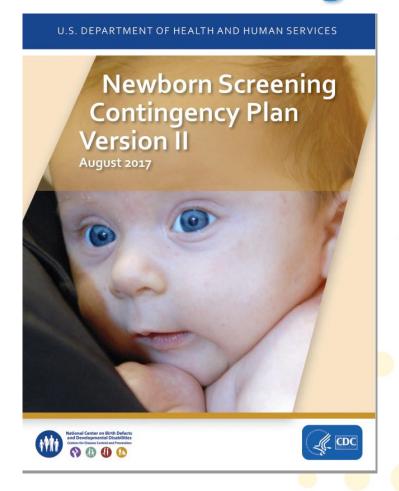


Newborn Screening

8/8/18



Newborn Screening Part 1





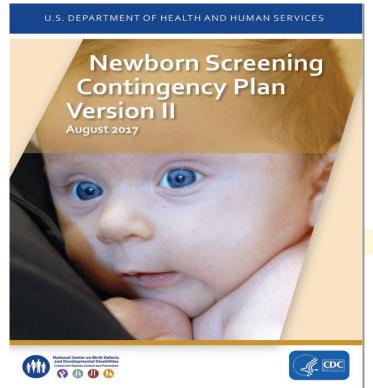
TA Opportunity

 Crisis Response Network and AHCCCS will be holding a joint TA session on the SMI eligibility system, including assessments, on Thursday, August 16th from 9-10am in the Gold room.



CDC

 https://www.cdc.gov/ncbddd/documents/S creening-Contingency-Plan-Version-II.pdf





Bloodspot Screening ADHS





https://azdhs.gov/preparedness/statelaboratory/newborn-screening/#healthcareproviders-disorder-info

ARIZONA'S NEWBORN SCREENING PANEL OF 31 DISORDERS Effective August 9, 2017

Endocrine Disorders (2) Congenital hypothyroidism (CH) Congenital adrenal hyperplasia (CAH)

Amino Acid Disorders (6)
Phenylketonuria (PKU)
Maple syrup urine disease (MSUD)
Homocystinuria (HCY)
Citrullinemia type I (CIT-1)
Argininosuccinic acidemia (ASA)
Tyrosinemia type I (TYR-1)

Fatty Acid Oxidation Disorders (5)
Carnitine uptake defect (CUD)
Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)
Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)
Long-chain La-3-hydroxyacyl-CoA dehydrogenase deficiency (LCHAD)
Trifunctional protein deficiency (TFP)

Organic Acid Disorders (9)
Isovaleric acidemia (IVA)
Glutaric acidemia (IVA)
Glutaric acidemia type I (GA-1)
3-Hydroxy-3-methylglutaric aciduria (HMG)
Multiple carboxylase deficiency (MCD)
Methylmalonic acidemia-cobalamin defect (Cbl A,B)
Methylmalonic acidemia-mutase deficiency (MUT)
3-Methylcrotonyl-CoA carboxylase deficiency (3MCC)
Propionic acidemia (PROP)
Beta-ketothiolase deficiency (BKT)

Hemoglobin Disorders (3) Sickle cell anemia (Hb SS) S, beta-thalassemia (Hb S/ß Th) S, C disease (Hb S/C)

Other Disorders (4)
Biotinidase deficiency (BIOT)
Galactosemia (GALT)
Cystic Fibrosis (CF)
Severe Combined Immunodeficiency (SCID)

Disorders not detected by bloodspot screening (2) Hearing Loss (HEAR) Critical Congenital Heart Defects (CCHD)



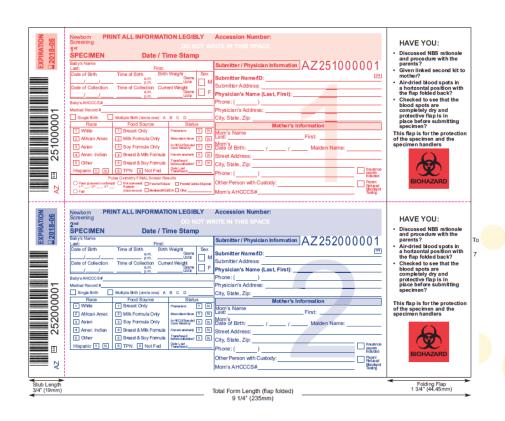


Current ADHS Program Overview

- AZ screens for 31 disorders, 29 from bloodspot collection and two point-ofcare tests—Hearing and Critical Congenital Heart Defect (CCHD) screening – Severe Combined Immunodeficiency (SCID) screening was added in August 2017
- A fee increase on the 1st screen was approved Nov. 2017 to partially offset costs associated with SCID testing 1st screen costs \$36. 2nd screen costs \$65.
- Two bloodspot screens are required for all newborns/infants The 1st one should be collected between 24-36 hours – The 2nd one should be collected between 5-10 days of life.
- The pulse ox screen should be collected at 24 hours and repeated up to 2 more times before discharge
- Initial hearing screening should be conducted at 24 hours of age or before discharge



