Newborn Screenings: Helping to PREVENT disabilities, "One Foot at a Time"

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Abstract

As Speech-Language Pathologists and Audiologists, we spend our lives working to make a difference in the lives of people with disabilities. But what if we could help to PREVENT some of those disabilities from occurring in the first place. We CAN!

What are Newborn Screenings?

"There are a number of potentially devastating diseases that can be present in a newborn, but hidden at the time of birth. These diseases, if undetected by newborn screening, have the potential to cause serious medical problems as the baby grows, severe disabilities that alter a life that could have otherwise been normal, or death (often sudden). One example of such an illness is phenylketonuria (PKU)," but there are many more. *www.savebabies.org*

How are Newborn Screenings Conducted?

Newborn screenings are simple heal prick/ blood spot tests that are taken on day 1 and often at week 2 of a baby's life. These blood spots are analyzed through tandem mass spectrometry to identify chemical markers of these disorders.

Photo with permission from March of Dimes.

What kinds of disorders are screened for?

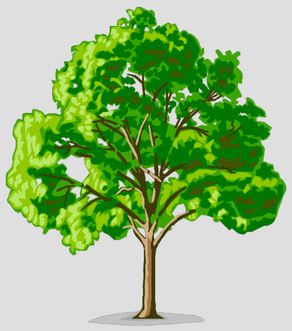
States vary in what disorders they screen for, but this is a typical list:

Arginosuccinic Acidemia (ASA)
Biotinidase Deficiency
Citrullinemia (Type I & II)
Congenital Adrenal Hyperplasia
Congenital Hypothyroidism
Cystic Fibrosis
Fatty Acid Oxidation (11)
Galactosemia
Hemoglobin S-Beta Thalassemia
Hemoglobin Variants
Homocystinuria
Hypermethioninemia
Hyperphenylalanimia
Maple Syrup Urine Disease
Organic Acidemia (12)
Phenylketonuria
Severe Combined Immune Deficiency (SCID)
Sickle Cell Disease
Tyrosemia (Type I,II,III)

www.slh.wisc.edu

Individually each of these disorders are quite rare, but collectively, 6000-7000 infants per year are diagnosed in the USA alone.
www.savebabies.org

Why are these disorders so deadly/ disabling?



Like a Tree without Chlorophyll
These children are lacking at least one of the chemicals necessary to metabolize food. Their bodies build up poisons instead.

Two ways to take the system down:
1) Slow and steady doesn't win the race.
2) Get sick and the whole system suddenly crashes.

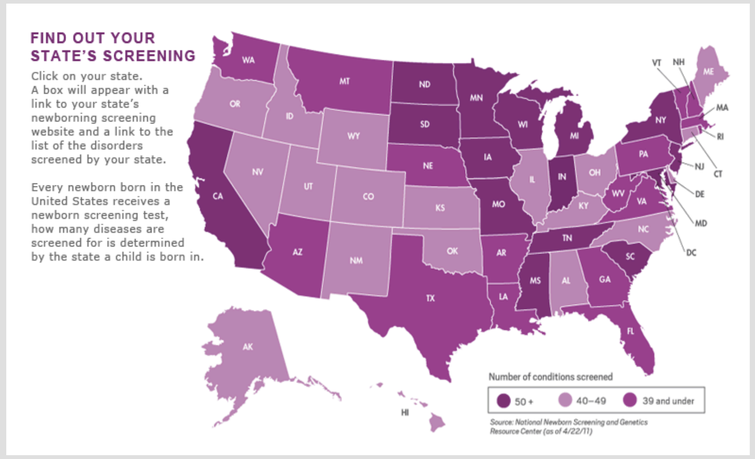
Not all of these disorders are metabolic disorders, but the analogy is often helpful.

What kinds of Problems can Result? (any of the following)

Global developmental delay
Intellectual disability
Autism
Cerebral Palsy-like symptoms
Spasticity/ Flaccidity/ Ataxia
Behavioral Challenges
Microcephaly/ Encephalopathy
Stroke
Speech- Language Delays
Juvenile Alzheimers/ Demensia/ Parkinsons
Sudden Infant Death Syndrome (SIDS)
Seizure Disorders
Failure to Thrive
Attention Deficit Disorder
And more

Saudubray et al. (2012)

What disorders does my state screen for?



www.savebabies.org

Why is it important to know the results?

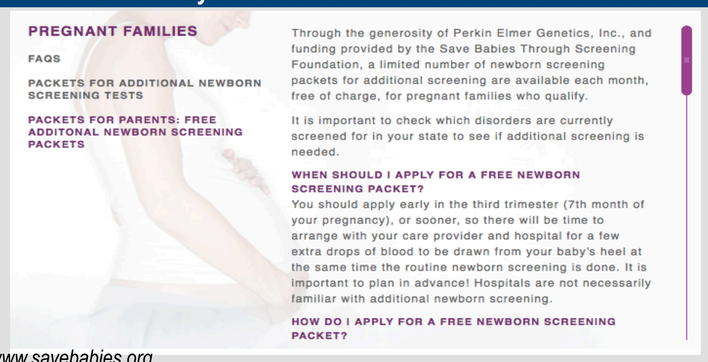
It is vital that you know the results from BOTH day 1 and week 2 tests!

