

## **Cholesterol-lowering drug shows promise against serious infections in sickle cell disease**

*St. Jude Children's Research Hospital scientists lead effort that identifies a novel way statins protect cells from a host of bacterial toxins*

**MEMPHIS, Tenn. – January 19, 2010** New research suggests a family of widely used cholesterol-lowering drugs might help protect individuals from serious illness following bacterial infection, including the pneumococcal infections that pose a deadly threat to those with sickle cell disease.

Research led by St. Jude Children's Research Hospital investigators reported that drugs called statins employ several methods to dampen inflammation and block pneumococcus and certain other bacteria from infecting cells and spreading throughout the body. Elaine Tuomanen, M.D., St. Jude Infectious Diseases chair, said those methods include a newly identified mechanism that statins use to protect healthy cells by blocking the toxicity of an entire class of bacteria. Along with pneumococcus, that class includes diphtheria, tetanus, listeria and group A streptococcus, which is also known as the flesh-eating bacterium. Tuomanen is co-senior author of the study with Carlos Orihuela, Ph.D., University of Texas Health Science Center at San Antonio (UTHSCSA). The work is published in the January 19 advanced, online edition of *The Journal of Clinical Investigation*.

The results provide the foundation for a possible future study to determine if statins, already widely used to lower cholesterol in adults, might protect children with sickle cell disease (SCD) from serious pneumococcal infection. SCD is an inherited blood disorder. The findings also suggest statins might protect others at high risk for pneumonia due to chronic inflammation of the lungs or blood vessels. In this study, scientists reported that statins prolonged the lives of mice with sickle cell disease following infection with the pneumococcal bacteria. Researchers also reported that a day after being infected, the treated mice had fewer bacteria in their lungs and blood, suggesting statins slowed the spread of the infection.

Full Text of the journal article

[http://www.jci.org/articles/view/39843?search\[article\\_text\]=&search\[authors\\_text\]=rosch](http://www.jci.org/articles/view/39843?search[article_text]=&search[authors_text]=rosch)

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## **National Library of Medicine Launches Mobile MedlinePlus to Meet the Health Information Needs of an On-the-Go Public**

Wondering what the side effects are for your new prescription? Go to Mobile MedlinePlus (<http://m.medlineplus.gov>) while you're waiting for the pharmacist to fill your order!

Or, instantly look up the symptoms of H1N1 flu if you're at the supermarket and your child's school calls you to tell you he doesn't feel well.

The National Library of Medicine's Mobile Medline Plus builds on the NLM's MedlinePlus Internet service, which provides authoritative consumer health information to over 10 million visitors per month. These visitors access MedlinePlus (<http://medlineplus.gov>) from throughout the United States as well many other countries, and use desktop computers, laptops and even mobile devices to get there.

The mobile Internet audience is large and growing fast, almost doubling from February 2007 to February 2009. Some experts predict that within the next five years, more people will connect to the Internet via mobile devices than via desktop or laptop computers. People use their mobile devices to accomplish a variety of tasks, including finding health information. With this in mind, NLM developed the mobile version of MedlinePlus to bring high-quality health information to users on the go.

"We know that a huge number of people are seeking good health information on the Web," noted NLM Director Dr. Donald A.B. Lindberg. "What better way to reach out to them than by offering this new mobile service, which delivers trustworthy, consumer-friendly information instantly, anywhere?"

Mobile MedlinePlus is available in English and Spanish (<http://m.medlineplus.gov/spanish>) and includes a subset of content from the full Web site. It includes summaries for over 800 diseases, wellness topics, the latest health news, an illustrated medical encyclopedia, and information on prescription and over-the-counter medications.

For instance, you could visit the "Talking With Your Doctor" page on Mobile MedlinePlus to learn how to get the most out of your doctor's visit.

Mobile MedlinePlus can also help you when you're trying to choose an over-the-counter cold medicine at the drug store.

