2016 Annual Report for New Mexico's Newborn Screening (NBS) Program

Felipe - A Family's Story

Inside this Edition

Felipe was born in January of 2015 at University of New Mexico Hospital (UNMH). Felipe's hearing was screened soon after he was born and he referred in both ears. I was given a rescreening appointment to return two weeks later. Based on the referral from the hospital, a Follow-Up Coordinator from the New Mexico Newborn Hearing Screening Program (NM NBHSP) checked in weekly by phone with me as well as professionals as needed throughout the process. Because Felipe still didn't pass, it was recommended that he see an audiologist. His pediatrician referred him to UNMH Audiology, where he was diagnosed with hearing loss at about 2-3 months of age.

I was told by the audiologist that they would begin with hearing aids and perhaps he'd be eligible for a cochlear implant if hearing aids weren't his best option. Upon receiving Felipe's diagnostic audiology report, the NM NBHSP Follow-up Coordinator contacted the New Mexico School of the Deaf (NMSD) Parent Infant Child Program (PIC). Before Felipe was 6 months, I chose to begin weekly PIC and deaf mentoring services from NMSD as well as speech-language services from

FYI Some Newborn Screening Blood Spot Kits will be expiring in February of 2018. ***Please check all kit (including those from Parents) EXPIRATION DATES are on back-left side corner of the blood spot cards.***

Felipe's hearing loss was progressive and by the time he was 14 months, he needed surgery for a cochlear implant in his left ear. PIC introduced me to a family with child who also had a cochlear implant before I decided on having a cochlear implant for Felipe. I learned about hearing aids and cochlear implants and learned to understand the significance of Felipe's hearing loss and how to help him. I also learned about deafness from the perspective of a Deaf adult. The most difficult decision for me was the cochlear implant, but my husband and I felt we had enough information to determine the benefits for Felipe. His Early Development Specialists for PIC even joined my family at the hospital during the surgery.

I am a Spanish speaker and I didn't feel there was a barrier for me at any point in the process. I was comfortable as possible at all stages of the screening process and treatment because the information shared with me along the way helped me make informed decisions. As needed, since the beginning, Spanish-speaking interpreters and professionals have allowed me fluid communications. I have been an integral part of all decisions regarding options available for Felipe. Because they were informed decisions I feel confident, competent, and supported.

Felipe is "El lo esta haciendo muy bien (He is doing very well)!" Felipe is now over 2 years old and continues to wear a hearing aid in his right ear. Felipe became eligible to attend NMSD Preschool at 18 months of age. He goes there in the morning and then joins me in the afternoons at the daycare where I work.

-Felipe and Monica

Why Screen Babies?

All babies are screened, even if they look healthy, because some medical conditions cannot be seen by just looking at the baby. It is only with time that some conditions discovered by newborn screening may affect your baby's brain, physical development, social skills, school performance, or cause other medical problems. By then the damage may be permanent. Finding these conditions soon after birth and getting treatment early can help prevent some serious problems (such as brain damage, organ damage, and even death) and promote normal growth and development of your child. Since treating your newborn soon after birth is so vital to their health, it is important that your doctor have a current phone number or address so they can reach you if your child needs follow-up care. If they do contact you, make sure to follow their instructions promptly in order to increase your child's chances of living a long, healthy life.

When and How is Newborn Screening Done?

Newborn Screening Blood Spot Test (For Endocrine and Inheritable Conditions)

A health professional will take a few drops of blood from the baby's heel. New Mexico requires two blood spots samples. The first blood sample is collected within 24-48 hours of birth and a second blood sample is collected at 10-14 days after your child's birth. The blood sample is sent to a newborn screening lab for testing.

Otoacoustic Emissions or Auditory Brainstem Response Test (For Hearing Loss)

Hearing screening is easy and not painful. The screen required by New Mexico plays soft sounds and your baby's responses to those sounds are measured. All babies should be screened for hearing loss no later than 1 month of age. It is best if they are screened before leaving the hospital after birth.

Pulse Oximetry Test (For Heart Defects)

Pulse oximetry is fast and easy test that does not hurt your baby. A small sensor is placed on a baby's right hand and one foot to measure the oxygen level in their blood. Pulse Oximetry should be done prior to leaving the hospital.

What is Newborn Screening Program?

The Newborn Screening program ensures that all babies with an abnormal screening results receives appropriate diagnostic testing and that confirmed cases receive early and timely care. The newborn screening program at the state level does this by coordinating information and medical care amongst doctors, nurses, laboratory technicians, social workers, and medical specialists. The program also works to keep costs affordable, reduces health disparities, and provides equal access to care and assistance.