Screening Card





Selecting Disorders for Screening

- Prevalence
- Cost effective and reliable testing
- Effective treatment/intervention
- No treatment = death /severe impairment
- Appears normal at birth





AZ Newborn Screening Advisory Committee: SB 1368

Newborn Screening Advisory Committee

Statutory Requirement ARS §36-694

Role	Position	Person
Endocrinologist—Physician #1	filled	Dr. Mark Wheeler
Pediatrician—Physician #2	NEW	Dr. Arturo Gonzalez
Neonatologist—Physician #3	NEW	Dr. Moira Richards
Family Practice—Physician #4	filled	Dr. Rachel Deatherage
Otologist—Physician #5	filled	Dr. Michael Fucci
Obstetrician—Physician #6	NEW	Dr. Laura Mercer- tentative
Metabolic GeneticistPhysician #7	Filled	Dr. Kirk Aleck
NNP	Filled	Deb Ledington
Audiologist	Filled	Lylis Olsen
Part C Provider	NEW	Jenée Sisnroy
Parent(s)	Filled (x2)	Deb Houck Monica Attridge
Insurance Industry	NEW	Dr. Leslie Paulus
AHCCCS	NEW	Dr. Eric Tack
Hospital/Healthcare Rep.	NEW	Debbie Johnston
Licensed Midwife	NEW	Olga Ryan





CDC RUSP

The Recommended Uniform Screening Panel (RUSP) 34 disorders recommended

Conditions listed on the RUSP are part of the comprehensive preventive health guidelines supported by HRSA for infants and children under section 2713 of the Public Health Service Act. Non-grandfathered health plans are required to cover screenings included in the HRSA-supported comprehensive guidelines without charging a co-payment, co-insurance, or deductible for plan years beginning on or after the date that is one year from the Secretary's adoption of the condition for screening. ¹

https://www.hrsa.gov/advisory-committees/heritable-disorders/rusp/index.html



How to Nominate a New Condition

 https://www.hrsa.gov/advisorycommittees/heritabledisorders/rusp/index.html

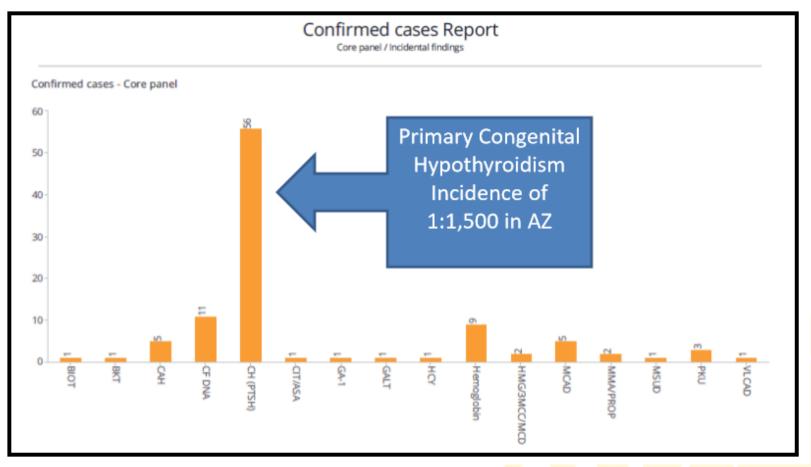


Data

- In 2017, > 155,000 bloodspot samples were processed in the lab, averaging about 13,000 per month representing about 84,000 babies 101 babies were diagnosed with genetic/metabolic disorders 282 (0.18%) samples were unsatisfactory for testing and repeat testing was necessary Another 3000 (2%) samples had insufficient blood on the card to complete all screening and therefore repeat testing was required.
- A high number of these quantity not sufficient (QNS) samples were attributed to the addition of SCID screening, which requires another punch from the spot of blood.
- A CQI project is underway with hospitals to improve outcomes. The total number of QNS samples have been reduced from 3.2% to about 1.70% within 6 months.



