

## WHAT IS THE HD ROSTER?

The National Research Roster for Huntington Disease Patients and Families is a unique nationwide information resource dedicated to assisting scientific research on Huntington disease. The goal of the Roster is to help researchers learn more about HD. To accomplish this goal, the Roster has involved nearly 3000 families in HD research.

The Roster collects the names of families, including information about the history of HD in the family (family trees) and other related data, received from two types of questionnaires. Information received from the questionnaires identifies HD patients and families who are interested in participating in research projects.

Scientists interested in studying HD may submit two types of requests for information contained in the Roster. The two types of information requested are anonymous information and identifiable information. Anonymous information is de-identified, meaning that names and personal identifiers are removed and can be given to researchers without having to contact Roster families. Identifiable information includes data such as names, dates of birth, and family structure. Identifiable information can also be used to help researchers find volunteers who are willing to participate in HD research projects nationwide. In any instance where identifiable information may be given to a researcher, the Roster will contact participants to ask if they are willing to share their identifiable information for a research project.

- The Roster is totally VOLUNTARY.
- You can withdraw from the Roster at any time.
- No information about you will be given to anyone without your WRITTEN permission.
- No one, including your own family members can find out if you are participating in the Roster without your WRITTEN permission.
- You decide if you want to participate in any available research opportunity. You do not have to participate if you are not interested.

We would like to thank the many families who have participated in the Roster by providing family history information. We believe the Roster is a valuable scientific resource which will assist in the discovery of a cure for HD. We would like to extend the invitation to join the Roster to you and your family.

**We are always eager to accept new participants!**

# REACH

Volume 4

Winter 2008

The National Research Roster for Huntington Disease Patients and Families

## HD MAPS STUDY

By **Richard H. Myers, PhD**

Boston University School of Medicine

The purpose of the HD MAPS study is to identify the genes that influence the age at which people will develop Huntington's disease as well as to identify genes that may influence other important features of HD, such as the rate of the disease progression. In addition to the HD Roster, the HD MAPS study has recruited people affected with HD from about a dozen US clinics as well as clinics in Canada, Australia, England, Ireland, France and Italy. To date 1,170 people have provided DNA and information on onset age to the study.

A lot of people recognize that the CAG repeat in the huntingtin gene influences onset age. That is especially evident for people with large repeats (above 55 or 60). It is also apparent however, that for people with mid-life onset of HD the repeat size is not a good predictor of onset age (the figure below shows how much onset age can vary for people with the same repeat size).

People with HD CAG repeats above 60 tend to have very young onset ages, often before age 20. However, individuals with repeats in the range between 40 to 50 CAG repeats, persons with the same repeat size can have onset ages that vary by 30 years or more.

For example if you look at people with 45 repeats, the person with the youngest onset was 20 years old and the person with the oldest onset was 50 years old.

Consequently, the repeat size is not a good predictor of onset age for the common midlife onset form of HD. (figure 1)

Studies of families with HD showed that it is very likely that other genes interact with the HD gene and influence how old people are when they develop HD. So the HD MAPS study set out to identify the genes influencing onset age with the hope that these findings might give us clues about how to postpone the onset for people who weren't fortunate enough to inherit genes that would do that for them.

### RESEARCH FINDINGS

The first phase of the study focused on families with affected siblings (brothers and sisters) and other affected relatives. With a sample of 836 people from families with at least two affected members participating, a study was performed to identify the chromosomes that most likely carry genes that modify HD onset age. The findings showed that there is a gene on chromosome 6 (specifically chromosome 6q23-24) that appears to be a very powerful modifier of HD onset age. A second modifier seems to be located very close to the HD gene itself on chromosome 4 (chromosome 4p16). A third area of interest is on chromosome 18 (chromosome 18q22). These findings were published in two papers (Li et al. American Journal of Human Genetics 2003; 73:682-687 and Li et al. BMC Medical Genetics 2006, 7:71).

