UCLA ASC NEWSLETTER, SPRING 2017
FACILITIES EDITION

NEWBORN SCREENING: ON-THE-SPOT

APRIL 4, 2017

UCLA AREA SERVICE CENTER (ASC 97)

MEET THE TEAM:

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PROGRAM SPECIALIST

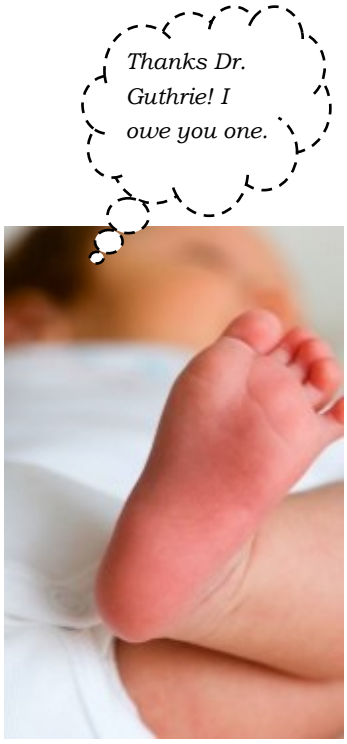
DIANE PAIETTA
ADMINISTRATIVE ASSISTANT

SPECIAL POINTS OF INTEREST:

- **50 yrs of newborn screening in California!**
- **Quality Improvement: timeliness matters**
- **New TRF forms: 31 vs. 30 series**
- **TPN and newborn screening: to draw or not to draw??**
- **New disorder added to NBS panel: ALD**

NEWBORN SCREENING CELEBRATES 50 YEARS IN CALIFORNIA!

For half a century, newborns in the state of California have been tested for various genetic and congenital disorders, trailing the National Program for newborn screening (NBS) by only one year. Thanks to the pioneering efforts of Dr. Robert Guthrie, who introduced the first newborn screening test for phenylketonuria (PKU) in the early 1960's, babies born in California have benefited from early detection and early treatment for five decades. Thousands of babies have been saved from the debilitating and—at times—deadly effects, such as metabolic crises, seizures, failure to thrive, developmental delays, and death, of genetic diseases. The methodology—collecting blood specimens via heel stick by using a filter paper card—was also inaugurated by Dr. Guthrie in the early '60s and has become the standard technique for NBS specimen collection. Although the California NBS program tested only for PKU in 1966, the panel has progressively expanded, adding hypothyroidism and galactosemia in 1980; sickle cell and other hemoglobinopathies in 1990; and congenital adrenal hyperplasia and a plethora of metabolic disorders in 2005, thanks to the development of tandem mass spectrometry (MS/MS). Now, in 2016, the state of California screens newborns for 80 different genetic disorders and more babies are living longer, healthy lives. A success for public health and a big win for babies and families across the state!



With one heel stick, many abnormalities are caught early. Lives are saved.

IMPROVING TIMELINESS, SAVING LIVES

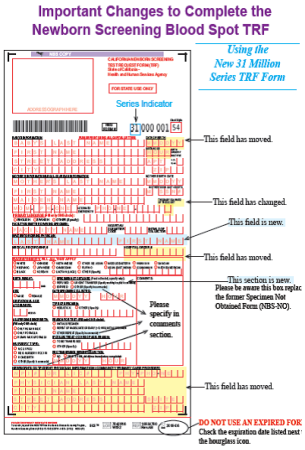
Now in Year 2 of the statewide Quality Improvement (QI) project, ASC 97 continues to emphasize the importance of timeliness in specimen collection and transit. With support from the newly revised state regulations, the QI project has 2 main goals for facilities: 95% of initial specimens must be collected within 12-48 hours of life and 85% of all initial specimens must be transported to the NAPS[1] lab within 2 business days of collection. Although many facilities appear to have difficulties achieving the specimen transit goal of 85%, improvements from baseline are evident. Stay tuned for updates as the fiscal year progresses!