And if you're traveling abroad, you can use Mobile MedlinePlus to learn about safe drinking water.

Mobile MedlinePlus puts reliable health information at your fingertips.

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## **Firm Brings Gene Tests to Masses**

By [ANDREW POLLACK](#) Published: January 28, 2010  
<http://www.nytimes.com/2010/01/29/business/29gene.html>

Counsyl, is selling a test that it says can tell couples whether they are at risk of having children with a range of inherited diseases, including cystic fibrosis, Tay-Sachs, spinal muscular atrophy, sickle cell disease and Pompe disease. Once informed, Counsyl says, couples can take steps like using in vitro fertilization with genetic testing of the embryos, to avoid bearing children who would have the diseases, many of which are incurable and fatal in childhood. Some genetic testing of prospective parents is done now, but only for a few diseases like cystic fibrosis and Tay-Sachs, and only for certain ethnic groups. Each test can cost hundreds or even thousands of dollars.

Counsyl's test, which analyzes DNA from saliva samples, costs \$349 for an individual or \$698 for a couple. Similar tests from others are on the way, experts say. The trend shows that new technology could make possible widespread screening for the risk of passing on rare diseases, something that was simply not practical before.

“As a genetic counselor, I've been waiting for this for a really long time,” said Elena Ashkinadze, who does prenatal genetic counseling at the Robert Wood Johnson Medical School in New Brunswick, N.J.

But some experts caution that it is too soon to know how accurate Counsyl's test actually is, in part because neither the company nor any outside reviewer has published papers on its approach and results. And some experts say the company's Web site overstates the case. The company calls its product the Universal Genetic Test, for example, even though there are thousands of genetic diseases, not just the 100 Counsyl tests detect. “Everyone hopes there is a test that will provide a perfect baby, but the reality is that that single magic bullet doesn't exist,” said Dr. Joe Leigh Simpson, a geneticist and obstetrician and dean at Florida International University College of Medicine.

Still, Counsyl executives say the company, which has been operating quietly for a few months, has already administered thousands of the tests. The test is already offered by more than 100 fertility clinics around the country, and Counsyl says some insurers are paying for it. “For the same price that we would be checking cystic fibrosis, they are checking cystic fibrosis but also 100 other diseases,” said Dr. R. Ian Hardy, medical director of the Fertility Centers of New England. Dr. Hardy said two couples at his clinic who had taken the Counsyl test had already found they could be at risk of having a child with a genetic disease. The diseases Counsyl screens for are rare and caused by a mutation in a single gene. For most of the disorders, people with one mutated version of the gene and one normal version do not have the disease and often are not aware that they are carriers of the mutation. But if both parents have one mutated gene, each of their children will have a one in four chance of inheriting two mutated copies and having the disease.

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## **Resources for Transitioning Children to Adult Services**

If you have a responsibility for or interest in the health and well being of young adults with chronic medical conditions and disabilities, you may be interested in receiving the "Transition Digest". This free e-mail monthly newsletter typically includes information about emerging trends, promising health care transition practices, lessons learned, materials for youth and parents, transition related publications (from a broad range of journals), websites, meetings, and questions from subscribers. It does NOT address medical management, but rather focuses on issues like improving systems of care, promoting teen's autonomy and medical decision making skills, assessing transition readiness and facilitating the transfer between pediatric and adult providers.

If you are interested in receiving the Digest (at no cost) send a request by e-mail to [jgr@ichp.ufl.edu](mailto:jgr@ichp.ufl.edu) .

Past issues of the Transition Digest (from 2009 and 2008) are available, as PDFs at <http://hctransitions.ichp.ufl.edu/listserv.html>

Health care materials developed by the Institute for Child Health Policy can be seen at: <http://hctransitions.ichp.ufl.edu/hct-promo/>

John Reiss, PhD, Associate Professor of Pediatrics Institute for Child Health Policy  
University of Florida Gainesville, Florida 32610-0147

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## News From The NIH

RFI (request for info). <http://bbsrresponse.com/> It asks for feedback on a new group of funding opportunities called Basic Behavioral and Social Science Opportunity Network (OppNet).