Bloodspot Disorders for 2017





Future Additions

SMA currently under review

AZ screens for 31 disorders and is missing

Pompe

X-ALD

MPS-1



ADHS needs your help!

Follow up Activities

- Help close the loop when referrals are written by doctor
- If we can reach out to MCH Coordinators directly it would help
 - Can we get current contact info for each health plan?
- Arizona is mandated to screen every newborn twice but there is no mechanism in place to ensure that infants who receive only one screen with normal results receive the required second screen

What the Data Shows

In 2016

Bloodspot

- 97.13% of infants had at least one bloodspot screen;
- Of them, 21 babies had out of range (positive) bloodspot results but no additional testing

Hearing

- 98.02% had a hearing screen
- 259 or 29.80% were lost to follow-up



Audiologic Testing

- AABR Automated Auditory Brainstem Response
- OAE Otoacoustic Emissions
- Incidence is 2-3/1000 live births
- NIH Fact Sheet https://report.nih.gov/NIHfactsheets/ViewF actSheet.aspx?csid=104



Risk Factors

Risk factors for neonatal sensorineural hearing loss

Family history of permanent hearing loss

Craniofacial abnormalities including those involving the external ear

Congenital infections including bacterial meningitis, cytomegalovirus, toxoplasmosis, rubella, herpes and syphilis

Physical findings consistent with an underlying syndrome associated with hearing loss

Neonatal intensive care unit stay >2 days OR with any of the following regardless of the duration of stay:

- Extracorporeal membrane oxygenation
- · Assisted ventilation
- Ototoxic drug use
- Hyperbilirubinemia requiring exchange transfusion



Hearing

- Newborn hearing screening is the standard of care in hospitals nationwide. The primary purpose of newborn hearing screening is to identify newborns who are likely to have hearing loss and who require further evaluation. A secondary objective is to identify newborns with medical conditions that can cause late-onset hearing loss and to establish a plan for continued monitoring of their hearing status (Joint Committee on Infant Hearing [JCIH], 2007). The EHDI guidelines include hearing screening completion by 1 month of age, diagnosis of any hearing loss by 3 months of age, hearing aid selection and fitting within 1 month of confirmation of hearing loss if parents choose that option, and entry into early intervention (EI) services by 6 months of age.
- In 2014, 96.1% of babies born in the United States had their hearing screened before 1 month of age (Centers for Disease Control and Prevention [CDC], 2016), and 6,163 infants were diagnosed with permanent hearing loss.
- Screening programs target permanent childhood hearing loss (PCHL) irrespective of type. However, some
 protocols are more effective at identifying types and degrees of hearing loss within different populations
 (i.e., well-baby nursery or neonatal intensive care unit [NICU]).
- Passing a screening does not mean that a child has normal hearing across the frequency range. Because
 minimal and frequency-specific hearing losses are not targeted by newborn hearing screening programs,
 newborns with these losses may pass a hearing screening. Because these losses have the potential to
 interfere with the speech, language, and psychoeducational development of children (Yoshinaga-Itano,
 DeConde Johnson, Carpenter, & Stredler-Brown, 2008), monitoring of hearing, speech, and language
 milestones throughout childhood is essential.



Principles

- All infants should have access to hearing screening using a physiologic measure before 1 month of age.
- All infants who do not pass the initial hearing screen and the subsequent rescreening should have appropriate audiologic and medical evaluations to confirm the presence of hearing loss before 3 months of age.
- All infants with confirmed permanent hearing loss should receive intervention services before 6 months of age. A simplified, single point of entry into an intervention system appropriate to children with hearing loss is optimal.
- The EHDI system should be family centered with infant and family rights and privacy guaranteed through informed choice, shared decision making, and parental consent.
 Families should have access to information about all intervention and treatment options and counseling regarding hearing loss.
- The child and family should have immediate access to high-quality technology, including hearing aids, cochlear implants, and other assistive devices when appropriate.

