AHS THE BLOODLETTER

Issue # 5 October, 2000

Official Online Newsletter of the American Hemochromatosis Society (AHS)
Editor: Sandra Spangler

In this Issue

"Baron"
AHS "Spokesdog"
announces that
October is "German HH Awareness Month"
See page 20 for the story.

HH "Poster Child" for Genetic Medicine

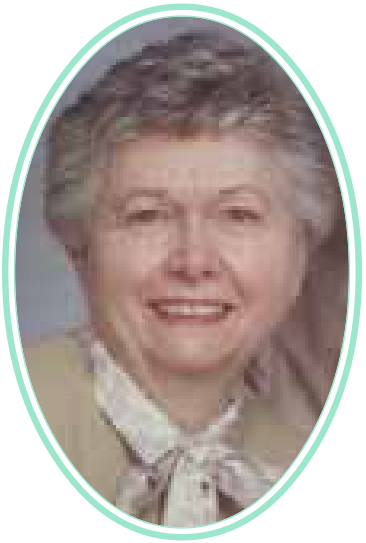
The announcement of the completion of the **Human Genome Project** (HGP) at the end of June was international news. In an article which appeared in the Pittsburgh Post-Gazette, June 25, HGP Director, Francis Collins, MD, stated that hemochromatosis may well be the "poster child for genetic medicine".

On June 20th, CBS Evening News producer, Sally Garner and reporter, Wyatt Andrews, flew from NYC to Delray Beach to interview AHS President Sandra Thomas for a segment on the HGP and genetic discrimination.This segment appeared on the CBS Evening News with Dan Rather on June 26th. Sandra, an HH carrier, said she had been denied health insurance based on a "genetic assumption." *(continued on next page)*

In Memoriam

Florence Cope

Born: August 23,1918, Denver, CO
Died: July 29, 1998, Denver, CO



Florence's heart started to fail in the early 1980's, but underlying hemochromatosis was not detected or treated, so the congestive heart failure and ventricular tachycardia worsened until she needed an internal defibrillator ten years later. Over the years, she visited many doctors complaining of chronic fatigue and stomach problems and was diagnosed with diabetes, a thyroid condition, and arthritis. In 1993 when she complained of abdominal pain, a gastroenterologist noticed that her liver was enlarged and ordered a biopsy. He diagnosed cirrhosis of the liver caused by hemochromatosis and started her on phlebotomies. Too much organ damage had already occurred. She had surgery for colon cancer in 1997 and in 1998 she became very ill and lost a lot of weight as her abdomen swelled. As her cirrhotic liver failed, she suffered extreme pain; her body was covered with a itchy rash. Two weeks after she entered the hospice she passed away July 29, 1998. She was not a drinker. Her father, who also was not a drinker, died of cirrhosis of the liver in 1957. Three brothers died of cancer. She is survived by three sisters, five children, eleven grandchildren, and two great-grandchildren.

Grandson Steven Huber honored her memory with an award winning Science Fair project on HH. See p 20.

Ann Landers Letter Draws International Response!

Following up the HGP events which brought hereditary hemochromatosis into the national spotlight on July 1, the letter to Ann Landers from Sandra Thomas was published. The popular, syndicated column appeared in newspapers all over the USA, Canada, and the U.S. Armed Forces military newspaper. AHS has been flooded with thousands of letters, emails, and phone calls as a result. Countless stories of missed, late diagnoses, diagnosis at autopsy, and organ damage and injury, including cirrhosis, have arrived at AHS. We have sent life saving information to all of these contacts and are expecting many new diagnoses to result. The letter also pointed out that the FDA has approved the use of HH blood as donor blood. Patients have contacted their local blood banks across the country to be told that the blood bank is not aware of this "new" ruling by the FDA. Others have said they know about it, but are not ready to adopt this variance at this time. For more details on how you can help your local blood bank be a part of the new FDA ruling, please go to the AHS web page under "Blood Banks" and read all about it! Also see updates on pp18-19 this issue. You may read a copy of the Ann Landers letter on our web page. Thank you, Ann Landers for spreading the word!!

In other HGP News *continued from page 1*

Media Features AHS Medical Advisor Geoffrey Block, MD

June 25th, the Pittsburgh Post Gazette featured AHS medical advisor, Geoffrey Block, MD, in several articles on the Human Genome Project and hereditary hemochromatosis. You may read these articles at http://www.post-gazette.com/healthscience/20000625genome1.asp & http://www.post-gazette.com/healthscience/20000625hemoSide3.asp

June 29th, Dr. Block, a hepatologist, and Sandra Thomas appeared on the Mediconsult web site for a live discussion and seminar of hereditary hemochromatosis. To view a copy of the discussion, go to the website at: http://www.mediconsult.com

October 7
U of Pittsburgh Medical Center UPMC will sponsor free HH SCREENING Lawrenceville Family Health Fair
9am - 3pm

http://www.upmc.edu/hemochromatosis

AHS Welcomes Two Doctors to Medical/Scientific Advisory Councils

AHS welcomes **Kenneth R. Bridges, MD,** Associate Professor of Medicine at Harvard University and **Young Yang, PhD** of the R.W. Johnson Pharmaceutical Institute, San Diego, CA to the Medical/Scientificl Advisory Council.

Dr. Bridges currently is a member of the Centers for Disease Control and Prevention (CDC) Expert Panel on Hemochromatosis.

His major research interest focus on therapeutic approaches to iron overload in the context of molecular and cellular biology.

He currently serves as a member of the Scientific Subcommittee on Iron and Heme, American Society of Hematology. For the year 2000 he is Chair, Scientific Subcommittee on Hemoglobin/Red Cell, American Society of Hematology. In 1999 he served on the Special Emphasis Panel, Iron Overload and Hereditary Hemochromatosis Study, NHLBI, NIH. He is also a Member, Ad Hoc Review Panel, Genetic Services Branch, Maternal and

cont. from page 2

Child Health Bureau, HRSA.

Dr. Bridges has also served as a Board member and Scientific Advisor to the American Cancer Society and is a member of the Peer Review Committee for Thrombosis for the American Heart Association.

He is the Director of the Joint Centers Sickle Cell and Thalassemic Disorders at Brigham and Women's Hospital in Boston.

Young Yang, PhD

Dr. Young Yang is presently a research group leader at the R. W. Johnson Pharmaceutical Research Institute, San Diego CA, and serves as an editor for *Modern Aspects of Immunobiology*.

His research interests include molecular and cellular biology of iron metabolism and major histocompatibility complex class I antigen processing and presentation.

His current research focus is to study the functional roles of the hemochromatosis protein HFE, transferrin receptor1/2, and iron transporters ferroportins and Nramp in the pathophysiology of hereditary hemochromatosis.

GeneSage To Empower Patients and Physicians

GeneSage is an Internet based health company co-founded by Paul Billings, MD, PhD, one of America's foremost medical geneticists. GeneSage has vast resources drawn from America's leading experts in the field of genetic medicine to provide genetic information, services and products to consumers and those who care for them. GeneSage publishes the online newsletter Geneletter **http://www.geneletter.com** for consumers and health care professionals. The June issue focuses on genetic therapy and has an article on genetic discrimination.

Geneletter featured the American Hemochromatosis Society in its April 3 issue. Watch for an upcoming issue which will feature a research project directed by Dr. Michael Nowicki, University of Mississippi, who has requested to work with the AHS Children *HH*elping Children screening project to find children eligible to participate in the study. S*ee page 23 this issue for more information about how you can enroll your children/grandchildren in the first HH children's study.*.

Addressing the American Association of Health Plans (AAHP) in Orlando, FL on June 5, 2000, Dr. Billings explained that he founded GeneSage to provide information and tools to translate the benefits of the genetic revolution in medicine directly to patients and their doctors. To read Dr. Billings' remarks see this URL: **http://www.genesage.com/professionals/aboutgenesage/ahhp.html**

He highlighted Hemochromatosis as a genetic condition for which good screening and DNA tests are available to illustrate how the genetic revolution will be able to save lives and money.

Billings believes that the future of genetics is now, not 3 to 5 years in the future. He said, "The tidal wave is on the immediate horizon with tens of thousands of tests, thousands of new drugs and treatments and major demand for tailored treatments, identifying the predisposed and avoiding medical errors with genetic technology...."

GeneSage will help customers by providing information for MDs designed to "empower and inform consumers and ease the burden on the health care system." According to Billings the mission of GeneSage is to "be the trusted advisor, bringing the revolution and expertise to the market place."

