

# CDS Newborn Screening Tests: Call for Samples

The Association for Creatine Deficiencies (ACD) would like to inform our community of the opportunity to participate in an important study involving newborn screen testing for Creatine Deficiency Syndromes (CDS) at the Mayo Clinic.

Creatine Deficiency Syndromes are inborn errors of metabolism, which interrupt the formation or transportation of creatine. Proper diagnosis and early intervention are critical to establish treatments needed to improve life quality and longevity for CDS patients.

Newborn screening for Creatine Deficiency Syndromes (CDS) has been limited by the absence of an effective screening test for all 3 disorders: Creatine Transporter Deficiency (CTD), Guanidinoacetate Methyltransferase Deficiency (GAMT) and L-Arginine: Glycine Amidinotransferase Deficiency (AGAT or GATM). This challenge may be overcome by the implementation of a new test under development at Mayo Clinic.

To determine the effectiveness of Mayo's test in identifying newborns with CDS, Dr. Dietrich Matern and his colleagues at the Mayo Clinic Biochemical Genetics Laboratory in Rochester, MN, are conducting an IRB-approved research study (#15-005393) to develop and validate a new screening test that will improve the identification of individuals affected with an inborn error of metabolism. The study also aims to establish and/or refine disease ranges in order to reduce false positive results and the unnecessary follow-up. An effective screening test would allow diagnosing and potentially treating affected infants prior to the onset of symptoms and development of irreversible damage.

## Call for Samples—Participate in Ongoing Research at Mayo Clinic

The Biochemical Genetics Laboratory is currently seeking volunteers who have a confirmed diagnosis of an inborn error of metabolism and/or their first degree relatives. We welcome samples from patients and their parents, siblings, and children.

You may be able to help in the following ways:

- 1) Allowing Mayo Clinic access to any leftover newborn screening samples that may still be available in the newborn screening laboratory located in the state where a patient or family member of a patient was born.
- 2) Donate a fresh urine sample or a dried blood spot collected by a finger stick or regular blood draw which will be used to measure analyte and/or enzyme levels and to identify relevant newborn screening biomarkers.

Your current or future medical care at the Mayo Clinic will not be jeopardized if you choose not to participate. If you have any questions or concerns, a biochemical genetic counselor from Mayo Clinic would be happy to discuss the study with you in more detail. If you would like to participate or have any questions, please contact Dr. Matern or a biochemical genetic counselor at 507-266-4996 or by e-mail at [biochemicalgenetics@mayo.edu](mailto:biochemicalgenetics@mayo.edu).

See the table on next page to find out if your NBS card is still available. Storage times vary by state.\*

## Newborn Screen Dried Blood Spot Retention Times by State

Colorado	6 mo
Washington	21 yr
Oregon	1 yr
California	indefinitely
Idaho	1 yr
Nevada	1 yr
Montana	1 yr
Wyoming	6 mo
Utah	90 d
Arizona	3 mo for normal; indefinitely for all others
New Mexico	1 yr
Alaska	3 yr
Hawaii	1 yr
Texas	2 yr; w/consent 25 yr
Oklahoma	42 d
Kansas	1 mo
Nebraska	3 mo for normal; indefinitely for all others
South Dakota	1 mo
North Dakota	18 yr
Minnesota	71 d
Iowa	5 yr
Missouri	5 yr
Arkansas	3-4 mo
Louisiana	1 mo
Mississippi	1 yr
Alabama	3 mo for normal; indefinitely for all others
Georgia	2 mo for normal ; 2 yr for positive

Florida	6 mo
Tennessee	1 yr for normal; indefinitely for all others
South Carolina	12 mo for normal; longer for others
North Carolina	5 yr
Kentucky	2 mo
Virginia	6 mo for normal 10 years for all others
Illinois	2-4 mo
Wisconsin	1 yr
Michigan	indefinitely
Indiana	23 yr
Ohio	2 yr
West Virginia	3 mo for normal; indefinitely for all others
Pennsylvania	8 mo
Maryland	25 yr
New Jersey	23 yr
New York	27 yr
Maine	indefinitely
Vermont	indefinitely
New Hampshire	6 mo
Massachusetts	21.5 yr
Connecticut	2 yr
Deleware	3 yr
Rhode Island	23 yr
Washington D.C.	1 yr
Puerto Rico	2 yr
Guam	unk