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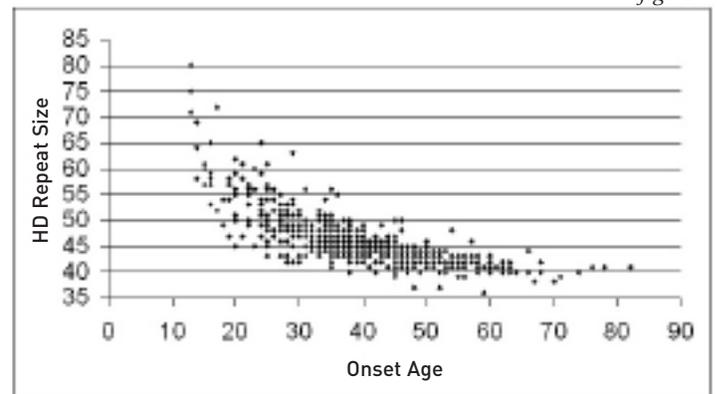


figure 1

## FUTURE RESEARCH

In order to identify the specific gene(s) that are influencing the onset age, a second study called a “genome wide association study” was initiated. In September of 2007, 1170 DNA samples, including the original 836, were sent to be genotyped for 1,000,000 genetic variants. It is expected that the results of the genotyping will be available for study by the end of 2007.

The investigators expect that it will take several months to try to make sense of all the information that will be forthcoming, but we hope to identify specific genes that are most likely to influence the onset age for Huntington’s disease.

## STUDYING MORE THAN JUST ONSET AGE

The HD MAPS investigators are also very interested in looking into some of the other important clinical presentations in HD. For example, we would like to evaluate whether there are genes that influence how rapidly the disease progresses or whether people will have depression or weight loss as a significant part of the way their illness is expressed. For this reason, we are in the process of collecting as much additional information on each HD MAPS participant as we are able. In this way we can test for genes that might influence these other important features of HD.

## UNDERSTANDING WHY SOME PEOPLE RESPOND TO TREATMENT AND OTHERS DO NOT

The HD MAPS study will have information on a very large number of

individuals who may also be taking part in clinical trials. In some of the clinical trials that have taken place for HD, it has been noticed that some people seem to respond to treatments better than others. One possibility for these different responses is that some people are genetically more capable of benefiting from a treatment while others are genetically resistant to those benefits. With information on a large number of people with HD, we may be able to evaluate which genetic differences are likely to interact with a given treatment and to have a better idea about whether someone is likely to be helped by certain medications.

## SUMMARY

The eventual goal of the HD MAPS study is to identify the genes that influence many different aspects of the way HD is expressed. With this information we hope to develop treatments that would be capable of postponing the onset for individuals who carry the HD gene. In addition to postponing onset, it may be possible to identify certain individuals who are prone to depression or rapid disease progression and to target the genes that may be responsible for these forms of HD.

The HD MAPS study appreciates all the cooperation of the HD Roster, and the Roster participants who have participated in the study. The study has been supported by PHS Grant P50NS016367 (Huntington's Disease Center Without Walls), NS16375, NS32765, the CIHR of Canada, the Huntington's Disease Society of America's "Coalition for the Cure", the Jerry McDonald Huntington's Disease Research Fund, and an Anonymous Donor. If you have questions about the HD MAPS study please contact Tiffany Massood at Boston University (617-638-5483) or [tmassood@bu.edu](mailto:tmassood@bu.edu).

## WELCOME OUR NEW HD ROSTER COORDINATOR!



**Shelley Burnham** joined The National Research Roster for Huntington Disease Patients and Families in March of 2006 as the new Research Coordinator. Shelley has several years

of health research experience having worked in both dental health and infectious disease research. As the new coordinator, Shelley is actively working to recruit new individuals and families to participate in the Roster and to promote awareness of the Roster among researchers interested in HD research.

## updates

### NEW ROSTER MAILING PROCEDURE

All letters and correspondence mailed to HD Roster members will now be addressed as “Dear Participant”. While this depersonalizes our mailings, we believe this change will prevent the risk for any letters being put into an envelope addressed to another Roster family which would cause a breach of subject confidentiality. The Roster believes confidentiality is very important and all identifiable information received by the Roster remains confidential. We hope our Roster contact members are not offended by this new procedure, but take comfort in knowing the Roster is working very hard to keep your information private.

### ADDRESS CHANGES

Has your address changed? If it has, please notify us. Having current address information on file will ensure that Roster contacts receive REACH newsletters without complications, as well as recruitment letters for potential research opportunities. Notification of change of address can be mailed to:

The National Research Roster for Huntington Disease Patients and Families  
Attention: Shelley Burnham  
Department of Medical and Molecular Genetics  
410 West 10th Street • HS 4000  
Indianapolis, IN 46202

Or email: [hdroster@iupui.edu](mailto:hdroster@iupui.edu)

# LIVING POSITIVELY WITH HUNTINGTON'S DISEASE

**By Phil Hardt**

Chief Volunteer Officer for the Arizona Affiliate  
phardt1@cox.net

Good afternoon, my name is Phil Hardt. I am the HDSA 2001 Person of the Year and have been the current Chief Volunteer Officer for the Arizona Affiliate and Director of their Annual HD and JHD Retreat for the last 6 years. I had the privilege of testifying about HD in front of the NIH's committee on Genetic Discrimination in 2004.

