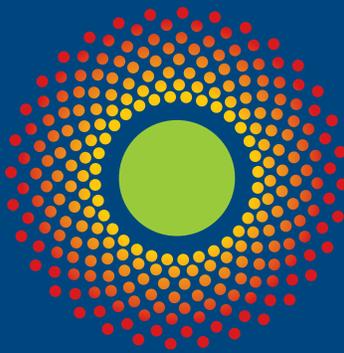


Enroll!

Updates from the Enroll-HD global community



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PORTRAIT OF A CLINIC: COLUMBIA UNIVERSITY

This HD center is one of the top recruiting sites for Enroll-HD. What's their secret?

The HD center at Columbia University Medical Center in upper Manhattan is not glamorous. The big old buildings on 168th Street are gloomy, and the HD clinic itself is modest—just a small waiting room and a handful of cramped exam rooms. But this clinic is one of the highest-recruiting sites for Enroll-HD, with 87 people in the study as of May 31. The reasons for this success are straightforward: Staff who are focused on HD, a wide range of expertise, and a decades-long commitment to the families who come to the clinic. “Part of the reason we’ve been able to recruit so many people is that these are people we’ve been with for a long time,” says neurological nurse practitioner Carol Moskowitz, who has worked with HD families since 1976. “They know that we’re always there for them. They know that if they call my cell phone, I will pick up.”



Neurologist Karen Marder, MD, helped get Columbia's HD clinic off the ground in 1991.

The clinic, formally known as the Columbia University HDSA Center of Excellence at the New York State Psychiatric Institute, is unusual in a number of ways. Two site coordinators work on HD full time, rather than splitting their time with patients who have other movement disorders. Patients are also seen for free, most-

ly because the HD clinic has arranged for free space at the Psychiatric Institute just down the block. That arrangement not only removes a burden from patients, it also adds another layer of privacy since people can be treated without getting an insurance company involved.

The staff also includes a diverse range of medical experts, from speech pathologists to neurologists and psychiatrists. “It’s amazing—they offer everything there,” says Edye, who

has been coming to the Columbia clinic for about eight years. (She asked that her last name not be used to protect her privacy.)

Like her, many people have been coming to this clinic for years—and the staff have come to know multiple generations in the same family. “The key is that we have relationships,” says neurologist Karen Marder, MD, MPH, the clinic director. “Because we’ve had long relationships with families, we’re always active, always seeing a new generation. We know them, so they feel comfortable with us.”

Getting started

In the early 1980s Moskowitz became more involved in HD care after meeting Marjorie Guthrie, a pioneering advocate for HD families. Guthrie would send patients to her for help, and Moskowitz began to realize that HD families needed a dedicated medical team that included expertise in multiple domains. At the time Columbia was conducting world-class genetic research under the leadership of Nancy Wexler, PhD, but there were no dedicated resources for HD patients. In 1991, Moskowitz proposed to behavioral neurologist Richard Mayeux, MD, that Columbia start a clinic devoted to HD. He brought the proposal to the chair of psychiatry, who immediately agreed. Mayeux paired Moskowitz with Marder, arranged for a social worker to be hired, and launched the clinic. All four of them are still on staff.

Since then the clinic has expanded to include two additional neurologists, a psychiatrist specializing in movement disorders, a social worker focused exclusively on HD, a genetic counselor, a speech pathologist, and an occupational therapist who also provides physical therapy. Two coordinators manage the clinic’s day-to-day operations: Ronda Clouse is a research coordinator and Paula Wasserman organizes both research studies and clinical care. A research assistant also helps translate for the 10-15% of the families who come to the clinic for treatment and speak only Spanish.

With this relatively large staff and dedicated space Wasserman and Clouse are able to set up efficient visits for patients and their caregivers, allowing them to see five specialists in a row



Clinical coordinator Paula Wasserman streamlines patients' visits so they can see various specialists in one day: "We try to make things very efficient."

rather than coming back for repeat visits on subsequent days. Specialists can also talk to patients together in what Moskowitz calls "sandwich" visits. "I'll start with a patient, then [occupational therapist] Ash Rao will come in, and I'll sit on the sidelines while he continues the interview," she says. It helps keep all the medical staff working together and means that patients like Edye get everything they need in one visit. "They have great services," she says. By comparison her sister, who lives far away in another state, goes to a clinic where the doctors and social workers don't fully understand the disease and its implications. Services are minimal and she has had difficulty both finding a psychiatrist who knows anything about HD and getting her disability applications approved.

