Oregon's Newborn Bloodspot Screening Program

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Newborn Screening in Oregon

Three screenings are offered to all newborns born in Oregon.

- Critical Congenital Heart Disease (CCHD)
- Hearing screening
- Newborn bloodspot screening (NBS)

What is Newborn Bloodspot Screening?

Newborn bloodspot screening is a **state public health program** that identifies infants with **treatable disorders**, which may otherwise go **unrecognized**, to avoid or **prevent adverse outcomes**.

"No child should die or suffer disabilities if a simple blood spot can prevent it."

Robert Guthrie, PhD, MD, 1916-1995
Developer of the first newborn screening test
(the Guthrie test for PKU)

Newborn Bloodspot Screening Statistics

- **97% of the nearly four million** newborns born in the United States each year are screened

https://www.newsteps.org/about-newsteps

- Saves or improves the lives of over **12,000 babies** in the United States each year

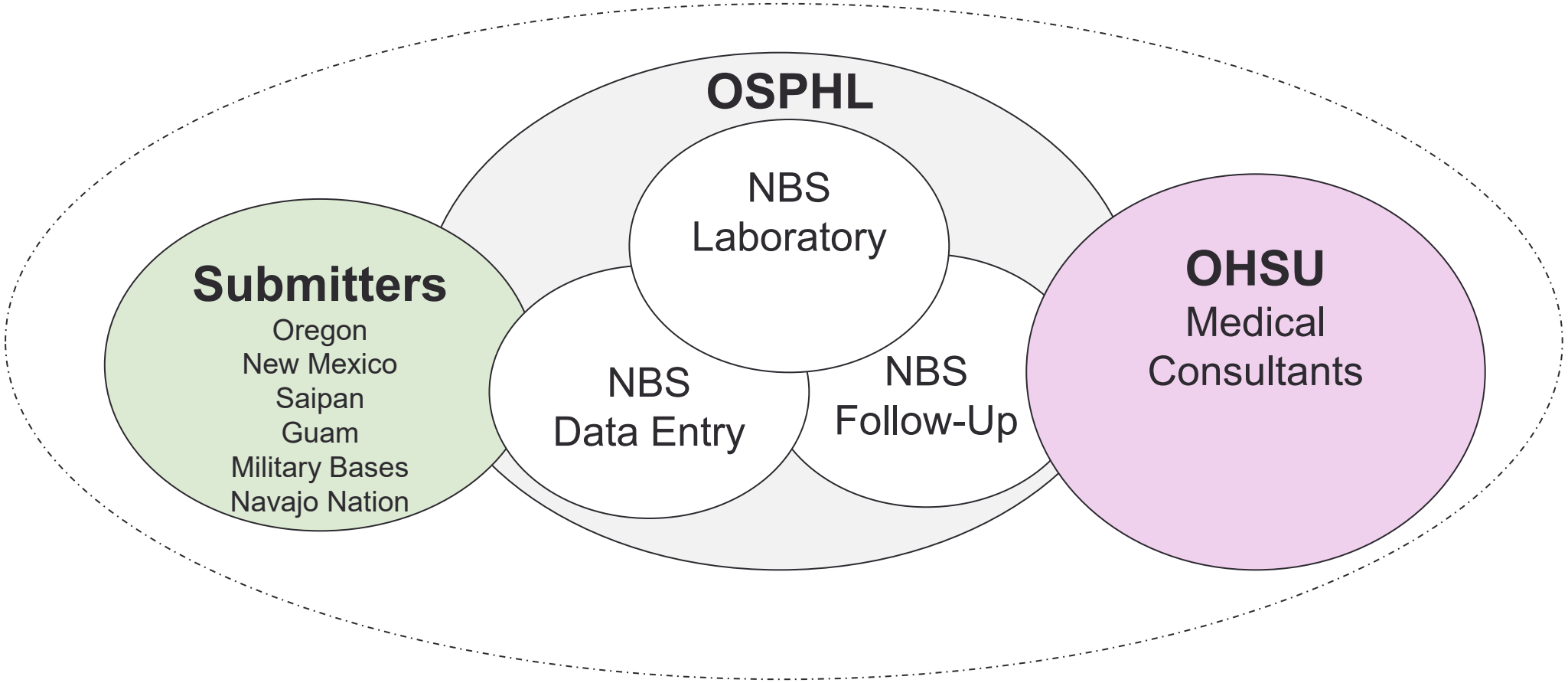
https://www.newsteps.org/about-newsteps

- In Oregon, approximately **40,000 babies are screened** each year and **more than 100** are diagnosed and treated for one of the disorders on the newborn screening panel.