One of the main purposes of the Newborn Screening Program is to educate parents and healthcare providers about newborn screening. New Mexico state law requires that all infants born either at birthing facility or with a midwife receive a newborn screen, NMSA §24-1-6 (2014) and NMSA §24-1-6.1 (2001). Parents can refuse screening by sending a completed refusal form to the state's newborn screening program.

New Mexico does not do research with blood spot samples. All samples are destroyed within a year of the child's birth.

Screened Conditions

Amino Acid Disorders

Argininemia (ARG); Argininosuccinic aciduria (ASA); Citrullinemia, type I (CIT); Citrullinemia, type II (CIT II)Phenylketonuria (PKU);Homocystinuria (HCY); Hypermethioninemia (MET);Tyrosinemia, type I (TYR I);Tyrosinemia, type II (TYR II); Maple syrup urine disease (MSUD);

Endocrine Disorders

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

Hemoglobin Disorders

Hemoglobinopathies (Var Hb); S, Beta-thalassemia (Hb S/ßTh); S, C disease (Hb S/C); Sickle cell anemia (Hb SS)

Screened Conditions Continued

Fatty Acid Oxidation Disorders

Carnitine acylcarnitine translocase deficiency (CACT);
Carnitine palmitoyltransferase I deficiency (CPT-IA),
Carnitine palmitoyltransferase type II deficiency (CPT-II);
Carnitine uptake defect (CUD); Glutaric acidemia, type II (GA-2);
Long-chain L-3 hydroxyacyl-CoA dehydrogenase deficiency (LCHAD);
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD);
Trifunctional protein deficiency (TFP);
Short-chain acyl-CoA dehydrogenase deficiency (SCAD);
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

Organic Acid Conditions

2-Methyl-3-hydroxybutyric academia (2M3HBA); Glutaric acidemia type I (GA1);
2-Methylbutyrylglycinuria (2MBG); 3-Hydroxy-3-methylglutaric aciduria (HMG);
3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC); Isovaleric acidemia (IVA);
3-Methylglutaconic aciduria (3MGA); Beta-ketothiolase deficiency (BKT);
Holocarboxylase synthetase deficiency (MCD); Isobutyrylglycinuria (IBG);
Malonic acidemia (MAL);
Methylmalonic acidemia (cobalamin disorders) (Cbl A,B);
Methylmalonic academia (methymalonyl-CoA mutase deficiency) (MUT);
Methylmalonic acidemia with homocystinuria (Cbl C, D, F);
Propionic acidemia (PROP)

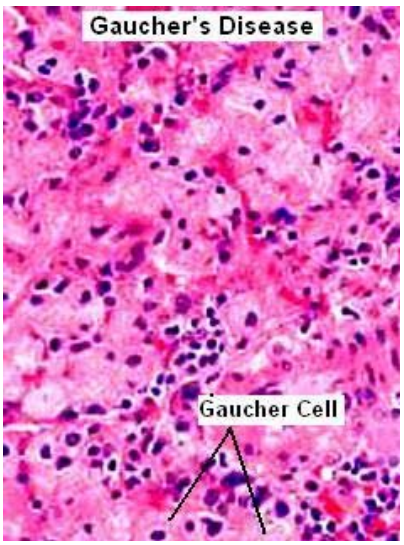
Other Disorders

Biotinidase deficiency (BIOT); Galactosemia (GALT); Cystic fibrosis (CF);
Severe combined immunodeficiency (SCID); Hearing Deficiency;
Critical Congenital Heart Disease (CCHD)—Hypoplastic left heart syndrome, Tetralogy of Fallot, Pulmonary atresia (with intact septum), Total anomalous pulmonary venous return, Tricuspid atresia, Truncus arteriosus, Transposition of the great arteries

Added to Newborn Screening Panel-LSD

New Mexico will be adding four lysosomal storage disorders (LSD): Gaucher disease, Fabry disease, Pompe disease, and Hurler/Scheie syndromes to the newborn screening panel. In lysosomal storage diseases, the intracellular digestive membranes called lysosomes are defective and toxins build up in the cell. Treatment for these four disorders has progressed over the past 2 decades and it is now possible to prevent long-term complications through early diagnosis and periodic enzyme replacement therapy.

Gaucher disease is due to defective function of lysosomal β-glucocerebrosidase. Defective function of this lysosomal enzyme results in accumulation of glucocerebrosidase. Signs and symptoms of this disorder include hepatosplenomegaly and bone marrow dysfunction. Thrombocytopenia and anemia in combination with splenomegaly can be mistaken for leukemia.