The robust infrastructure at Columbia also makes it easier to recruit large numbers of people into Enroll-HD. "Recruiting is not difficult," says Wasserman, but for many clinics it can be difficult to find the time to schedule the visits required to register new participants. Says Wasserman: "We are able to handle the numbers." Both Edye and her husband have signed up (he as a control).

Wasserman says that most people do want to participate in the study, as do their spouses. "They find it's a good way to contribute to science, a good way to give back," she says. Many were already part of previous studies, such as COHORT or the UHDRS-99, so they're familiar with the tests and evaluations that long-term studies require. But the staff at Columbia is careful to make it clear that getting involved in Enroll-HD is purely optional. Anyone is free to opt out. "If they don't want to participate, regardless of the reason, we fully respect that," says Wasserman. "We'll still love you and take care of you. Any contribution is wonderful."

Practical answers

The core staff at Columbia has about 80 years of collective experience in HD between them, which translates into a wealth of practical knowledge—tricks and tips that are specific to HD and can make families' lives easier. The clinic acts like a clearinghouse for knowledge; the staff learn from patients and families and they pass along that new information to others, says

Moskowitz. Much of it takes the form of down-to-earth advice on living well with HD. She gives a few examples: Don't eat olives because it's too easy to choke. Make sure you get lots of sleep. If you want to go out to a restaurant, eat a snack first and review the menu online at home because you may have to wait.

Some of the knowledge can also be transformative, such as the best way to get a policy from a long-term care insurance company. Just because someone has HD doesn't mean they can't improve their quality of life, says Moskowitz. "When the



Spouses and caregivers often sign up as controls for Enroll-HD, says research coordinator Ronda Clouse: "They think: What can I do to help?"

person leaves the clinic, they've got their diagnosis, but they've also got [a list of] four things they have to do," she says. "Eventually—not necessarily on the first visit, but eventually—they're empowered to change situations so they feel more comfortable."

While the first business of the clinic is treating patients and helping families, they consider studies like Enroll-HD a high priority, says Wasserman: "Everybody on the team helps with recruitment." Anyone who comes to the clinic who is cognitively able to understand the informed consent process is offered the opportunity to become involved, and the staff try to make it as quick and convenient as possible for them to enroll in the study. "It's a tremendous gift when people participate in research," says Marder. "You need to do what works for them."

Because the study involves follow-up visits at least once a year, it can be good for patients as well. "We follow up with people," says Wasserman. "That's what is great about studies like COHORT and Enroll-HD—it's a form of outreach, it's a way to keep people connected to the clinic that might not otherwise stay connected, or for people to come in once a year and deal with HD." Regular yearly follow-ups means that sometimes the clinic staff notice changes or problems that might have otherwise been overlooked. For people who have mild or no symptoms and no ongoing medical issues, the study is a reminder to check in on their health.

Particularly for spouses and caregivers, joining a study can also be a way to do something constructive in the face of a terrible disease. "I like to participate in whatever way I can," says Edye. "This way, hopefully other people can benefit." 

BEV HEIM-MYERS

The head of the Huntington Society of Canada on advocacy, research and Enroll-HD



As Enroll-HD gets up and running in Canada, one of the leaders in the HD community explains how the study will take shape in such a diverse, sparsely-populated nation. Bev Heim-Myers is CEO and executive director of the Huntington Society of Canada, a non-profit that supports research and family services across the country.

How does your organization get involved in research?

Our mission is half research and half family services. We invest in biomedical and clinical research. We also advocate for Canadians living with HD, to enhance services for them, increase the reach to rural communities, and increase social work support. HD takes out generations of a family. What we do is advocate so it doesn't have as much of an impact. We also advocate for genetic fairness. In a nutshell, we increase awareness of HD.