Why Newborn Bloodspot Screening is Important:

- Affects one in 500 newborns in Oregon.
- Approximately 20 disorders can kill or severely harm a baby if untreated in the first two weeks of life.
- Oregon is a 2-screen state. The second screen helps to detect later onset conditions.

Who are Partners of the Oregon NBS Program?



History of the Oregon NBS Program

1963	Oregon initiates NBS for Phenylketonuria
1975	Screening for MSUD, Galactosemia, and Congenital Hypothyroidism
1985	Screening for Biotinidase Deficiency
1995	Screening for Hemoglobinopathies
2002	Screening for amino acid disorders, organic acidemias, and fatty acid oxidation defects
2003	Screening for Congenital Adrenal Hyperplasia
2006	Screening for Cystic Fibrosis
2014	Screening for SCID
2018	Screening for Pompe, MPSI, Gaucher, Fabry
2022	Screening for SMA
2023	Screening for XALD
2025	Screening for Infantile Krabbe Disease

Oregon NBS Panel: 46 Disorders

Organic Acidemias

- Propionic acidemia (PA)
- Methylmalonic acidemia (MMA)
- Isovaleric acidemia (IVA)
- 3-methylcrotonyl CoA carboxylase deficiency (3MCC)
- 3-hydroxy-3-methylglutaryl CoA lyase deficiency (HMG)
- Multiple carboxylase deficiency (MCD)
- Beta-ketothiolase deficiency (BKT)
- Glutaric acidemia, Type I (GA-I)
- Malonic acidemia (MAL)
- Isobutyryl-CoA dehydrogenase deficiency (IBD)
- 2-methylbutyryl CoA dehydrogenase deficiency (2MBC)
- 3-methylglutaconyl CoA hydratase deficiency (3MGH)
- 2-methyl-3-hydroxybutyryl CoA dehydrogenase deficiency (2M3HBA)

Amino acid disorders

- Argininosuccinate lyase deficiency (e.g. Arginosuccinic aciduria or ASA)
- Citrullinemia, Type I (CIT)
- Maple syrup urine disease (MSUD)
- Homocystinuria (HCY)
- Phenylketonuria (PKU)
- Tyrosinemia, Types I, II, and III
- Arginase deficiency (ARG)

Endocrine disorders

- Primary congenital hypothyroidism (CH)
- Congenital adrenal hyperplasia (CAH)

Lysosomal storage diseases

- Mucopolysaccharidosis Type I (MPS I)
- Fabry (alphagalactosidase A deficiency)
- Gaucher (glucocerebrosidase deficiency).
- Pome (glycogen storage disease Type II)
- Infantile Krabbe Disease

Severe combined immunodeficiency disease (SCID)

Spinal Muscular Atrophy

Cystic fibrosis

Fatty acid oxidation disorders

- Carnitine uptake defect (CUD)
- Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
- Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
- Long chain 3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
- Trifunctional protein deficiency (TFP)
- Short chain acyl-CoA dehydrogenase deficiency (SCAD)
- Glutaric acidemia Type II (GA2)
- Carnitine palmitoyl transferase deficiency, Types I and II (CPT I and CPT II)
- Carnitine acylcarnitine translocase deficiency

Biotinidase deficiency

Classic Galactosemia

Hemoglobinopathies (Sickle cell disease)

X-linked Adrenal Leukodystrophy (XALD)

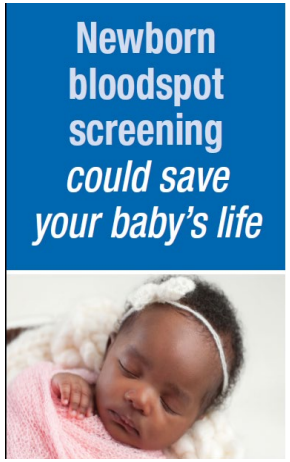
Oregon Newborn Bloodspot Screening Program