Fabry disease is a lysosomal storage disorder due to defective function of lysosomal α-galactosidase A. This results in accumulation of globotriaosylceramide. This compound accumulates in vascular endothelial cells resulting in dysfunction in many different organs. Signs and symptoms of this disorder include acroparesthesias, renal failure, cardiomyopathy, and strokes.

Pompe disease, also known as glycogen storage disease type II, is due to defective function of lysosomal α-glucosidase. This results in accumulation lysosomal glycogen. There are multiple clinical presentations of this condition including infantile, juvenile, and adult. The infantile form is characterized by severe cardiomyopathy and hypotonia; the juvenile and adult forms are characterized by skeletal muscle myopathy with weakness.

Hurler and Scheie syndromes are also known as mucopolysaccharidoses type I. They are due to defective function of lysosomal α-iduronidase. This results in accumulation of glycosaminoglycans (commonly known as mucopolysaccharides) in many tissues and organs. Signs and symptoms include hepatosplenomegaly, joint contractures, corneal clouding, cardiac and pulmonary involvement and variable intellectual disability.

-Dr. Randal Heidenreich, MD at UNM Pediatrics

Is There Support for Families?

One of the concerns many families have when they first learn that their baby has one of the disorders detected by the Newborn Screening Program is the possibility of increased health care costs. Fortunately, the state of New Mexico has various policies and programs in place that can assist families in obtaining the best care for their child.

Under the Affordable Care Act, both public and private health insurers (which does not include grandfathered plans) are required to fully cover the costs of diagnosing your baby with a metabolic or genetic disorder that is listed on Secretary's Advisory Committee on Heritable Diseases in Newborns and Children. Most health insurers will cover treatment (including clinical services, medical prescription drugs, nutritional management and medical foods), although durational limits, caps, deductibles, coinsurance and copayments may apply. If your baby is identified as having one of the disorders detected by newborn screening, the Affordable Care Act makes it illegal for your health insurer to deny health coverage to your baby. For families that have difficulty paying for newborn screening services (including treatment) or who do not have health insurance, please contact the New Mexico Newborn Screening Program for additional resources.

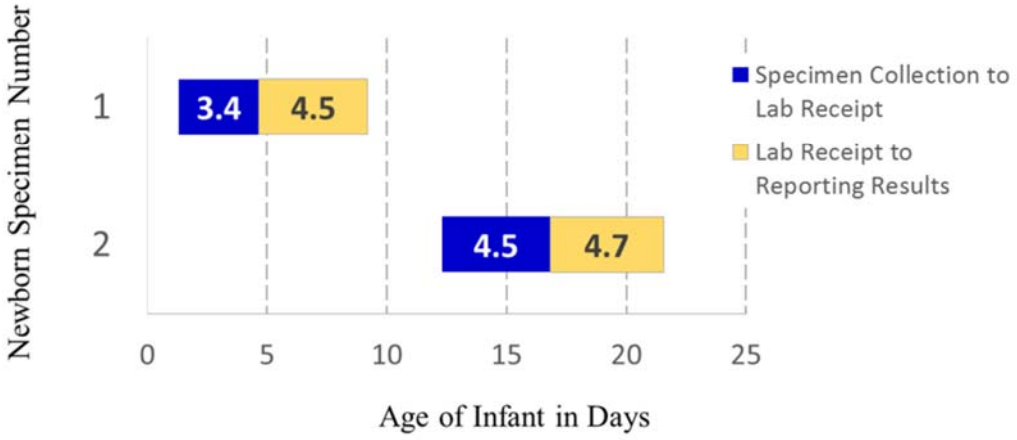
The New Mexico Newborn Screening Program is housed within the Children's Medical Services (CMS), another program in the Department of Health. Families who have a baby identified by newborn screening can receive care coordination from a CMS Social Worker to help them obtain the care they need for their baby and identify other community resources. CMS sponsors community-based pediatric specialty clinics around the state for metabolic, genetic, endocrine, and cardiology conditions. For families who qualify for income support, CMS will cover the costs of diagnosis, treatment, and provision of health insurance to your child. CMS also partners with family support groups such as Parents Reaching Out (PRO), Education for Parents of Indian Children with Special Needs (EPICS), Hands and Voices, Community Outreach Program for the Deaf (COPD), and the Sickle Cell Council of New Mexico in addition to other national parent support groups such as the National PKU Alliance. For further information, see their website at http://nmhealth.org/about/phd/fhb/cms/cyshcn/

Overview of Newborn Genetic Screening

Ninety-seven percent of all babies born in New Mexicans, excluding babies that did not survive their first month of life, received a newborn blood spot screen in 2016. This means that 22,759 New Mexican babies were screened for potentially crippling or deadly inheritable conditions. Most babies received their first screen at one day old and their second screen at twelve days of age. Doctors will typically know the results of a babies screen about a week after the collection of the specimen. Refer to Figure 1.

