

Newborn Screening for Thalassemia

By Kimberly Bouchard, BA, and Lauren Ebe, BA

What is newborn screening?

Newborn screening is a way to test infants for treatable diseases. Early identification of babies born with treatable diseases makes it possible to begin health-enhancing interventions right away.

Newborn screening began in the 1960s. Over the years, new tests have been added. Diseases included in newborn screening have the following in common:

- They can be detected within 24 to 48 hours after birth
- They have a treatment
- There is known benefit in screening for the disease.

Newborn screening started in the 1960s

Newborn screening started in the 1960s with a test to detect phenylketonuria (PKU). Those with this disease lack an enzyme needed to break down phenylalanine, an amino acid found in food.

Affected children may be treated with a special diet. But if the disease is not detected and treated early in life, it can impair cognitive development and later cognitive functioning.

PKU fits the criteria for useful newborn screening because:

- A test of the baby's blood or urine within 24 to 48 hours after birth determines a baby's phenylalanine level; if the level is high, further testing can be done

to determine if the baby has PKU

- PKU can be treated with a modified diet
- Treating PKU prevents cognitive impairment, which occurs if the disease is left untreated.

PKU is on the list of disorders screened for in all 50 states. Beyond this core list, that also includes congenital hypothyroidism and galactosemia, each state decides what other disorders to test for. Many states test for up to 30 illnesses; some test for as few as 10.

In California, newborn screening includes hemoglobinopathies—diseases of the blood—such as **thalassemia** and sickle cell disease.

How does screening work?

A blood sample is taken by pricking the baby's heel, typically while the baby is still in the hospital. The blood sample is tested for specific diseases.

If the test is normal, testing ends. But if the test is abnormal, additional testing confirms whether or not a newborn has a particular disease. If an infant is diagnosed with a disease, a medical team works with the family to begin treatment immediately.

Screening newborns for thalassemia

Newborn screening that tests for several varieties of thalassemia begins by testing for abnormal hemoglobin. If it's

found, more testing determines if the infant has thalassemia, and which type of thalassemia the baby may have.

Remember, thalassemia disease can also be detected later in life. People can also be tested for thalassemia trait by having a simple blood test.

Because newborn screening for thalassemia is not universal, in California it only began in the early 1990's, there are many people who were not screened for the disease at birth. However, testing is available for children and adults.

Kimberly Bouchard and Lauren Ebe are first-year medical students at Drexel University College of Medicine in Philadelphia. They are interning with the Thalassemia Outreach Program at Children's Hospital Oakland for the community health component of their seven-week primary care practicum.

Lauren and Kimberly are interested in specializing in pediatrics. Looking forward to their careers in medicine, they realize how important it is to provide education to the greater community. They hope to learn how to identify community needs and tailor outreach to specific patient populations.

For more information on newborn screening and what tests are done in your state please refer to the following Web sites:

www.kidshealth.org/parent/system/medical/newborn_screening_tests.html

www.nlm.nih.gov/medlineplus/newbornscreening.html

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*Forever
in our hearts!*

COMING SOON

The Standards of Care for
Thalassemia, 2nd Edition
In the past year, Dr. Vichinsky
and many experts in the field of
thalassemia have been working to
update the 2001 Standards of
Care. It will be available Sept. 1 at
www.thalassemia.com. If you
would like a hard copy, please
email LLevine@mail.cho.org with
your request.

Highlights



On Feb. 9, **Jon Miller** and **Maricar Boyle** from the Children's Hospital & Research Center Foundation, **Elliott Vichinsky, MD**, and **Laurice Levine**, presented the Italian Catholic Federation (ICF) Central Council with the Bertha Wright Award. This award honors donors who have contributed a minimum of \$100,000 to programs in the hospital. Since 1999, the ICF has contributed more than \$480,000 to the thalassemia program at Children's Hospital Oakland. We thank the federation for their years of dedication, generosity and support.

EVENTS CALENDAR

If you would like to volunteer in
the thalassemia outreach program
please call **Eve Alley**, **Thalassemia
Outreach Coordinator**, at 510-428-
3885, ext. 4398.

July 10-13: Painted Turtle Camp, Lake
Hughes, Calif.

July 12-13: Lotus Festival, Los Angeles

July 17: Children's Hospital Oakland
Multicultural Health Fair, Oakland,
Calif.

Aug 29-31: ICF Annual Convention,
Anaheim, Calif.

Oct/Nov: Date TBA – Thal Retreat

Dec: Date TBA – Thalassemia Annual
Holiday Party



Huythong Nguyen celebrates his
40th birthday!



Lydia Salib graduates with her MBA
from Case Western Reserve
University in Cleveland.

Thalassemia trait characteristics

People with thalassemia may be found anywhere in the world, but it's most commonly found among particular ethnicities, including those whose ancestry is:

- Italian
- Greek
- Transcaucasian, including people from Georgia, Armenia, and Azerbaijan
- Southeast Asian, including people from Vietnam, Laos, Thailand, Singapore, Cambodia, the Philippines, Indonesia, Burma, and Malaysia
- Chinese
- South Asian, including people from India, Pakistan and Bangladesh
- Middle Eastern.

Thalassemia is common in parts of the world where malaria is prevalent, because the thalassemia trait helps protect those with it against malaria.

The two main types of thalassemia trait are alpha-thalassemia trait and beta-thalassemia trait. There are four alpha-globin genes and two beta-globin genes.

Four sub-types of alpha-thalassemia trait

- *Silent alpha-thalassemia trait* occurs when one of four alpha-globin genes is missing. No symptoms are associated with silent alpha-thalassemia trait.