Knowing the results from Both day 1 and week 2 tests can save your baby's life. Some of the chemical markers of these disorders build up slowly through time. Over the course of days or weeks, these poisons build up in the system. Therefore, the test on day 1 may not show a positive result, but the test on week 2 may.

What if the state doesn't screen for my child's disorder?

Watch for Red Flags: Progressive neuro-degeneration, developmental regression or plateauing, significant deterioration in behavior, unexplained movement disorder, siblings died suddenly/ multiple miscarriages, recurrent and unexplained vomiting, unexplained causes or early onset of any of the disorders listed. *www.tidebc.org*

How can a baby be screened for ALL of the known disorders?



www.savebabies.org

Most of these disorders are TREATABLE!

Galactosemia (GAL). Galactose-1-Phosphate Uridyltransferase (GALT) Deficiency - Failure to metabolize the milk sugar galactose results in GAL and occurs in about 1 in 50,000 U.S. newborns. The classical form detected by newborn screening can lead to *cataracts, liver cirrhosis, mental retardation and/or death.* Treatment is elimination of galactose from the diet usually by substituting soy for milk products. (1)

Homocystinuria (HCY). HCY is caused by an enzyme deficiency that blocks the metabolism of an amino acid that can lead to *mental retardation, osteoporosis, stroke, and other problems* if left undetected and untreated. The incidence is approximately 1 in 350,000 U.S. newborns. Treatment may involve a special restricted protein diet and supplemental medicines, including Vitamin B6. (1)

Maple Syrup Urine Disease (MSUD). MSUD is a defect in the way that the body metabolizes certain amino acids and is present in about 1 in 200,000 U.S. newborns. Early detection and treatment with a special restricted protein diet *can prevent death and severe mental retardation.* *www.dshs.state.tx*

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What should I be doing to make a difference?

1) Become educated about newborn screenings and treatable metabolic disorders.
2) Educate others about the importance of newborn screenings.
3) Watch for RED FLAGS. Know the signs of metabolic disorders. If you have a patient, friend, family member who is showing these signs, refer them to the TIDEBC website.
4) Refer to genetics. If you suspect that a patient (child or adult) has a metabolic disorder, refer them to a genetic/ metabolic clinic.

What should I do if my child has a Positive screening?

1) Don't freak out. These screenings are designed to be very sensitive. They don't want to miss a single child. Consequently, false positives happen.
2) Take it seriously. Each state has a special genetic/ metabolic clinic that can do further testing do determine if your baby truly has a disorder.
3) Follow up. If the child does have a disorder, the genetic/ metabolic clinic will provide specific recommendations. Follow through is crucial.

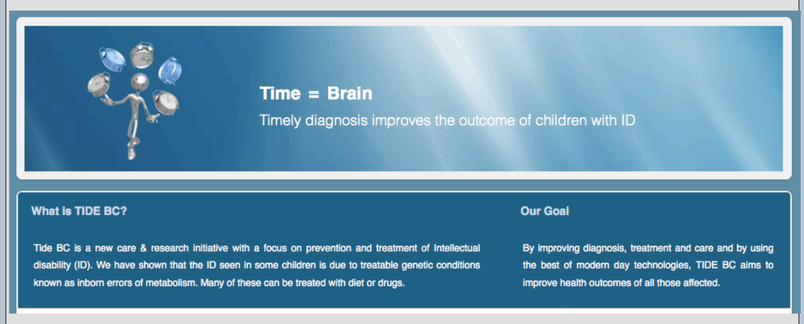
What if I notice these problems in a person who is older than newborn?

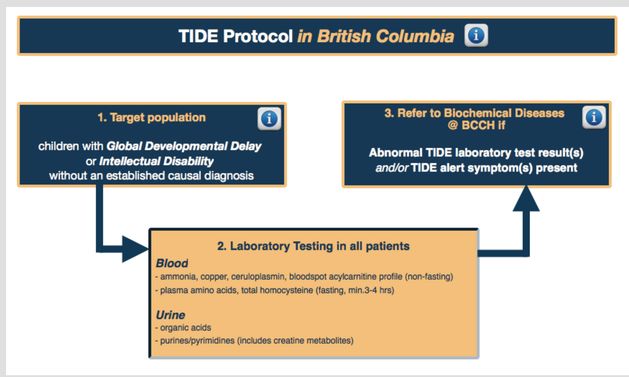
Document the changes and refer the patient to Genetics/ Metabolics.

An example of a thorough metabolic protocol is at the website below.

A recent meta-analysis found a total of 81 TREATABLE causes of intellectual disabilities. Up to 14% of individuals with intellectual disabilities were diagnosed with treatable metabolic disorders following through metabolic testing. *(van Karnebeek & Stockler, 2012)*

TIDE BC
Treatable Intellectual Disability Endeavor in B.C.





www.tidebc.org

Conclusion

Knowledge is power. We can make a difference and improve outcomes!

References, continued

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