During the course of the year, Newborn screening identified, diagnosed, and assist in providing early treatment to 102 New Mexican babies that had potentially crippling or deadly inheritable conditions. These identified conditions included: primary congenital hypothyroidism, cystic fibrosis, critical congenital heart disease, DiGeorge Syndrome, hearing loss, isovaleric acidemia (IVA), medium chain acyl co -A dehydrogenase (MCAD), short chain acyl-CoA dehydrogenase (SCAD), thyroxine-binding globulin deficiency (TBG), sickle cell anemia, and severe combined immunodeficiency (SCID). During the screening process, 192 additional infants were identified as having a blood disorder. On average, babies with an identified newborn genetic condition began receiving treatment at 24 days of age and

Table 1: The average time between the collection of New Mexico Newborn Screening Blood Spot samples and laboratory reporting for 2016.



Specimen Problems for NBS

The newborn screening laboratory received a total of 43,073 newborn screening blood spot samples (which includes first and second specimens) in 2016. State Newborn Screening Program piloted overnight holiday delivery service as well as a number of hospitals did quality improvement projects around newborn screening which may partially explain the slightly better transit times in 2016. See Figure 2. The large spike in blood spot errors around September 2016 was result of facilities submitting expired blood spot cards to the newborn screening laboratory. See Figure 3. Also in 2016, there has been a slight increase in demographic errors when

Figure 2: Percent of New Mexico newborn screening samples that spent more than 5 days in transit from date of collection to laboratory receipt, 2015-2016.

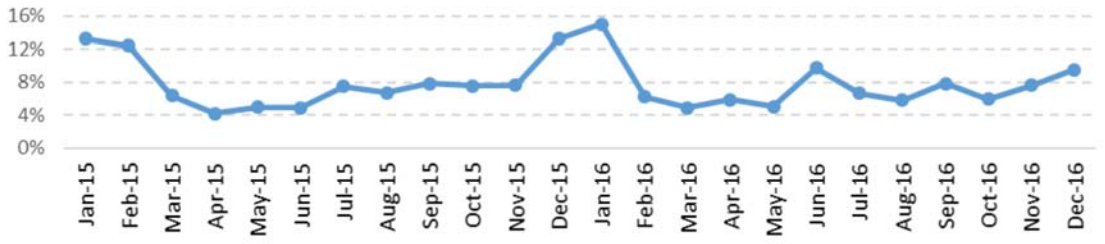


Figure 3: Percent of New Mexico newborn screening samples that had any errors in the collection of the blood spot specimen, 2015-2016.

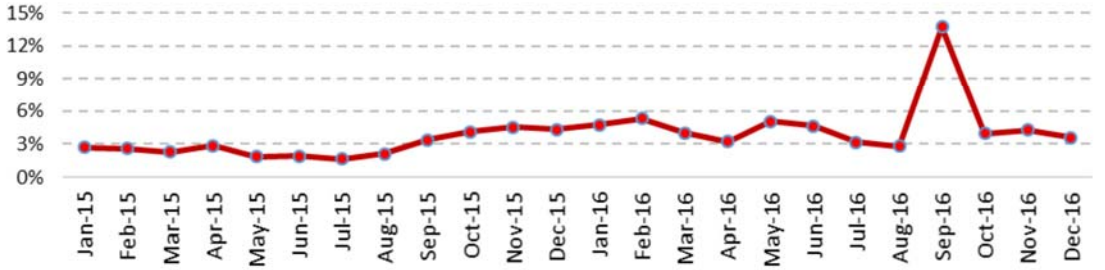
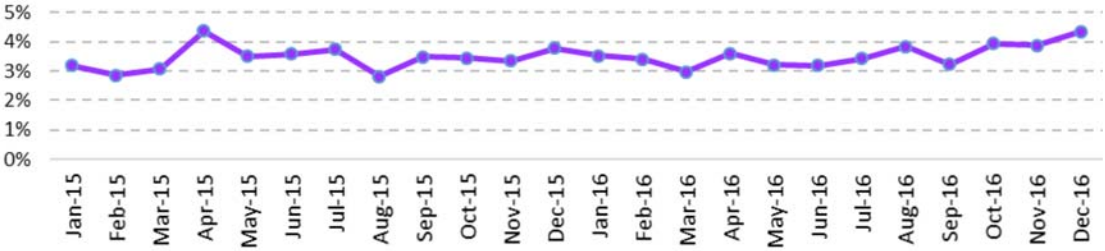