**Responses are due Feb 19, 2010.**

The NIH Basic Behavioral and Social Science Opportunity Network (OppNet) is seeking input from the scientific community, health professionals, patient advocates, and the general public about current and emerging priorities in *basic* behavioral and social sciences research (b-BSSR) that offer the greatest potential for improving the Nation's health and well-being. This input will inform the OppNet strategic planning process and help OppNet meet its mission of pursuing opportunities for strengthening b-BSSR at the NIH while innovating beyond existing investments.

Through this RFI information is sought to identify priorities for b-BSSR activities that are consistent with the mission and goals of OppNet. Ideas for both short (1-2 years) and long (3-5 years) term activities that focus on humans or model animals are welcome. The scientific community, scientific organizations, health professionals, patient advocates, and the general public are invited to respond to the following:

1. The Challenge: Describe what you believe to be the most critical, health related challenges that will benefit from b-BSSR.
2. The Opportunity: Indicate the most promising b-BSSR targets that will inform solutions to those problems. Targets may include scientific research, research training or core/shared resources (e.g., databases, assessment tools) in the basic behavioral and social sciences.
3. Outcome Indicators: Describe what measures could be used to indicate whether the proposed activities were successful in meeting the Challenge(s).

We especially welcome suggestion of priority areas that are relevant to the missions and public health challenges of multiple NIH Institutes, Centers and Offices and as such, are appropriate activities for OppNet.

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## Articles in the Medical Literature

[Increased levels of the inflammatory biomarker C-reactive protein at baseline are associated with childhood sickle cell vasocclusive crises.](#)

Krishnan S, Setty Y, Betal SG, Vijender V, Rao K, Dampier C, Stuart M.

Br J Haematol. 2009 Dec 8.

. [Sickle cell disease at the dawn of the molecular era.](#)

Abboud MR, Musallam KM.

Hemoglobin. 2009;33 Suppl 1:S93-S106.

. [Pathophysiology of transfusional iron overload: contrasting patterns in thalassemia major and sickle cell disease.](#)

Porter JB.

Hemoglobin. 2009;33 Suppl 1:S37-45.

[Steps toward the prevention of hemoglobinopathies in the kingdom of saudi arabia.](#)

Al-Shahrani M.

Hemoglobin. 2009;33 Suppl 1:S21-4.

. [Prevention of hemoglobinopathies in egypt.](#)

El-Beshlawy A, Youssry I.

Hemoglobin. 2009;33 Suppl 1:S14-20.

. [Current understanding in the management of sickle cell disease.](#)

Inati A, Chabtini L, Mounayar M, Taher A.

Hemoglobin. 2009;33 Suppl 1:S107-15.

. [Hydroxyurea for children with sickle cell disease.](#)

Heeney MM, Ware RE.

Hematol Oncol Clin North Am. 2010 Feb;24(1):199-214.

. [Effects of a single sickling event on the mechanical fragility of sickle cell trait erythrocytes.](#)

Presley TD, Perlegas AS, Bain LE, Ballas SK, Nichols JS, Sabio H, Gladwin MT, Kato GJ, Kim-Shapiro DB.

Hemoglobin. 2010;34(1):24-36.

. [Hemoglobinopathies in North Africa: a review.](#)

Haj Khelil A, Denden S, Leban N, Daimi H, Lakhdhar R, Lefranc G, Ben Chibani J, Perrin P.

Hemoglobin. 2010;34(1):1-23.

. [Burden of influenza-related hospitalizations among children with sickle cell disease.](#)

Bundy DG, Strouse JJ, Casella JF, Miller MR.

Pediatrics. 2010 Feb;125(2):234-43. Epub 2010 Jan 25.

