

What are the disorders?

In Michigan, blood spot screening looks for over 50 disorders that may affect:

- Blood cells
- Brain development
- How the body breaks down nutrients from food
- Lungs and breathing
- Hormones
- How the body fights infection

Congenital hypothyroidism, sickle cell disease, and cystic fibrosis are some of the most common disorders. For a complete list, visit: www.michigan.gov/newbornscreening. NBS may also find some babies who are healthy carriers of these disorders.

What happens if my baby has one of these disorders?

Help is available if your baby is found to have a disorder. Treatment usually begins early and continues through life. Each year, newborn blood spot screening finds about 270 Michigan babies with these medical disorders.

How is the cost of NBS covered?

If your baby is born in a hospital, the cost is part of the hospital charge. If your baby is born in a non-hospital setting, the NBS card may be purchased online at www.michigan.gov/nbsorders or by calling 1-866-673-9939. Some homebirths may qualify for a free screening.

What happens to my baby's blood spots after screening?

All of the blood spots are not always needed for screening. The lab saves one full blood spot for future use by you or your child, if it is ever needed. The remaining blood spots are sent for storage.

Remaining blood spots from newborn screening may be made available for future medical research with a parent's consent. To learn more, please read the Michigan BioTrust for Health pamphlet or visit www.michigan.gov/biotrust.

State law allows you to ask that a second blood spot sample be taken for your safekeeping. If you would like a second sample, please talk to your healthcare provider.

Forms are available if you want your child's blood spots destroyed after newborn screening is complete. Please call 1-866-673-9939 for more information or visit www.michigan.gov/newbornscreening.

- ✓ Can be ordered through our NBS Online Ordering Module in English, Spanish, and Arabic
- ✓ Gives an overview of the newborn blood spot screen
- ✓ Great for new parents

Newborn Screening for CCHD

- ✓ Can be ordered through our NBS Online Ordering Module in English, Spanish, and Arabic
- ✓ Gives a summary of newborn screening for critical congenital heart disease (CCHD)
- ✓ A space is provided for the birthing attendant to write the baby's CCHD screening results.
- ✓ Great for new parents

Newborn Screening – Information for Expectant Parents

Critical Congenital Heart Disease Screening

Pulse oximetry screening can help find babies with critical congenital heart disease (CCHD), which occurs when a baby's heart or major blood vessels do not form properly. A "critical" heart defect is one that needs urgent treatment in order to have the best outcome for the baby. Treatment can include medical and surgical procedures.

Babies are screened for CCHD around 24 hours of age. Pulse oximetry is fast, easy and does not hurt. A small sensor is placed on a baby's right hand and one foot to measure the oxygen level in the blood.

Most babies are born with healthy hearts, and have enough oxygen in their blood. Although babies are checked very carefully by a doctor after they are born, some babies with CCHD may not have symptoms until later. Screening can help find babies with CCHD shortly after birth, before they have any serious complications.

For more information on CCHD screening, visit Michigan.gov/CCHD.

What parents can do after their baby is born

ASK Hospital staff or your midwife if:
- Blood spots were collected
- Pulse oximetry screening was done
- Hearing screening was done
- BioTrust decision was recorded

BE SURE Your current contact information is on file at the hospital so you can be reached if needed.

CHECK With your baby's health care provider or midwife about NBS results.

FOLLOW-UP With your baby's provider if more testing or appointments are needed.

Telephone: 866-673-9939 (toll-free)
Fax: 517-335-9419
Email: NewbornScreening@Michigan.gov

PO Box 30195 • Lansing, MI 48909
Michigan.gov/NewbornScreening

MDHHS

The Michigan Department of Health and Human Services will not exclude from participation in, deny benefits of, or discriminate against any individual or group because of race, sex, religion, age, national origin, color, height, weight, marital status, gender identification or expression, sexual orientation, partisan considerations, or a disability or genetic information that is unrelated to the person's eligibility.

MDHHS-PUB-1232 (Rev. 4-21)

Newborn Screening

Information for Expectant Parents

Saving babies through screening because every baby deserves a healthy start…

…ning?

…orders may …become very

…reened for over 50 disorders in three ways:

1. Blood spot screening
2. Hearing screening
3. Critical congenital heart disease (CCHD) screening

Whether your baby is born in the hospital or at home, newborn screening should be done. If not found early, many of the disorders can cause serious and permanent health problems. To learn more about newborn screening, visit Michigan.gov/NewbornScreening.

Newborn screening can be life-saving!

Blood Spot Screening

Newborn blood spot screening finds babies with diseases such as phenylketonuria (PKU), cystic fibrosis, and sickle cell disease to ensure early treatment.

A few drops of blood are taken from a baby's heel to fill five or six small spots on a card. The card is then sent to the State Newborn Screening Laboratory for testing.

In Michigan blood spot screening looks for more than 50 conditions that may affect:

- Blood cells
- Brain development
- How the body breaks down food
- Immune system
- Lungs and breathing

BioTrust for Health

The BioTrust is a program to oversee blood spots that remain after screening; any unused whole and partial spots are stored up to 100 years at a secure site. Stored spots can be used for approved research to better understand diseases or improve health. Parents get more details after delivery and can choose whether to allow their baby's blood spots to be used for future research. Be sure to mark your decision on the BioTrust consent form after your baby is born. For more information, visit Michigan.gov/BioTrust.

Hearing Screening

Hearing screening can detect hearing loss present at birth and is done before your baby leaves the hospital, or a few days after birth if born at home. This screen is mandated because hearing loss is invisible and babies with hearing loss look and behave just like other babies. Ninety-four percent of babies with hearing loss have two hearing parents.