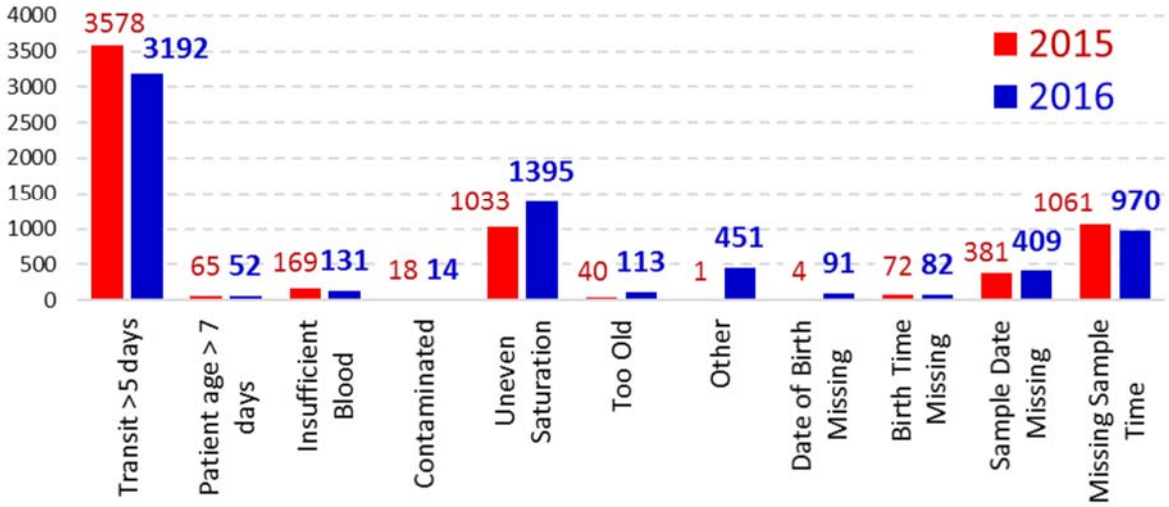
Figure 4: Percent of New Mexico newborn screening samples that had any demographic errors in filling out the blood spot form, 2015-2016.



Types of Specimen Errors

The most common problems submitting blood spot specimens to the laboratory in 2016 were long transit times (which is time between sample collection and laboratory receipt), uneven saturations, and missing sample times. See Figure 5. For more tips on sampling and mailing specimens correctly, please see the Practitioner's Manual posted at http://nmhealth.org/about/phd/fhb/cms/nbgs/

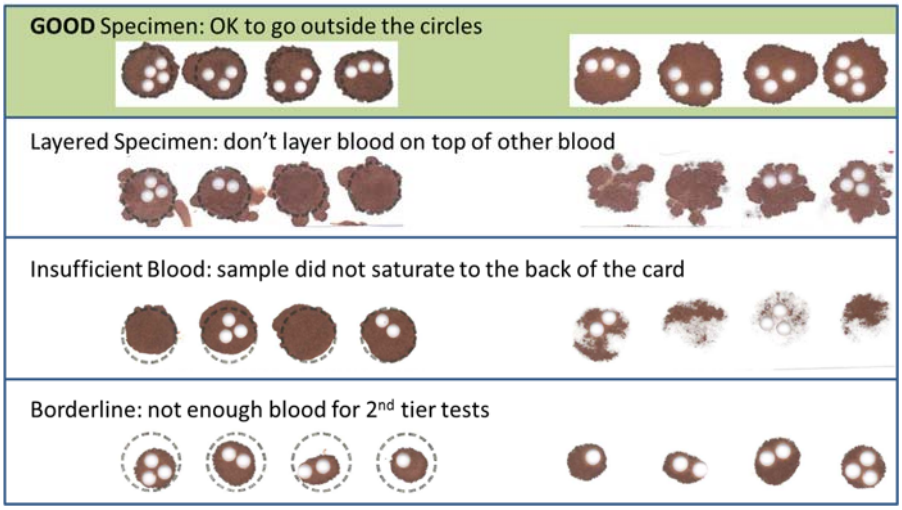
Figure 5: The number of incorrect blood spot specimens in New Mexico by type of error, 2015-2016.



Unsatisfactory specimen Errors	Possible causes
Uneven saturation: Blood spot appears layered and/or scratched or abraded.	•Application with a capillary tube or needled syringe. •Applying blood to both sides of the filter paper. •Touching the filter paper with fingers or gloves (Sweat and oils from fingers or powder residue from gloves affect absorbency of filter paper). •Layering several blood drops on top of each other. •Hanging filter paper to dry. •Compression of the filter paper.
Insufficient blood (QNS): Blood spots do not look the same from both sides of the filter paper.	•Failing to open a large enough capillary bed resulting in insufficient blood flow from the infant's heel to allow complete absorbency through to the second side. •Removing the filter paper before blood has completely soaked through to the second side.
Specimen too old: Specimen received in the laboratory more than 14 days after collection date	•Specimen was not mailed within 24 hours after collection.