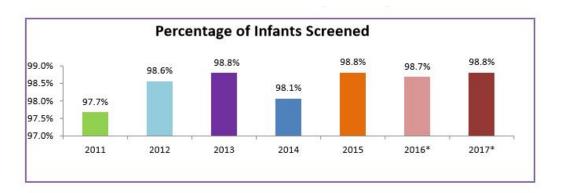


AMPM 430

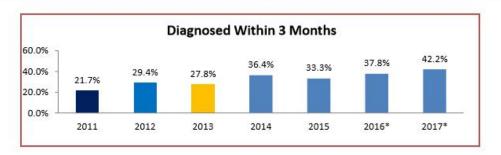
- AMPM 430 B 10
- Contractors must ensure that the Bloodspot Newborn Screening Panel and hearing tests are conducted, including initial and secondary screenings, in accordance with 9 A.A.C. 13, Article 2.
- Appropriate vision, hearing, and speech screenings are covered during an EPSDT visit. EPSDT covers eye examinations as appropriate to age according to the AHCCCS EPSDT Periodicity Schedule (AMPM Exhibit 430-1) and as medically necessary using standardized visual tools. Payment for vision and hearing exams, (including, but not limited to CPT codes 92015, 92081, 92285, 92551, 92552, 92553, 92567, 92568, 92285, 92286, 92587, 92588, 95930, and 99173) or any other procedure that may be interpreted as fulfilling the vision and hearing requirements provided in a PCP's office during an EPSDT visit, are considered part of the EPSDT visit and are not a separately billable services.



Hearing Screening AZ Data



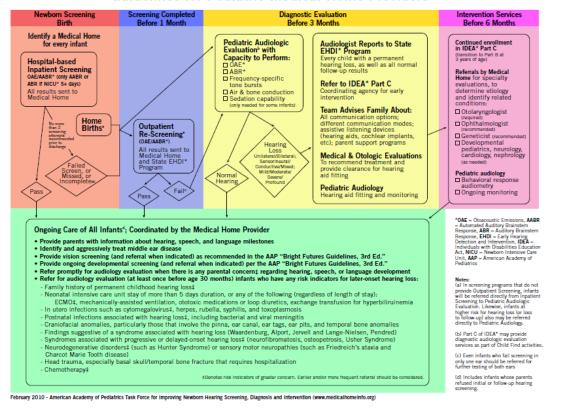
CALENDAR YEAR 2017	Total Screened: 82,035	
Total with Permanent Hearing Loss (Initial Diagnosis)	162	
Permanent Hearing Loss ID Before 3 Months of Age	125	
Permanent Hearing Loss ID After 3 Months of Age but Before 6 Months	23	
Permanent Hearing Loss ID After 6 Months of Age	14	





Hearing Screening Flow

Early Hearing Detection and Intervention (EHDI) Guidelines for Pediatric Medical Home Providers





Resources



Early Hearing Detection & Intervention - Pediatric Audiology Links to Services (EHDI-PALS)

Welcome to EHDI-PALS!

- ▼ Home
- Find Audiology Facilities
- Resources about hearing
- Resources about early intervention
- Other Helpful Websites
- Audiologists:
 Create/Update
 Facility Profile
- Professional Resources
- EHDI-PALS Advisory Group
- ▶ EHDI Program Log-in

⊠ Contact us

Welcome to EHDI-PALS, Early Hearing Detection & Intervention - Pediatric Audiology Links to Services, a web-based link to information, resources, and services for children with hearing loss. At the heart of EHDI-PALS is a national web-based directory of facilities that offer pediatric audiology services to young children who are younger than five years of age.

LEARN MORE about childhood hearing loss, hearing testing, and important questions parents can ask when making appointments. This contains great web resources for parents and professionals.

 $\label{lem:continuous} \textbf{Find OTHER HELPFUL WEBSITES, including national and state parent support organizations and other resources. }$

You can also find out more about the EHDI-PALS Advisory Group.

Looking For A Facility?

Find a Facility for hearing services for children from birth to 5 years old:

Find An Audiology Facility

List Or Update Your Facility

Are you a provider interested in listing your facility in the EHDI-PALS directory? If so, enter here:

List or Update Your Facility

Informatics support by the UMaine Developmental Epidemiology and Biobehavioral Informatics Group (DEBBI)



Resources

- https://www.asha.org/PRPSpecificTopic.asp x?folderid=8589935234§ion=Key Issu es from the American Speech-Language-Hearing Association.
- https://www.cdc.gov/ncbddd/hearingloss/r ecommendations.html from the CDC



CCHD

- https://www.cdc.gov/ncbddd/documents/S creening-Contingency-Plan-Version-II.pdf
- HB 2491 (enacted April 2014)
- Incidence = 1% (~40,000) of all live births in US
- VSD most common
- ~4.2 % of neonatal deaths due to CCHD