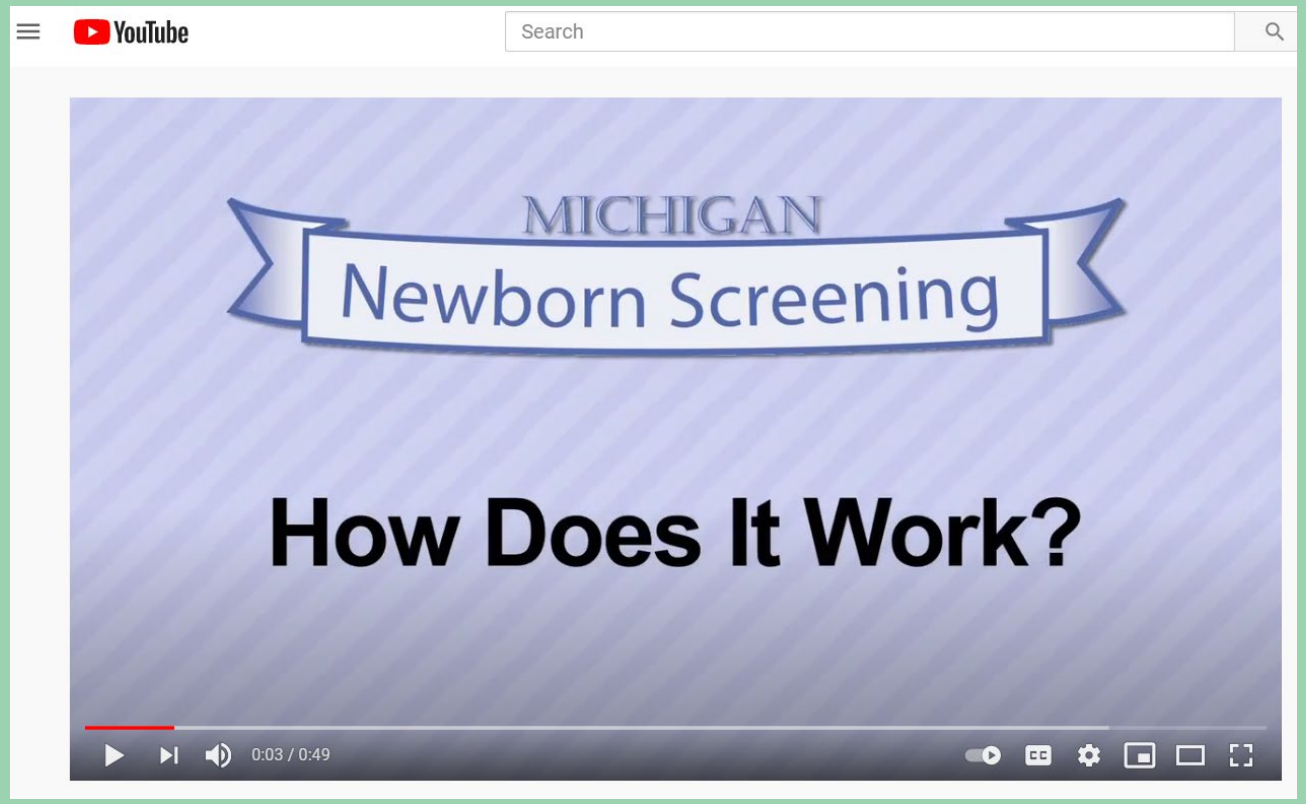
Hearing screening is not painful in any way to your baby and is best done when a baby is quiet or sleeping. The test is quick, simple and safe. Using a specialized machine, the test takes a few minutes and checks for the baby's ability to hear sounds within the speech range.

Babies are born ready to learn about the world around them. Each of the five senses contributes to a baby's knowledge. Any change in sense, including hearing, will impact how a baby learns. Hearing is important for speech and language development. If hearing loss is found, help can be started right away for your baby.

For more information about the Michigan Early Hearing Detection and Intervention Program, visit Michigan.gov/EHDI.

- ✓ Can be ordered through our NBS Online Ordering Module in English, Spanish, and Arabic
- ✓ Gives a quick summary of the newborn screen, including the two point of care screens
- ✓ Has a summary of the BioTrust Program
- ✓ Great for prenatal education

NBS YouTube Video



https://www.youtube.com/watch?v=c1w2Vow9UUo

- ✓ Can be viewed using the link above
- ✓ Gives an animated overview of the newborn screen
- ✓ Great for new and expecting parents

BioTrust YouTube Video

https://youtu.be/UMcvVskMaiY

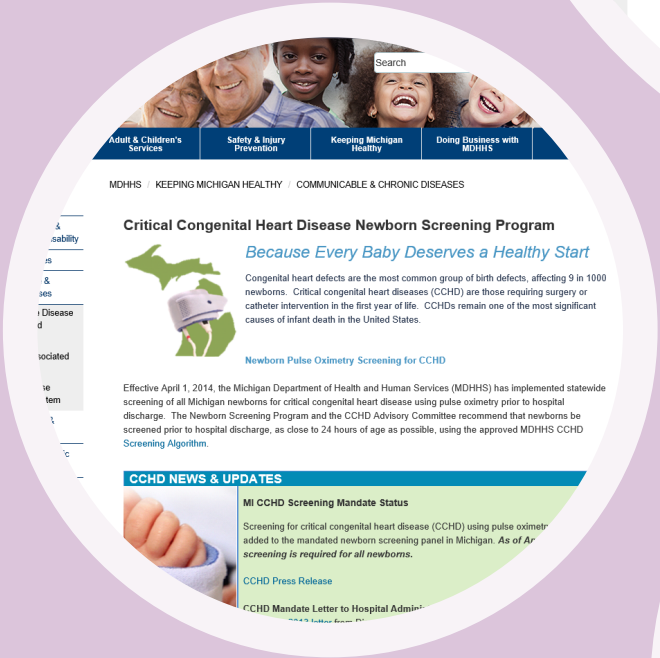
- ✓ Can be viewed using the link above
- ✓ Gives an animated overview of the BioTrust and parents options for DBS after screening
- ✓ Great for new and expecting parents

