Newborn Screening On-the-Spot

Announcements:

Congratulations are in Order!

After 34 years of outstanding service to the California Newborn Screening Program (NBS), UCLA Area Service Center (ASC), Project Director, Kathy Jew, RN, MPH has retired in February 2015. During her career, the NBS Program has expanded from screening two disorders to 80 disorders including 40 metabolic conditions. Additionally, Kathy's expertise has influenced program design and her experience has gained statewide recognition in the field of Genetic Disease. Through her leadership and commitment, she has made innumerable contributions to our ASC, the Children's Hospital of Los Angeles, the Department of Pediatric Genetics at UCLA and to the state NBS Program. Lastly, in her tenure positions as a Coordinator and Project Director, Kathy's presence not only touched our lives but had a significant impact on many lives in the community.

We congratulate Kathy on her new journey and wish her the very best in her retirement!

Need More Information on Health Information Exchange HIE/HL7?

As highlighted in our 2014 ASC Spring Newsletter, the CA NBS Program has introduced a new project to provide facilities with newborn screening results electronically. This HIE/HL7 project was developed to meet the federal guidelines of "meaningful use", which enforces health care entities to move away from paper-based manual processes to improve patient safety. The first wave of this project was implemented in July of last year and the second wave is expected to start late 2015 or early 2016.

To learn more about how to sign up, or to get up to speed on the content of HIE/HL7, please visit us on the web at :

www.cdph.ca.gov/programs/nbs/Pages/HIE.aspx

Did You Know that Every Year in April, Public Health Week and Minority Health Awareness Month are Celebrated Nationally?

In remembrance of these two anniversaries, our ASC has highlighted Biotinidase Deficiency, also known as BD disorder as our feature story. Please turn to pages 2-3, to learn about this very rare but easily treatable metabolic disorder and the characteristics of BD distribution throughout our ASC population over the past seven years.

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

A Brief look at Biotinidase Deficiency

Biotinidase Deficiency (BD) is an inherited metabolic disorder that if not treated early can cause neurological, cutaneous and biochemical conditions. Often these conditions can become severe or even life threatening.

Individuals with BD have a defective biotinidase enzyme in which the biotin vitamin cannot be recycled. Moreover, BD deficient individuals fail to release biotin from dietary products. Although there may be related conditions there are only two types of BD; profound deficiency or partial deficiency.

Individuals with profound deficiency have very little biotinidase activity in the serum (<10% of the normal mean activity value) and if left untreated can exhibit seizures, feeding or breathing difficulties, rashes of the skin, alopecia, developmental delays, visual and hearing problems including: pinkeye and sensorineural hearing deficit. On the other hand, individuals with partial deficiency have more biotindase activity in their serum ranging between 10-30% and are often asymptomatic. Symptoms if they do occur develop upon illnesses, stress or poor diet.

BD is treated by life-long ingestion of medically prescribed biotin supplements. Over-the-counter biotin vitamins will not treat BD and should not be used as an alternative.

The genetic makeup and demographics of BD are quantified in every racial and ethnic group. However, the prevalence of BD is highest among those with origins of Greek, Middle Eastern and Hispanic roots. In California, the incidence rate is highest among Hispanics.

Family support for BD is available. For more information please visit http://biotinidasedeficiency.20m.com/