Two months ago I had the bitter-sweet experience of traveling to Colombia, South America, to the city of Juan de Acosta to help impoverished families struggling with HD. In case you haven't heard of Juan de Acosta, it has the distinction of having the second largest concentration of those with HD in the world, right behind Lake Maracaibo in Venezuela.

I was privileged to be able to take over 500 pounds of clothing, the first information they had ever received on Huntington's Disease (in Spanish of course) and over two and a half million pesos which was used to provide assistance to 20 families for two months each (not hard when the average monthly income there is only \$40 US dollars). In addition to the normal complications we have in dealing with our loved ones with HD, they are doing it, most without running water, transportation and medications and some without even running water and electricity! I hope to be able to return to help more families this September. During the years I have come up with a list of recommendations to help each of us live positively with HD.

I'd like to start with a true story called Three Strings. It's an attitude and a lifestyle that I commend to all of you:

## ONE

On November 18, 1995 Itzhak Perlman, the violinist, came on stage to give a concert at Avery Fisher Hall at Lincoln Center in New York City. If you have ever been to a Perlman concert, you know that getting on stage is no small achievement for him. He was stricken with polio as a child, and so he has braces on both legs and walks with the aid of two crutches.

To see him walk across the stage one step at a time, painfully and slowly, is a sight. He walks painfully, yet majestically, until he reaches his chair. Then he sits down, slowly, puts his crutches on the floor, undoes the clasps on his legs, tucks one foot back and extends the other foot forward. Then he bends down and picks up the violin, puts it under his chin, nods to the conductor and proceeds to play.

By now, the audience is used to this ritual. They sit quietly while he makes his way across the stage to his chair. They remain reverently silent while he undoes the clasps on his legs. They wait until he is ready to play.

But this time, something went wrong. Just as he finished the first few bars, one of the strings on his violin broke. You could hear it snap - it went off like gunfire across the room. There was no mistaking what that sound meant. There was no mistaking what he had to do.

People who were there that night thought to themselves: 'We figured that he would have to get up, put on the clasps again, pick up the



“Each of us has lots of beautiful music left in us and all we have to do is start learning how to play it!”

crutches and limp his way off stage - to either find another violin or else find another string for this one.'

But he didn't. Instead, he waited a moment, closed his eyes and then signaled the conductor to begin again. The orchestra began, and he played from where he had left off. He played with such passion and such power and such purity as they had never heard before. Of course, anyone knows that it is impossible to play a symphonic work with just three strings. I know that, and you know that, but that night Itzhak Perlman refused to know that. You could see him modulating, changing, and recomposing the piece in his head. At one point, it sounded like he was de-tuning the strings to get new sounds from them that they had never made before.

When he finished, there was an awesome silence in the room. And then people rose and cheered. There was an extraordinary outburst of applause from every corner of the auditorium. We were all on our feet, screaming and cheering; doing everything we could to show how much we appreciated what he had done. He smiled, wiped the sweat from his brow, raised his bow to quiet us, and then he said, not boastfully, but in a quiet, pensive, reverent tone, 'You know, sometimes it is the artist's task to find out how much music you can still make with what you have left.' (Jack Riemer, Houston Chronicle)

Think about what he said one more time- "You know, sometimes it is the artist's task to find out how much music you can still make with what you have left." Each of us has lots of beautiful music left in us and all we have to do is start learning how to play it!