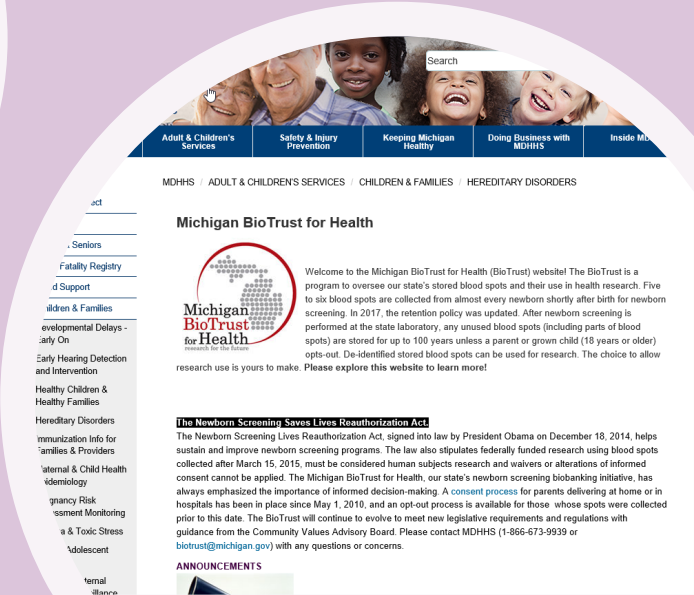
Our Websites

Michigan.gov/newbornscreening

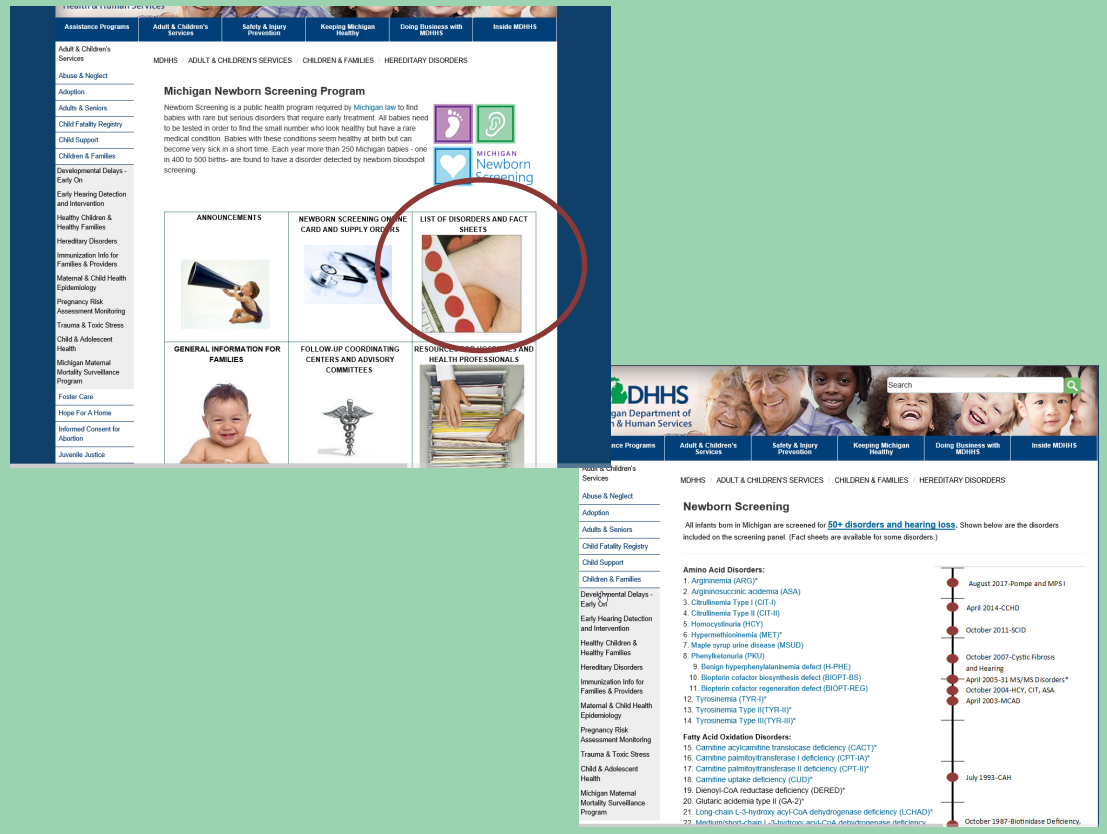
Michigan.gov/CCHD

Michigan.gov/biotrust





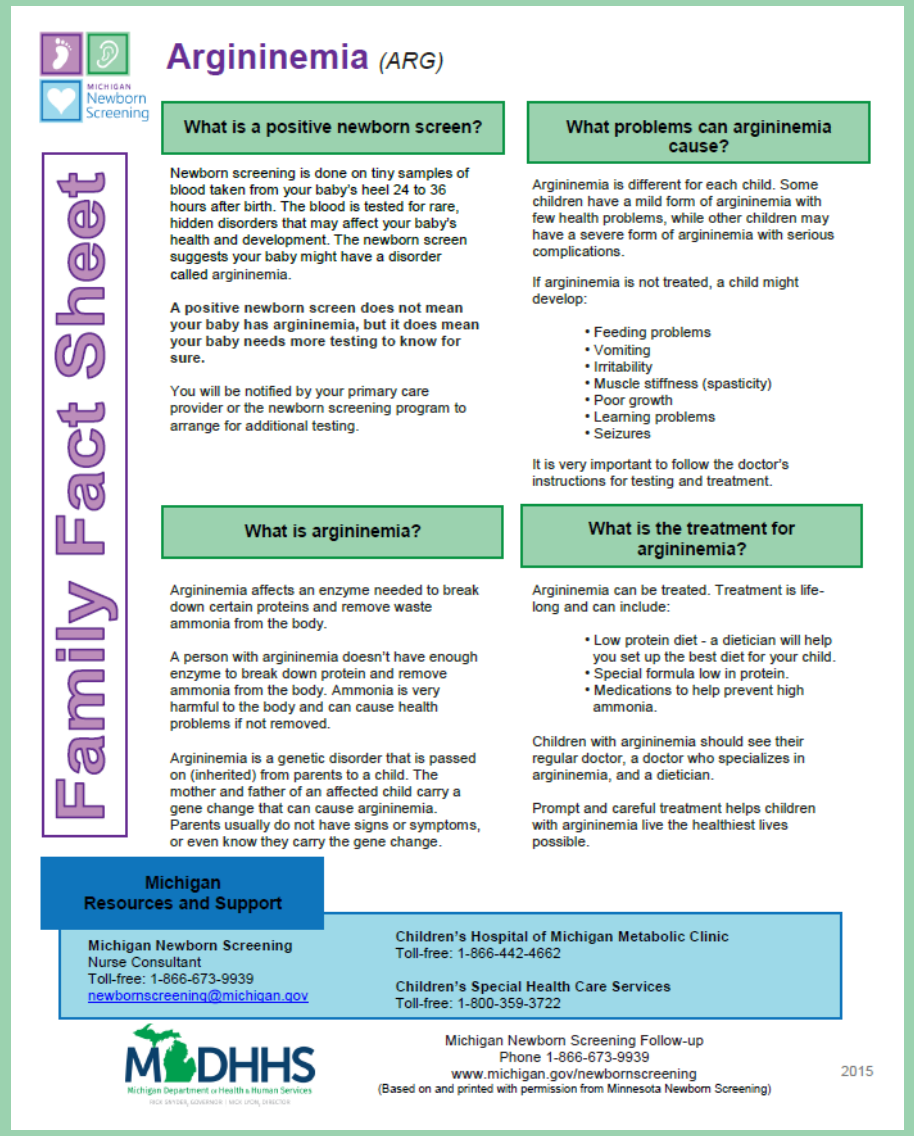
List of Disorders



- ✓ A list of the disorders included on the Michigan NBS panel can be found on our website Michigan.gov/NewbornScreening
- ✓ Includes a timeline of when disorders were added to the panel
- ✓ Most disorders have a fact sheet for families with an explanation of what a positive screen means and a summary of the disorder.

Family Fact Sheets

- ✓ Great for families who have an infant with a positive newborn screen
- ✓ Can be printed from our website
- ✓ Can be downloaded as a PDF for electronic distribution
- ✓ ADA Compliant



MICHIGAN Newborn Screening

Family Fact Sheet

Argininemia *(ARG)*

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 36 hours after birth. The blood is tested for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called argininemia.

A positive newborn screen does not mean your baby has argininemia, but it does mean your baby needs more testing to know for sure.

You will be notified by your primary care provider or the newborn screening program to arrange for additional testing.

What problems can argininemia cause?

Argininemia is different for each child. Some children have a mild form of argininemia with few health problems, while other children may have a severe form of argininemia with serious complications.

If argininemia is not treated, a child might develop:

- Feeding problems
- Vomiting
- Irritability
- Muscle stiffness (spasticity)
- Poor growth
- Learning problems
- Seizures

It is very important to follow the doctor's instructions for testing and treatment.

What is argininemia?

Argininemia affects an enzyme needed to break down certain proteins and remove waste ammonia from the body.

A person with argininemia doesn't have enough enzyme to break down protein and remove ammonia from the body. Ammonia is very harmful to the body and can cause health problems if not removed.

Argininemia is a genetic disorder that is passed on (inherited) from parents to a child. The mother and father of an affected child carry a gene change that can cause argininemia. Parents usually do not have signs or symptoms, or even know they carry the gene change.

What is the treatment for argininemia?

Argininemia can be treated. Treatment is life-long and can include:

- Low protein diet - a dietician will help you set up the best diet for your child.
- Special formula low in protein.
- Medications to help prevent high ammonia.

Children with argininemia should see their regular doctor, a doctor who specializes in argininemia, and a dietician.

Prompt and careful treatment helps children with argininemia live the healthiest lives possible.