Newborn Genetic Screening Tips

Newborn Bloodspot Specimen Examples: Front & Back

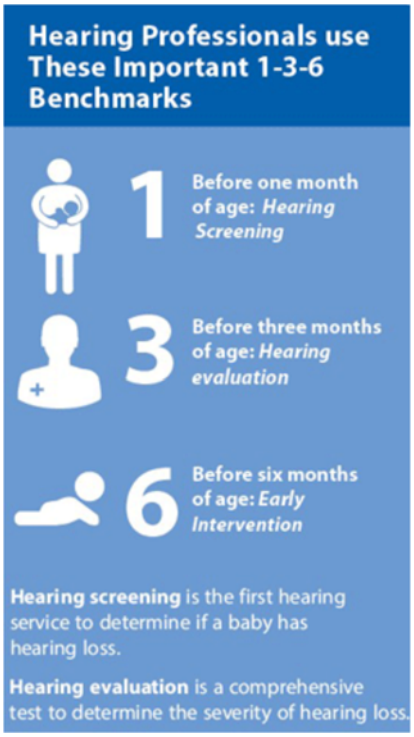


The Newborn Genetic Screen program recommends the following actions to reduce the amount of screens and help the program follow up with abnormal screen results.

- Always check the expiration date on the back-left side corner of the newborn screening form. **Do NOT use expired forms.**
- For repeat screens due to unsaturation (i.e. poor sample collection), use the next screen card parent brings to visit or a single kit for the repeat. If possible, wait until the baby is at least 24 hours old.
- Single kits are available to parents at Public Health Offices if they misplace their screening forms. Whenever possible, please use the 2nd form given to parents at hospitals, because this makes linking all of the child's screening results easier
- Completely fill out your clinic's information (including physician name, phone number and address) at the top right hand side.
- Please verify parent's contact information (including address, phone and second contact) verbally. This information is very important in case we need to follow up with an abnormal screening result.
- Newborn screening specimens take about 4 hours to dry. **Dry ONLY at room temperature in a horizontal position with the bloodspots exposed.**
- **NEVER place newborn screening specimens in plastic bags prior to sending out.**
- All bloodspots must be sent out within 24 hours of collection. **Do NOT batch shipping.**
- DVD's are available on how to properly collect newborn screening specimens free of charge. To check out a copy, please contact 505-476-8868

Overview Hearing Screening

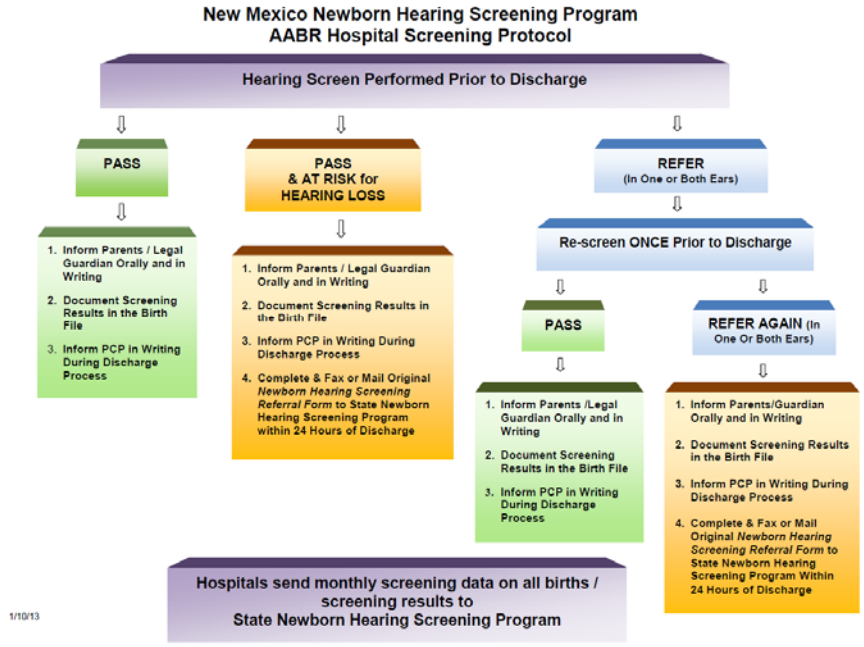
In 2015, 23,152 New Mexicans were screened for hearing loss at birth. Of those screened, 97% passed on their first newborn hearing screen. The 740 New Mexicans babies that did not pass their hearing screens were assisted by the Newborn Hearing program to get confirmatory testing. for Of those babies with an abnormal hearing screen result, 48 New Mexican babies were diagnosed with permanent hearing loss and another 15 babies were identified with transient hearing loss. Eighty-three percent of babies diagnosed with permanent hearing loss were enrolled in early intervention services (such as Part C) within six months of birth.