[1]Neonatal and Prenatal Screening

Newborn Screening Area Service Center designation and funding are provided by the California Department of Public Health, Genetic Disease Screening Program

NEW TEST REQUEST FORMS—31 SERIES

Starting in October 2016, the California Department of Public Health, Genetic Disease Screening Program has issued new Test Request Forms (TRF) for newborn screening and distribution of these new forms—the 31 series—is underway. Although similar to previous forms, key differences exist between the 31 and 30 TRF. For example, there are two fields for physician information: one field for the *inpatient/ordering physician* (in the middle of the 31 TRF) and another field for the *outpatient physician/PCP* (at the bottom of the 31 form). This change will hopefully provide sufficient information, should additional follow-up be needed while the baby is still inpatient or after discharge. Unlike the 30 TRF, there is no field on the 31 form to indicate if a baby is NPO or on TPN. In this case, we urge the collector to write this information in the *Comments* field, located towards the bottom right of the 31 TRF. Additionally, if your facility is using 31 TRFs and a specimen is not collected for whatever reason, please complete the *Specimen Not Obtained* field and write pertinent information in the *Comments* field—this replaces the NBS-NO form. Lastly, if your facility still has 30 forms, don't dispose of them! 30 TRF forms do not expire until December 2017.



Several fields have changed on the 31 form vs. 30 form

SPECIAL FOCUS ON TPN CASES

It is no surprise that many babies in the NICU receive Total Parenteral Nutrition (TPN) at some point during their stay in the hospital. Whether newborns are in critical condition, being prepped for surgery, working on increasing their feeds, or simply just extremely premature, babies on TPN cannot tolerate enteral feedings and must acquire some or all of their nutrition intravenously. At the same time, an initial newborn screen (NBS) specimen must be collected within 12-48 hours of life, per state regulations. As some of you might have observed, babies who received TPN around the same time that the initial NBS specimen was collected may have test results that are inconclusive, as TPN—and specifically, the amino acids in the intravenous solution—confounds the interpretation of the amino acid panel on the MS/MS portion of the newborn screen test; a redraw is then required. *So why collect a NBS specimen if a baby is on TPN?* Because if abnormalities related to the baby's hemoglobin, acylcarnitine, or other non-amino acid analytes are present, early detection and treatment can save the baby's life, despite TPN.

"Next to creating a life, the finest thing a [person] can do is save one." - Abraham Lincoln

CALLING ALL NBS CHAMPIONS!

Are you a Newborn Screening champion? Do you prioritize newborn screening and make your best effort to ensure that your facility excels at specimen collection and transit time? Know somebody who fits in this category? As part of the statewide Quality Improvement project, ASC 97 wants to identity healthcare providers and professionals—RNs, Care Coordinators, PAs, NPs, MDs, technicians, assistants, specialists, etc.—who lead by example and encourage their teams to meet the collective goals of the newborn screening program. If you exemplify this role and/or have a fellow colleague that fits this description, please contact us! By highlighting key success factors and sharing best practices regarding newborn screening, we hope that *all* facilities within the UCLA Area Service Center—and across the state—will achieve the principle goals of 95% and 85% in specimen collection and transit time. We look forward to finding our NBS champions!

ASC 97 Welcomes Kristina to the Team!

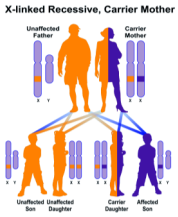
In September 2016, the UCLA Area Service Center welcomed Kristina Parkins to the team, as the Program Specialist. Ms. Parkins comes to UCLA, after 2 years working for The Hunger Project, an international non-profit organization based in NYC aimed at eradicating chronic hunger and poverty in various countries in Sub-Saharan Africa, South Asia, and Central and South America. Prior to working for The Hunger Project, Ms. Parkins earned her Master of Public Health (MPH) degree from EHESP School of Public Health in Paris, France, where she took special interest in research and programs aimed at improving health outcomes for women, newborns, and young children from low-income and culturally diverse backgrounds. When she's not preoccupied with newborn screening, Ms. Parkins enjoys reading, traveling, photography, and spending time with loved ones. We are very excited to have Ms. Parkins on the team!