What kind of research are you doing?

Our focus is to enhance clinical research across Canada, and to set the stage for research going forward. That way, if and when a treatment becomes available, we have the clinical sites ready and we have a mechanism to connect them.

We also support research that takes us closer to treatment. We invest about C\$1,000,000 a year. It's almost like seed funding, to take promising hypotheses that could lead to treatments, and bring them to a stage to access bigger money. For example,

Simonetta Sipione at the University of Alberta has recently reversed the effects of HD in a mouse, and brought the mouse back to normal physically. That's exciting research.

You also help the Canadian HD clinics share good ideas—how does that work?

The clinical sites don't necessarily talk to each other, and I see that as our responsibility to make that work. One way is through regular teleconferences.

Or, for example, when the multidisciplinary HD clinic at North York Hospital in Toronto was moving into a clinical trial, they didn't know where to go to get information on the next steps. They connected with the clinic in British Columbia to help them. We're now planning a round table discussion for September to bring HD clinic reps from across Canada to discuss laying the foundation for clinical trials, best practices, and mechanisms to ensure ongoing conversations. I find that in the HD community, we're pretty good at sharing our best practices.

Canada is the second largest nation in the world, but it has only 33 million people. How does that affect what you do?

Part of our strategy is to reach out to individuals in rural areas, to be more accessible. For families, accessing care can be difficult. To fly all the way to Toronto from a rural area for medical treatment just doesn't make sense. Some specialists, like [Toronto neurologist] Mark Guttman, actually travel to the northern areas once or twice a year. In between he can set up some visits via Skype. That model is important.

We've also funded a project for "telehealth" predictive testing in British Columbia that provides access to experts by phone. It can work in a rural area that might have only a nurse practitioner locally. A healthcare professional does the biosample locally, which is then sent in. The results are then discussed over the phone with the expert, but the person being tested also has the healthcare professional there with them—they don't have to go into a clinic. Not only did we find that people in rural areas were interested in that mechanism, but also people in metropolitan areas who didn't want to go into a hospital. I'd like to roll that out across Canada.

Is there anything unique about HD in Canada?

We have a significant native population, and some groups have a high incidence of HD because of the founder effect [see box]. It's up to us to be accessible to them. I have to understand what will entice, and how to build relationships, so people are not left with inaccurate or flawed information. That's true not just of our First Nations communities, but other cultures that may not necessarily come to us. We need to figure out how to make that

happen. One way is to have a more diverse board. We now have a voting member from our youth group on our board, and an elder from a First Nations community as well.

HSC is also focused on helping young people—why is that?

Young people can feel incredibly isolated if they don't have anybody to turn to. It felt important to us for the upcoming generation to have a voice. At our conference in November, we had our first-ever youth day, planned by youth for youth. One young woman said that prior to that meeting the only people she'd ever known with HD were her grandfather, who died of HD, and her father who recently had to be put in a home. She said she was at risk and she'd never been able to talk to anybody about it. It's incredibly important for people like her to have a place to turn to get the right information.

We're now working on a youth peer mentorship program, so that a few young people who have experienced a lot with this disease have been trained and are available to young people coming into the organization. Youth mentors are also paired with a social worker, to help them mentor somebody else. It's really a young person coming to a young person, with a professional to turn to who can answer questions.

In Canada, genetic privacy is not protected by law. Does that make it harder to recruit for Enroll-HD?

Yes, although Enroll-HD does give participants the option to sign up and not know their results.

You don't need to know your genetic status to participate. You can provide your biomaterial and tell the [site staff] that

you don't want to know. If somebody [such as a potential employer] asks if you've had your genome done, you say yes, but the information wasn't disclosed to you. That way, you are protected. We help people get their insurance in a row before they sign up, we coach them. But it is a barrier.

What hopes do you have for Enroll-HD, and what do you tell people about the study?

Families and the community can be involved in this. You can be engaged in the future of treatment.

