August 2014

Snohomish County Child Health Notes

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

Distributed by: Children with Special Health Care Needs and Snohomish County Early Intervention Program

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

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?

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

<table>
<thead>
<tr>
<th>Referral Centers</th>
<th>Diagnostic and follow-up services for disorders identified by the WA State Newborn Screening Program:</th>
</tr>
</thead>
</table>
| **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 |
| **Cystic Fibrosis: CF Clinics at:** | Seattle Children’s Hospital, Seattle ……………………………………………………………………….. 206.987.2024  
Mary Bridge Children’s Hospital, Tacoma……………………………………………………………. 253.792.6630 |
| **Hemoglobin Disorders (i.e. Sickle Cell Disease):** | Odessa Brown Children’s Clinic, Seattle …………………………………………………………………… 206.987.7232  
Mary Bridge Children’s Hospital & Health Center, Tacoma……………………………………… 253.792.6630 |

**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; 1.866.660.9050  
  [http://www.doh.wa.gov/YouandYourFamily/InfantsChildrenandTeens/NewbornScreening](http://www.doh.wa.gov/YouandYourFamily/InfantsChildrenandTeens/NewbornScreening)  
  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.  
  [https://newsteps.org/](https://newsteps.org/)

- **Save Babies Through Screening Foundation**  
  Provides general information about newborn screening disorders, family stories, resources, and frequently asked questions.  
  [http://savebabies.org/](http://savebabies.org/)

**Local:**

- **Children with Special Health Care Needs**  
  Snohomish County Early Intervention Program  
  425.339.8652  
  425.388.7402

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[Insert images of logos for Children with Special Health Care Needs, Snohomish County Early Intervention Program, Snohomish Health District, and Washington State Department of Health]