

HIND-Sight

Huntington's Information & New Discoveries

HDSA Center of Excellence
University of Iowa

The Human Genome Project and Beyond

Researchers from around the world united in 1990 to launch the Human Genome Project. Their goal was to determine the sequencing of the billions of building blocks or letters that form our unique genetic code or instruction manual. This genetic information is stored in 23 pairs of chromosomes. They speculated that the genomes of any two people are more than 99% the same. This tiny fraction of difference makes each of us unique. The HD community is well aware of how important the sequencing of DNA has been to finding the huntingtin gene on chromosome 4 which led to an accurate blood test for the HD gene.

A further collaboration of scientists, publishing in Nature 2007, are examining that 99% sameness or what has been referred to as "junk DNA". They believed that the large percentage (95-99) must have some other function. They now believe that this "junk" is involved in regulating how genes build and maintain the body. One scientist used the analogy of musical instruments as DNA and the regulating of genes

as the instructions needed to bring the symphony together.

Even Francis Collins, the geneticist in charge of the genome project, is impressed with how much the scientific community has learned since completing the genome project in 2003. Scientists have discovered 1700 genes linked to disease, including HD. Collins believes that "the best hope of curing disease comes out of genomics, because it points to the problem of disease at the molecular level rather than at symptoms". This will lead us to having more accurate ways of diagnosing patients for their risk of developing specific diseases.

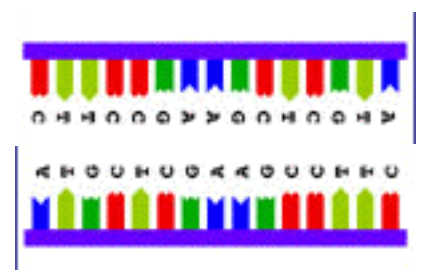
An example of this was the co-founder of Google, Sergey Brin, who invested in a company that tests for certain diseases. He has a family history of Parkinson's but he now knows that he carries the gene that increases his risk for the disease. (Though this ability to test for a certain disease may concern the average person) the HD community has been facing these decisions for at least 15 years. Do I want to know my genetic make up? People who choose to test can feel empowered with this information. They can obtain more education and get involved in research. For others, testing may create more anxiety and fear. Involving a genet-

ic counselor is a good way to sort out information before proceeding with testing.

The HD scientific community benefits from these collaborations. Understanding HD at the molecular level continues to be a focus of HD researchers like James Gusella, PhD, Massachusetts General/Harvard University. As he explained at the HDSA meeting in Pittsburgh in June, basic research at the molecular level is organized in four teams that are continuing to learn about the structure and function of the huntingtin protein.

The collaboration happens on many levels. Scientists continue to work together to encode the DNA elements, HD researchers work on teams to learn more about the huntingtin protein, drug development happens at the applied level and HD families are needed for research. Thank you to all the groups moving knowledge forward.

For more information, check out: www.genome.gov



Iowa Hope Dinner

The Celebration of Hope Dinner took place on November 1, 2008 at the Hotel Fort Des Moines in Des Moines, Iowa. The 2008 honorees were:

Excellence in Medicine

Anne Leserman

HDSA Achievement Award

Ben Easter

Giving a Voice to HD Award

KMA radio and Tim Wayne

HD Advocate of the Year

State Senator Thomas Courtney

Corporate Leadership

Best Buy

Individual Leadership

Karen Brown

HDSA Family of the Year

Brian and Christy Walker

Dinner guests enjoyed both a silent and live auction, live music and a lovely dinner. Thanks to both attendees and sponsors for continuing to support the HDSA Center of Excellence at the University of Iowa. Please see the Iowa Connection website for pictures from the event. <http://hdsaiowa.org/>



Iowa
Celebration of Hope Dinner

GINA Update from HSG

The Genetic Information Non-discrimination Act (GINA) was discussed at the November HSG (Huntington Study Group) meeting in St Pete, Florida. Lewis Maltby, President of the National Workrights Institute, spoke about the recently signed legislation.

GINA was signed in May by President Bush and will go into effect May, 2009 or the month that your employer's health plan goes into effect.

The bill specifically covers genetic discrimination in health insurance. Employees cannot be treated differently because of their genetic status, and an employer cannot acquire or ask for genetic information from its employees. Genetic information includes genetic tests of an individual and that person's family members (including a disease or disorder that are in the family). It does not include provisions for discrimination in life insurance or long term care insurance.

Though not a perfect solution, GINA begins to address issues of a genetic nature. It is common that scientific discoveries precede social policy. It will continue to be important to modify and improve social policy as the science of genetics expands and the ability to know our future health concerns are readily available.

Tetrabenazine Approved and Available

Tetrabenazine is the first drug to be approved by the FDA (Federal Drug Administration) for the treatment of chorea associated with HD. The medication will become available after November 24, 2008.

The pharmaceutical company, Ovation, will distribute the medication through specialty pharmacies throughout the country. Patients and their physicians will fill out a one page form that will be faxed to Ovation for review. The company will work with patients to receive the medication with few finan-

cial barriers to patients and their families. The company's REACH (resource and education) program will look at individual patient's pharmacy coverage.

Please note that tetrabenazine is a medication that treats only severe chorea. It is a medication with some serious side effects. Any person taking this medication will be carefully monitored by their treating physician. The most common side effect is depression.