At this moment, there may not be something in it for the individual. But if you look down the road there may be. Having a longitudinal study with a robust international cohort is really, really important. If we have a huge registry then, when we do have a treatment, we do know who to go to [to test it]. Signing up means answering a lot of questions, but once you're in the system you're in the system. If you move, or you disengage for a while, your information is still there when you re-engage.

When you're faced with a devastating disease, you want to do something. By enrolling in Enroll-HD, you are doing something.

Founder Effect

When many people from a tight-knit community share a common ancestor, the group as a whole tends to be more similar genetically than typical populations of that size. If one of the founders of that community had a genetic disease like HD, it can become very common in that population.

Protecting Canada

Heim-Myers and HSC are pushing to get legislation passed to protect Canadians from genetic discrimination. Canada is currently the only G8 nation that does not protect genetic information, which means that insurance companies and employers are legally permitted to ask about any disease genes you might carry before hiring you or offering you health or life insurance. "That's not fair," says Heim-Myers. "People have enough to worry about."

Heim-Myers is chair of the Canadian Coalition for Genetic Fairness, a coalition of 15 organizations campaigning for policies and laws that would prevent businesses and employers from accessing or requesting genetic information, and restrict its use for health and research purposes only. In April, a non-discrimination bill was introduced before the Senate—the first step. This law would prevent employers from requiring potential employees to get a genetic test or reveal their genetic status. It would keep genetic information private, with some exemptions. It would also amend the Canadian Human Rights Act to make discrimination based on genetic status illegal. At the provincial government level other efforts are underway to change the laws that regulate how much information insurance companies can access. "You shouldn't be fearful of somebody being able to get ahold of that information and discriminating against you," says Heim-Myers.



The Enroll-HD trainers in Buenos Aires.

MASTER CLASS IN ARGENTINA

With Enroll-HD now officially underway in Latin America—the first participant was enrolled in Argentina at the end of May—medical staff from across the region convened at a workshop in Buenos Aires for expert training on how to administer Enroll-HD. More than 35 neurologists, neuropsychologists, site coordinators and others from Argentina, Peru and Chile attended the two-day session in April. It was the first state-of-

WHY IS THIS FORM SO LONG?

How informed consent works

Anyone who signs up for Enroll-HD must first read a consent form that spells out why the study is being done, what it involves, and how privacy will be protected. The form is long and detailed. It explains what personal information will be collected, who is allowed to see it, and how it is de-identified so that your identity (your name and other personal factors) can't be connected to the biological data and test results. The form describes Enroll-HD's cognitive and motor tests, explains that participation is voluntary and that anyone can withdraw at any time, and gives participants the option to be contacted for future drug or therapy trials. All of it must be explained clearly in non-technical language that anyone can understand.

The purpose is to protect the rights and privacy of participants, provide all the information necessary so that potential participants can make an informed decision, and make sure that decision is freely made and not under any pressure (called coercion). "It's really important that people know their rights," says Jamie Levey, chief operating officer of the European Huntington's Disease Network. "All of this takes a lot of explanation," adds Levey, who is also an HD family member who herself participates in research. "You want to keep it simple, but that doesn't necessarily mean shorter."

the-art training workshop for HD in Latin America, says Claudia Perandones, MD, PhD, a clinical geneticist with the Argentinean National Administration of Laboratories and Institutes of Health.

The workshop is designed to standardize the way the study is done: For Enroll-HD to succeed, the tests given to participants must be administered the same way around the world, and the data must also be collected in the same way. The training workshops are also helpful for patients because the clinicians who attend learn the best and most up-to-date information about recognizing and evaluating the symptoms of HD. "They are being trained by the best people in the world," says Perandones. "It's critical not only for research, but also clinically, to make decisions about patients."

For instance, part of the workshop explains how to distinguish between dystonia and chorea, two similar movement disorder diagnoses. Another focus was on identifying suicidal thoughts, which are common in HD. "We have very well-trained physicians now who can detect alarm signs in their patients," says Perandones. "They can make better clinical decisions." A similar training program is planned for Brazil after the World Congress on Huntington's Disease in September in Rio de Janeiro. 