## TWO

Become familiar with what I like to call the "softer" symptoms of HD. I define the "softer" symptoms as the behavioral, emotional and cognitive symptoms of the disease. These are, beyond a doubt, the most damaging to the Person with HD (Phd) and their loved ones. Note: I like to call the person with HD a Phd instead of the sick one, ill one, crazy one, etc. as I believe it renders dignity where it is deserved. If you will notice, it's also spelled a different way- I always capitalize the

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“P” and make the “hd” in small letters because we are bigger than it and will eventually beat it one day! Although these are the most important, they are unfortunately still the least understood, talked about, researched, and sometimes the hardest to recognize. These softer symptoms are the ones that cause the Phd to get fired by their employer, arrested, divorced, and that destroy family relationships with siblings, children and spouses. An abbreviated list of them includes:

- A) loss of short term memory
- B) impulsivity
- C) loss of inhibitions which cause the Phd to say hurtful things, get arrested for public masturbation, walk outside naked, have affairs, get caught shoplifting or embezzle millions of dollars
- D) loss of forward or consequential thinking which causes the Phd to not see the consequences of rash purchases, hitting a policeman who has falsely arrested them for being drunk in public (can you see how dangerous it would be to combine loss of inhibitions with loss of consequential thinking and impulsivity! Three strikes before you get started!)
- E) loss of emotional gating which causes rage attacks and/or an increase in emotional blunting which makes the Phd seem cold and uncaring
- F) unawareness - I prefer to call this “unawareness” rather than “denial” because I feel this is another HD symptom and not the Phd purposely trying to be an SOB. Unawareness causes the Phd to not know that they are unable to continue driving (despite their car looking like it took last place at a demolition derby) or that they need to take their prescribed medications.

Other “softer” symptoms include:

- G) the inability to notice lowered work quality
- H) the loss of ability to care about personal hygiene
- I) impaired judgment and reasoning
- J) self-centeredness
- K) obsessive and compulsive behaviors
- L) loss of ability to initiate conversations. It’s also important to note that if you are experiencing any of these HD symptoms they will usually be “uncharacteristic” of your prior behavior and demeanor.

### THREE

While you are still able, involve yourself in the service of others so you don’t have time for a pity party. One of the best ways to do this is volunteering. Volunteering can fill the void left from not being able to drive and work any more and can restore your feelings of self-worth and meaning to your life again.

### FOUR

Participate in ALL research studies. I hear everyone complaining that we don’t have a cure yet and then when we have the opportunity to participate in research studies we don’t. PHAROS is a sad example of what I’m talking about- approximately 250,000 at-risk individuals who could have participated and the study could barely get 1000 participants. This is appalling. Our numbers are small and so everyone really counts in these types of studies. Participate to make a difference.

### FIVE

Wrap up things in your life and do some things you’ve always wanted to (within reason), so you have no regrets later.

### SIX

Learn to understand researcher’s jargon. When they say they’ve created “HD-like” symptoms or “HD-similar symptoms” in a mouse and made them go away, remember this is not the same as actual HD symptoms and a mouse’s brain or the eye of a fruit fly are not the same as a human brain- it’s a start that will lead to more discoveries later but learn not to get too excited and then depressed later if the studies don’t pan out. I think you get my point.



Phil Hardt zip lining through the jungle in Costa Rica on his honeymoon, May 2007.

### SEVEN

Learn to have hope today in improved CARE. As Dr. Edmund Chiu said in 1999 in his talk in New York City: “We hope and pray for a cure but there is not a cure for Huntington’s Disease, not yet. Good people are working on it of course. In the meantime, other good people are working on the only alternative available – care. The kind of care that doesn’t abandon the Huntington’s Disease person, the kind of care that tries to make full use of every single brain cell the Huntington’s Disease person can command [sounds like Three Strings], the kind of care that understands occasional erratic behavior.”

When Dr. Jane Paulsen was speaking to our retreat two years ago a caregiver asked her how to tell if their spouse’s behavior was HD-affected or if they were just trying to be an SOB. I will never forget her answer- “I would error on the side of compassion and then it really doesn’t matter, does it?” So, caregivers, in order to allow your loved ones to continue making as much beautiful music as they can with only three strings I would also ask you to “error on the side of compassion” too and Phds, I would strongly encourage you not to take advantage of this!

### EIGHT

Focus on what you can still do instead of what you can’t.

### NINE

If you are “at-risk” go to [www.hdlighthouse.org](http://www.hdlighthouse.org) and download my “At-Risk Preparation Checklist.” It will help you prepare like never before and then you can follow my admonition: “Prepare like you will get HD and then live like you won’t!” If you are totally prepared and still get HD it really won’t matter will it as you and your family

will be better prepared financially, medically, and emotionally to minimize the devastation than those who persist on being ostriches with their heads in the sand pretending that if they ignore HD it will ignore them. This worn-out philosophy hasn't worked yet because your 50/50 chance is not good odds. We owe it to ourselves, our families and our friends to prepare early while we can, long before HD-affected judgment and reasoning taint or preclude our true wishes and desires from being documented.