It is good news that tetrabenazine has been approved by the FDA. We are hopeful that, in the near future, other medications will also gain FDA approval for HD symptoms. Please be aware that there are other medications used to treat HD symptoms even if they are not exclusively approved for treating HD.

HD Support Groups:

DES MOINES
Valley View Village Conference
2571 Guthrie Ave
3rd Sunday at 1:30 pm
Mark Hillenbrand
(515) 208-3511

OMAHA, Nebraska
Perkins Restaurant
108 L St
2nd Monday at 6:00 pm
Cathy McNeil
(402) 537-0739

IOWA CITY
University of Iowa Hospitals and
Clinics
Della Ruppert Conference Room
4th Sunday at 1:00 pm
Anne Leserman
(319) 353-4307

FONDA
Fonda Nursing and Rehab
Wilma Frey
(712) 288-4441



RESEARCH

2CARE

Coenzyme Q10 in HD

The Huntington Study Group (HSG) is conducting a multi-center, randomized, double-blind, placebo-controlled study of coenzyme Q10 (CoQ) in individuals with HD to assess the effects of CoQ on the progression of functional decline in HD, as well as the long-term safety and tolerability at the dosage studied. Coenzyme Q10, a naturally occurring substance in the body, is available for purchase over the counter as a nutritional supplement. CoQ is being studied as an investigational drug at a higher dosage than is currently available for purchase.

CoQ has been used to treat a variety of human disorders, including those involving the heart and circulatory system, cancer, muscular dystrophy, a muscle coordination disorder called ataxia, and other disorders. The most marked results seem to have occurred in patients with a preexisting inherited deficiency of CoQ, although the studies reporting this information are limited because they were not controlled clinical trials. No serious safety issues have been reported. Recent preliminary studies of CoQ in neurologic disorders such as Parkinson's disease, Amyotrophic Lateral Sclerosis, and HD confirm the safety and tolerability of CoQ in daily dosages up to, including, and exceeding the

dosage planned for the 2CARE study when used for a short time.

The 2CARE study will be the largest therapeutic clinical trial to date in HD. Six hundred eight research participants will be enrolled at approximately forty-six clinical sites in the US, Canada, and Australia. For information about participating at the University of Iowa, please contact William H. Adams at 319-353-4411 or email william-h-adams@uiowa.edu.

BESEVBCH



Eye Tracking:

Establishing a Biomarker for HD

Eye Tracking is a study designed to utilize advanced eye-tracking methods to establish a biomarker for HD by investigating subtle motor and cognitive eye tracking problems in individuals tested for HD. Participants 18 years of age or older who have completed an HD gene test are invited to participate in this 60 minute eye tracking session. Participants will include people who have tested both positive and negative for the HD gene and those with and without symptoms of HD. Compensation is available to study participants. For more information call Anne Leserman at (319)353-4307 or email anne-leserman@uiowa.edu.

RESEARCH

CIT-HD

Citalopram in HD

Do you know someone that is gene positive and/or diagnosed with HD? Is that person NOT take an anti-depressant medication? The HD Center at the University of Iowa is looking for 40 such people. The CIT-HD study assesses results of thinking tasks in participants with early symptoms of HD. This is a double blind study of citalopram, an FDA approved medication for depression. For more information call Bill Adams at (319) 353-4411 or email william-h-adams@uiowa.edu.

COMING SOON

HART

ACR16 Versus Placebo for the Symptomatic Treatment of HD The Huntington Study Group (HSG) is conducting a study of the research medication ACR16 in persons 30 years of age and older who have clinical features of HD. HART is designed to determine the general safety and tolerability and an effective dose of ACR16 as well as the effect of ACR16 on motor (movement) and cognitive (thinking) abilities in subjects with HD. Approximately 35 research centers in North America will enroll up to 220 subjects for 16 weeks each. Anticipated enrollment will begin at Iowa late in 2008. This study is sponsored by NeuroSearch Sweden AB.

BESEVBCH RESEARCH



Anne Leserman
1-317 MEB
Iowa City, Iowa 52242
(319) 353-4307
anne-leserman@uiowa.edu

Mental Health Parity

Congress incorporated the Mental Health Parity Act into the Emergency Economic Stabilization Act (the “bailout” bill). It requires health insurance providers to NOT distinguish between mental and physical health benefits in terms of co-pays, deductibles, and coverage limits for plans provided to companies with more than 50 employees.

The bill was passed after 12 years of advocacy by friends and relatives of people with mental illness and addiction disorders. The law is an effort to end insurance discrimination and to reduce the stigma of mental illness.

Insurance group health plans will need to adjust their benefits to comply with the new law, which requires equivalence, or parity, in the coverage. For decades, insurers have set higher co-payments and deductibles and stricter limits on mental health treatment. The law will encourage insurers to integrate coverage for mental health care with medical and surgical benefits.

The drive for mental health parity was led by Senator Pete Domenici, Republican of New Mexico, who has a daughter with schizophrenia, and Paul Wellstone, the Democrat from Minnesota who was killed in a plane crash in 2002. Mr. Wellstone had a brother with severe mental illness. The mental health parity law was put forth in the Congress in a consensus building process.

Mental illness is quite prevalent in the current US population with about 58 million Americans with a diagnosable mental disorder. Mental illness has also been associated with more than 30,000 suicides each year. An estimated 16 percent of all inmates suffer from mental illness and about 20-25 percent of the homeless population meet criteria for a mental disorder.

Nytimes.com
Abcnews.com
National mental health information center