GeneSageRX, a subscription service announced June 5, 2000, is set up to provide health care professionals with fact sheets on genetic conditions, and immediate access to specialists and continuing medical education. The service is targeted to primary care physicians and traditional medical specialists to keep them up to date on the latest testing and treatment options.

President's Report

Sandra Thomas
-- AHS Founder

2000 has certainly been a busy year so far and it will only get more exciting with each new step forward. On March 31st, 2000, the American Hemochromatosis Society observed its 2nd anniversary! We are pleased that AHS has thrived and grown since its founding, even though this was a time of adversity since my mother was diagnosed with advanced primary liver cancer in July 1998 due to hereditary hemochromatosis only a few months after I founded AHS.

In February AHS launched an internet discussion forum, **Families *HH*elping Families** (FHHF) at FHHF@egroups.com moderated by AHS Board Member Cindy Munn, RN. This active list now has been joined by nearly 200 members who are finding immediate answers to their questions about HH testing and treatment. Almost 3000 messages have been posted and archived. Members can elect to receive a daily e-mail digest, individual e-mails or read posts directly from the web. To sign up go to: http://www.egroups.com Thank you Cindy for working on the behalf of HH families and being there for your family through difficult times. AHS would also like to extend our sincere sympathy to Cindy and her family on the loss of her father-in-law to cancer this summer.

1st Annual AHS Genetic Screening Week Honors Memory of Josephine Bogie Thomas

My involvement with the HH cause, founding AHS, and helping others with HH would never have happened, had my mother not had HH, died from HH, and encouraged me to help others throughout her life. The thanks go to her, not to me, for anything that AHS or I may do to assist anyone else who has HH. To thank her and to honor her memory, I planned the "1st Annual Genetic Screening for Hereditary Hemochromatosis Week" around these past events , making the observance from May 13th (the date she died in 1999) to May 20th (the date of her funeral in Louisville, Kentucky). This awareness week will be incorporated into an awareness "month" next year. On May 17th, WPTV Channel 5 in West Palm Beach did a segment on hereditary hemochromatosis for our 1st Annual Genetic Screening Awareness Week. Thanks to medical reporter, Roxanne Stein for airing this segment.

We placed memorial ads in several newspapers, detailing the symptoms of HH, and pleading with the public to be tested in memory of Josephine Bogie Thomas. We had a significant response to these ads, which appeared in the Delray Beach News/Boca Raton News, Sun Sentinel, Palm Beach Post, and the Mt. Sterling (Kentucky) Advocate (where my mother was born). A copy of this memorial may be seen on our web page at: http://www.americanhs.org And so, the month of May completed the one year cycle since my mother's death. It has been a year of great reflection, and sorrow by our family, but we look to the future with optimism, which was one of her greatest traits, and hope that we can truly make a difference this year in saving lives of others from HH.

Promoting Awareness

I am pleased to report that AHS has taken advantage of opportunities to raise awareness for Hereditary Hemochromatosis (HH) through participation in government seminars, community activities, presentations to physicians' groups, media appearances and

participation in a pediatric research project conducted by Dr. Michael Nowicki, MD, University of Mississippi. *See page 23 this issue.*

On April 18th, AHS Vice President and Executive Director David Snyder met with Geoffrey Block, MD who is on our medical advisory board. They discussed the University of Pittsburgh (UPMC) Hemochromatosis Center and its plans for the future. The center has attracted many new HH patients since we left Pittsburgh last September after my mother's death last May; we are thrilled to see that it is doing so well. Dr. Block will continue to present his free public seminars in Pittsburgh. For a schedule of seminars check the UPMC web site at

http://www.upmc.edu/hemochromatosis/

For an appointment with Dr. Geoffrey Block

Please call his clinic at: 412-647-1500. Dr. Block sees patients on Mondays and Wednesdays. **Note: Due to the large number of patients going to see Dr. Block, please do not send him medical records if you are not scheduled for an appointment with him in his clinic, or call him and expect to speak with him about your case. He wishes he could help each and every one of you, but an appointment in his clinic is essential to being evaluated by him. Your understanding is greatly appreciated.**

Throughout the year AHS has promoted HH awareness through letters to the editor, TV and newspaper interviews, community fairs and family reunions.

L to R Jutta Hess,Kathy Fleming, VP of Communications, HealthScreen America, Sandra Thomas

German TV Interviews Sandra Thomas for Monalisa Show

At HealthScreen America, I was interviewed by Jutta Hess with the ZDF German TV station for a show ***Monalisa,*** which aired on August 13th in Germany. Jutta flew in from her NYC office to do the interview for a show about genetic discrimination. *(See page 6 of this issue for a story about HealthScreen America).*

Palm Beach Post/ WPTV Feature HH Gene Testing

On April 16th, Palm Beach Post reporter, Ron Wiggins published an article on iron overload/hemochromatosis, and even took the DNA genetic test himself. Although he was negative on the gene test, he still brought greater awareness of HH to the community and his syndicated article resulted in many phone calls to AHS.

On August 1st, I was interviewed by Kelly Dunn, at WPTV Channel 5 in West Palm Beach, Florida. We discussed hemochromatosis and she even used the DNA test kit on the air with my demonstrating to her how to use it! It was a great segment and she will be announcing the results of her testing on the air as well. Thank you, Kelly!!

"Delray Affair"

On April 30th, David Snyder and I attended the "Delray Affair", a popular annual outdoors arts and crafts festival held in Delray Beach, Florida, where AHS is based. Such fairs/festivals are excellent places to spread the word about HH. You can wear a recognition ribbon for HH (green sticky ribbons are available from AHS for $20.00 for a roll of 100), or bring up the subject with everyone you can. You'd be surprised at how many people you can inform about it as well as potentially diagnose with it!

Family Reunion Opportunity To Increase HH Awareness

David Snyder attended his family reunion on June 24th in New Jersey and brought with him brochures, cards, and information about hereditary hemochromatosis. Please remember when family gathers together, this is a great time to increase awareness and distribute AHS

materials. Family reunions, which are popular in the summer months, are a perfect time to help other family members learn about HH and get the proper testing that they need.

St. Paddy's Day

Those of us of Irish/Celtic heritage celebrated St. Patrick's Day on March 17th. My mother, Josephine Bogie Thomas, was of Irish and Scottish ancestry and she carried the double (homozygous) cys282 gene mutation. People of Celtic/Irish/British/Scottish/Welsh ancestry are at very high risk for carrying the HH (HFE) gene mutations. The carrier rate in the USA is 1 in 8, however, in Ireland, it is 1 in 4. The double gene status in the USA is 1 in 100-200, but in Ireland, it is 1 in 65. That is why we call hereditary hemochromatosis (HH), the "Celtic Curse"

We used St. Patrick's Day as an opportunity to alert the Irish-American community about the dangers of HH. City Limits Newspaper of Ft. Lauderdale, Florida did a comprehensive story on HH and its Irish roots which appeared on March 15th. David Snyder and I attended several local community Irish Festivals where we distributed flyers about the "Celtic Connection" with HH. These are fun events, with great food, arts and crafts with a Celtic flavor. Don't wait until next St. Patrick's Day to consider alerting the Irish-American people in your community. How can you reach them? Through the civic groups, churches, and shops that they use. AHS can send you literature to distribute to Irish shops as well as Celtic festivals throughout the year. Be on the lookout for reporters and others who are prominent in your community who have Irish names; they might have HH and not know it, but you can help them get a diagnosis and perhaps spread the word on HH/AHS at the same time! You will note that the AHS "awareness ribbon" is green because of the strong association between those of Celtic ancestry and hemochromatosis.

In August, we gained a new family member, "Baron", a 93-pound black Shutzhund German Shepherd. He will be the official mascot of the American Hemochromatosis Society. Read more about him on page19.

So, as you see, the year 2000 has been a very active one for AHS. We have evolved, grown, and are serving even more patients, family members/friends, than ever and look forward to helping even more people in the future. I personally have come full circle this year, experiencing the firsts of the loss of a loved one. As it always seems, everything, eventually, and inevitably, returns to my mother, the underlying cause, raison d'etre for my involvement with the hemochromatosis cause. I now start a new chapter in my life, moving past that first very painful year of "anniversaries" of important dates and celebrations in our family, and look forward to what can be done to help others avoid what our family has experienced. We have many exciting activities in the next three months of 2000, and these will be reported in the next BloodLetter.