The New Mexico Newborn Hearing Screening Program has made great strides to screening every baby born in New Mexico. As part of this goal, the Newborn Hearing Screening Program:

- Partners with Dar a Luz to hold a monthly hearing clinic in Albuquerque for any family that needs a hearing screen or rescreen and includes home birthed babies as well as babies born in hospitals in need of hearing screening or rescreening. http://www.daraluzbirthcenter.org/
- Partners with New Mexico Hands and Voices to provide information, education materials and parent support for families with children who are Deaf or Hard of Hearing. http://www.hvnm.org/
- For hospital referrals, immediately sends information to parents regarding follow-up services, audiology facilities and child development information to families who may be in need of assistance with hearing rescreening and audiological testing.

Hearing Screening Tips



The Newborn Hearing Screen program recommends the following actions to reduce the amount of screens and help the program follow up with abnormal screen results.

- Complete **ALL** fields on the Newborn Hearing Screening Referral Form http://nmhealth.org/publication/view/form/1061/
- Minimize noise (including electrical equipment) or confusion in the screening area
- Wait until the baby is at least 24 hours old, if possible
- Screen the baby when they are quiet (preferably sleeping), well-fed, and comfortable
- For OAE, choose a proper tip that fits snugly in the baby's ear and is seated properly on the probe
- For ABR, remember better contact = better impedance = better and faster screening
- Screen **BOTH** ears of the child at the same time and no more than three times
- If the baby doesn't pass on the first try, remove the probe and inspect the baby's ear and probe for debris (such as wax, blood, vernix)
    - Check placement and seal of earphones
- A child is considered to Pass without referral when **BOTH EARS PASS IN THE SAME SCREEN**
- If the baby still doesn't pass in one or both ears, wait a few hours before rescreening

Infant Hearing Testing Facilities

ALBUQUERQUE

Ear Associates
Office: 505-224-7610
Fax: 505-224-7619

Presbyterian Ear Institute (PEI)
Office: 505-224-7020
Fax: 505-224-7023

Presbyterian Hospital
Office: 505-563-6391
Fax: 505-563-6390

University of New Mexico Hospital
Office: 505-272-3535
Fax: 505-272-0300

University of New Mexico Newborn Hearing Screening
Office: 505-272-9613
Fax: 505-272-0300

Indian Health Services
Office: 505-248-4052
Fax: 505-764-0446

Wendy Gallegos Audiology
Office: 505-898-2474
Fax: 505-899-8172

Albuquerque Hearing and Balance
Office: 505-890-0003
Fax: 505-890-3330

ALAMOGORDO

Alamogordo Ear, Nose and Throat
Office: 575-437-4533
Fax: 575-437-5009

DEMING

Hearing Healthcare Clinic
Office: 575-544-4041
Fax: 575-546-7210

FARMINGTON

San Juan Regional Medical Center
Office: 505-609-6079
Fax: 505-609-6841 (fax)

Animas Valley Audiology
Durango, CO
Office; 970-375-2369
Fax: 970-375-9054

GALLUP

Rehoboth McKinley Christian Health Care Services-College Clinic
Office: 505-827-5995
Fax: 505-863-1898

HOBBS

Texas Tech Hearing & Balance Center
Lubbock, TX
Office: 806-775-9305
Fax: 806-775-9302