Pre-analytic

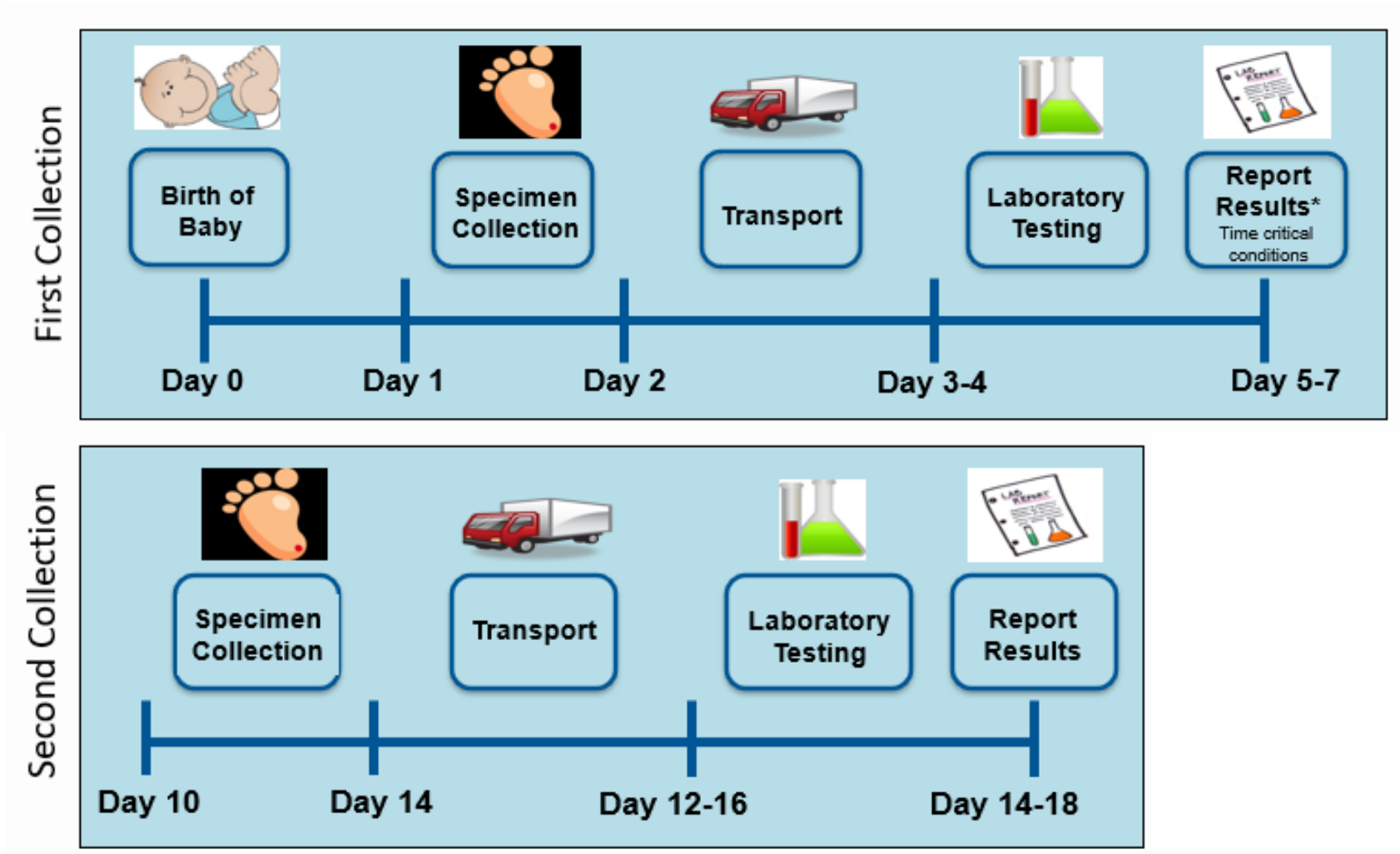
Analytic

Post-analytic

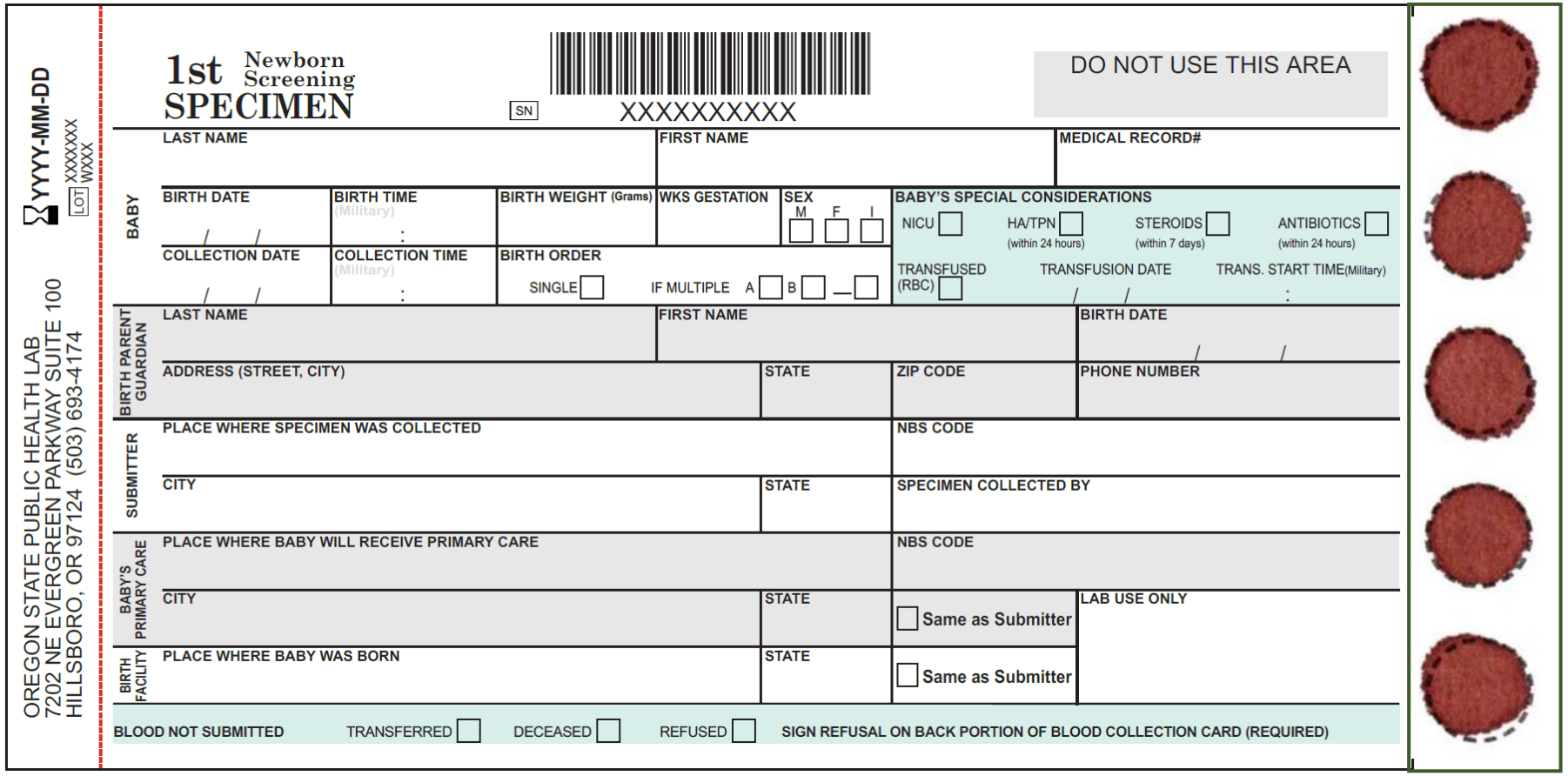
- Prenatal Education
- Specimen Collection
- Transport
- Laboratory Testing
- Short-term Follow-up
- Long-term Follow-up



Newborn Bloodspot Screening Timeline



Collection of the Bloodspots



Newborn Bloodspot Screening Case Study

- Maisie was born in Oregon 2 summers ago. This was the parent's third child after two healthy children.
- Mom was aware of and thankful for newborn screening but did not have any concerns. "*The things the NBS test for were rare and not likely to be anything I ever had to worry about*".
- Maisie's NBS screening was positive for MCAD Deficiency (Medium-chain acyl-CoA dehydrogenase deficiency).

Newborn Bloodspot Screening Case Study

- Screening results were available when Maisie was 5 days old.
- Family was notified by the PCP that day of screening results and scheduled confirmatory testing.
- Because of newborn bloodspot screening, Maisie can grow and thrive.

Newborn Bloodspot Screening Education Video for Parents

NBS video for parents

How can the WIC Nutritionist Support the NBS Program?

- Review video with parents or have it available to view in clinic waiting room.
- Offer NBS education pamphlet to parents during pregnancy.
- Support families to pick a primary care provider during pregnancy.
- Refer family to educational resources for additional information.

How can the WIC Nutritionist Support the NBS Program?

- Ask families if their baby has completed both NBS screenings and any repeat or additional testing that was ordered. Was any follow up indicated?
- Refer families to their PCP if they have not had newborn bloodspot screening completed. We can screen babies up to 6 months of age.
- Remind families to ask their baby's PCP for screening results.

NBS Specimen Retention Policy

- Specimens are stored at the state lab for 12 months and then securely destroyed.
- Specimens with unused bloodspots may be used for internal lab quality control purposes.
- Parent/guardian can request specimens be returned to them after screening is complete.
- Screening results are retained for six years.

Additional information for families

Babysfirsttest.org

- Information about which conditions are screened for in Oregon.
- Information about each disorder.
- Living with a disorder.
- The newborn screening process.

NBS video for parents

bit.ly/NBSnewvideoforparents

www.healthoregon.org/nbs

- The Oregon NBS website has a section for parents that includes helpful information about newborn blood spot screening.

Thank you

The Oregon NBS program and the Oregon WIC program have the same goal to support the health of babies in Oregon.

Thank you for all that you do for families.

Questions?

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