Please revisit our web site at: http://www.americanhs.org We have made some changes and updates. Don't forget to become a member of AHS if you aren't one already, we need your support; and, if you can, support our work with a donation in honor of, or in memory of, a loved one.
—Sandra Thomas, *President & Founder, American Hemochromatosis Society.*

The mission of the American Hemochromatosis Society (AHS) is to educate and support the victims of hereditary hemochromatosis and their families and educate the medical community on the latest research on HH. AHS' aim is to identify through genetic testing, the 35 million+ Americans who unknowingly carry the single or double gene mutation for HH which puts them at risk for loading excess iron.

AHS
TOLL FREE
INFORMATION
HOTLINE
1-888-655-IRON (4766)

HealthScreen America Center Offers Consumer "Testing On Demand"

When David Snyder, AHS VP, pointed out an article in USA Today about HealthScreen America, I responded to this story with a letter to the editor, supporting this consumer oriented, pro active health center based in Jacksonville, Florida. My letter, published May 25, 2000 by USA Today, was the beginning of an alliance between AHS and HSA. This alliance will help increase awareness of HH and help promote HH comprehensive testing, including DNA testing, at their facility. See letter at http://www.usatoday.com/life/health/hcare/lhhca094.htm

HealthScreenAmerica embodied all of the concepts that I had long thought would be perfect for a medical center. It offers "testing on demand" for the medical consumer who is busy, wants to be in control, and wants a pleasant and attractive environment in which testing takes place.

My letter to the editor expressed my support for HealthScreen America and their concept of medical care for the public. I contacted this company and encouraged them to offer both iron studies (serum iron, TIBC, and serum ferritin) and the DNA test kit for hereditary hemochromatosis. We are pleased that they are now offering these tests at their facility. Their web site is:http://www.healthscreenamerica.com HH testing is performed onsite only. New locations will be opening soon around the country. Dr. Ed Balbona is the medical director and is very enthusiastic about HH screening.

You simply walk in and request the tests; you are then tested; and you pay at the door. Some results are given to you directly and immediately. Others are mailed to you when they are ready. You do not have to go through a doctor to get the testing, or the results. Just that simple.

My father, Joe Thomas, Dave and I went to Jacksonville on July 27th to take a closer look at this facility and to experience this kind of healthcare for ourselves. We were very impressed with their airy, bright and colorful decor, friendly staff, cleanliness, and speedy, no waiting style of testing. They offer a wide variety of testing, including bloodwork, CT scans of the lungs and/or heart (to detect early heart disease), ultrasound, and much more. Prevention is the key word at this medical center!

Chris Fey, President, HealthScreen America
David Snyder, VP/Exec. Dir., AHS
Sandra Thomas, Pres./Founder, AHS
Fred Fey, VP, HealthScreen America
in the lobby of the Jacksonville, FL Center

"I only wish my mother could be here to see the progress AHS has made and experience the exciting things she worked so hard for to become reality." —*Sandra Thomas*

Sandra Thomas & Dad Joe
Remember Josephine Bogie Thomas

The year following a loved one's passing is filled with many "firsts" remembering and reliving the dates that held significance for a family. March 14th would have been my parents' 60th wedding anniversary. This year we recalled their 59th anniversary spent at the University of Pittsburgh Medical Center's Presbyterian Hospital in Pittsburgh, PA. My mother spent the entire month of March in that hospital, with complications from hemochromatosis. It was a very stressful and solemn time, yet, we still tried to observe this important date, which would be the last they would observe together.

April 25 marked my mother's birthday, the first since she died of HH on May 13, 1999. She would have been 79 years old. and this was another important one. I only wish my mother could be here to see the progress AHS has made and experience the exciting things that she worked so hard for to become reality.

See related Delray Beach/Boca Raton News article "Will of Iron"

http://www.americanhs.org/News%20Articles/Boca_Raton_News_07012000.htm

Biotech Researchers Developing New HH Screening Technologies

As a single gene carrier (heterozygote) for the cys282 gene mutation, I donated blood to a biotech company for research on new technologies to be used for screening HH patients. Anyone who has the HFE gene mutations should contact AHS if they are willing to donate their blood to biotech companies for research of this type. We will place your name in our research database as someone who is willing to participate in research and studies.

Sandra Thomas

David Snyder Represents AHS at Medical Conferences

AHS Vice President and Executive Director, David Snyder has traveled extensively during this year to represent the society at health meetings and conventions including the National Heart Lung and Blood Institute NHLBI-NIH meeting in Bethesda, MD on February 9th. On February 15th, he went to the Florida capital in Tallahassee to discuss hereditary hemochromatosis with legislative representatives.

May 8th-10th, David attended a workshop sponsored by the Centers for Disease Control and Prevention (CDC) in Atlanta, GA entitled, **" What We Want Physicians to Know about Hereditary Hemochromatosis."** He was invited by the CDC to make a presentation to a select group of expert physicians and CDC representatives. The PowerPoint presentation outlined what an advocacy group like the American Hemochromatosis Society would like to see the CDC emphasize with America's physicians when educating them about hereditary hemochromatosis.

On May 23rd, he lectured about pediatric hereditary hemochromatosis for grand rounds, to 75 doctors in Ft. Worth, Texas along with Dr. Jeffrey Murray, Cooks Children's Hospital. The next day, Dave went to Dallas, where he met with Dr. Dan DeMarco, a gastroenterologist at Baylor University Medical Center.

Jeffrey Murray, MD and David Snyder Grand Rounds, Cooks Children's Hospital

On September 15, David exhibited at the 13th annual meeting of the American Society of Pediatric Hematology/Oncology (ASPH/O) at the Marriott City Center in Toronto.

David Snyder at the AHS Exhibit American Society of Pediatric Hematology/Oncology

The conference and meetings gave AHS an opportunity to talk to pediatric hematologists about newborn genetic DNA screening and neonatal hemochromatosis issues.

The American Hemochromatosis Society is scheduled to exhibit at the following medical conventions:

American College of Gastroenterology, New York, NY, October 14-19, 2000
American Association of the Study of Liver Disease, Dallas, TX, October 26-31, 2000
Southern Medical Association, Orlando, FL November 1-5, 2000
American Society of Hematology, San Francisco, CA December 1-5, 2000

Reprints of the journal article "**Use of HFE Mutation Analysis for Hereditary Hemochromatosis: The Need for Physician Education in the Translation of Basic Science to Clinical Practice"** will be distributed with permission of the author, Clive S. Zent, MB, BCh, at the meeting of the Southern Medical Association in Orlando in November. *See p. 24 for a review of this article.*

EDITORIAL:

Sandra Spangler
Editor

Does Gene Testing Increase Risk of Genetic Discrimination for Hereditary Hemochromatosis?

Fear and controversy swirl around the debate over genetic testing as the human genome project reaches its goal of mapping the human genome. People who have a diagnosis of Hereditary Hemochromatosis (HH) worry if they or their family members will be denied health or life insurance or a job if insurance companies find out that they have had a gene test that confirms the HH gene mutation which puts them at risk of developing iron overload. Anxiety about gene testing paralyzes parents and forces them into an impossible dilemma. Of course they want to shield their children from discrimination, yet they fervently desire to protect their children's health and privacy at the same time. **AHS has learned that gene testing alone may pose no added risk of genetic discrimination. However, you could be denied insurance because of hemochromatosis even if you have not had a gene test.**

> ***"A major misconception is that by avoiding the gene test you can prevent genetic discrimination."***
>
> ***—Sandra Spangler, Editor***

A major misconception is that by avoiding the gene test you can prevent genetic discrimination. Some people believe that the clinical diagnosis of HH without organ damage based simply on elevated iron levels with subsequent iron removal will not likely be used to deny insurance or discriminate. The current debate can be understood better from the perspective of whether it is necessary to diagnose HH or not before symptoms arise in order to avoid discrimination, not whether to take a genetic test or not. It is the diagnosis of HH by clinical iron studies and liver biopsies which increases risk of discrimination, not the gene test. Current laws, which focus on preventing genetic discrimination arising only from a gene test are not strong enough protection. It is not likely that someone positive for HH gene mutations would be discriminated against because of the gene alone. To learn how you can get direct confidential clinical blood and genetic tests from HealthCheckUSA, LabAmerica or Health Screen America see p. 20 this issue.