. [Blood transfusions for treating acute chest syndrome in people with sickle cell disease.](#)

Alhashimi D, Fedorowicz Z, Alhashimi F, Dastgiri S.

Cochrane Database Syst Rev. 2010 Jan 20;(1):CD007843.

. [Drugs for preventing red blood cell dehydration in people with sickle cell disease.](#)

Nagalla S, Ballas SK.

## Ask the Experts

### Q: WHAT'S THE NEWS IN SICKLE CELL BONE MARROW TRANSPLANTATION?

A: Bone marrow transplantation offers a cure for sickle cell disease, but is only a good option for a small group of people. That group just got a little bigger when researchers at the National Institutes of Health announced early success with a new way to make bone marrow transplantation (BMT) available for adults with sickle cell who were too sick for the standard ways of doing BMT.

Q: What are the details?

**WHY LOOK AT BMT FOR ADULTS WITH SICKLE CELL DISEASE, NOT CHILDREN** Using the standard BMT approach was fatal for the majority of adults with sickle cell because their organs were damaged by years of living with sickle cell disease. BMT programs excluded adults from this type of BMT, but the increasing life expectancy in sickle cell means there are probably more adults than children with sickle cell in the USA. Many doctors have been looking for new ways for adults with sickle cell disease to have safe and successful BMT.

The new approach from NIH features medications that heavily suppress the immune system - like kidney transplantation – instead of using the standard ways of doing BMT. Nine out of ten adults had successful transplants with this new approach and they appear to be cured of sickle cell. The other one adult had Graft Rejection (the transplanted cells did not grow) and still has sickle cell disease but survived.

**HOW GOOD IS THIS BREAKTHROUGH?** This is very good news because it offers hope for adults to have cure by BMT, where standard BMT were often fatal for adults with sickle cell. The 100% survival and 90% cure in these adults with the new immune suppression approach are similar to the success rates in children with the standard BMT approach.

**HOW COULD IT BE BETTER?** There is caution because the long-term immune suppression medicine may carry a higher risk of infections. Also, we need to watch for problems with graft rejection years later. Lastly, this new approach to BMT still requires a donor who is a full brother or sister and a very good match on tissue typing (HLA-match) plus either sickle trait or no sickle gene at all – the genetic probability of this combination happens in 18.8% of siblings.

**HOW DOES BONE MARROW TRANSPLANTATION CURE SICKLE CELL DISEASE?**

Because sickle red blood cells are made in the bone marrow, one way to cure sickle cell disease is to replace your own bone marrow with bone marrow from somebody else who does not have sickle cell disease. The major steps in transplanting bone marrow are:

- (1) finding the right donor (The best donor is a full brother or sister who is a complete match on tissue typing (HLA-match) and has either sickle trait or no sickle gene at all )
- (2) preparing the body to accept the new bone marrow cells,
- (3) giving the new bone marrow cells into the bloodstream like a blood transfusion (not transplanted by surgeons). These new cells find their way to the bone marrow space.
- (4) waiting for the new cells to be accepted by the body's immune system, then grow and produce normal red blood cells instead of sickle cells.

In the standard preparation for bone marrow transplantation (BMT), medications wipe out the old bone marrow and make room for the transplanted new marrow cell to grow. Children can tolerate these medications and have successful cures by BMT about 90% of the time.

### **WHY CAN'T EVERYBODY HAVE BMT? WHAT HAPPENS IF BMT IS UNSUCCESSFUL?**

Steps (1) and (2) are huge barriers that prevent most people from having BMT.

- (1) The probability that your brother or sister has the right combination of genes to be a matched donor is only 18.8%, and not everybody has siblings at all. Doctors are exploring ways to use less-matched relatives to be donors, or find acceptable unrelated donors from the National Bone Marrow Registry or Cord Blood Banks.
- (2) The preparation to make your body ready to accept the transplant is very challenging, and doctors are still working to make the risks of BMT to be less than the risks of living with sickle cell disease. Currently, the standard BMT approach is offered for children who have had very severe sickle cell complications such as stroke or frequent acute chest syndrome or pain despite hydroxyurea treatment.