CCHD Screening For

CHDs & CCHDs

Critical Congenital Heart Defects

- · Hypoplastic Left Heart Syndrome
- Pulmonary Valve Atresia and Stenosis
- · Tetralogy of Fallot
- Transposition of the Great Arteries
- Tricuspid Valve Atresia and Stenosis
- Total Anomalous Pulmonary Valve Return
- Common Truncus/Truncus Arteriosis

Congenital Heart Defects

- Coarction of Aorta*
- Ebstein Anomaly*
- Endocardial Cushion
 Defect/Atrioventricular Canal
 Defect
- Interrupted Aortic Arch
- Single Ventricle
- Aortic Valve Stenosis
- Atrial Septal Defect
- Ventricular Septal Defect



Health and Wellness for all Arizonans

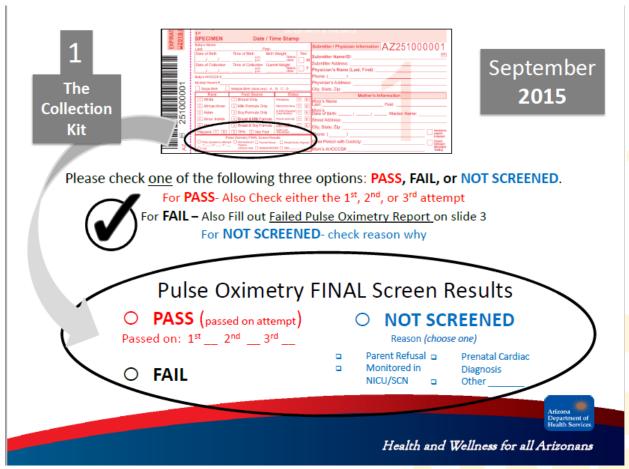


Bloodspot Collection Form

IRAT	1st SPECIMEN	Date / Tir	DO NOT WI	RITE IN THIS SPACE
EXPIRA (2018)	Baby's Name Last: Date of Birth	First:	Weight Sex Grams Lb/oz ent Weight	Submitter Address: Physician's Name (Last, First): Phone: ()
001	Medical Record # Single Birth Race	Multiple Birth (circle one) A	B C D	Physician's Address: City, State, Zip: Mother's Information
000	1 White 2 African Amer.	Breast Only Milk Formula Only	Promoture Y N Meconium lieus Y N	Mom's Name Last: First:
251	3 Asian 4 Amer. Indian.	3 Soy Formula Only Breast & Mile Formula 5 Breast & Soy Formula	Known anomaly Y N	
	Pass (passed on attemp	6 TPN 0 Not Fed	Date Last Transfused	City, State, Zip: Phone: () Other Person with Custody:
A 2	9 Fal	Reason (choose one) Northred NOUS		Mom's AHCCCS#



Documentation



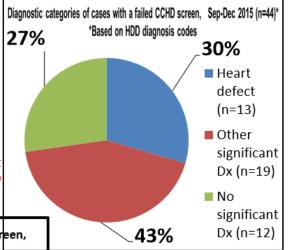


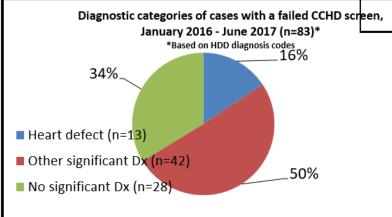
CCHD Screening Results

Year 1 (07-2015/06-2016) data analysis from 44 babies who failed the screen by diagnostic categories:

- 30% with heart defects;
- 43% with other clinically significant Dx;
- 27% false positive—no clinical findings

A noted limitation for both years was that about 10% of babies have no documented screen





Year 2 data from 83 babies who failed the screen by diagnostic categories:

- 16% with heart defects;
- 50% with other clinically significant DX;
- 34% false positive—no clinical findings