Until the discrimination issues are resolved concerning genetic testing for all genetic conditions, screening for hemochromatosis is not likely to be recommended by public health agencies. The genetic testing debate, when it includes HH as part of a general debate on the ethical dilemmas of gene testing, is used to justify postponement of screening recommendations and treatment guidelines and becomes the rationale for more scientific studies. AHS advocates universal screening for HH. No other approach will be able to protect the public as effectively from suffering and dying needlessly from delayed symptom-based HH diagnoses. A compelling argument can be made for adding a gene test for HH as part of the screening panel for a number of newborn diseases which are not nearly as common or treatable as HH.

Because many gene-linked diseases discovered by the genome project are incurable and even untreatable, people understandably don't want to know if they will face this future fate and also fear they will be denied job or insurance coverage if they learn they have genes for breast cancer, Alzheimer's or Huntington's.

Hereditary Hemochromatosis (HH) is extremely different. The medical literature confirms that HH may manifest at any age, in children, adolescents, young adults as well as older adults. Early death and disease is totally preventable. Yet many people are still being diagnosed too late, some at autopsy. The sooner you know you have the gene(s), the better the chance to prevent organ damage. Yet because the policy makers have followed a one-size-fits-all approach, HH gets lumped together with incurable and untreatable diseases and conditions. The result is delaying the benefits of gene testing to the public while people are suffering and dying needlessly.

Anticipating the expansion of gene testing arising from the Human Genome Project, the states have written protection from genetic discrimination into law. More than thirty states have such laws, but usually these laws are written very narrowly to prevent discrimination arising only from a gene test itself. So, ironically, at least in California which has very tough anti-discrimination in insurance laws, the law might protect you better if you had a gene test first and then discovered through blood tests that you had started to load iron.

The laws were designed mainly to ensure that someone who learned that they had a gene for breast cancer or Huntington's disease would never be discriminated against as long as they presently had no symptoms or manifestations of disease. Even if you have HH and are asymptomatic, (have no symptoms), if you have had blood tests or liver biopsy in your medical history that indicatd excess iron storage at one time, one strictly legal interpretation would allow the insurer to deny coverage based on a pre-existing life-threatening condition, not taking into account whether you have been treated and have a prognosis for a normal life expectancy. However, Paul Billings, MD, Ph.D., an authority on genetic discrimination and editor of GeneLetter *http://www.geneletter.com* , *(See page 3 this issue)* and AHS Medical Advisory Council member Victor Herbert, M.D. JD, of Mount Sinai School of Medicine in New York disagree. Both firmly believe this prevailing opinion can be challenged successfully in court.

Obviously, government policy and practice, insurance companies and physicians have not been able to keep up with the breathtaking pace of scientific and technological genetic advances. Until there are laws in place that clearly prohibit genetic discrimination for asymptomatic HH, the burden is on the patients on a case by case basis to educate insurance companies and prove that it is unjust to deny insurance simply on the basis of a clinical diagnosis of HH if otherwise we can prove they are in good health. Fortunately some insurance companies are taking a progressive approach to asymptomatic HH. Read the following article and learn how you can assertively plead your case.

" It is the diagnosis of HH by clinical iron studies and liver biopsies which increases risk of discrimination, not the gene test."

—Sandra Spangler, Editor

How to Build A Strong Case for HH Insurability

by Sandra Spangler, Editor,

When the genes for Hereditary Hemochromatosis (HH) were discovered in 1996 by Mercator Genetics in the race to map the human genome , the Centers for Disease Control and Prevention (CDC) held a conference in March 1997 to focus on hemochromatosis as the most common genetic condition in the US. A consensus of panelists agreed that early diagnosis was the key to preventing early death and disease from excess iron storage.

Consequently more people who are asymptomatic are being diagnosed early with HH, either with a gene test because a family member is known to have HH, or by iron studies that indicate high serum iron, transferrin saturation % or ferritin.

Concern about obtaining health/life insurance and fears about employment discrimination is growing. Newly independent young people who have not previously held insurance on their own and who as yet do not have group coverage, those who are self-employed or been laid off or have elected early retirement, are being intimidated. Insurance companies are automatically saying they will deny applications from anyone who has HH period.

Commonly, these people are told that HH is in the highest risk category. Some have learned, when turned down for life insurance, that the reason is because they have a chronic liver disease. Insurance company databases contain information collected from patients who got a late diagnosis and went on to develop serious and costly deadly diseases. There are not yet enough cases on record of asymptomatic HH. So we need to be assertive to educate the insurance companies.

Because the current genetic discrimination laws were designed to protect individuals who had genes for, but no clinical evidence of diseases like breast cancer, Alzheimer's or Huntington's, there is some doubt about whether the genetic anti-discrimination laws can be interpreted to protect people who know they have HH if the first evidence of HH was high iron storage.

> ***"An important precedent was set recently when a young person with HH successfully obtained individual standard risk insurance. . ."***

If you are told that you are not insurable because of HH or excess iron, do not give up! The agents who discourage you from applying do not make the final decisions. You must apply for individual insurance and obtain the official denial in writing. Then you have rights of appeal for reconsideration as well as further appeal rights given to you under state law. Appeals should first be directed to your state's department of insurance.

An important precedent was set recently when a young person with HH successfully obtained individual standard risk insurance after a long struggle despite such discouraging tactics by following these steps which AHS recommends.

1. Consult the Genetic Alliance http://www.geneticalliance.org email: info@geneticalliance.org **Helpline: (800) 336-GENE** for information about the genetic anti-discrimination laws in your state. The Genetic Alliance has an insurance expert on staff to assist you and send you information on state high-risk insurance pools which you may be eligible to take advantage

of, usually at higher cost while you are appealing.

2. Have a comprehensive health checkup at one of the Hemochromatosis Clinics to establish good health and normal life expectancy. Be willing to travel and pay for the exam.
3. Include a letter from one or more recognized authorities on HH based on the results of your checkup.
4. Include a letter from your personal physician stressing that clinical iron studies are not symptoms of disease, making reference to the Annals of Internal Medicine December 1998 Supplement on HH. http://www.acponline.org/journals/annals/01dec98/mgmthemo.htm
5. Disclose Hemochromatosis rather than excess iron or iron overload on the initial application if you have had a gene test confirming one of the HFE mutations.
6. Refer specifically to the law and let them know that you are aware of your rights. Word this strongly to imply that you are not afraid to go as far as necessary to prove your case.
7. Apply to multiple insurance companies.
8. Keep detailed copies of all medical records, insurance application and correspondence and phone calls.

Be sure to incorporate the following supporting references:

Vincent J. Felitti, MD of Kaiser Permanente Department of Preventive Medicine, San Diego, CA , is managing a program which screens patients within the HMO every year for iron overload. Dr. Felitti is proving that the damage due to undiagnosed hemochromatosis can be totally prevented. Reporting in HealthNews/ January 5, 1999, a journal of the Massachusetts Medical Society, Felitti reports that "When diagnosed early, a simple and inexpensive treatment program can prevent significant, debilitating and potentially fatal illness."

More representatives in the health care industry are becoming aware of an unparalleled opportunity to save lives and money by being able to diagnose hemochromatosis early enough to prevent complications. The CDC has identified hereditary hemochromatosis (HH) as the most common genetic condition in America. Demonstrating the growing support for early diagnosis of hemochromatosis, in testimony before the Committee on Labor and Human Resources of the US Senate May 21,1998, Mary Nell Lehnard, Senior Vice President Policy and Representation, Blue Cross and Blue Shield Association, singled out hemochromatosis as one genetic condition that can be prevented through testing. She recognized that early diagnosis is a win-win situation, which prevents suffering and keeps costs down. "Once identified, individuals can undergo occasional bloodletting procedures and thereby avoid the liver cirrhosis, heart deterioration, and early death that would otherwise result."

The following academic references document the projected longevity of healthy young homozygotes.

Semin Hematol 1998 Jan;35(1):72-76
Screening for hemochromatosis: phenotype versus genotype
Edwards CQ, Griffen LM, Ajioka RS, Kushner JP
Department of Medicine University of Utah College of Medicine, Salt Lake City 84132

Edwards et.al. report that “Iron-depletion therapy of homozygotes before the development of disease-related morbidity results in normal longevity.”

New Eng. J. Med 328 1993:1616-20
Screening for Hemochromatosis
Edwards, C, and Kushner J
Department of Medicine University of Utah College of Medicine, Salt Lake City 84132

Edwards et.al. argue that “since the genetic definition of hemochromatosis does not imply the presence of disease, healthy homozygotes will not be disqualified from health or other insurance. Edwards and Kushner include in this category of healthy homozygotes individuals whose hemochromatosis is controlled by phlebotomies since those individuals who have neither cirrhosis, nor diabetes have no morbidity due to their condition and have normal life expectancies.