## TEN

Learn the warning signs of depression so you can help yourself and your loved ones. Did you know that the suicide rate for those with HD is 10-12 times the national average? This is unconscionable and should not be allowed to happen. The reason so many Phds end up as casualties of suicide is two-fold:

- 1) The biological cell death in the brain causes a chemical imbalance
- 2) Phds also experience situational depression at the same time, wondering how much longer they are going to be able to work, to be intimate with their spouse, to walk or talk. Please realize that almost everyone else around us is already on anti-depressants just trying to cope with situational depression, let alone the biological-caused depression at the same time.

## ELEVEN

Find humor in HD. Look closely at the bag I've been carrying around with me yesterday and today. It has a picture of the Scarecrow from the

*Wizard of Oz* on it with his famous saying: "If I Only Had A Brain." Once you become familiar with the "softer" symptoms make a game out of trying to stay ahead of HD by compensating at work by doing such things as using 3x5 cards to take notes, using Post-It-Notes all over your computer screen, putting footers on everything you work on to help you remember the next day where your documents are saved. When you forget how to shift your car, switch to a car with an automatic transmission and when you are unable to drive it any more, buy a big tricycle so you can still get out and go to the store. See what I mean? Keep telling your HD: "You think you've got the best of me because I can't do this any more, well think again because now I'm going to do it this way!"

In closing I would like to encourage every one of you to continue making beautiful music with what you have left. Remember, you might have to recompose or rethink things in your head a little differently. You may have to do things in a different way today than you did yesterday or adjust your attitude a little bit more towards the positive side but I promise you that the music you make while you have HD will be more memorable and more sacred than any that you have ever made before. May God bless everyone with HD!

June 10th 2006, HDSA National Convention in Milwaukee, Wisconsin.

*Printed in 21 May 2007*

*Find this article at: <http://www.hdac.org/>*

*Article posted to [www.hdac.org](http://www.hdac.org) on: 07-27-2006*

# invitation

Do you have a question you would like to ask of a doctor familiar with HD? Do you have a personal HD experience that you would like to share? You are invited to submit questions to be answered in our "Ask the Doctor" column and/or to share your personal experiences, whether a care giver, patient with HD, or person at risk for HD. If you are interested, please send your questions or story to:

The National Research Roster for Huntington Disease Patients and Families  
Attention: Shelley Burnham  
Department of Medical and Molecular Genetics  
410 West 10th Street • HS 4000  
Indianapolis, IN 46202

Or email: [hdroster@iupui.edu](mailto:hdroster@iupui.edu)

## Become an HD Roster Contact

The National Research Roster for Huntington Disease Patients and Families (HD Roster) was founded over 25 years ago. The participation of HD Roster patients and families was critical in the discovery of the initial marker for Huntington disease and the later localization of the huntingtin gene. The HD Roster has provided clinical data and helped recruit subjects for nearly 200 research studies and has helped researchers improve the understanding of the symptoms of HD.

The Roster currently has over 3,700 individuals who are listed as contacts. A contact is a member of an HD family who has consented to be the source person for information from the family. Contacts are often persons at-risk for HD, a spouse of a person with HD, the parent of a person with HD or the child of a person with HD. Contacts receive the HD Roster Reach newsletter you are reading and also receive mailings about ongoing research studies which are seeking to recruit subjects.

It is very exciting news to announce that the HD Roster has been asked to help recruit subjects for several new clinical trials. It is our hope that these clinical trials will lead to the discovery of drugs that improve the symptoms of HD. In order to most effectively provide potential research participants for the upcoming clinical trials, we would like to increase the number of individuals listed as contacts for the HD Roster. In this way, we hope to reach more members of HD families directly and to provide more of you with the opportunity to participate in research. We encourage all members of each HD family to become an HD Roster contact.