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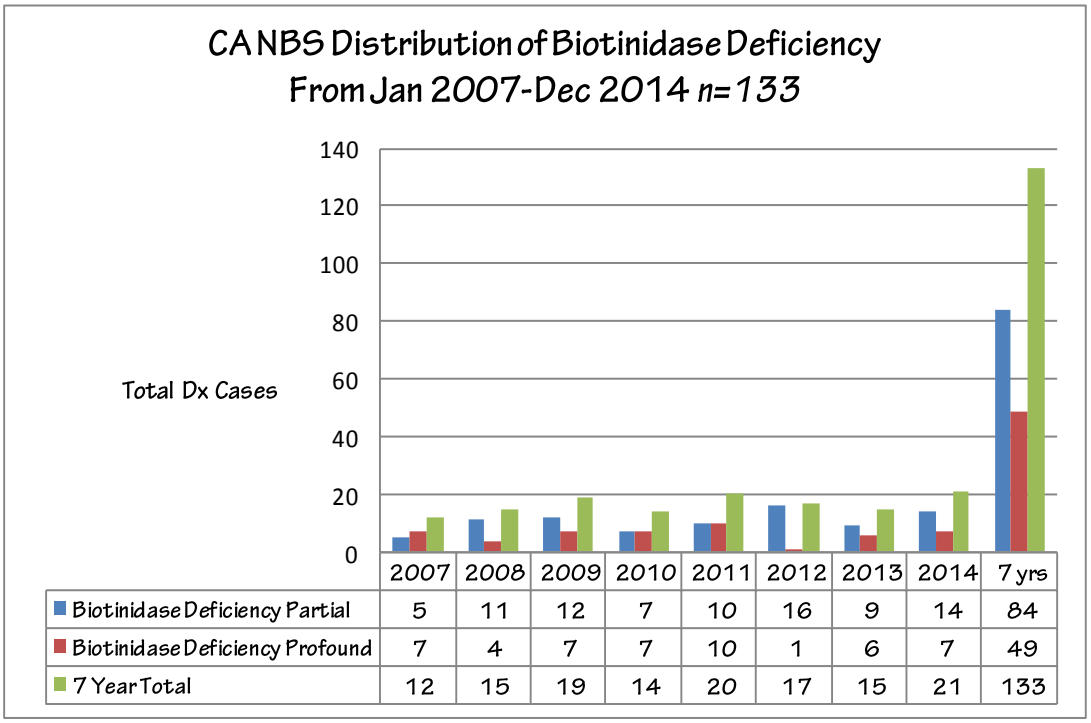
Looking for a Community Event Related to Newborn Screening in your Area?

Luckily there are several events that take place year around. Most of these local sponsored events provide education or awareness to health professionals and the community at large. Here are two upcoming events that you may find of interest:

1. 2nd Annual National Sickle Cell Disease Walkathon (Sickle Cell Disease Foundation of CA), Saturday, August 29th, Central Park, 11200 Baseline Road, Rancho Cucamonga, CA . Learn more at: www.scdfc.org

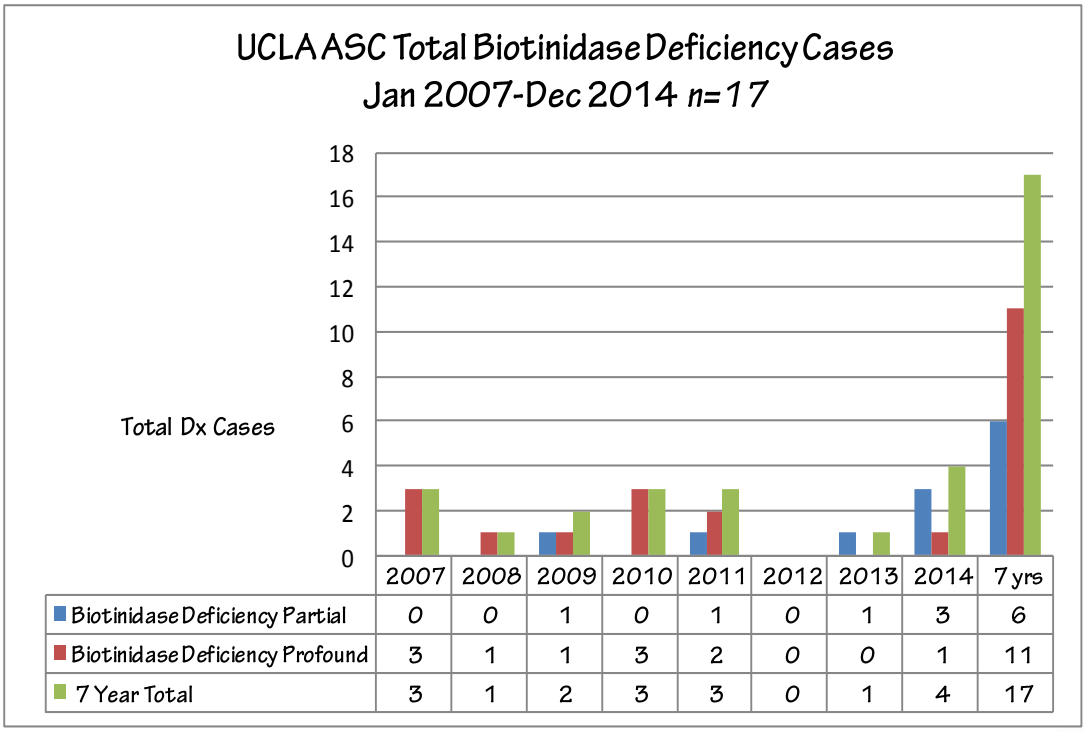
2. 2nd Annual Xtreme Hike (Cystic Fibrosis Foundation, Los Angeles) September 11-13th, Sequoia National Park. Register online at: www.cff.org/Chapters/losangeles/