Gastroenterology 1996:110-1107-1119
Long-term Survival in Patients With Hereditary Hemochromatosis
Niederau C, Fischer R, Purschel A, Stremmel, W, Haussinger D and G Strohmeyer
Division of Gastroenterology, Hepatology and Infectious Disease
Department of Medicine, Heinrich-Heine-Universitat Dusseldorf, Dusseldorf;
Department of Medicine, Heinz Kalk-Klinik, Bad Kissengen, Germany

Niederau et.al. report that “Early diagnosis and therapy largely prevent the adverse consequences of iron overload.”

J Public Health Policy, 1994 15 (3): 345-357
Alper J, Geller N, Barah C, Billings P, Laden V and M Natowicz
Center for Genetics and Public Policy, University of Massachusetts-Boston 02125

Alper et.al. report that “Screening for presymptomatic [asymptomatic or controlled hemochromatosis] individuals who are homozygous for hemochromatosis should result in decreased medical costs and increased years of healthy life for those individuals who develop clinical hemochromatosis and are treated at an early stage of the disease.”

Liver, 1999 Apr 19:2 73-80,
Olynyk JK
Hereditary haemochromatosis: diagnosis and management in the gene era.
N Engl J Med 1999 Sep 2;341(10):718-24

Olynyk reports that the Cys282 gene accounts for 90-95% of the subjects found to be homozygous for hereditary hemochromatosis. The His63 mutation also has been identified, but has not shown to produce as much iron overload as the Cys282. 20% of the subjects heterozygous for both mutations, compound heterozygotes, can express HH. The paper gives evidence that asymptomatic patients with early iron overload, diagnosed

promptly with iron studies and genetic testing, can enjoy normal life expectancy with proper treatment. Olynyk further states that asymptomatic individuals with no evidence of liver disease, diagnosed under age 40, can begin phlebotomy without liver biopsy and recommends that first degree relatives should be screened for genotypye and phenotype.

Please let the American Hemochromatosis Society know if you are asymptomatic, are deironed and on maintenance. and have been unjustly discriminated against in your employment
or by official written denial of coverage for health, life or long-term care insurance

OR
you have successfully been granted standard risk individual health,life or long-term insurance.

Email: Sandra.Spangler@americanhs.org

Support Genetic Nondiscrimination in Health and Employment Act

Call your member of Congress TODAY to support S.1322/ H.R.2457: The US Capitol Switchboard (202) 224-3121 can connect you to your members' offices. Ask to speak to the Health Care Legislative Assistant

Dear

_____ _____

Date

I urge you to become a co-sponsor of the Genetic Nondiscrimination in Health Insurance and Employment Act (S.1322/H.R. 2457)

Unless a national law is passed, citizens will avoid the lifesaving diagnostic clinical and diagnostic testing for Hereditary Hemochromatosis (HH), the most common genetic disorder in the United States that causes people to load excess iron and destroy vital organs.

One in eight, over 32 million Americans are silent carriers who will pass gene mutations on to their childrens. One in 100-200 (1.5 million) Americans have the disorder. Late diagnosis means they will already have cirrhosis, heart failure, arthritis, cancer.

Dr. Francis Collins, Director of the Human Genome Project told the Pittsburgh Post-Gazette June 25, 2000 that HH may well be the "poster child" for genetic medicine.On the CBS News with Dan Rather on June 26 he said "If we don't put protections in place to avoid breaches in privacy and to protect against discrimination, and do it soon… this revolution in medicine could be stillborn."

I join the many voices represented by the American Hemochromatosis Society in support this legislation. Visit http://www.americanhs.org (561-266-9037)

_____ _____

Signature *Address*

Legislative Alert

The Genetic Nondiscrimination in Health Insurance and Employment Act (S.1322/H.R. 2457) enjoys broad bipartisan support. The House Commerce Committee chairman Rep. Bliley (R, VA) has tabled the bill so that it cannot come out of committee unless a majority of co-sponsors vote to bring the bill to the house floor. The Senate has held hearings on a the Senate version of the bill House sponsor Louise Slaughter, MPH D (NY), has tried for five years to get this bill passed.

The election campaign season is a good time to speak out and ask your representatives to become cosponsors of the bill(s) in both the House and Senate.

When I wrote to Sen. Jon Kyl,(R. AZ) I received an enthusiastic response. His health care legislative assistant telephoned to inform me that my letter was one of the best the Senator had received on this subject. The Senator himself had read my letter and underlined a key point—that people will not get genetically tested and treated if they fear discrimination.

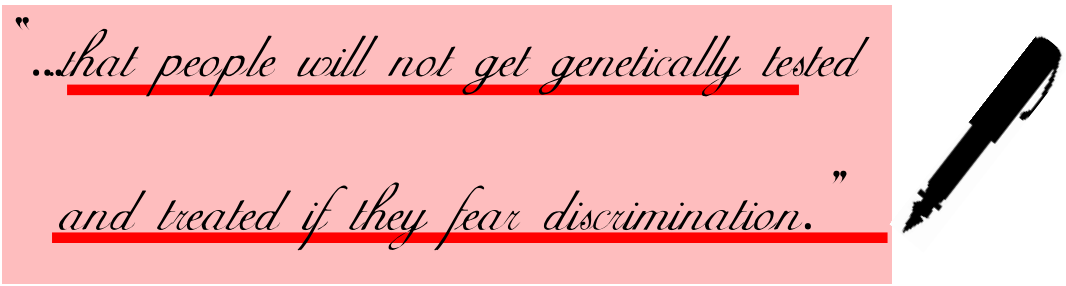
That's why a national law is needed to cover conditions like hemochromatosis which require early diagnosis and treatment in order to prevent premature disability and death.

I urge all of you to write, call and fax your representatives in Congress now to register your support for this bill.This is the most important legislation that will protect people with HH from unjust discrimination.

Emphasize that support for nondiscrimination in health insurance and employment must go together. It is easy for you to weigh in on this issue. Either clip or copy the postcard printed in this issue or if you have time write a longer letter including your personal story. A modified sample letter will appear soon on the AHS web site http://www.americanhs.org

Sandra Spangler,Chair
AHS Legislative Action Committee

The Senator himself [Jon Kyl,R AZ] had read my letter and underlined a key point—



Medical Record Privacy: A Hot Topic For Campaign 2000

In Los Angeles on September 19, Vice President Gore, addressing an audience of seniors and health care professionals, proposed legislation to protect the privacy of medical records. The Gore plan contains three key elements:

a. Legally requires anyone who handles medical data, including employers and insurers, to keep that information confidential;
b. Imposes tougher penalties for improper use of data;
c. Emphasizes passage of tougher genetic antidiscrimination laws to protect people and encourage them to get medical screenings when appropriate.

Gore attacked drug and insurance companies that share and sell private medical information without patient consent and frequently without patient knowledge. He stressed that the mapping of the human genome could raise the risks for people who are genetically predisposed to certain diseases. See http://www.cnn.com/2000/ALLPOLITICS/stories/09/19/campaign.wrap/index.html

In a press release dated September 19, Chip Kahn, President of the Health Insurance Association of America (HIAA) denied that health insurers sell private information that risks patient confidentiality. See http://www.hiaa.org/news/news-current/press-releases/release1.html

No comprehensive federal law offers privacy protection for personal medical records. The 1996 Health Insurance Portability and Accountability Act (HIPAA) included deadlines to plug the gap in federal rules. HIPAA mandated that if Congress failed to pass a health privacy law by August 2, 2000, Donna Shalala, Secretary of Health and Human Services is required to issue final health privacy regulations. Since Congress was unable to agree upon a comprehensive privacy law, the responsibility was given to the Clinton administration to write guidelines for medical privacy protection. The House of Representatives passed the bi-partisan Norwood-Dingell bill that would have afforded privacy protection, but the Senate and House could not agree upon a compromise bill. It is expected that these guidelines will be unveiled this fall prior to the presidential election.

A draft of the Administration's proposed regulations issued in October 1999 is summarized at: http://www.healthprivacy.org/latest/RegSum.fin.html

The watchdog Institute for Health Care Research and Policy at Georgetown University posts legislative updates for health care consumers on its web page at http://www.healthprivacy.org

by Sandra Spangler

Labs Offer First Confidential "Do-It-Yourself" HH Blood and DNA Screening

HealthCheck USA, an accredited laboratory based in San Antonio, TX, www.healthcheckusa.com is the first lab to offer confidential screening for hereditary hemochromatosis for most areas of the country. HealthScreen America also offers HH screening only at its on site location in Jacksonville FL, although the medical center plans to expand soon to other cities (See story page 8). LabAmerica https://labamerica.com will also offer HH screening in some cities. Contact LabAmerica at (877) 900-3625 for information.