Almost all medical studies require that participants read and sign an informed consent form, to prevent the kind of exploitation that sometimes happened in the past. In the US, the most famous example is the Tuskegee experiment, which began in the 1930s; black men who had syphilis were not told they had the disease, not told why they were being studied, and were not treated even when an effective drug became available in 1947. Informed consent, which spells out the purpose and the process of the study, is one way to ensure that this kind of abuse does not occur.

Today there is no single international authority that dictates what an informed consent form must include. Each nation has its own regulations. Some nations have one central ethics committee that grants a blanket approval, but in the US each university, hospital or research institute has its own committee, called an institutional review board (IRB). Each IRB may require different points to be addressed or have its own requirements for wording. The details of the informed consent form for Enroll-HD must be negotiated with dozens of different IRBs in the US alone.

Because the form is long and complex it's essential anyone interested in becoming part of Enroll-HD have time to read it thoroughly and think it over. The study site coordinator or site investigator should be able to answer any questions. In the end, the goal is for anyone thinking of joining the study to have all the information to make a truly informed decision. 

QUALITY OF LIFE

How Enroll-HD plans to improve health care

Making day-to-day health care better for people with HD is one of the central goals of Enroll-HD. By comparing the hundreds of clinical centers around the world that will be involved in the study, it should be possible to single out the ones that consistently have healthier patients and identify what they are doing right. These “best practices” can then be applied everywhere, improving health care for everyone involved in the study. “We have the opportunity to understand what aspects of treatment work best, and ultimately to encourage practitioners around the world to use these strategies,” says Martha Nance, MD, medical director of the HDSA Center of Excellence at Hennepin County Medical Center, who is the co-chair of Enroll-HD’s Clinical Care Improvement Committee.

To begin the process CHDI held a meeting at its New York office in April to learn from care quality improvement projects in areas like Parkinson’s disease and the inherited lung disease cystic fibrosis (CF). Two common ingredients for success: making data freely available, and getting patients actively involved. The Cystic Fibrosis Foundation, for example, noticed that patients at some CF centers were healthier and lived longer, so they began collecting key measures of patient health such as lung function and nutritional status at every CF center. These numbers are made public, so that each center’s performance can be compared against national benchmarks—strong motivation to provide better care. A quality improvement “toolkit” also teaches CF patients and families how to advocate and collaborate for better local care.

Barbara Vickrey, MD, MPH of the University of California Los Angeles found that simply making sure that clinics follow treatment guidelines can make a big difference. She studied dementia clinics that began following guidelines emphasizing coordinated care and connections with community agencies. For example, dementia clinic staff began talking directly to community agencies that offer services like financial planning advice and home-delivered meals, rather than expecting caregivers to make the appointments. Within a year, these changes improved patient quality of life and the overall quality of care, in comparison to clinics that did not adopt guidelines.

In HD the first step will be to define what success should look like and what should be the best way to measure the quality of life. One project in the US and one in the UK are now testing how to measure health-related quality of life for people on the HD spectrum, potentially including such factors as mood, satisfaction with social activities, cognition, mobility, difficulties with speech and swallowing.

In the next year the committee hopes to partner with patient organizations and experts in quality of care improvement. Potential partners should contact Nance at 952-993-6592 or martha.nance@parknicollet.com. Eventually a system might be set up so that each Enroll-HD site gets a regular report comparing its performance to a global average. The ultimate goal is to help all people with HD live as well as possible, says Nance. “What matters most is the health of the patients.” 

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PEOPLE
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AS OF MAY 31, 2013

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UPDATE: ENROLL-HD AROUND THE WORLD

LATIN AMERICA: The first Latin American participant was enrolled at the end of May in Argentina. Chile is expected to be next to join: One site was discussing ethics approval at the end of May, and could have participants by the end of June or early July. Peru will probably be next, followed by Brazil.

EUROPE: Site investigator meetings have been held for Finland, Italy, Germany and Spain. The first European site is expected to be officially launched in September.

PACIFIC: Two Australian sites may become active as soon as July, following an investigator meeting scheduled for mid-June in Melbourne. 