WASHINGTON STATE MEDICAL HOME PARTNERSHIPS PROJECT
CHILD HEALTH NOTES – DECEMBER 2013

Promoting early identification and partnerships between families, primary health care providers & the community.

This newsletter provides physicians, nurse practitioners, primary health care providers, public health centers and community partners with current information regarding identification and management of special health issues for children. Contributing to this issue: Sheila Weiss & Mike Glass at the Washington State Department of Health Office of Newborn Screening, Donna Borgford-Parnell, RN Public Health Seattle & King County and Katherine TeKolste, MD of the Medical Home Partnerships Project at the University of WA Center on Human Development & Disability

Saving lives with a simple blood spot

Newborn Screening Panel Updates for Washington State

Newborn screening detects health problems in newborn babies that would not otherwise be known without screening. If left untreated, these conditions can lead to brain damage, life-long disability, and in some cases, even death. New laboratory techniques and enhanced technology make it possible to screen for more congenital disorders in newborns. Early detection prevents chronic disability and helps affected children and their families' access support and services to assure the best health possible.

Currently, 27 disorders are on the Washington State panel of newborn screening tests. Each year, approximately 175 infants (about 1 in 500 births) in Washington are diagnosed with one of these disorders. Beginning January 2014, severe combined immunodeficiency disorders (SCID – a.k.a. Bubble Boy disease) will be added to the mandatory panel.

Successful newborn screening requires collaboration between the Washington State Department of Health Newborn Screening Program, health care facilities (hospitals, local health departments, clinics), health care providers (pediatricians, family practice physicians, nurse practitioners, midwives), and families of newborns.

Who Is Screened?

Washington State law requires that every newborn be tested prior to discharge from the hospital or within five days of age.

In addition to the required first specimen, it is strongly recommended that every baby born in Washington have a second screening specimen collected between 7 and 14 days of age.

A third screen between 4-6 weeks of age or prior to discharge is recommended for sick and premature infants.

Which Disorders Are Screened In Washington State?

Disorder:	Possible Outcomes if Treatment Delayed
Metabolic Disorders:	
Amino acid disorders Phenylketonuria (PKU) & 5 others	intellectual disabilities, coma, seizures, & death
Biotinidase deficiency	metabolic crisis, coma & death
Fatty acid oxidation disorders Medium chain acyl-CoA dehydrogenase (MCAD) deficiency & 4 others	serious damage affecting the brain, liver, heart, eyes, muscle, & death
Galactosemia	liver failure, sepsis & death
Organic acid disorders Glutaric acidemia type 1 (GA-1) & 7 others	severe nerve & physical damage & death
Disorders of Endocrine System:	
Congenital hypothyroidism (CH)	severe neurological & developmental damage
Congenital adrenal hyperplasia (CAH)	life-threatening salt loss leading to death
Cystic fibrosis (CF)	chronic lung & digestive system disease
Other Disorders:	
Sickle cell disease (3 forms) & other hemoglobinopathies	splenic enlargement, severe anemia, susceptibility to bacterial infections

Things to Consider For Your Practice:

- Assure that all infants in your practice have a second screening test and document the screening status of every child
- Respond quickly to information and specimen requests from the Newborn Screening Program
- Be familiar with the disorders screened in Washington State
- Use the 'Information and Resources' below to find information about the disorders
- Talk with families about newborn screening and assure referral to speciality care clinics as necessary

What Happens If Screening Results Are Positive?

All specimens that are determined to be presumptive positive through the Newborn Screening Program are followed up immediately through direct telephone contact with the child's primary health care provider. This is to ensure that diagnostic testing and treatment, if indicated, is initiated as quickly as possible. Following a definitive diagnosis, a long-term, disease-specific medical management program is implemented.

INFORMATION AND RESOURCES:		
Referral Centers	**Diagnostic and follow-up services for disorders identified by the WA State Newborn Screening Program:** **Metabolic Disorders:** University of WA - PKU/Biochemical Genetics Clinic – CHDD..... 1-877-685-3015 or 206-598-1800 Seattle Children's – Biochemical Genetics Clinic ..... (206) 987-3012	
	Endocrine Disorders: Congenital Adrenal Hyperplasia and Congenital Hypothyroidism Endocrine Clinic – Seattle Children's ..... (206) 987-2640 Endocrine Clinic - Mary Bridge Children's Hospital, Tacoma ..... (253) 792-6630 Congenital Hypothyroidism Clinic – Univ.of WA – CHDD ..... (206) 598-1800 Madigan Army Medical Center (military) ..... (253) 968-0109	
	Cystic Fibrosis: CF Clinics at- Seattle Children's Hospital, Seattle ..... (206) 987-2024 Mary Bridge Children's Hospital, Tacoma..... (253) 403-4141 Providence Physician's Group at Sacred Heart, Spokane ..... (509) 474-6960 Oregon Health Sciences Center, Portland, OR..... (503) 494-8023 Madigan Army Medical Center (military) ..... (253) 968-1980	
	Hemoglobin Disorders (i.e. Sickle Cell Disease): Odessa Brown Children's Clinic, Seattle ..... (206) 987-7232 Mary Bridge Children's Hospital & Health Center, Tacoma..... (253) 594-1415 Sacred Heart Children's Hospital, Spokane ..... (509) 474-2777	
State:	**Washington State Newborn Screening Program** Washington State Department of Health Newborn Screening website. Provides separate pages for health professionals and parents.	(206) 418-5410; Toll free 1-866-660-9050 http://www.doh.wa.gov/YouandYourFamily/InfantsChildrenandTeens/NewbornScreening/HealthcareProfessionals E-mail: nbs.prog@doh.wa.gov
National:	**NewSTEPs (Newborn Screening Technical assistance and Evaluation Program)** Provides information about NBS laboratory practice, quality improvement, education and training, program evaluation and policy. **Save Babies Through Screening Foundation** Provides general information about newborn screening disorders, family stories, resources, and frequently asked questions.	https://newsteps.org http://www.savebabies.org