You can order the same lab tests ordered by physicians and analyzed by an accredited medical reference laboratory. This easy option empowers patients to learn on their own without going through a doctor or an insurance company if they have clinical iron overload/hereditary hemochromatosis.

According to HealthCheck USA President George Vaughn, PhD, "All results are kept entirely confidential and distributed directly to the patient Although the laboratories do keep patient's records on a secure archive system for seven years, only HealthcheckUSA has exclusive rights to accessing these records. No other outside source has access to these records without the expressed written consent of the patient"

From the web site select a state from the menu to locate a list of participating local laboratories. HealthCheck USA plans to expand availability to most communities in the nation. Call the toll free number at **1-800-929-2044** for updates.

When you prepay online through a credit card, Health Check USA sends you a order which you take to your local laboratory. Results usually available within 24 hours are sent to the Medical Director at HealthCheck USA and then are reported directly to you.

HealthCheck USA offers a Hemochromatosis Screening Panel with the correct blood tests: serum iron, TIBC/total iron binding capacity, % saturation and serum ferritin for $59.95.

If you add the genetic test to the basic panel for Hereditary Hemochromatosis, the cost is $175.00. Kimball Genetics provides the DNA testing for these companies.

"Testing On Demand" Ensures Privacy, Reduces Discrimination Risk

by Sandra Thomas

Have you ever had a doctor refuse to order a test for you which you thought was important? Have you ever had to argue with a doctor to get the testing that you think might diagnose a medical problem? Has a doctor ever refused to test you, even though you offered to pay out of pocket for the test? I personally have heard countless accounts from distraught and angry patients who wanted comprehensive hemochromatosis testing but were unable to get it from their doctors, even though they were willing to pay out of pocket for it! I have experienced this situation myself and it is a very unpleasant one. What can a patient do? Well, the answer to your requests to us to "do something" about this tragic situation is becoming a reality.

Two medical companies are already offering consumer oriented medical services, putting the patient "in the driver's seat" long enough to get the tests that will give them peace of mind. Results are reviewed by a doctor at the facility and then mailed directly, and only, to the patient. This affords the patient confidentiality and the assurance that they do not need to fear genetic discrimination from test results which will never be seen by insurance companies or employers.

These pioneering companies are at the vanguard of a new wave of medical care which caters to the consumers' needs and offers them a no hassle, no argument option for medical care. So far, we have heard only rave reviews of these companies and patients are thrilled to be able to get the HH screening that they need without having to confront their doctor or risk genetic discrimination by paying for the tests through insurance companies.

(continued page 20)

Lifeblood to Accept Hemochromatosis Patients as Blood Donors

Lifeblood, **http://www.Lifeblood.org** a blood bank in Memphis, Tennessee kicked off the American Hemochromatosis Society's first annual Genetic Screening and Awareness Week May 13-20, by publicly announcing that it will accept hemochromatosis patients as regular blood donors.

Lifeblood joins the Mt. Sinai blood bank in New York City as one of two blood banks in the nation that are now accepting blood from healthy HH donors. AHS Medical Advisory Council Member Victor Herbert, MD, JD, has announced that Mt. Sinai now has met the New York state health department and FDA certification.

According to Lifeblood Director Gordon Wilson, "Lifeblood provides hemochromatosis phlebotomy as a community service at no charge and joins this national effort to raise awareness on a little known hereditary condition that if left untreated can cause premature death."

Wilson cited that the recent FDA ruling that "the HH condition alone does not pose a threat to the safety of the blood supply if the HH blood donor meets general blood donor suitability criteria." Lifeblood has met all FDA requirements to provide HH blood donors with the opportunity to participate in the community blood program in accordance with the guidelines and criteria for all volunteer blood donors."

Lifeblood is a non-profit, tax-exempt community service agency sponsored and endorsed by the Memphis/Shelby County Medical Society and the Memphis Hospital Council and is a member of the American Association of Blood Banks - America's Blood Centers and the Tennessee Association of Blood Banks.

News Bulletin

Read

"U.S. Blood Shortage Puts Surgeries On Hold"

AHS President Sandra Thomas says HH blood would ease shortage.

http://dailynews.yahoo.com/h/nm/20000919/ts/health_blood_dc_5.html

Update on Blood Banks and HH Donor Blood

by Sandra Thomas

We continue to hear from patients all over the country who indicate that they have contacted their local blood banks about the FDA variance. The blood banks are pleading ignorance on this issue (which may be true in some cases, but surely not in all of them) and therefore are simply doing nothing. This is a battle which must be fought by all concerned. It will not be easy or simple. It will take hounding and calling and writing by MANY for this to work. There is power in numbers and that is why it is important for chapters of AHS to be formed around the country. If one person calls the blood bank, ho hum. If 50 call, they start to listen; if 100 write and call, they are going to have to listen.

Without organization of chapters, getting numbers of HH patients together and focused, we cannot achieve the changes that we need as soon as we need them. You must write to the editor of your newspaper, not once, but over and over again. If they don't publish, write again, call. Ditto for the political representatives and the blood bank director. Also get your doctor enlisted in the efforts if he or she is of that mind. MD's have power too.

This is not going to be handed over on a silver platter. We must fight for change. If you are willing, call your local newspaper news desk and offer to be the subject of an article on HH. Give the AHS phone number for a press kit and further info: 561-266-9037. Local newspapers have done great stories on HH, with terrific photos. This heightens awareness and creates more " pressure" on the blood banks to get with the program. As long as there are only a few voices crying out about issues concerning HH, they will be swept under the rug. If you look at other causes, consider what makes them a success. One of those things is NUMBERS. We have the numbers, but not the organization of people together in cities around the country. Would you picket the blood bank? If you were able to organize 100 people to peacefully picket the blood bank, call in the local news tv cameras; you might make a change. A lot of work? You bet! But if you want it, you are going to have to go through these efforts to get changes made.

"There is power in numbers and that is why it is important for AHS Chapters to be formed around the country."

— SandraThomas

(continued from page 18)

Because an increasing number of reports are coming in to us at AHS from patients who are having their test results for genetic testing misinterpreted by the reporting doctor, they want to see their reports themselves. Unfortunately, some doctors are resistant to the sharing written lab/test results with their patients, creating a tense situation which sometimes ends up terminating the physician-patient relationship. These are difficult and challenging times for the HH patient. With companies like Healthcheck USA and Health Screen America, better times are on the horizon for hemochromatosis patients.

"With companies like HealthCheck USA and HealthScreen America, better times are on the horizon for hemochromatosis patients"

—Sandra Thomas

New German mascot kicks off awareness campaign

October 2000 is "German HH Awareness Month"

The American Hemochromatosis Society (AHS) has designated October as "German Hereditary Hemochromatosis Awareness Month."

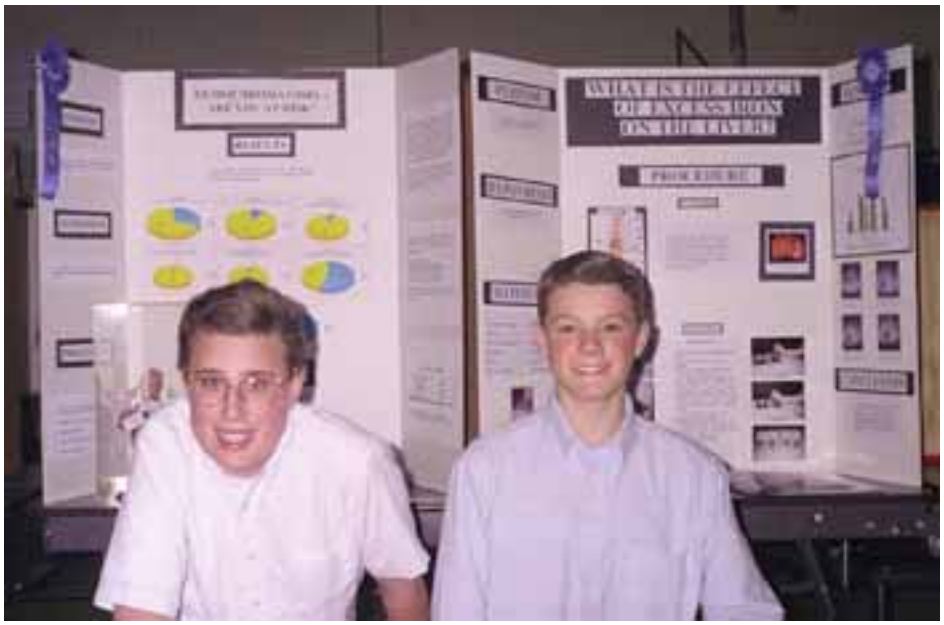
Most Germans, and those of German descent, are unaware of their high genetic risk for hereditary hemochromatosis" (HH), states Sandra Thomas, president/founder of AHS.