- *Alpha-thalassemia trait* occurs when two of four alpha-globin genes are missing. No symptoms are associated with alpha-thalassemia trait.

- *Hemoglobin H disease* occurs when three of four alpha-globin genes are missing. Symptoms associated with Hemoglobin H disease include becoming severely anemic when exposed to certain medicines and other agents.

- *Hydrops fatalis* occurs when all four alpha-globin genes are missing. Hydrops fatalis may cause stillbirth or death soon

after delivery, unless it is diagnosed in utero, and the infant is transfused in utero or at birth.

Beta-thalassemia trait

Beta-thalassemia trait occurs when there is one normal beta-globin gene and one altered beta-globin gene. People with this combination produce little to no beta-globin.

While no scientific evidence shows a person with beta-thalassemia trait has any symptoms, women with this trait may be more likely to develop anemia during pregnancy than women without the trait.

Misdiagnosing thalassemia as iron deficiency

Most types of thalassemia trait cause red blood cells to be slightly smaller than usual, leading to a condition called microcytosis. Since iron deficiency is the most common cause of microcytosis, doctors often mistakenly misdiagnose thalassemia trait as iron deficiency and prescribe iron supplements.

But thalassemia trait and iron deficiency are different problems. Before prescribing iron supplements, doctors should rule out thalassemia trait and/or perform lab tests to evaluate iron levels.

While a person with thalassemia trait can also be iron deficient, if they aren't, taking iron supplements may lead to excess body iron. This iron may be deposited in many areas of the body and over time, may cause organ damage.

Testing for thalassemia trait is easy

A blood sample is drawn and screened with:

- Hemoglobin electrophoresis with quantitative hemoglobin A2 and hemoglobin F

- Complete Blood Count (CBC)
- Iron studies, including free erythrocyte, proto porphyrin, and ferritin.

Sometimes it takes more than these screening tests to determine trait status. If iron deficiency is detected, the screening must be repeated after a course of iron supplement therapy. If iron deficiency exists, iron supplements should improve this condition. If there is no improvement, the person may have thalassemia trait.

Sometimes DNA testing that directly examines the alpha- and/or beta-globin genes is necessary. DNA testing is the only way to confirm the silent alpha-thalassemia trait and hemoglobin H-Constant Spring (another mutation of Hemoglobin H Disease).

Potential parents: Know your status

Potential parents need to know their thalassemia trait status, especially if their ancestry is one in which thalassemia is common. And because many types of thalassemia trait have no associated symptoms, testing is the only way to find out one's trait status.

If both parents have the thalassemia trait, there is a 25 percent chance their child will be born with thalassemia major. Unfortunately, parents who aren't tested sometimes only find out they carry the thalassemia trait when their child is born with thalassemia major, a severe form of the disease.

If you learn you have thalassemia trait and would like more information, please contact the Thalassemia Outreach Program at Children's Hospital Oakland, at 510-428-3885, ext. 4398, or visit our Web site at www.thalassemia.com.



PERSPECTIVES

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The Painted Turtle

A life-enhancing summer experience for children with transfusion-dependent thalassemia

by Ben Meisel, MD, FAAP, Medical Director, The Painted Turtle

The Painted Turtle, located in Lake Hughes, Calif., is a member of the Association of Hole in the Wall Camps founded by actor Paul Newman. The camps, which are American Camp Association Accredited, are considered the finest multi-disease medical camps in the world.

This year, in collaboration with California's 16 nonprofit children's hospitals the Painted Turtle offers a six-day camp experience for children who have transfusion-dependent thalassemia, from July 10 to July 15. Campers must be 7 to 16-years-old to attend. All campers and families attend camp free of charge.

The Painted Turtle's mission is to provide a year-round, life-changing environment and authentic camp experience for children with chronic and life-threatening illnesses. Activities range from aquatics to woodshop. The Painted Turtle supports children's medical needs,

inspires them to reach beyond their illnesses, and provides care, education, and respite for their families. The main goal of the Painted Turtle is to ensure that campers have a fun, safe, and empowering experience while fostering the following outcomes:

- **Developmental outcomes:** Self-confidence, communication skills, teamwork, responsibility, tolerance, technical skills, and creativity.

- **Program participation:** Programs are all "Challenge by Choice." The Painted Turtle champions the courage of all campers who try something new, whether it is embarking on the ropes course adventure in a wheelchair or performing on Stage Night in silly costumes.

- **Medical compliance:** During the summer, the nursing staff distributes the campers' medications during mealtimes in the dining hall. This establishes a sense of community and camaraderie among

peers. The Painted Turtle applauds all newly acquired medical-care skills through staff and peer recognition of a camper's accomplishment.

The Painted Turtle welcomes children from all ethnic, racial, and socioeconomic backgrounds. All camp sessions are illness-specific. To date, the Painted Turtle has served almost 5,500 campers and family members.

"It's the coolest place ever where you are not afraid to make a fool of yourself...and have a great time and be understood."

—Painted Turtle camper

To learn more about Painted Turtle:

www.thepaintedturtle.org

Camper Recruiter: Jill Mellor
jillm@thepaintedturtle.org or (661)
724-1768, ext. 02.

PERSPECTIVES

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Children's Hospital & Research Center Oakland
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Address change requested



Children's Hospital &
Research Center Oakland
Thalassemia
Outreach

Perspectives Newsletter is produced by the thalassemia outreach coordinator, Laurice Levine, MA, CCLS, and the Communications department at Children's Hospital & Research Center Oakland. For questions regarding the newsletter or for more information on thalassemia, call 510-428-3885, ext. 5427, or visit www.thalassemia.com.