

## Newborn Screening Success Story

A few months ago, the Office of Newborn Screening (NBS) welcomed Steve, Angela, and their two children Danica and Braeden Taylor to the Public Health Laboratory (PHL) to share their NBS story. Shortly after Danica was born in 2008, she had a small specimen of blood collected from her heel as a part of her routine NBS testing. Upon reviewing her results, NBS Chemist Bill Hoffman noticed an abnormal laboratory result. Despite the result not indicating any of the disorders screened for at the time of Danica's birth, Bill recognized a pattern consistent with a rare, metabolic disorder. The follow-up team contacted her doctor and advised them of the concerning results.

Steve, a DOH IT employee of about 20 years, recalled receiving a call from Danica's doctor who informed him of the abnormal laboratory results. With the help of the NBS program and Seattle Children's Biochemical Genetics team, Danica had diagnostic testing which led to a diagnosis of a rare genetic metabolic disorder: methylmalonic acidemia Cobalamin C (cbIC) deficiency. With this early diagnosis, Danica was able to receive early intervention and treatment. Because the doctors were aware of his sister's diagnosis, Braeden was promptly tested at birth. His

testing confirmed cbIC deficiency as well. Both Taylor children take dietary supplements and injection medicine to treat their cbIC in addition to following up with a genetic specialist. Danica, who enjoys playing soccer and being on her school's track & field team, remarked how amazing it is that no one would know she and her brother have cbIC by simply looking at them. Now fifteen, she will soon start high school and learn how to give injections to herself. Braeden enjoys soccer, basketball, and karate. Braeden shared that cbIC affects his vision but was excited that his doctors told him that he'll be able to drive some day! Both the Taylor family and the NBS staff benefited from their visit to the PHL.

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### Practice Guidelines

The following practice guidelines have been developed by the Clinical Laboratory Advisory Council. They can be accessed at the [LQA website](#).

Acute Diarrhea	Lipid Screening
Anemia	PAP Smear Referral
ANA	Point-of-Care Testing
Bioterrorism Event Mgmt	PSA
Bleeding Disorders	Rash Illness
Chlamydia	Red Cell Transfusion
Diabetes	Renal Disease
Group A Strep Pharyngitis	STD
Group B Streptococcus	Thyroid
Hepatitis	Tuberculosis
HIV	Urinalysis
Infectious Diarrhea	Wellness
Intestinal Parasites	

**Training Classes:**

- 2023 Northwest Laboratory Symposium (NWMLS)**  
**October 26, 27 (Virtual)**
- 2023 30th Annual Clinical Laboratory Conference**  
**November 13, 2023 (Virtual)**

The Calendar of Events is a list of upcoming conferences, deadlines, and other dates of interest to the clinical laboratory community. If you have events that you would like to have included, please email the information to Chuck Talburt at email listed on page 1. Information must be received at least one month before the scheduled event. The editor reserves the right to make final decisions on inclusion.

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Secretary, DOH: Umair A. Shah, MD, MPH  
Acting Health Officer: Scott Lindquist, MD  
Director, PHL: Romesh Gautam, PhD  
Program Manager, LQA: Jessica Holloway  
Editor: Chuck Talburt  
Circulation: Chuck Talburt

Comments, letters to the editor, information for publication, and requests for subscription can be directed to:

*ELABORATIONS*  
1610 NE 150th St  
Shoreline, WA 98155

e-mail address: [chuck.talburt@doh.wa.gov](mailto:chuck.talburt@doh.wa.gov)

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