LAS CRUCES

Rio Grande Hearing
Office: 575-521-3025
Fax: 575-521-3565

RATON

Greenwood Ear, Nose and Throat
Pueblo, CO
Office: 719-544-7115
Fax: 719-544-6242

ROSWELL

Audio Acoustics
Office: 575-623-8474
Fax: 575-623-8220

SANTA FE

Southwestern Ear, Nose and Throat (SWENT)
Office: 505-946-3955
Fax: 505-946-3945

SHIPROCK

Northern Navajo Medical Center
Office: 505-368-6365
Fax: 505-368-7078

SOCORRO

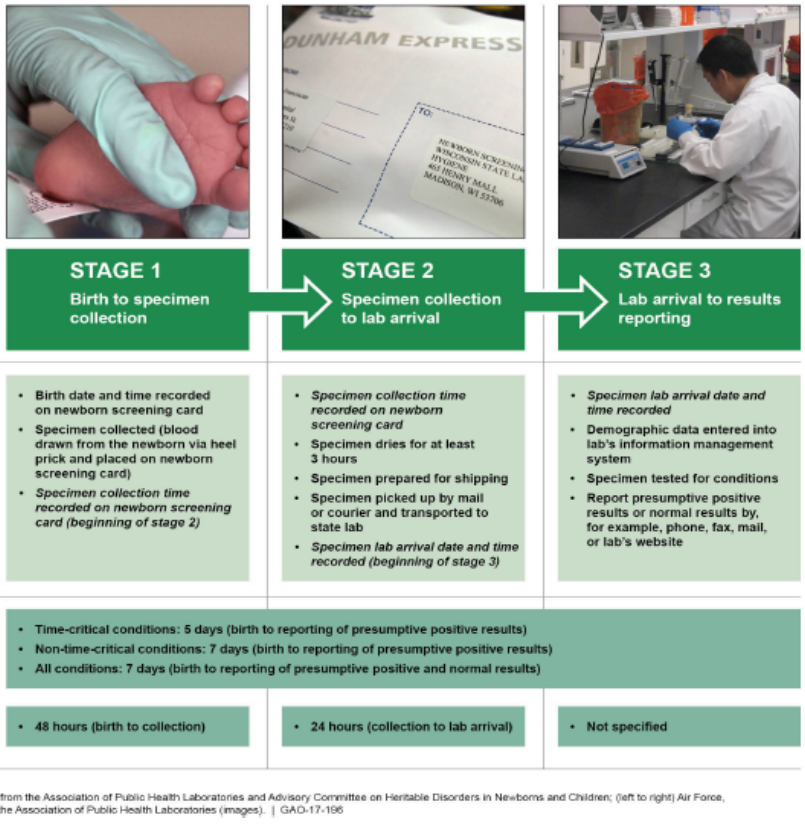
Marianne Cramer
Office: 575-835-8769
Fax: 575-835-8772

Facility Process Improvement

Newborn screening involves many partners to provide our newest citizens with a high quality newborn screen. Without these elements in place, the system would fail to provide the excellent service that New Mexicans have come to expect from newborn screening, such as screening is done in a expedited manner (see video every hour counts https://www.youtube.com/watch?v=30qbkhp1jQ8). These services include:

- Education of healthcare providers and parents
- Collection of a quality sample
- Timely transport of specimen to the state's designated laboratory
- Rapid and reliable testing methodology
- Timely notification of healthcare providers and parents of any unexpected result
- Timely repeat sample collection
- Appropriate referral of babies to specialists for diagnosis, treatment and counseling services
- Assuring access to programs that help our most vulnerable citizens

Figure 2: Activities Included in the Newborn Screening Process and Related Time-Frame Goals Set by the Advisory Committee on Heritable Disorders for Newborns and Children in 2015



Sources: GAO analysis of information from the Association of Public Health Laboratories and Advisory Committee on Heritable Disorders in Newborns and Children; (left to right) Air Force, © Milwaukee Journal Sentinel, and © the Association of Public Health Laboratories (images). | GAO-17-196

National Goals

Timeliness of specimen collection, transit, and laboratory reporting are all critical components to saving the lives of newborns affected by the disorders for which the Program tests. The Newborn Screening Saves Lives Act of 2014 included timeliness as an explicit goal and included a provision for the Government Accountability Office (GAO) to review newborn screening timeliness.

1. **Collection specimen 48 hours after birth**
2. **Specimen arrive at lab 24 hours after specimen collection**
3. **All newborn specimen results reported within 7 days (168 hours) after birth**

Changes to Hospital Practice Profiles

Hospital Practice Profiles are a way to provide individualized feedback about timeliness and specimen submission quality to your facility or practice. Recently the following changes have been made to them:

1. Goals for transit and collection times have been updated to the National Standard
2. Timeliness of specimen collection and transit 'errors' have been changed to 'goals'

Transit Time Supports

The state has a number of resources to help facilities meet these national goals.

1. Newborn Screening QI Toolkit has been developed to help hospitals identify suboptimal processes around newborn screening. An electronic copy can be found (https://www.surveymonkey.com/r/NMNBSHospitalSurvey.)
2. Selected sites have Next Day Delivery on Holidays
3. Newborn Screening Laboratory in Oregon is open Saturday mornings.

First specimens need to be shipped via UPS to:

Oregon State Public Health Laboratory
7202 NE Evergreen Parkway
Hillsboro, OR 97124