Newborn Bloodspot Screening in Maine

A databrief for providers

Newborn bloodspot screening can detect rare but serious medical conditions. Infants who are diagnosed and treated early can avoid serious complications.

All newborns delivered in Maine (about 12,000 per year) are required to be tested for certain congenital genetic disorders that can cause intellectual and developmental disability, serious illness, or death if left untreated. The newborn bloodspot screening test helps identify infants in need of follow-up testing.

Maine currently tests for 51 conditions and will be adding 4 new conditions in 2020. Types of conditions include endocrine, metabolic, hematologic, pulmonary, neuromuscular, and immune system disorders.

Maine Screening Data

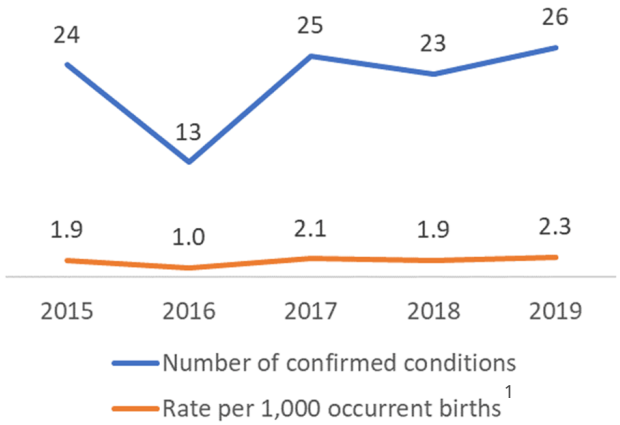
99.97%

of births in Maine in 2019 received a newborn bloodspot screening.

- **95%** had results in the normal range
- **5%** required follow-up testing
- **0.2%** of all births in Maine had a confirmed condition upon additional testing

Maine Diagnostic Data

The rate of confirmed conditions per 1,000 Maine births has been stable for the past five years. The overall number of conditions confirmed as positive is small.



The most frequent conditions are:

- Endocrine Disorders that disrupt the release of specific hormones necessary for important energy and growth functions
- Cystic Fibrosis in which the body produces thick and sticky mucus that can clog the lungs and obstruct the pancreas
- Inborn Errors of Metabolism in which the body cannot properly turn food into energy

Data by Condition

Number of confirmed cases *and presumptive positive screens*[2]**, Maine, 2015-2019**

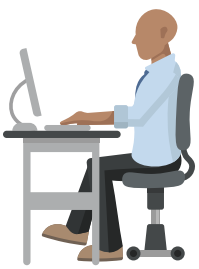
	2015		2016		2017		2018		2019		Average	
Occurrent births	12,527		12,471		12,072		11,998		11,510		12,116	
Percent receiving at least one screen	98.5%		99.4%		99.4%		100.0%		99.9%		99.4%	
Congenital Hypothyroidism (Primary)	**14**	*206*	**9**	*181*	**12**	*145*	**11**	*142*	**9**	*132*	**11.0**	*161*
Congenital Adrenal Hyperplasia	**0**	*90*	**1**	*82*	**1**	*59*	**0**	*67*	**0**	*55*	**0.4**	*71*
Cystic Fibrosis	**6**	*75*	**2**	*56*	**7**	*50*	**5**	*57*	**8**	*59*	**5.6**	*59*
Phenylketonuria (PKU) (Classical)	**1**	*59*	**0**	*42*	**4**	*37*	**1**	*29*	**2**	*46*	**1.6**	*43*
MCAD	**0**	*5*	**0**	*8*	**0**	*4*	**1**	*10*	**1**	*8*	**0.4**	*7*
Galactosemia (Classical)	**0**	*0*	**0**	*2*	**0**	*1*	**1**	*1*	**2**	*4*	**0.6**	*2*
Biotinidase Deficiency	**0**	*2*	**0**	*5*	**0**	*2*	**0**	*0*	**0**	*2*	**0.0**	*2*
Maple Sugar Urine Disease (MSUD)	**0**	*42*	**0**	*18*	**0**	*32*	**0**	*80*	**0**	*78*	**0.0**	*50*
Homocystinuria	**0**	*121*	**0**	*98*	**0**	*79*	**0**	*75*	**0**	*114*	**0.0**	*97*
CPT II	**0**	*0*	**0**	*0*	**0**	*0*	**1**	*1*	**0**	*0*	**0.2**	*0*
Other Metabolic	**1**	*59*	**1**	*32*	**0**	*32*	**1**	*1*	**3**	*11*	**1.2**	*27*
Sickling Disorders	**2**	*2*	**0**	*0*	**1**	*1*	**2**	*2*	**1**	*204*	**1.2**	*42*
Severe Combined Immuno Deficiency (SCID)	**0**	*27*	**0**	*28*	**0**	*20*	**0**	*27*	**0**	*37*	**0.0**	*28*

Data Collection and Use

Maine providers are a vital link between parents and test results.

Providers help ensure initial testing takes place and conduct follow-up testing if necessary.

Birth attendants help ensure initial bloodspot collection takes place



The Newborn Bloodspot Program monitors lab results and notifies primary care providers

Pediatricians and Pediatric Specialty Care **providers** perform follow-up testing and care

Maine CDC Newborn Bloodspot Screening Program

For more information, contact us at: www.mainepublichealth.gov\bloodspot
Tel: (207) 287-8188

1. Occurrent births are those occuring in Maine, regardless of whether the mother is a Maine resident.
2. Presumptive positive screens were out of the normal range upon initial testing, but not yet confirmed by follow-up testing.