- • • If you would like to receive **HD Roster mailings** to learn about new research studies and to have the HD Roster Reach newsletter delivered directly to your mailbox, please contact us and we will be glad to help you become an HD Roster contact. Please contact us by phone at: 1-866-818-0213, or by email: [sburn@iupui.edu](mailto:sburn@iupui.edu) or at our website <http://hdroster.iu.edu>

# IDENTIFYING NEW BIOMARKERS FOR HD

By Jason D. Rupp

MD/PhD Candidate, Indiana University, School of Medicine

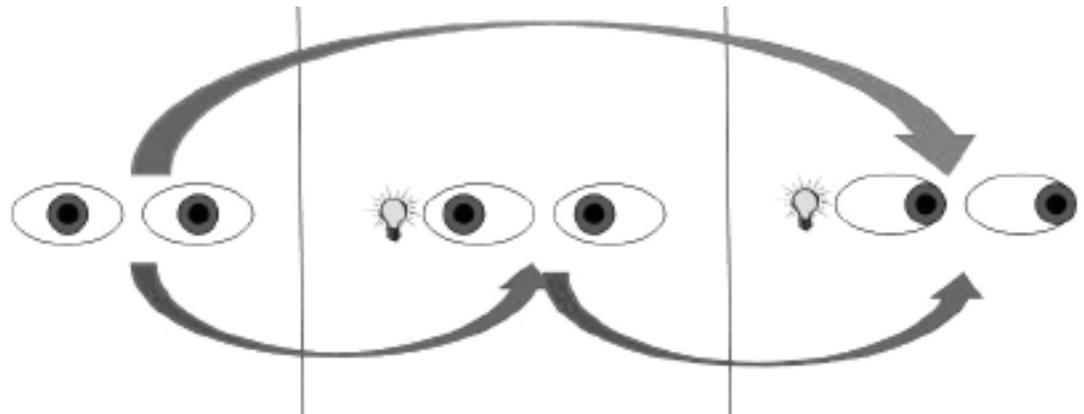
## WHAT IS A BIOMARKER?

Biomarkers are used in medicine in a number of different ways. One use is to help lead physicians to a diagnosis. A good example of this is the use of prostate specific antigen (PSA) when evaluating the likelihood of prostate cancer. Cancer can only be confirmed by biopsy, but an elevated PSA indicates a higher risk. Another use of biomarkers is to help follow the effectiveness of a drug. Following cholesterol levels is a very common example of this practice. Many drugs, including statins, are used to decrease a person's risk of coronary artery disease (CAD). Unfortunately that risk can't be measured directly, so another measure – cholesterol – is used. If a certain drug is effective, then cholesterol levels will drop, indicating a decreased risk of CAD.

Finding a biomarker for Huntington disease (HD) is a current research goal for a number of studies. The hope is to find a biomarker that is an early marker of HD. This marker would be abnormal long before any symptoms of HD can be found during a neurological examination. Dr. Tatiana Foroud and her colleagues have been carefully evaluating eye movements. They have shown that a certain type of eye movement, called a saccade, appears to be abnormal before an individual shows signs of disease that can be detected by a neurologist.

## WHAT IS A SACCADE?

Saccades are very rapid eye movements that we use to shift our gaze from one object to another. They can be involuntary or voluntary. An example of an involuntary saccade is when something out of the corner of our eye catches our attention and we immediately move our eyes to focus on the object; we don't consciously think about looking at the object, it's simply a reflex. On the other hand, we often consciously decide to make a saccade. An example of such



**Figure 1.** In the research laboratory, a person sits in a chair and looks straight ahead. The person is told that a light will flash either to the right or to the left, and he is instructed to look away from the light when it does appear. Correct performance of this task is indicated by the top arrow – the person immediately looks away from the light. Incorrect performance of this task is indicated by the bottom arrows – the person initially looks toward the light, then corrects himself and looks away from the light.

a voluntary saccade occurs when we are watching TV and look at the clock on the wall to check the time; a saccade rapidly moves our gaze from the action on the screen to the clock.

People with HD have difficulties performing certain types of saccades (see figure 1 for an explanation of one type of difficulty), and it appears that these difficulties get worse as the disease progresses. Some of the measurements are so sensitive that they can detect abnormalities more than 10 years before an individual is predicted to onset with HD.

Dr. Foroud's group is conducting many studies to find out which types of saccades are most affected early in disease progression, and to establish how rapidly these saccades worsen as a person gets closer to onset.