Out of NBS materials? Need forms?

To order official forms, supplies or education materials for newborn screening, ***call California State Dept. of Public Health*** *at 510-412-1542,* ***fax request*** *to 877-984-9650, or send an* ***email*** *to* ***NBSOrders@cdph.ca.gov***

SPOTLIGHT ON ADRENOLEUKODYSTROPHY (ALD)

Following the addition of ALD onto the federal Recommended Uniform Screening Panel (RUSP), California has begun screening all babies for adrenoleukodystrophy (ALD), as part of the state-mandated newborn screening program. A progressive neurological disorder with an x-linked inheritance pattern, ALD mainly affects the nervous system and adrenal glands and is characterized by the gradual destruction of myelin and high levels of saturated, very long chain fatty acids (VLCFA) in the brain and adrenal cortex. As this is an X-linked disorder, ALD is expressed predominantly and more severely in males and can lead to varying degrees of symptoms—from mild to life-threatening—across the child's lifespan. Females are also affected by ALD, as carriers can develop symptoms in adulthood and pass the defective *ABCD1* gene to their children. Now that ALD is part of the newborn screening program, all babies born after September 2016 (with backlog testing for infants born after February 2016) in California have been tested for this disorder, in three tiers. For ALD testing, the fatty acid that is measured is C26. If C26 levels are flagged as *high* in Tier 1, a second test—Tier 2—is then performed. If Tier 2 screening is positive, *ABCD1* gene sequencing is then conducted to test for mutations, in Tier 3. The UCLA Area Service Center ensures that all babies—those who test positive for *ABCD1* gene mutations and/or who have elevated C26 levels in Tier 2 testing—receive follow-up care at a Metabolic Specialty Care Center. As this is a new experience for many providers, please let us know if you have any questions or concerns about ALD testing!



ALD is rare, x-linked neurological disorder, meaning that is affects males more severely than females. Females are ALD carriers.

QUESTIONS OR COMMENTS? LET'S HEAR FROM YOU

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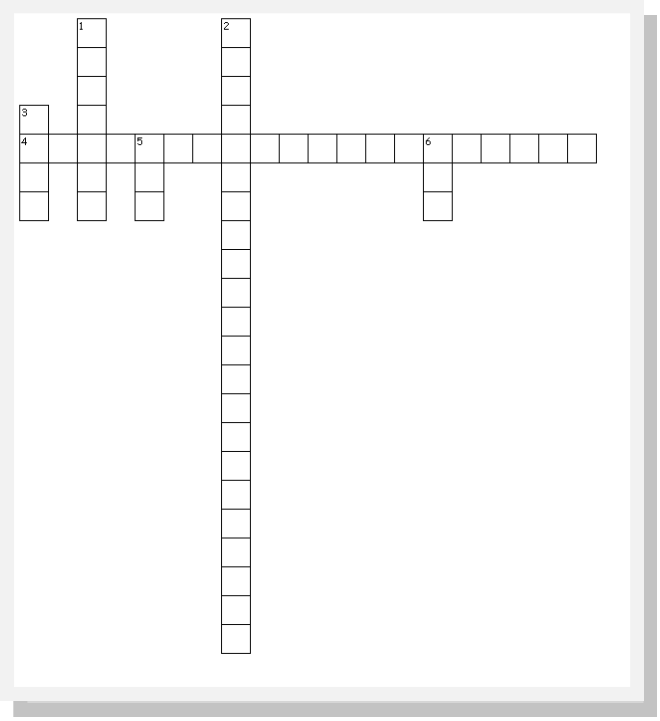
NEWS flash!