Michigan Resources and Support

Michigan Newborn Screening
Nurse Consultant
Toll-free: 1-866-673-9939
newbornscreening@michigan.gov

Children's Hospital of Michigan Metabolic Clinic
Toll-free: 1-866-442-4662

Children's Special Health Care Services
Toll-free: 1-800-359-3722

MDHHS
Michigan Department of Health & Human Services

Michigan Newborn Screening Follow-up
Phone 1-866-673-9939
www.michigan.gov/newbornscreening
(Based on and printed with permission from Minnesota Newborn Screening)

2015

Michigan Department of Health and Human Services Virtual Baby Fair

REGISTER NOW!

Michigan Department of Health and Human Services

Virtual Baby Fair

Are you a new or expecting parent? Join us for a virtual baby fair to learn how to be better prepared for your best adventure yet! Learn about resources available to you, important information, and ask questions to experts.

Tuesday, December 6, 2022, from 9:00-11:00 a.m.

Monday, December 12, 2022, from 9:00-11:00 a.m.

Wednesday, December 14, 2022, from 6:30-8:30 p.m.

Register at

https://www.eventbrite.com/e/virtual-baby-fair-tickets-446197848777

Participating MDHHS Programs:

- Newborn Screening
- BioTrust for Health
- Infant Safe Sleep
- Childhood Lead Poisoning Prevention Program
- Women, Infants, & Children (WIC)
- Vital Records
- Unintentional Injury Prevention
- Immunizations
- Early Hearing Detection and Intervention
- Maternal Infant Health Program
- Michigan Home Visiting Initiative
- Birth Defects Education and Outreach
- Breastfeeding Support
- Children's Special Healthcare Services
- Eat Safe Fish

MDHHS Michigan Department of Health & Human Services

Scan here to register!

For more information please contact Kristen Thompson

ThompsonK23@michigan.gov

517-284-4992

- Department wide educational initiative
- Free to registrants
- Good for new or expecting parents, or anyone who wants to know more about resources available to new or expecting parents.

After Newborn Screening (NBS): Engaging families in the Michigan BioTrust for Health

Shelby Atkinson, MPH

Genomics and Newborn Screening Research Coordinator

Michigan Department of Health and Human Services (MDHHS)

Michigan BioTrust for Health
research for the future

Presentation Objectives:

- Provide overview of the BioTrust for Health program
- Provide relevant details to share with expectant resources about:
  - The BioTrust for Health Program
  - Choices families can make for blood spots after newborn screening
- Provide resources for parents and providers

What happens to DBS after NBS?

- 5-6 blood spots are collected to ensure that enough specimen is available to complete all tests included in the screen
- Most specimens are normal and whole or parts of blood spots are left over
- All residual DBS are stored for *up to 100 years*
- Current archive of DBS dates back to 1987
- DBS are stored at the Michigan Neonatal Biobank, located at Wayne State University

How are left over DBS used?

1

MDHHS laboratory use for NBS QA, test improvement, and test development

2

Family directed use. Examples include future genetic testing and to diagnose an untimely death

3

De-identified medical and public health research through the BioTrust

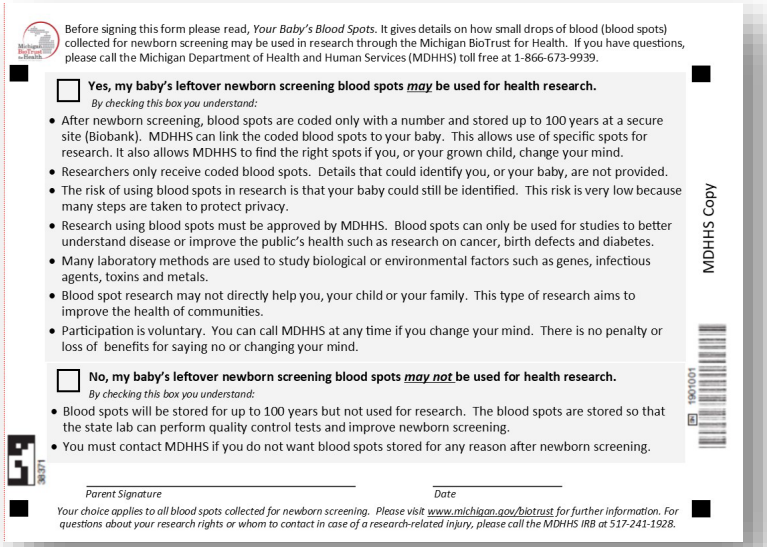
Michigan BioTrust for Health

- The BioTrust is an MDHHS program created in 2009 that oversees the research of blood spots left over after newborn screening (NBS)

- The goals of the BioTrust are:
  1. To make blood spots leftover after NBS more useful in medical and public health research
  2. To store blood spots to better preserve the samples
  3. To use blood spots in a manner acceptable to the public
  4. **To inform the public and allow personal decision making**

BioTrust Consent Process

- Statewide prospective consent process implemented in May of 2010
- Executed by birthing attendants in both the hospital and home birth settings
- Allows families to choose if their child's residual specimens can be used in de-identified research studies



Before signing this form please read, *Your Baby's Blood Spots*. It gives details on how small drops of blood (blood spots) collected for newborn screening may be used in research through the Michigan BioTrust for Health. If you have questions, please call the Michigan Department of Health and Human Services (MDHHS) toll free at 1-866-673-9939.

☐ **Yes, my baby's leftover newborn screening blood spots *may* be used for health research.**

By checking this box you understand:

- After newborn screening, blood spots are coded only with a number and stored up to 100 years at a secure site (Biobank). MDHHS can link the coded blood spots to your baby. This allows use of specific spots for research. It also allows MDHHS to find the right spots if you, or your grown child, change your mind.
- Researchers only receive coded blood spots. Details that could identify you, or your baby, are not provided.
- The risk of using blood spots in research is that your baby could still be identified. This risk is very low because many steps are taken to protect privacy.
- Research using blood spots must be approved by MDHHS. Blood spots can only be used for studies to better understand disease or improve the public's health such as research on cancer, birth defects and diabetes.
- Many laboratory methods are used to study biological or environmental factors such as genes, infectious agents, toxins and metals.
- Blood spot research may not directly help you, your child or your family. This type of research aims to improve the health of communities.
- Participation is voluntary. You can call MDHHS at any time if you change your mind. There is no penalty or loss of benefits for saying no or changing your mind.

MDHHS Copy

☐ **No, my baby's leftover newborn screening blood spots *may not* be used for health research.**

By checking this box you understand:

- Blood spots will be stored for up to 100 years but not used for research. The blood spots are stored so that the state lab can perform quality control tests and improve newborn screening.
- You must contact MDHHS if you do not want blood spots stored for any reason after newborn screening.

3571

Parent Signature *Date*

Your choice applies to all blood spots collected for newborn screening. Please visit www.michigan.gov/biotrust for further information. For questions about your research rights or whom to contact in case of a research-related injury, please call the MDHHS IRB at 517-241-1928.

Michigan BioTrust for Health
research for the future

Why use DBS in health-related research?