On German-American Day, October 6, "We want to emphasize to everyone, and especially Germans all through the month of October, the importance of taking a genetic/DNA test kit for hemochromatosis by using a painless, "do-it-yourself" test kit you can order yourself."

"Baron", a German Shepherd is the new mascot helping Sandra kick off the awareness campaign. "Born in Alsdorf, Germany, Baron is a hit wherever he goes—he gets people's attention, and that's when I have the opportunity to tell them about HH," Thomas confides about her new "staff member".

During October, "Oktober Fests" feature German food, culture, arts, and crafts. AHS wants to alert and educate people of German ancestry about how to diagnose and treat HH. Although HH can cause diabetes, chronic fatigue, liver disease and heart attack, Thomas points out that HH is totally preventable and treatable with early genetic diagnosis.

Science Fair Project Promotes HH Awareness



Left to right Steven, 13, Scott, 14

By Steven Huber, grandson of Florence Cope

As eighth graders at Annunciation School in Albuquerque, NM, Scott Prokesch (14) and I, Steven Huber (13), did a team Science Fair project on hemochromatosis that won first place at the school science fair February 9, 2000. I wanted to do the project because I saw how my grandma, Florence Cop, suffered during her last years of life and knew that the suffering was not necessary if she had been diagnosed earlier. My best friend Scott has juvenile diabetes and is adopted. He wanted to learn how hemochromatosis causes diabetes. I have had the DNA test, and am a carrier of the gene. One of my brothers is also a carrier, and my other brother does not have the gene. My mother is also a carrier.

Our project received honorable mentions at NM Regional Science Fair (Junior Division) at the University of New Mexico in Albuquerque, NM on March 16 - 18, 2000 from the Albuquerque Science Teacher's Association, and Top of Category Award for team projects. At the State Science Fair April 14-15 at New Mexico Tech in Socorro, New Mexico, we received an honorable mention in Top of Category for team projects. The judges recognized us for superior work, thoroughness, and the tremendous amount of time and effort it took for us to complete the project. One judge said that she had heard of hemochromatosis for the first time only a few weeks before when one of her co-workers was diagnosed.

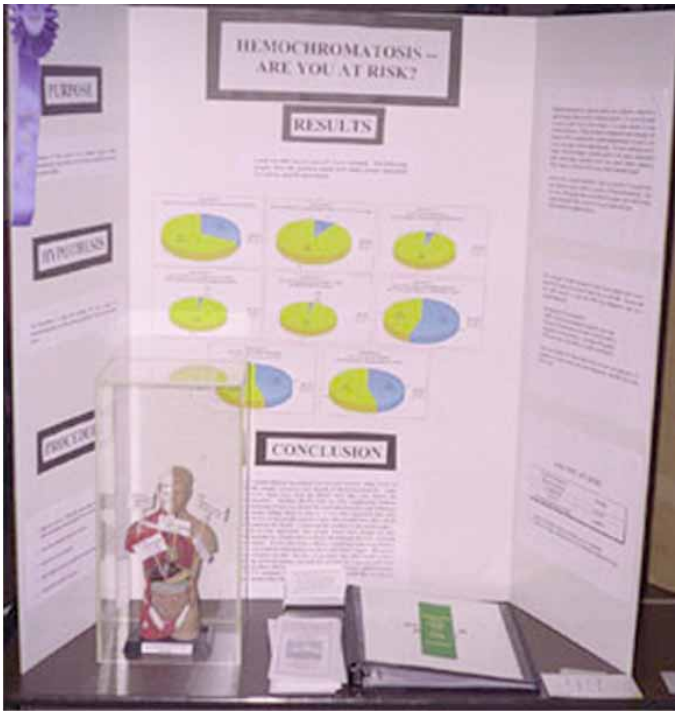
Our purpose was to educate people how common the disorder is and how easy it is to control if diagnosed soon enough. We also showed how excess iron damages the liver.

First we sent out a survey with a brief description of hemochromatosis and asked the following questions:

1. Before you received this questionnaire, had you ever heard of this genetic defect?
2. Has anyone in your family died of liver cancer?
3. Have you ever had a transferrin saturation and iron binding capacity blood test?
4. Has anyone in your family ever been diagnosed with hemochromatosis?
5. Has anyone in your family died from complications of hemochromatosis?
6. After reading this information, do you think you will ask your doctor to do the two blood tests during your next physical?

7. Do you or any member of your family take a multi-vitamin containing iron or an iron supplement without being told to do so by a physician?
8. Do you think that the iron level in your blood could show that you are anemic, but you could still have hemochromatosis?

Only 31.6% of the people surveyed had ever heard of hemochromatosis, and only 5.1% had ever had the diagnostic blood tests necessary to diagnose hemochromatosis.



The survey educated the participants:

After reading the information on hemochromatosis, 54.7% of the people surveyed are considering having the diagnostic blood tests during their next visit to the doctor. This is an increase of almost 50% over the number tested before the survey.

Of the people surveyed, 45.3% take an iron supplement without being told to do so by a doctor. If you take iron that your body does not require, you could be damaging your liver and other organs. If you are screened for this disorder at an early age, and are found to have HH, you will live as long as a person who doesn't have HH as long as you have the necessary phlebotomies.

Experiments showed liver developing cirrhosis

We also did an experiment to show the effects of iron on the liver. We purchased a fresh beef liver and cut it into 12 sections. Three sections were placed in distilled water and left alone. Three sections were placed in distilled water and the minimum daily requirement (18 mg) of pure iron. Three sections were placed in distilled water and 3 times the daily requirement of iron. The last three sections were placed in distilled water and 10 times the daily requirement of iron. The samples placed in the distilled water remained unchanged, as did the samples with the minimum daily requirement of iron. The samples placed in 3 times the daily requirement of iron developed mild cirrhosis. The samples placed in 10 times the daily requirement developed cirrhosis, and started falling apart. (We compared the samples with pictures of livers with cirrhosis, and our liver looked the same. It developed black spots and bumps.) We repeated the test, and got the same results.

Children/Teens Needed for First HH Pediatric Study

The first HH Children's Study of its kind is now officially open! This is an excellent opportunity for you to have your young family members tested for the genotype and phenotype for HFE associated hereditary hemochromatosis for FREE! This could include genetic testing for the cys282 and his63 gene mutation as well as annual transferrin saturation percentage (TS) and serum ferritin testing for the next five years.

The child may be any age, up to the 18th birthday, and can live anywhere in the USA. Blood samples would be shipped from the child's doctor to the doctor in Mississippi who is the lead investigator on this study. Dr. Michael Nowicki of the University of Mississippi, has requested that I work through my Children *HH*elping Children screening project to find children who are eligible for this study. The children must be compound heterozygotes or homozygotes for the cys or his mutations to be eligible to enroll. The children may have already been genetically tested, and if they are compound heterozygous or homozygous, are automatically eligible for the study IF, they have never received treatment (bloodletting).

If they have already had treatment, then they would fall into another category, that of a compilation of children who have had treatment for hemochromatosis.

All information will be confidential, consent forms will be signed by parents, and a full protocol will be given to anyone considering enrollment of their child(ren).

If the parents have not genetically tested the children, but know that one or both of them is a confirmed homozygote or compound heterozygote, or is a heterozygote for (one mutation) then the chances are high (or certain) that the child(ren) will be homozygous or compound heterozygous. If this is the genetic configuration of the parents, then the study will test the children GENETICALLY for free also in the hopes of finding more children who are eligible for the study who need to be screened and protected.

This study will help establish documented cases of HFE mutations in children and give doctors and researchers the information that they need. I am very pleased that the American Hemochromatosis Society has been asked to provide children from the C*HH*C project I founded three years ago as well as to recruit new candidates from our database.