A Brief look at Biotinidase Deficiency Continued...



	2007	2008	2009	2010	2011	2012	2013	2014	7 yrs
Biotinidase Deficiency Partial	5	11	12	7	10	16	9	14	84
Biotinidase Deficiency Profound	7	4	7	7	10	1	6	7	49
7 Year Total	12	15	19	14	20	17	15	21	133

"The genetic makeup and demographics of BD are quantified in every racial & ethnic group"



	2007	2008	2009	2010	2011	2012	2013	2014	7 yrs
Biotinidase Deficiency Partial	0	0	1	0	1	0	1	3	6
Biotinidase Deficiency Profound	3	1	1	3	2	0	0	1	11
7 Year Total	3	1	2	3	3	0	1	4	17

Current Events on Online Specimen Tracking (OST)

Every licensed perinatal hospital throughout the state of California is required to use Online Specimen Tracking (OST) as part of your daily practices to ensure all babies born at your facility received the Newborn Screening Test. This online tool, allows your facility to verify within 2-5 days after collection that each specimen sent from your facility was received by our state laboratory for processing.

Additionally, your facility is required to complete the following tasks:

* Report missing specimens by the newborns seventh day of age
* Report specimens not collected at your facility

You can also identify data entry errors before a baby's results mailer is generated, and request duplicate results mailers to be sent if needed. You may request duplicate results mailers for babies up until six months of age. Moreover, OST users can be designated from any department related to the NBS processes; HIM, LAB, Nursery or NICU. Every facility is required to have at least one main user and a back-up. As well, facility restrictions of three maximum users has been cancelled by GDSP. You may now request additional user passwords. Please do not share your passwords!

In support of this state requirement, the UCLA ASC has developed an OST Report Card for your quarterly review. This report card was created to assist your facility with complying with state demands and with educating your facility on your OST performance. For reference, your facility's Quarter 1 and Quarter 2 OST Report Cards were sent out at the end of October 2014 and January 2015. Your latest scores can be reviewed on your facility's Quarter 3 Report Card sent on April 30, 2015.

Furthermore, the UCLA ASC has developed a training to educate/support your facility in understanding all OST processes using the Screening Information System (SIS) software, and with introducing your facility to the GDSP HIE/HL7 Project. UCLA ASC scheduling for your facility's training will take place over this year and is separate from your Bi/annual site visit meetings.

All UCLA ASC facilities, regardless of your quarterly report card scoring will receive training!

As a *Reminder ...*

Our ASC is still conducting a contest for HEPP Super Stars!

We are looking to reward facilities that receive 100% compliance on the "Big Three Fields"

For more information, please refer back to our Fall 2014 Newsletter

Think You Know Newborn Screening? Let's See… Try Unscrambling These Terms.

1. OLBDO TOSSP _____
2. RTETNMATE _____
3. TIODIAIEBSN DEIYECNCFI _____
4. FLIAYM _____
5. GEEITCSN _____
6. TRABYLOAOR _____
7. ETDAEQAUIN _____
8. LBOONGEIHM _____

[1] Blood Spots, [2] Treatment, [3] Biotinidase Deficiency, [4] Family, [5] Genetics, [6] Laboratory, [7] Inadequate, [8] Hemoglobin