- May lead to new screening tests
- May provide clues about factors that impact health or cause diseases not only in Michigan but worldwide
  - Contain genetic information and other biological markers
  - May contain evidence of exposure to environmental agents such as infections, toxins or chemicals
- Can conduct studies of large numbers of people w/o collecting new samples
- DBS samples are no longer needed for clinical purposes
- Because samples have been stored since 1987, multigenerational research is possible

What type of research is done?

- The BioTrust Community Values Advisory Board (CVAB) reviews and formulates policies about the acceptable use of blood spots in research.
  - *MDHHS Policy 114: Guidelines for Research Use of Dried Blood Spots*
- Research must be related to improving the public's health and priorities include prenatal, childhood and adult-onset disorders and environmental exposures.
- Under the BioTrust consent, DBS cannot be used for:
  - *Whole genome or whole exome sequencing*
  - *Chemical, biological or nuclear warfare*
  - *Cosmetics or non-health related ventures*

What type of research is done?

- All studies must be approved by two MDHHS boards:
  - MDHHS Institutional Review Board
  - BioTrust Scientific Advisory Board

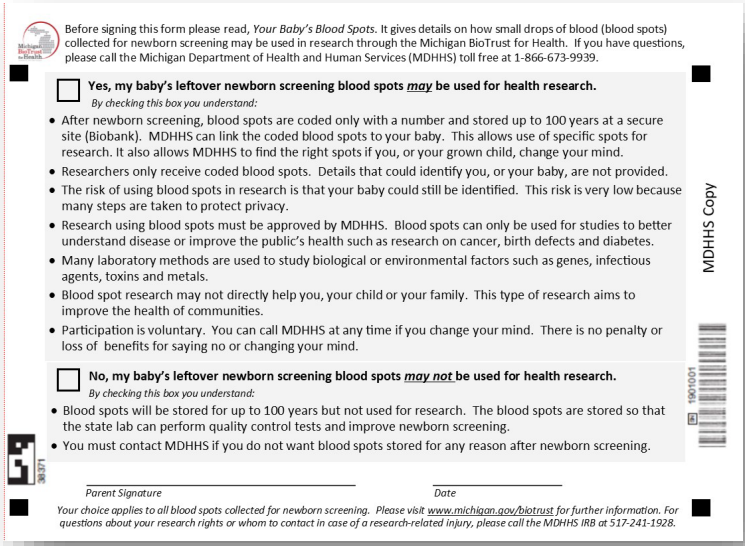
- All projects covered by the BioTrust consent process are de-identified and researchers do not know whose blood spots are being used.

- Since 2010, **77** research projects have been approved to use leftover blood spots through the BioTrust

Consent Options

Around the time of newborn screening, families receive the BioTrust consent form and check either the “Yes” or “No” box:

- By checking “Yes”:
  - Blood spots left over after newborn screening will be available for research studies
- By checking “No”:
  - Blood spots will be stored but will not be used for research studies.



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- You must contact MDHHS if you do not want blood spots stored for any reason after newborn screening.

_____ _____

Parent Signature *Date*

Your choice applies to all blood spots collected for newborn screening. Please visit www.michigan.gov/biotrust for further information. For questions about your research rights or whom to contact in case of a research-related injury, please call the MDHHS IRB at 517-241-1928.

Families can contact MDHHS if they do not want blood spots stored for any reason after newborn screening is done.

Michigan BioTrust for Health
research for the future

BioTrust Prenatal Education

- Goal: Inform parents about the BioTrust program and the consent decision they will be asked to make before they arrive to the hospital for delivery.

- Parents who learn about the program during pregnancy may have more time to explore BioTrust resources and may be more prepared to document their consent choice.

Michigan BioTrust for Health
research for the future

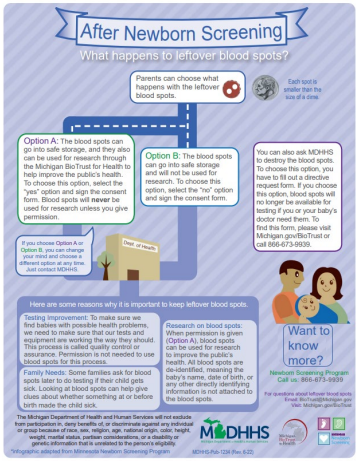
BioTrust Prenatal Education

- MDHHS is actively working to engage partners who work with families during pregnancy to distribute BioTrust information. Potential partners include:
  - Hospitals providing prenatal classes or tours
  - OB/GYN offices
  - Midwife practices
  - Local health departments
  - And you!

Michigan BioTrust for Health
research for the future

BioTrust Prenatal Education

- Brochures and infographics designed for the prenatal period are available to order online, free of charge
  - Available in English, Spanish and Arabic



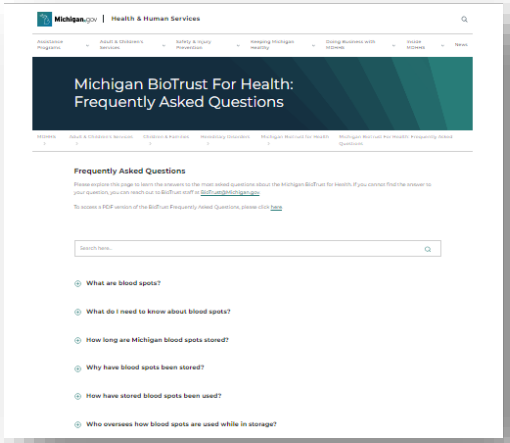
Michigan BioTrust for Health
research for the future

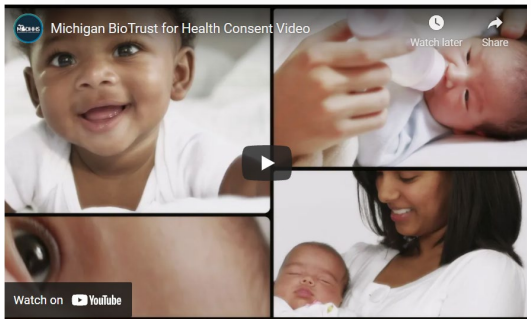
Additional Resources for Parents

Website: Michigan.gov/BioTrust

Resources include:

- A 6-minute informational video
- Frequently asked questions page
- BioTrust research report
- Page dedicated to consent options
- Contact information for BioTrust staff





Michigan BioTrust for Health
research for the future

Key Messages for Parents

Before delivery:

- Newborn screening is required by law because of its important health benefits. **Participation in the BioTrust is optional!**

- Parents will receive a brochure and the BioTrust consent form from their birthing team around the time that blood spots are collected for newborn screening. This form allows them to document if their child's left over blood spots can be used for research.

- No extra blood is collected for the BioTrust. If parents say "yes" to the BioTrust consent, researchers can only use what may be left over after screening. Researchers do not receive identifying information on the parent or child.

- Parents can learn more about the BioTrust program before giving birth at www.Michigan.gov/BioTrust

Michigan BioTrust for Health
research for the future

Messages for Parents

After delivery:

- Regardless of BioTrust consent decision, all left over DBS are stored for up to 100 years. Parents can request that leftover DBS be destroyed by contacting MDHHS.

- Parents can change their BioTrust decision at any time by contacting MDHHS.