## HOW CAN SACCADES BE USED TO HELP HD RESEARCH?

One of the most exciting advancements in the treatment of HD is the development of neuroprotective drugs. As these drugs enter clinical trials, their effectiveness must be measured. Many clinical trials will follow biomarkers as a way to test how

“Finding a biomarker for Huntington disease (HD) is a current research goal for a number of studies. The hope is to find a biomarker that is an early marker of HD. This marker would be abnormal long before any symptoms of HD can be found during a neurological examination.”

well the drug is working. The goal of Dr. Foroud's research is to determine whether the evaluation of saccades could be used as a way to determine whether a drug might be acting to delay the onset of HD or to slow down the progression of disease. For example, if saccades were measured as part of a clinical trial of a new drug, researchers might test whether subjects who received the drug had fewer abnormalities in their saccades or if the drug seemed to slow the rate at which an individual's saccades worsened.

## HOW DO YOU EVALUATE A BIOMARKER?

Dr. Foroud is currently completing several studies designed to determine how rapidly an individual's saccades worsen as they approach the onset of HD.

To perform this study, Dr. Foroud has been studying saccades in groups of individuals at-risk for HD, some of whom will develop HD and some who will not. She is also studying whether changes in saccade function can be determined over a 1 to 2.5 year interval. The answer to this question is very important in evaluating whether these saccadic eye movements will be useful in a clinical trial.

# Research Opportunities

• • • • • If you are interested in any of the research studies described below, please contact the HD Roster:  
Phone: 317-274-5744  
Toll-free: 866-818-0213  
Email: sburn@iupui.edu

## **COHORT: Cooperative Huntington's Observational Research Trial**

COHORT study is designed to collect observational information from individuals who are affected by HD and from those who are part of an HD family, in order to learn more about HD, potential treatment and to plan future research studies of experimental drugs aimed at postponing the onset or slowing the progression of HD.

### **Inclusion/Exclusion Criteria**

- 18 years of age and older
- Individuals who have HD or tested positive for the HD gene
- Parents, children and siblings of individuals who have HD or tested positive for the gene
- Grandparents and grandchildren of those individuals participating in COHORT who have HD or tested positive for the gene
- Spouses of those individuals participating in COHORT who have HD or tested positive for the gene
- HD family members who have tested negative for the HD gene.
- For individuals under age of 18, only individuals who have HD are eligible to participate

## **Identification of Novel Biomarkers in HD**

The Identification of Novel Biomarkers in HD study is seeking to identify potential biomarkers of early HD progressions. Biomarkers are any sign or symptom of disease which can be used to measure the onset of disease even in the earliest stages. This study will test whether individuals with an expanded number of CAG repeats have progressive worsening of their saccadic eye movements, olfactory identification and neurocognitive performance as they approach clinically recognized, disease onset. The identification of such biomarkers is critical to studies of neuroprotective agents which could be used in future treatments to delay or eliminate the onset of the symptoms of HD.

### **Inclusion/Exclusion Criteria**

- Between ages of 18 and 65
- Gene status must be known
- Have a biological parent previously diagnosed with HD
- Not be clinically diagnosable or claim to be experiencing symptoms of HD
- Not pregnant

- Not been diagnosed with a central nervous or neurological disorder
- Never had head trauma that caused the loss of consciousness and required hospitalization
- Do not have any significant abnormal eye impairments

## **Functional Magnetic Resonance Imaging (fMRI) in HD**

Dr. Jane Paulsen's lab has contacted the Roster for help in recruiting patients for the fMRI study. The purpose of the fMRI study is to gain a better understanding of brain functioning in HD. fMRI scans will be used to identify areas of the brain that are active during certain cognitive tasks. This will provide useful data to researchers about the disease process. The HD Roster advisory committee is currently reviewing this study. Once approved, Roster staff will contact all Roster participants to tell them more details of the study and ask if they would like to participate.

### **Inclusion Criteria**

- Between ages of 18 and 65
- Currently enrolled in Predict-HD
- Gene positive, pre-symptomatic individuals
- Gene negative individuals
- At risk for HD and tested for the HD gene

### **Exclusion Criteria**

- Unstable medical or psychiatric illness
- Active substance abuse in the last year

## **Predict-HD**

PREDICT-HD is an observational study; the goal of PREDICT-HD is to determine the earliest sign of active disease in persons inheriting an expanded number of CAG repeats in the HD gene. This information will allow future clinical trials of new drugs delaying the onset of HD to be administered at the earliest possible moment. This study is still recruiting for gene positive/pre-symptomatic and gene negative individuals.

### **Inclusion Criteria**

- 18 years and older
- Able to commit to a minimum of 4 yearly evaluations
- Commitment of a companion to attend visits or complete surveys via mail
- Able to undergo a MRI
- Gene negative at risk individuals
- Gene positive individuals who have not been diagnosed with symptoms of HD

## **WE'VE MOVED!**

In January 2007, the HD Roster moved into our new offices in the Health Information and Translational Sciences building.