BMT can be unsuccessful in 3 ways:

- a) Graft Rejection - you go through the BMT process but at the end your own bone marrow grows back and you still have sickle cell disease
- b) Graft Versus Host Disease (GVHD) - the transplanted marrow attacks the rest of your body as foreign tissue and can cause great damage
- c) Death – infection, bleeding, failure of bone marrow to re-grow, damage to major organs (liver, lungs, kidneys)

-Lewis Hsu, MD, PhD  
Pediatric Hematologist

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### **Web Links**

Recently, **Genetic Alliance** has gone through a process of redeveloping [www.geneticalliance.org](http://www.geneticalliance.org) in the efforts to make quality content more accessible to everyone. As a part of this process, the links to many of our pages and resources have changed. So please, update your bookmarks and any links to [www.geneticalliance.org](http://www.geneticalliance.org) that may be embedded in your organization's website and together we can make this transition as seamless as possible.

**Sickle Cell Mom** - at <http://sicklecellmom.wordpress.com/> Sickle Cell Mom is a mother parenting someone with Sickle Cell or a mother who has Sickle Cell herself. I've long needed a place to journal and express my thoughts and experiences as a Sickle Cell Mom. I plan to keep it real, informative and definitely inspirational

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### **Conferences and Activities of Interest to the Sickle Cell Community**

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**February 14 - 19, 2010** Hollywood , FL **4th Annual Sickle Cell Disease Research and Educational Symposium & Grant Writing Institute and Annual National Sickle Cell Disease Scientific Meeting PROGRESS AND PROMISE: SICKLE CELL DISEASE AT 100 YEARS** The Westin Diplomat Resort & Spa, Hollywood , Florida 3555 South Ocean Drive, Hollywood , FL 33019 Web <http://floridasickle.org/>

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**March 25, 2010 - Capitol Hill – Save the Date – SCDA** \* More details to follow \*\* Sincerely, the Sickle Cell Disease Association of America, Inc. 231 E. Baltimore Street, Suite 800 Baltimore, MD 21202 800-421-8453 <http://www.sicklecelldisease.org/>

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**March 27, 2010, New York, NY** .Sickle Cell Thalassemia Patient Network 17th Annual Dinner & Dance Benefit SATURDAY, MARCH 27, 2007 GRAND PROSPECT HALL Park Slope, Brooklyn, New York For Tickets and Information please call: 877-812-4216 or Email: [dinnerdance@sctpn.org](mailto:dinnerdance@sctpn.org)

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**March 27, 2010 Quebec, Canada** Association D'ANEMIE FALCIFORME DU QUEBEC AAFQ GALA BENEFICE DU CENTENAIRE Chateau Royal, Laval Quebec, Canada March 27, 2010 Phone: 514-830-4782 [www.anemie-falciforme.org](http://www.anemie-falciforme.org)

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**March 26 - 27 Sacramento CA** - 7th annual Sickle Cell Disease - Services for Children and Families in California Conference - For a PDF of the brochure [Click Here](#)

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**May 14, 2010 New York, NY** An All Day Symposium : Sickle Cell Disease at 100 The latest Advances in Treatment May 14, 2010 Sponsored by the Comprehensive Sickle Cell/Thalassemia Program At New York Methodist Hospital Brooklyn, New York 718-857-5643

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**April 14 - 16, 2010 Leicester, UK** Sickle Cell: The Next 100 Years International Conference on Social Research for Sickle Cell and Thalassaemia De Montfort University, Leicester, United Kingdom **Keynote Speaker:** Professor Kwaku Ohene-Frempong

*Sickle Cell: The Next 100 Years* will mark the 100<sup>th</sup> year anniversary since James Herrick published his first observations on ‘peculiar elongated cells’, what is now known as Sickle Cell Disease. This unique and highly distinctive 3 day conference will bring together a selection of papers offering delegates the chance to explore the social research being carried out around the world, now and for the next 100 years. This conference invites papers on the social aspects of Sickle Cell and Thalassaemia from academics and practitioners in the disciplines of: social medicine, public health, genetic counselling, nursing, social work, sociology, social policy, politics, health services research, social history, anthropology, cultural psychology, human geography, and law and ethics.