- Program Contact information:
  - Email address: BioTrust@Michigan.gov or newbornscreening@Michigan.gov
  - Phone number: 517-335-4181
  - Fax: 517-335-9419 or 517-335-9739

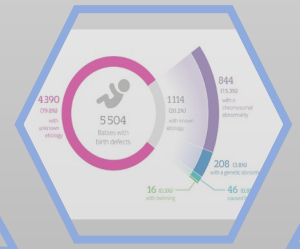
Michigan BioTrust for Health
research for the future

THANK YOU!

Shelby Atkinson, MPH
Genomics and Newborn Screening Research Coordinator
atkinsons2@michigan.gov
517-335-6497

BIRTH DEFECTS
EDUCATION & OUTREACH

MICHIGAN DEPARTMENT OF HEALTH AND HUMAN SERVICES



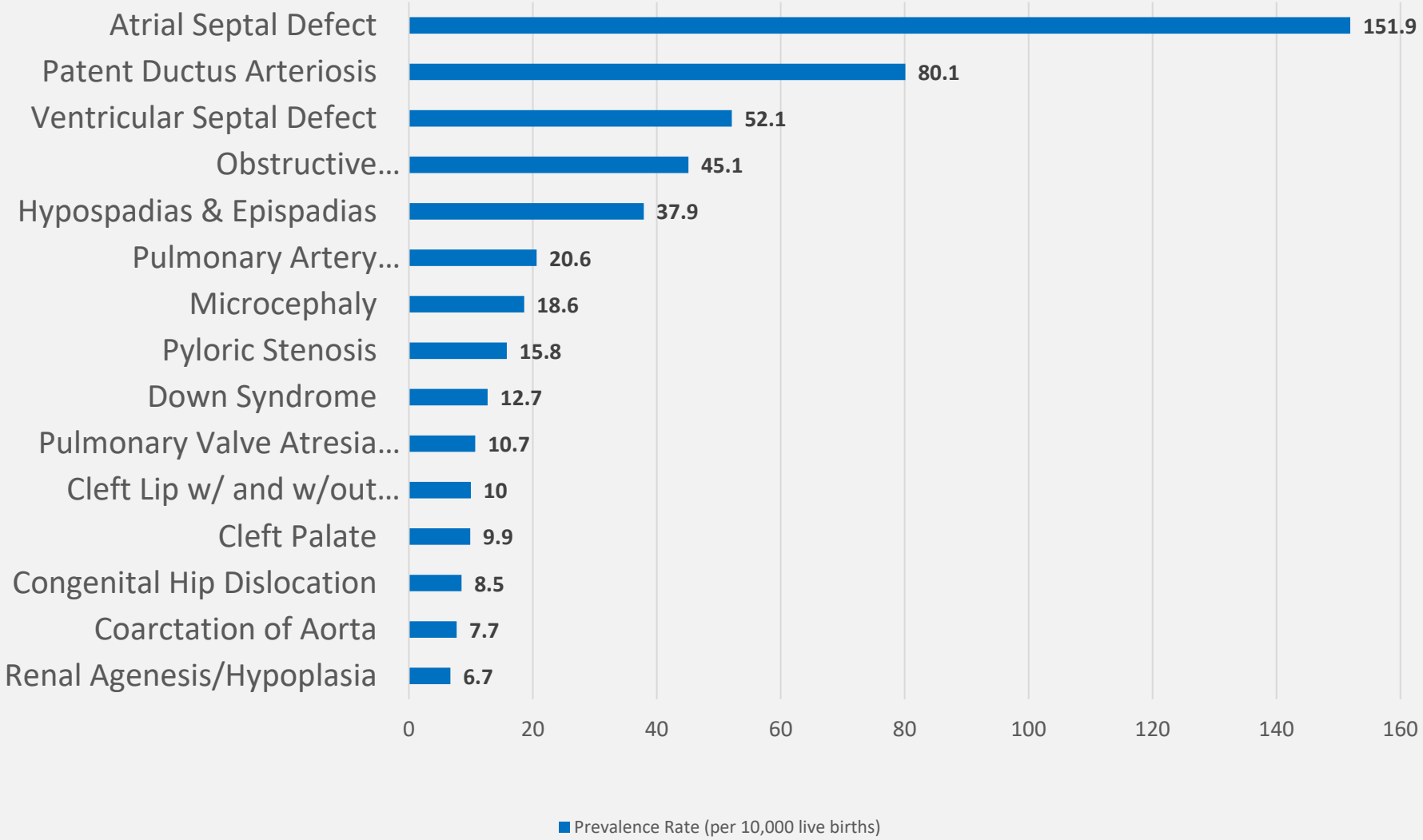
Michigan Birth Defects Registry (MBDR)

- By law, hospitals and medical labs report certain health conditions affecting Michigan born babies up to 2 years of age.
- MBDR information is confidential. It is used to:
  - Calculate baseline rates
  - Analyze trends
  - Identify and respond to public health concerns
- MBDR data helps us understand health and developmental needs, identify causes, and share prevention strategies.
- Birth Defects Reporting Forms (michigan.gov)

Public Health Impact of Birth Defects in Michigan

- Birth defects have a serious impact on the health of children in Michigan and the entire United States.
  - 15,561 children with birth defects reported to the MBDR in 2019.
  - This corresponds to approximately 14.4% of Michigan newborns.
- Birth defects contribute significantly to infant and child mortality.
  - The case fatality rate for children born in 2019 with a birth defect was 13.2 deaths per 1,000 births. This compares to an infant death rate of 6.4 per 1,000 live births for all infants born in Michigan during the same year.
- Birth defects also significantly contribute to infant and child morbidity and long-term disability.

Most Prevalent Birth Defects



- [Birth Defects & Early Hearing Detection and Intervention (EHDI) (michigan.gov)]

Birth Defects Prosperity Region Factsheets

Birth Defects Profile in Michigan Prosperity Region 1

(Alger, Baraga, Chippewa, Delta, Dickinson, Gogebic, Houghton, Iron, Keweenaw, Luce, Mackinac, Marquette, Menominee, Ontonagon, and Schoolcraft counties)

Michigan Birth Defects Registry (MBDR)

- By law, hospitals and medical labs report certain health conditions affecting children at birth or up to 2 years of age.
- MBDR information is confidential. It is used to:
  - Calculate baseline rates.
  - Analyze trends.
  - Identify and respond to public health concerns.
- MBDR data helps us understand health and development needs, identify causes, and share prevention strategies.

Birth Defects in Region 1 (2013-2019)

- An average of 2,708 infants were born in Region 1 each year.
- Approximately 225 of these infants were born with a birth defect each year.
- Hypospadias and Trisomy 21 were the most prevalent birth defects in Michigan and Prosperity Region 1 (Table 1).
- The prevalence of hypospadias is **lower** in Prosperity Region 1 [21.6 (95% Confidence Interval (CI): 15.0-28.2)] than Michigan as a whole [38.9 (95% CI: 37.6-40.4)].
- The prevalence of all other birth defects was similar between Prosperity Region 1 and Michigan as a whole.