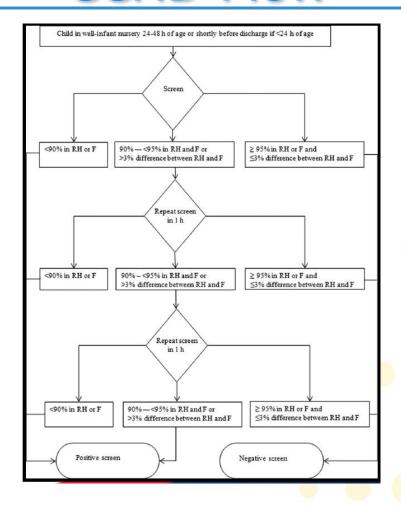


CCHD Flow

Complete within 30 days of a failed screen an	d return via fax to ADHS Office of Newborn Screening at: 602-364-1495	
Newborn Demographics		
Last Name:	First Name:	
Date of Birth (mm/dd/yyyy):	Time of Birth (military time):	
Mother's Name:	Mother's DOB:	
Birth Hospital / Facility/ or Site:		
Pulse Oximetry Test Results		
Screening Facility:	Infant's Medical Record Number (MRN):	
Screening Results/Values:		
First attempt		
Date and Time of screen:	R Hand% R / L Foot%	
Second attempt		
Date and Time of screen:	R Hand% R / L Foot%	
Third attempt		
Date and Time of screen:	R Hand	
Referral and Diagnosis		
1. Was the infant referred and/or transferred	to another facility? Yes No If yes, where?	
Did the infant have a known or suspected C	Congenital Heart Defect prior to screening? (Chicke one) Yes No Unknow	
If yes, provide diagnosis and/or any pertinent	information:	
3. Does the infant have other congenital diagr	noses? (circle cire) Yes No Unknown	
If yes, provide diagnosis and/or any pertinent	information:	
4. Did the infant receive an echo and/or addit	ional testing following the Pulse Ox Screen: (Circle one) Yes No Unknow	
If yes, provide diagnosis and/or any pertinent	Information:	
5. Final cardiac and/or additional diagnoses.	If known. Or, other explanation of failed screen:	



CCHD Flow





Raising Arizona Kids Demonstration

 https://www.google.com/url?sa=t&rct=j&q =&esrc=s&source=web&cd=11&cad=rja& uact=8&ved=2ahUKEwjch8Hwmq7cAhUnrl QKHVf2C6sQwqsBMAp6BAgEEBY&url=http s%3A%2F%2Fwww.youtube.com%2Fwatc h%3Fv%3D098PUs2WxGg&usg=AOvVaw2 BcrunhMY5dDwWCxfvk7ww



Vision Screening Ages 3 – 6 Years

- WHO data:
- 19M worldwide have some sort of vision impairment:
 - 12M with a refractive error
 - 1.5M irreversible blindness



Early Signs and Symptoms

Most **babies** start to focus on faces and objects by 4-5 weeks of age. By about 6-8 weeks, most babies will start smiling at the familiar faces and things they see. But if a baby has vision impairment, you might notice she has trouble doing this.

Other signs that a baby might have a problem with his vision are if his:

- eyes move quickly from side to side (nystagmus), jerk or wander randomly
- eyes don't follow your face or an object, or he doesn't seem to make eye contact with family and friends
- eyes don't react to bright light being turned on in the room
- pupils seem white or cloudy rather than black
- eyes don't line up but look towards his nose or turn outwards.



Older Children

An older **child** might:

- hold things up close to her face
- rub her eyes a lot
- turn or tilt her head or cover one eye when looking at things up close
- get tired after looking at things up close for example, reading, drawing or playing handheld games
- seem to see better during the day than at night
- say she has tired eyes
- seem to have misaligned eyes or a <u>squint</u>
- seem clumsy for example, she might knock things over or trip often.



AMPM 430 B 10

- Ocular photoscreening with interpretation and report, bilateral (CPT code 99177) is covered for children ages three to five as part of the EPSDT visit due to challenges with a child's ability to cooperate with traditional vision screening techniques. Ocular photoscreening is limited to a lifetime coverage limit of one. This procedure, although completed during the EPSDT visit, is a separately billable service.
- NOTE: Automated visual screening, described by CPT code 99177, is not recommended for or covered by AHCCCS when used to determine visual acuity for purposes of prescribing glasses or other corrective devices.
- Vision CPT codes with the EP modifier must be listed on the claim form in addition to the preventive medicine CPT codes for visit screening assessment.
 With the exception of CPT code 99177, no additional reimbursement is allowed for these codes.