While our phone numbers remained the same, our address has changed. Our new address is 410 West 10th Street, HS 4000, Indianapolis, IN 46202-3002



in the spotlight

# JACQUELINE GRAY JACKSON



When I came to Indiana University in 1985, I was hired by Dr. P. Michael Conneally to oversee the National Research Roster for Huntington Disease Patients and Families. I had not heard of HD before coming to my new job and certainly didn't think I knew anyone with HD. Little did I know how much this job would affect my life. Twenty three years later, I am still involved in the HD Roster

project and am also working on several other HD research studies. While many of these projects have been within the United States, my work with HD has also taken me many times to Maracaibo Venezuela, where I worked alongside a team of neurologists and neuropsychologists led by Dr. Nancy Wexler in our common goal of understanding the cause of HD.

Over the course of the past 23 years working with HD has become much, much more than a job. It is a sincere passion driven by the constant desire to help in the search for a treatment or cure for this disease that affects people, many of whom I now call friends. I've come to view HD as a "contagious" disease. Not of course because it's really

contagious in the usual sense of an illness, but because the more you work with HD, the more families you meet, the closer you become to those who are affected by this disease, the more you understand what the disease does to not only the patient but their family, the more you are pulled into the disorder and the more desire you have to find a treatment or cure. I've attended many Huntington Disease Society of America (HDSA) national conventions and numerous HDSA state conventions both in Indiana and other states. The conventions became for me not only a way to spread the word about the Roster but also a way to keep up with families who are now regarded as friends.

In the spring of 2007, I received the HDSA Indiana Center of Excellence award. It was such a fabulous honor to receive this award for service and dedication to HD. I must admit though that the honor of working with HD families has been all mine. HD research has allowed me to meet many new friends, taken me to the most isolated of places and given me the challenge of a lifetime. As with anyone who has gotten to know HD families, there will be no end to our dedication until a treatment or cure for this disease is found. It is my sincerest hope that this will happen in the near future.

## Internet Information on Huntington Disease

### **National Research Roster for Huntington Disease Patients and Families**

<http://hdroster.iu.edu>

### **Huntington Disease Society of America**

[www.hdsa.org](http://www.hdsa.org)

### **NINDS – National Institute of Neurological Disorders and Stroke**

Phone: (800) 352-9424  
[http://www.ninds.nih.gov/health\\_and\\_medical/disorders/huntington.htm](http://www.ninds.nih.gov/health_and_medical/disorders/huntington.htm)

### **HDF – Hereditary Disease Foundation**

<http://www.hdfoundation.org>

### **WE MOVE – Worldwide Education and Awareness for Movement Disorders**

Provides comprehensive educational materials for patients and health care professionals.  
<http://www.wemove.org>

### **YGYH – Your Genes Your Health**

Multimedia website guide for genetic disorders.  
<http://www.ygyh.org>

### **Stanford HOPES – Huntington's Outreach Project for Education at Stanford University**

Provides information on the scientific aspects of Huntingtons disease.  
<http://www.stanford.edu/group/hopes>

### **Caring for People with Huntington Disease?**

Information about caring for people with HD; developed for patient's families and professionals.  
[www.kumc.edu/hospital/huntingtons](http://www.kumc.edu/hospital/huntingtons)

### **Huntington Disease Advocacy Center**

Information, question and answers, problem-sharing, personal experiences and articles on HD.  
[www.hdac.org](http://www.hdac.org)

### **International Huntington Association**

Federation of national voluntary health agencies.  
[www.huntington-assoc.com](http://www.huntington-assoc.com)

### **Nutrition for People affected by Huntington Disease**

Nutritional information and resource links on aspects of nutritional care for people with HD.  
[www.lib.uchicago.edu/~rd13/hd/nutritn.html](http://www.lib.uchicago.edu/~rd13/hd/nutritn.html)

### **Huntington Disease Lighthouse**

Current information on treatments, drugs, support and patient resources.  
<http://hdlighthouse.org>

### **Guide for Healthcare Proxies**

<http://www.abanet.org/aging/toolkit/tool9-lock.pdf>

### **How to select your health care agent or proxy**

<http://www.abanet.org/aging/toolkit/tool11-lock.pdf>