Table 1. Prevalence of Selected Birth Defects within Prosperity Region 1 and the State of Michigan, 2013-2019

	Prosperity Region 1		Michigan	
Birth Defect	**Number of Cases**	**Prevalence (per 10,000 live births)**	**Number of Cases**	**Prevalence (per 10,000 live births)**
Anencephaly	--*	--*	69	0.9
Cleft Lip with Cleft Palate	16	8.4	517	6.6
Cleft Lip without Cleft Palate	--*	--*	264	3.4
Cleft Palate without Cleft Lip	10	5.3	541	6.9
Gastroschisis	11	5.8	321	4.1
Hypoplastic Left Heart Syndrome	7	3.7	308	3.9
Hypospadias	41	21.6	3052	39.0
Limb Deficiencies	7	3.7	308	3.9
Spina Bifida	9	4.7	326	4.2
Tetralogy of Fallot	--*	--*	406	5.2
Transposition of the Great Arteries	--*	--*	298	3.8
Trisomy 21	20	10.5	1010	12.9
Total Number of Live Births	18,958		782,227	

* Data suppressed due to small birth defects counts (1 ≤ N ≤ 5),

MBDR Data Limitations and Cautions

- Data are based on passive reporting, which means MBDR relies on facilities to identify and report cases of birth defects. Getting all facilities to report complete and timely data can be a challenge.
- Some facilities report children with a birth defect that is later "ruled out" resulting in an overcount of the actual number of cases.
- Children diagnosed and treated in facilities in other states may be missed, which will significantly affect the completeness of data for Michigan's border counties.

Birth Characteristics among all Mothers and Babies

Between 2013 and 2019, roughly **2,708** babies were born in Prosperity Region 1 each year.

During an average year in Prosperity Region 1:

Mother's Race and Ethnicity

- **2,300**, or **85 percent** of, infants were born to **white, non-Hispanic** mothers.
- **10**, or **less than 1 percent** of, infants were born to **Black, non-Hispanic** mothers.
- **340**, or **13 percent** of, infants were born to **all other non-Hispanic** mothers.
- **50**, or **2 percent** of, infants were born to **Hispanic** mothers.

Mom's Socioeconomic and Educational Status

- **1,280**, or **47 percent** of, all births reported **Medicaid as the intended payer**.
- **210**, or **8 percent**, of infants were born to **mothers with less than a high school education**.
- **670**, or **25 percent** of, infants were born to **mothers who had a high school education**.
- **1,080**, or **40 percent** of, infants were born to **mothers who had some college or an associate degree**.
- **700**, or **26 percent** of, infants were born to **mothers who had a college degree**.

Mom's Age

- **150**, or **5 percent** of, infants were born to **teenage** mothers (15 to 19 years).
- **2,300**, or **81 percent** of, infants were born to mothers **20 to 34 years of age**.
- **330**, or **12 percent** of, infants were born to mothers **35 years of age** or older.

Data source: Michigan resident live birth files, MDHHS, Division of Vital Records and Health Statistics, 2013-2019.

Mom and Baby Health Before, During, and After Pregnancy

- **1,470**, or **54 percent** of, infants were born to mothers who were **overweight** or **obese**.
- **750**, or **28 percent** of, infants were born to mothers who **used tobacco** during pregnancy.
- **280**, or **10 percent** of, infants were **born prematurely**.
- **180**, or **7 percent** of, infants were born at a **low birth weight**.
- **20**, or **less than 1 percent** of, infants were born to mothers who had **pre-pregnancy diabetes type 1 or type 2**.

MBDR MDHHS Michigan Department of Health & Human Services

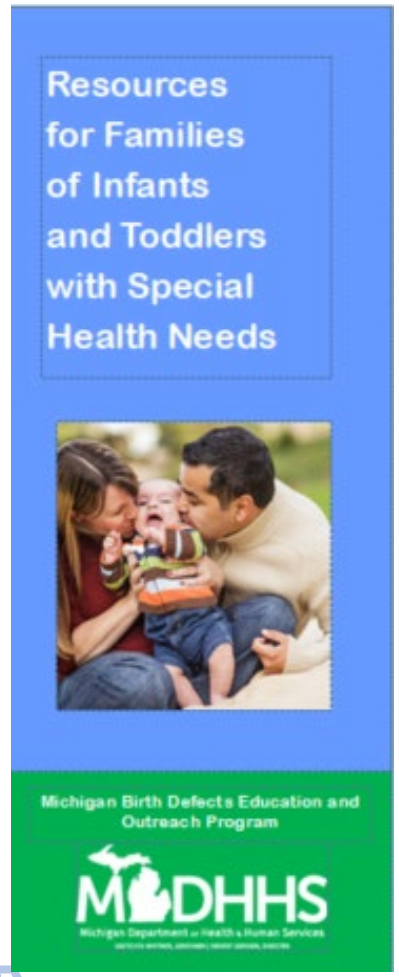
Birth Defects Education & Outreach (BDEO)

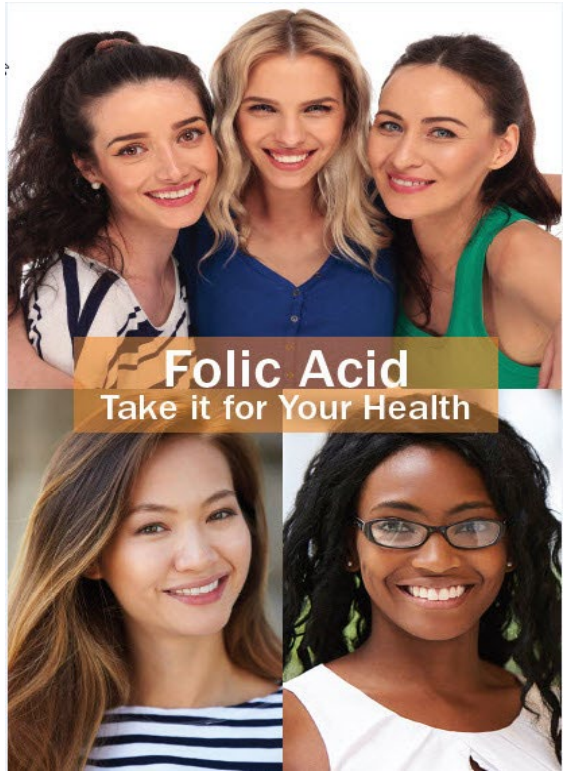
- Assess, develop, and disseminate MBDR surveillance data and education materials reflective of at-risk populations.
- Collaborate with partners to implement birth defects prevention initiatives with an aim to increase referrals.
- Inform our work through direct outreach to families.
- Birth Defects Prevention (michigan.gov)

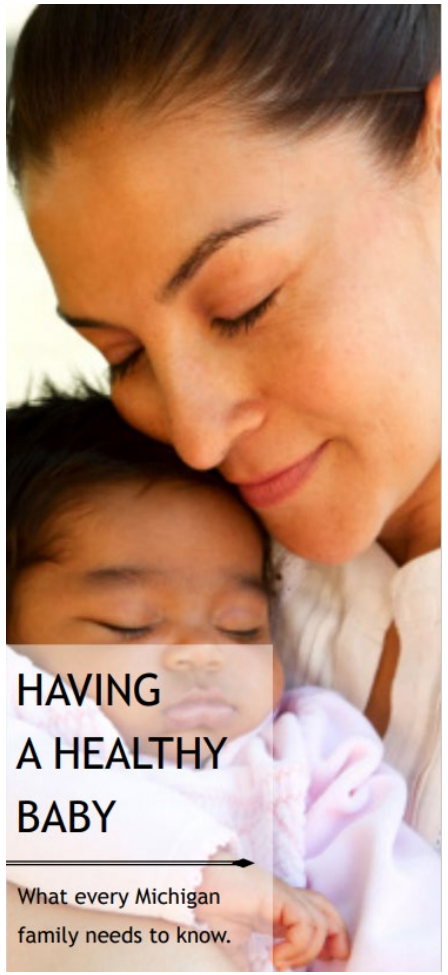
Not all birth defects can be prevented. We may not have total control over behavioral, environmental, health and hereditary factors that contribute to birth defects. But we also know that women can increase their chances of having a healthy baby by managing health conditions and by adopting healthy behaviors before and during pregnancy.