Not State-of-the-Art





Ocular Photoscreening

Photoscreening is a form of vision screening for children. It uses a camera to take images of a child's undilated eyes. By looking at the configuration of the crescents of light returning after a flash (red reflex). The devices can estimate refractive error and determine which children are at risk of amblyopia (<u>lazy eye</u>). These images can be analyzed by a human interpreter, or by software incorporated into the equipment to evaluate the alignment of the eye and estimate refractive error. If significant refractive error or a misalignment appears to be present it can indicate amblyopia risk factors. If amblyopia risk factors are felt to be present a referral should be made for the child to be seen by a pediatric ophthalmologist for a cycloplegic examination. Photoscreening has advantages to more traditional eye chart acuity screening, and is particularly useful on younger (age 3-5), preverbal children (under age 3) and non-verbal children. Photoscreening usually takes less than a minute to obtain the necessary images on a child. The only cooperation required is for the child to briefly look at the camera.



Ocular Photoscreening

<u>iScreen</u> – The iScreen photoscreener was first introduced in 2006. The first generation of the device was an off-axis binocular photoscreener taking one image, which is electronically transmitted for remote interpretation. The first generation iScreen was a tabletop device in which the child rests their head against a chin rest. The iScreen 3000, introduced in 2011, has been significantly redesigned and miniaturized [5]. It is now a hand-held device, which takes 2 photos in rapid succession in 2 axes with a separation of 90 degrees. Aiming beams placed on the child's forehead focus the camera. A blinking fixation light, and sound attract the child's attention. The images are then taken in such rapid succession that the child perceives just one flash prior to a blink. This is an improvement over the analog MTI photoscreener which required lens rotation prior to taking the second photo, and prevents differential accommodative effort between photos. The photo can be reviewed immediately on the device and if the child is not fixating properly can be re-taken. The final image is sent electronically to the company for interpretation, which provides the advantage of a consistent, expert interpretation of the images. A report is emailed securely which shows the results of the test and reason for the referral and also includes a picture of the child's eyes.



iScreen





Demonstration



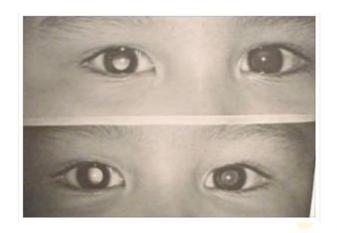


Equipment - MTI

• MTI – The MTI photoscreener was first introduced in 1995(5)^[4]. The camera uses special high resolution black and white polaroid style instant film with an off axis flash which rotates 90 degrees between images. Focus and fixation are similar to the iScreen with aiming beams and a flashing fixation target and noise to attract the child's attention. Two consecutive photos are taken on each patient resulting in a photograph showing the first image on the top and the second image on the bottom printed on a special high-resolution instant film. Based on the shape, size and location of the crescents, a determination can be made as to whether the child has significant refractive error or strabismus. The MTI is no longer being manufactured but is still in use.



Documentation



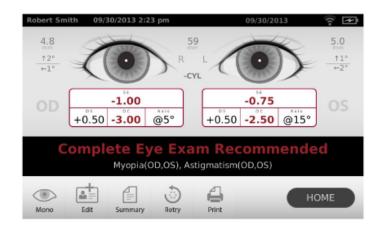


Equipment - Spot

Spot – The Spot is a new hand-held infrared digital photoscreener developed and marketed by Pediavision. It functions similarly to the plusoptiX, but has been significantly miniaturized. Rather than having the infrared elements on the face of the unit flanking the camera aperture, the Spot places the infrared elements below and in front of the camera aperture. The infrared light is then reflected off of a 45% two-way mirror. The design allows for eye tracking. The device creates a diagram with the location of the eyes, demonstrating any strabismus present as well as an autorefractive reading. Spot creates a report giving referral criteria based on preset criteria which can be printed. Visiscreen – The Visiscreen is a 35-mm camera with a databack recorder attached to a 500-mm telephoto lens. There is an electronic strobe light located below the lens and a flashing light-emitting diode located above the lens. The camera is mounted on bars ensuring it is exactly 8 feet and 3 inches away from a headrest the patient places their chin in.



Demonstration



Accurate detection

Spot accurately* detects the potential indication of these common vision problems:

- · Myopia (nearsightedness)
- · Hyperopia (farsightedness)
- · Astigmatism (blurred vision)
- Anisometropia (unequal refractive power)
- · Strabismus (eye misalignment)
- · Anisocoria (unequal pupil size)
- · *Accuracy refers to the sensitivity and specificity of the screening referral recommendation.



Advantages/Disadvantages

- Most accurate
- Patient Friendly
- Quantitative Results
- Training/Usage
- Cost Issues



Reminder: NO Back To Basics 8/15/18





Thank You.