The best papers will be published in the international journal *Ethnicity & Health* <http://www.tandf.co.uk/journals/carfax/13557858.html> to be edited by Karl Atkin, Hannah Bradby, Seeromanie Harding and Simon Dyson.

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**2nd World Sickle Cell Day, June 19, 2010 - Multi city world wide**

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**July 15 - 18 Kiawah Island Resort near Charleston, South Carolina - 10th Annual Using Transcranial Doppler, MRI/MRA and Transfusion to Prevent Stroke in Sickle Cell Disease.** This activity has been approved for AMA PRA credit. For more information, contact: Office of Continuing Medical Education Medical University of South Carolina, Charleston, SC 29425 Phone: 843-876-1925 • Email: [maxwells@musc.edu](mailto:maxwells@musc.edu)

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**July 20-23, 2010 Accra, Ghana** within the *First Global Congress on Sickle Cell Disease*, July 20-23, 2010, co-sponsored by the Sickle Cell Center at Children’s Hospital

of Philadelphia and The Sickle Cell Foundation of Ghana. For information email [sicklecellsymposium@email.chop.edu](mailto:sicklecellsymposium@email.chop.edu)

Please submit your abstract as an email attachment to [sicklecellsymposium@email.chop.edu](mailto:sicklecellsymposium@email.chop.edu) by **March 1st**, 2010. In the subject line, put "Abstract for 1<sup>st</sup> Global Congress on Sickle Cell Disease." A confirmation reply will be sent to you. **In the body of your email, please include:** Name and degree(s) Position Title, Department, Affiliation Mailing address Telephone and Fax

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**22 - 27 November 2010— Raipur (Chhattisgarh) India - Sickle Cell Disease International Organization in collaboration with Centre for Genetic Diseases & Molecular Biology Department of Biochemistry, Pt. J.N.M. Medical College , Raipur (C.G.) INDIA are organizing the Fourth International Congress 2010 Sickle Cell Disease International Organization**

1.Simple models of survey/ screening. 2. Methods of counseling for :a. General population b. Youth & marriageable age group (premarital counseling) c. Post marriage counseling for carriers and sufferers including antenatal checkup, family planning, MTP, adoption of child. d. Counseling for sufferers of the disease. e. Counseling for the parents of sickle cell disease affected children. 3. Models of treatment plan at primary, secondary and tertiary level including plan for sickle cell clinics at village level, district level and super specialty clinic at medical college level. 4. Scope of research in developing countries. 5. Advocacy for financial support, scope of a network and linking the various NGOs working in the field of sickle cell disease. **KEY DATES 22<sup>nd</sup> November 2010** - Preconference briefing:to be attended by NGOs, Doctors and Technicians working in the field of sickle cell anemia. **23<sup>rd</sup> -24<sup>th</sup> November 2010-** Scientific Sessions. **25<sup>th</sup> November 2010-** General/Executive body meeting of the congress & draft presentation of the proceedings. **26<sup>th</sup> November 2010-** Sight Seeing.**27<sup>th</sup> November 2010-** Valedictory Function; Conclusion note, approval of proceedings. **FIRST ANNOUNCEMENT & CALL FOR PAPERS**  
<http://4scongress.co.in>

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If you would like to obtain a monthly e-mail newsletter about the latest website changes and sickle cell news, leave the listserv or read past archives go to:  
<http://listserv.emory.edu/archives/sicklecell.html>