A birth defect may affect the health or development of a child and require special medical care. Resources are available. Early diagnosis can help babies and children get the health care and services they need to live their best life.

Birth Defects Education and Outreach Pamphlets
English, Spanish and Arabic



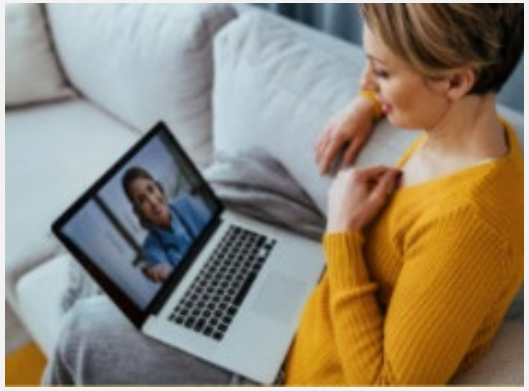




https://migrc.org/order-materials/

Birth Defects Prevention Focus Areas

- Increasing uptake of folic acid
- Promoting preconception health
- Visiting a healthcare provider
- Caring for the body and mind
- Avoiding harmful substance



Folic Acid Awareness Week

- FAW is recognized the second full week in September.
- Celebrate the success of folic acid supplementation and fortification in preventing serious birth defects of the spine and brain.
- Promote folic acid to be taken BEFORE and DURING pregnancy.
- National Folic Acid Week resources

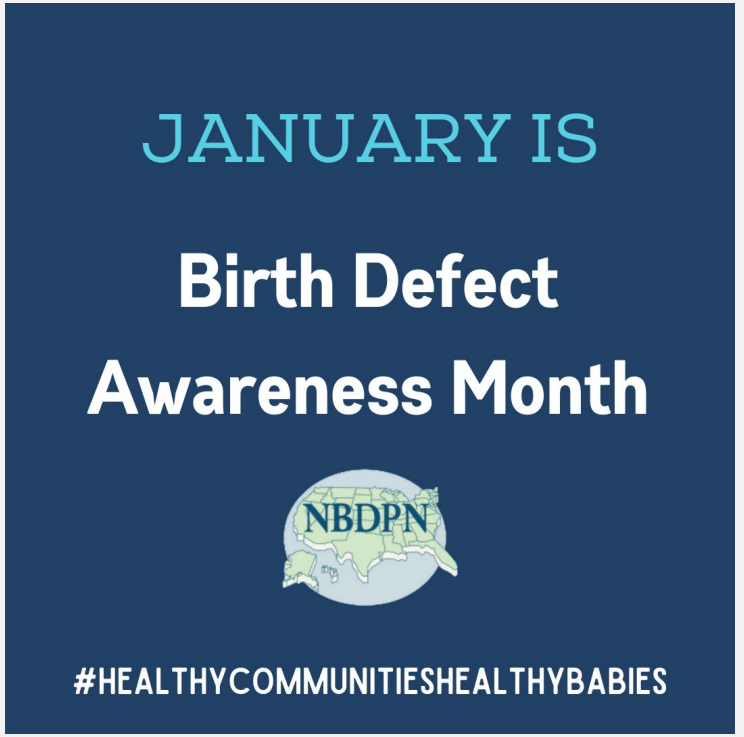
CDC Resources

- Folic Acid
- General Information about NTDs, Folic Acid, and Folate
- MTHFR Gene, Folic Acid, and Preventing Neural Tube Defects
- Free Materials & Multimedia

Birth Defects Awareness Month

- 5 Prevention Tips for Healthy Communities and Healthy Babies
- National Birth Defects Awareness Month Tips Resource Map
- Social Media, Press Release, and Proclamation templates available at

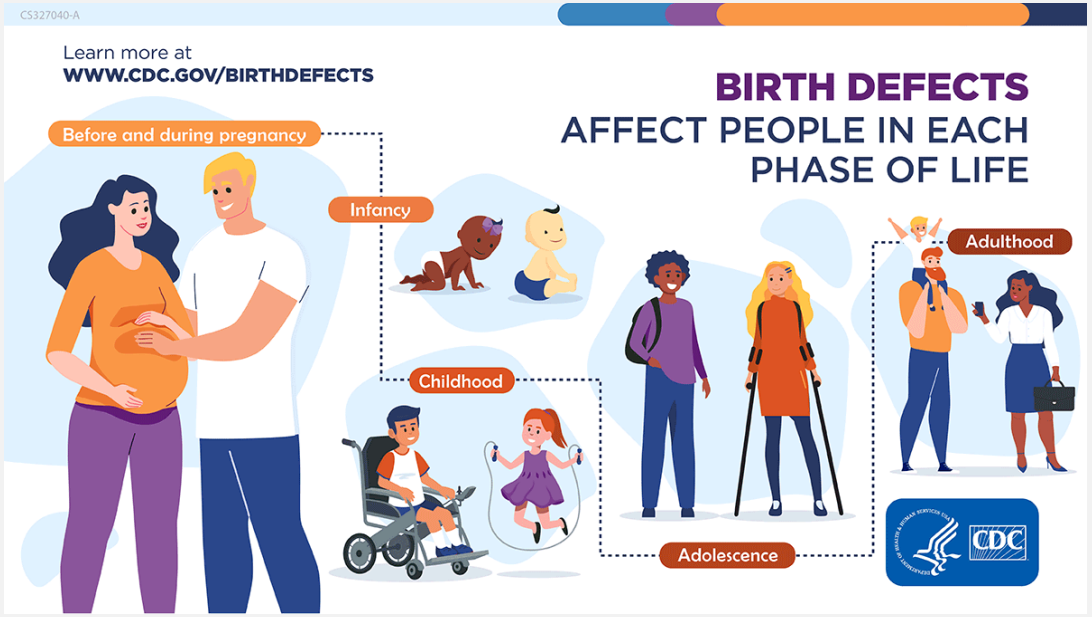
  https://www.nbdpn.org/bdam.php



Birth Defects Across the Lifespan

CDC toolkit describes some of what scientists have learned:

- Birth defects prevention **before and during pregnancy**
- Survival and health of **infants** with birth defects
- Educational needs and special healthcare needs for **children** with birth defects
- Transition from pediatric to adult medical care for **adolescents**
- Planning for pregnancy, recurrence prevention, and genetics and family history for **adults** with birth defects
- [National Birth Defects Awareness Month | CDC]
- [Learn More about Birth Defects | CDC]



Thank you!

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