Do you know how your facility is doing in the QI goal of specimen transit time? What changes can be made to improve outcomes?

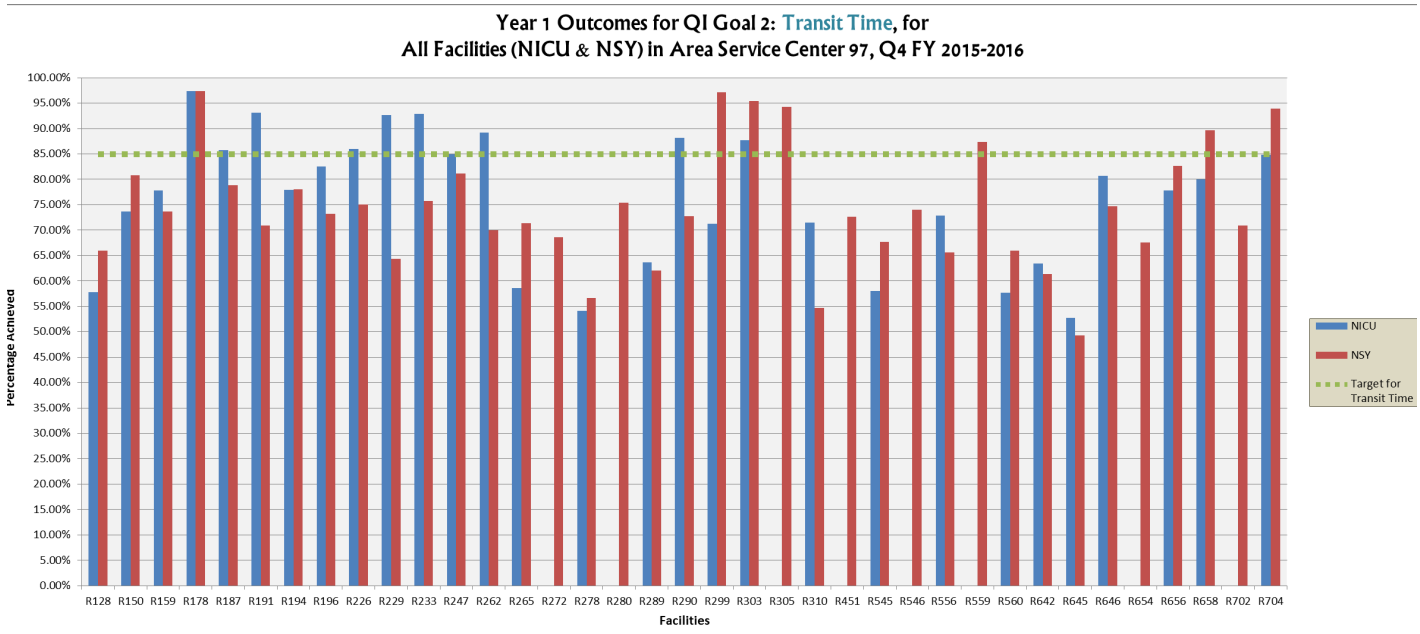
Newborn Screening: Do You Know Your Stuff?



Across:
4. Rare genetic disorder that causes a progressive destruction of myelin (hint: it's newly added to NBS panel in California)

Down:
1. Medical doctor credited for introducing population-level newborn screening, which originally tested only for PKU
2. Technology that allowed for massive expansion of the NBS panel, allowing us to test for a myriad of metabolic disorders (hint: it's abbreviated MS/MS)
3. Abbreviation for the Neonatal and Prenatal Screening lab
5. Abbreviation for "nothing by mouth" often used in medical care
6. Intravenous nutrition that infants receive in the NICU

SPECIMEN TRANSIT TIME ACROSS OUR AREA SERVICE CENTER



Answers to crossword: *across* 4. adrenoleukodystrophy, *down* 1. Guthrie, 2. tandem mass spectrometry, 3. NAPS, 5. NPO, 6. TPN