

ARIZONA NEWBORN SCREEN

1. What type of screening is done for newborn hearing?

Hearing loss is the most frequent birth defect in the US (2-4/1000 live births). Otoacoustic emissions test (OAE) is usually the test used in the hospital prior to discharge. Failed tests can be caused by too much external noise, debris in the ear canal, or true poor cochlear response. OAE does not detect 8th nerve neuropathy. If a newborn fails the OAE, auditory brainstem response test (ABR) can be done which measures evoked responses from the cochlea to the brainstem.

2. What diseases are screened for under the Arizona newborn screen?

- 6 amino acid disorders (including PKU)
- 5 fatty acid oxidation disorders (including MCADD)
- 9 organic acid disorders
- Biotinidase deficiency
- Classic galactosemia
- Congenital hypothyroidism
- Congenital adrenal hyperplasia
- 3 hemoglobin diseases
- Cystic Fibrosis (new June 2007)

See below for complete list

Many new disorders were added to the screening profile because of advances in detection by mass spectrometry.

3. What should the timing be for newborn screens?

1st screen at 24-72 hours

2nd screen at 5-10 days or first PCP visit

ARIZONA'S NEWBORN SCREENING PANEL OF DISORDERS

Same list recommended by the American Academy of Pediatrics

Amino Acid Metabolism Disorders

Phenylketonuria (PKU)
Maple syrup urine disease
Homocystinuria
Citrullinemia
Tyrosinemia type I
Argininosuccinic acidemia

Fatty Acid Oxidation Disorders

Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
Very long-chain acyl-CoA dehydrogenase deficiency
Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
Trifunctional protein deficiency
Carnitine uptake defect

Organic Acid Disorders

Isovaleric acidemia
Glutaric acidemia type I
3-OH 3-CH₃ glutaric aciduria
Multiple carboxylase deficiency
Methylmalonic acidemia
Methylmalonic acidemia (mutase deficiency)
3-Methylcrotonyl-CoA carboxylase deficiency
Propionic acidemia
Beta-ketothiolase deficiency

Hemoglobin Disorders

Hb S/Beta-thalassemia
Hb S/C disease
Sickle cell anemia

Other Disorders

Congenital hypothyroidism
Congenital adrenal hyperplasia
Biotinidase deficiency
Galactosemia
Hearing Loss
Cystic Fibrosis (added in October 2007)

Cost:

1st screen = \$30

2nd screen = \$40

List of Resources

1. Website that reviews the expanded newborn screen for Arizona :
<http://azaap.org/Common/Files/Newborn%20Screening%20Training%20Update%202011-06.ppt>
2. List of the disorders included in expanded newborn screening:
http://www.azdhs.gov/phs/owch/pdf/disorder_list.pdf
3. Arizona Department of Health Services Home Page on Newborn screening for reference:
<http://www.aznewborn.com/>