If you have any questions, please email me directly so we can discuss them. Thank you for your help with this historic study!

Sandra Thomas, President, AHS
Founder, "Children HHelping Children"
Screening & Awareness Project (CHHC)
ahs@emi.net
561-266-9037

HH Screening "Cost Effective" Study Reports

Annals of Internal Medicine
15 February 2000 Volume 132 Number 4 Pages ***261-269***
http://www.annals.org/issues/v132n4/full/200002150-00003.html

Screening for Hereditary Hemochromatosis in Siblings and Children of Affected Patients: A Cost-Effectiveness Analysis

Hashem B. El-Serag, MD, MPH; John M. Inadomi, MD; and Kris V. Kowdley, MD

The authors compared the cost-effectiveness of screening siblings and children of the proband, the first person diagnosed in the family, with results of no screening.

Screening strategies included serum iron studies as well as genetic testing.

If the proband tested homozygous for the C282Y gene (two copies of the mutant gene) , the spouse underwent gene testing. If the spouse was heterozygous (a carrier of the single mutation of the C282Y gene), the children underwent gene testing. If the proband was homozygous, then relatives underwent direct gene testing.Results showed that overall these strategies proved cost-effective; strategies using gene testing were less costly than serum iron studies.

Survey Finds that Physicians Need Education On Gene Testing for HH

[South Med J 93(5):469-471, 2000. Southern Medical Association]

Use of HFE Mutation Analysis for Hereditary Hemochromatosis: The Need for Physician Education in the Translation of Basic Science to Clinical Practice

http://www.medscape.com/SMA/SMJ/2000/v93.n05/smj9305.05.kohl/smj9305.05.kohl-01.html

Manish Kohli, MB, ChB, Steven A. Schichman, MD, PhD, Louis Fink, MD, Clive S. Zent, MB, BCh, Department of Internal Medicine, Division of Hematology/Oncology and the Department of Pathology, John L. McClellan Memorial Veteran's Hospital and University of Arkansas for Medical Sciences, Little Rock.

The researchers discovered that the HFE genetic tests for Hereditary Hemochromatosis were underutilized in Arkansas. Mail surveys were sent out to all physicians in the state to determine (1) if the physician was aware that a genetic test existed; (2) knew that the test was available to physicians in Arkansas (3) whether the physician had used the test. The results indicated that of 40% of the surveys returned, only 21%of the doctors knew that a gene test existed, only 10% knew that the test was locally available, and less than 3% had ever used the test. Only one doctor had used the test to screen first degree relatives of a C282Y homozygote proband. The study found that specialists (gastroenterologists and hematologists) were more likely than primary care physicians to know about and utilize the tests.

Study Reports Screening Results

Annals of Internal Medicine

5 September 2000 Volume 133 pp. 329-337

The Effect of *HFE* Genotypes on Measurements of Iron Overload in Patients Attending a Health Appraisal Clinic

Ernest Beutler, MD; Vincent Felitti, MD; Terri Gelbart, BS; and Ngoc Ho, MS

This important recent article reports the results of the screening of 10,198 patients at the Kaiser Permanente Health Appraisal Clinic in San Diego, CA.

The purpose of the study was to relate the frequency of the three mutations that cause hemochromatosis to variables of clinical iron studies.

Data showed that transferrin saturation levels for all three mutations were elevated even for heterozygotes. Although the mean hemoglobin level differed significantly among persons with each of the three genotypes, the study concluded that for those carrying any of the HFE mutations, the hemoglobin was slightly higher.

The study also documented similar increase in serum ferritin levels in compound heterozygoes (C282Y/H63D), but that heterozygotes for either of these mutations showed no significant increase in serum ferritin. However, male compound heterozygotes for the (C282Y/S63C) mutations showed a similar increase in serum ferritin as compound heterozygotes for the (C282Y/H63D) mutation. The study also reported that "In addition the effect of HFE mutations on the mean corpuscular volume [MCV] was significant even in heterozygotes."

The primary conclusion of this study was that screening for TS% and ferritin levels is insufficient for detecting all homozygotes for the major hemochromatosis mutation and that heterozygotes for all mutations had a lower risk for iron deficiency anemia.

On the other hand, the study reached no solid conclusions on the relationship of HFE mutations to the expression of disease, but it raised many questions and proposed more studies.

Read the full text of this statistically based article at:

http://www.annals.org/issues/v133n5/full/200009050-00008.html

AHS Basics for Diagnosis and Treatment

by Sandra Thomas

*Do not simply ask your doctor to test you for iron overload/ hereditaryhemochromatosis, but rather ask for these blood tests by name: serum iron,TIBC (total iron binding capacity), and serum ferritin (blood taken indoctor1s office from your arm not your finger.) Remember hematocrit (hct)and hemoglobin (hgb) are not tests to confirm HH.

*Percent of saturation is calculated by dividing the numerical result of the serum iron by the numerical result of the TIBC. A percent of saturationgreater than 40% (>40%) and/or a serum ferritin greater than 150 ng/ml (>150ng/ml) is suggestive of HH and should be further investigated by a qualified physician.

*DNA (genetic) testing by a lab which tests for both HFE mutations (cys282 & his63) can help confirm the diagnosis in the presence of high iron levels or determine genetic risk for HH in patients with normal iron levels who may be at high risk of storing iron in the future. Patients who have normal iron levels but who are positive for the HH mutations should be put in a surveillance program by their doctors.

*Always get copies of your own lab results and medical records and read them yourself. Keep a master set of medical records in a home medical file; never give out your original master set records, make copies instead.

*Always consider getting a second or multiple, medical opinions, especially if you have an advanced case of HH with serious medical complications.

*Avoid eating or handling raw seafood, especially if you have liver disease due to HH (or any cause) cooked seafood is fine; do not take vitamin C supplements (natural sources such as orange juice are fine);and do not take iron supplements or vitamins with iron; do not cook food in cast iron cookware.

*Do not eat a diet void of iron, it is unpleasant, unhealthy, and you will end up weak! (one patient reported eating nothing but water and cottage cheese the first two weeks after her diagnosis she was so fearful of eating anything with iron in it). Remember our diet motto: "All take heed, you may eat the food that you need, but be willing to bleed!"

*Aggressive bloodletting (therapeutic phlebotomy/TP) should be the treatment of choice with 1 unit of blood removed usually once or twice a week, until serum ferritin is less than 20ng/ml (<20) and then maintain it there performing 3 to 4 treatments per year for the lifetime of the patient. Treatments should be individualized to each patient, taking into consideration the age, size, weight, and stage of HH of the patient, so that frequency of treatments and amount of blood removed can be adjusted accordingly.

*Liver biopsy may be necessary to determine extent of advanced liver disease if liver function tests (LFT's) are significantly elevated and there are signs of liver distress. Discuss the need for this procedure carefully with your physician. Those with early, asymptomatic HH with normal LFT's, probably will be able to forego this invasive, and potentially risky procedure. The earlier the diagnosis, the less chance you'll need a liver biopsy, so early screening is important. If you do have a liver biopsy, request that it be done with "guidance" such as ultrasound or CT and don't take aspirin type medications a week prior to the procedure. Liver biopsy is used to diagnose, not screen for, primary liver cancer (PLC) . Patients at high risk for PLC should be screened every three to six months with alpha fetoprotein and PIVKA-II blood tests for the rest of their lives.

*Warn all family members to be tested and keep a computer file of family members' results. Don't forget the elderly and the kids when screening family members. Remember, men, women, and children can get iron overload/HH.

*Download a copy of the ***Annals of Internal Medicine***, December 1998 supplement on the **"Diagnosis and Management of Hereditary Hemochromatosis"** at http://www.acponline.org/journals/annals/01dec98/mgmthemo.htm and show your doctor.

*Take an active part in your health care. Ask questions, take notes, research information, keep records, and educate yourself. It's your life!

HH Centers Featured in past issues

University of Pittsburgh, Geoffrey Block MD, Director, 412-647-1500.

University of Pennsylvania, Chris Friedrich MD, Director, 1-800-789-PENN (7366)

University of Washington, Kris V. Kowdley MD, Director, 206-598-3339.

Kaiser Permanente, San Diego Vincent Felitti MD, Director Contact Naomi Howard, at Naomi.X.Howard@kp.org

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Want to Help Raise *HH* Awareness?

Form an AHS Support Team in your state or city.

How?

Look for Guidelines to be posted at the AHS web site

Contact:

Sandra.Spangler@americanhs.org

to enroll your Chapter

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