HORIZON

RESEARCH • SERVICE • EDUCATION No. 160 Winter 2020

Predicting Age of Onset

New Research Shows a Single DNA Letter Makes a Big Difference

By Julie Stauffer

When will I start showing symptoms? For anyone who tests positive for the Huntington disease (HD) gene mutation, it's a burning question. Meanwhile, people who have the reduced penetrance version of the gene mutation wonder *will I develop HD or not?*

A new study from the Hayden Lab at the University of British Columbia (UBC), recently published in the *American Journal of Human Genetics*[1], brings some answers.

When it comes to age of onset, we know CAG count plays a big role. The higher the number of these repeated DNA triplets someone has, the earlier HD is likely to develop. It's not the only factor, however. That's why Jane and Joe can have the same CAG count, but Jane starts showing symptoms at the age of 30 and Joe's symptoms don't appear until his 60s.

So what else is going on? According to Dr. Galen Wright, a research associate in Dr. Michael Hayden's lab at UBC, a subtle difference in the HD gene can make a big impact.

As Dr. Wright explains, DNA serves as a recipe book that tells cells which amino acids to put together to create different proteins. Each three-letter "word" in a recipe corresponds to specific protein building blocks called amino acids. For example, the CAG triplet corresponds to the amino acid glutamine. So does CAA.

In most people who have the HD gene mutation, the string of CAG triplets that causes the disease includes one CAA, just before the final CAG. But in approximately one per cent of persons, that second-last triplet is changed from CAA to CAG. For many years, scientists didn't pay much attention to that difference. After all, in both cases the DNA is instructing the cell to make the same amino acid, glutamine.

However, as Drs. Hayden and Wright (along with their colleagues) recently discovered, having a CAG in that spot means someone is likely to develop HD symptoms earlier than someone with the same CAG count who has a CAA there – sometimes as much as decades earlier.

They also found the second-last triplet makes a big difference for people with the reduced penetrance gene mutation (e.g., people with 36-39 CAG repeats). Persons with this number of CAG repeats may or may not develop HD in their lifetime. When that triplet is CAG rather than CAA, the chances of developing HD in their lifetime go up significantly.

Scientists don't fully understand why this particular triplet is so important, but they suspect it's linked to DNA repair. The cells in our body are constantly exposed to factors that cause damage to our DNA. During the repair process, errors in the DNA can creep in. Without CAA in the second-last spot, it's easier for bits of DNA to get repeated by mistake – and that's especially true in areas of the brain most affected by HD.

There are other factors that also influence age of onset, but the second-last triplet makes a substantial difference. Unfortunately, checking for it is currently a complicated and time-consuming process. Once researchers have developed a faster, simpler method, Drs. Wright and Hayden expect it may become a standard part of genetic testing.

That means people who may have this CAG instead of CAA will have a better prediction of when their HD symptoms are likely to appear. Meanwhile, people with the reduced penetrance gene mutation will get a better sense of their chances of developing the disease in their lifetime.

Dr. Wright foresees even more insights into age of onset in the coming years. "It's quite an active field in HD research at the moment," he says. And what makes this work possible is the blood and brain tissue samples that families affected by HD have donated to Dr. Hayden's HD Biobank at the University of British Columbia over the last 30 years. "Without the gift from the families for research, we wouldn't be able to answer any of these questions," he says.

According to Dr. Galen Wright of the Hayden Lab at UBC, a subtle difference in the HD gene mutation can make a big impact on age of onset.

To learn more about the Huntington disease BioBank at UBC, visit https://cmmt.ubc.ca/facilities-services/centre-for-huntington-disease/dna-and-tissue-bank/.

1. Wright GEB, Collins JA, Kay C, et al. Length of Uninterrupted CAG, Independent of Polyglutamine Size, Results in Increased Somatic Instability, Hastening Onset of Huntington Disease. *American Journal of Human Genetics* 2019.

INSIDE

HORIZON

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Horizon is the newsletter of the Huntington Society of Canada. Published throughout the year, its purpose is to convey information to individuals with Huntington disease and their families, health care professionals, friends and supporters.

Huntington disease (HD) is a hereditary, neurodegenerative illness with physical, cognitive and emotional symptoms. Symptoms vary from person to person and at different stages of the disease but may include involuntary movements and difficulty with focus and thought. Symptoms usually appear between the ages of 35 and 55, and gradually worsen over the 10-20 year course of the disease. But HD can also appear in youth (under 20 years – Juvenile HD) or older adults (Late Onset HD). As yet, there is no meaningful treatment.

The Huntington Society of Canada is a national non-profit charitable organization founded in 1973 to help individuals with Huntington disease and their families.

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Horizon welcomes your comments, ideas and suggestions for future articles (see page 6 for survey link).

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Message from the Board Chair

Welcome to a New Year! The past few months have brought several changes at the Huntington Society of Canada (HSC), so I want to take this opportunity to bring you up to date.

Last October, Robin Markowitz stepped down as chief executive officer (CEO). When Robin joined HSC, her home in Thornhill, ON was listed for sale. More than a year later, it still had not sold, so Robin made the decision to move back and find employment closer to home.

On behalf of HSC's board of directors, I want to thank Robin for her contributions and dedication to the organization over the past 15 months and to wish her well in her future endeavours.

Since her departure, our staff have pulled together to keep everything moving smoothly during this period of transition. Meanwhile, I'm delighted that our former CEO, Bev Heim-Myers, agreed to serve as interim CEO while we search for our new leader. With Bev now in place, we can take our time to find the right leader to guide HSC through our next phase of evolution.

At the board level, we have some new faces around the table. Thank you to outgoing directors Bob Scriven, Jonathan Genest-Jourdain, Doris Ramphos and Christian Lejeune for their valued contributions and welcome to incoming members Cameron Barrett, Geneviève Bélanger and Diane Tullson. Also, thank you to outgoing chair, Brenda Nowakowski, for leading the board so capably. I have big shoes to fill!

The final big piece of news is that HSC has a new office. When we received notice that our old building was being sold – and probably demolished – we knew it was time to move. Our new location in Waterloo, ON is just steps from the uptown area and its new light-rail (ION) station, creating more accessibility for volunteers, interns and the local Huntington disease (HD) community.

As you will read in the pages that follow, HSC achieved much in 2019. Our redesigned Community Education Forums proved very successful, with more than 20 events hosted across the country and a virtual forum that attracted participants both nationally and internationally. We are very grateful to the generous sponsors who made it all possible: Roche, uniQure, Vaccinex and Wave Life Sciences.

Roche also helped fund one of our first two clinical fellowships: Dr. Ragini Srinivasan in Toronto, ON. The second clinical fellowship, Dr. Fabricio Pio in Vancouver, BC was generously funded by the Manning Family and Beckman Family Fellowship. Now, we're looking to support one or two more young neurologists this year, continuing to train the next generation of HD clinical experts.

Speaking of the next generation, we held our biggest YPAHD Day yet in November. Last year's event - sponsored by TD Securities, SickKids Foundation and Kindred Home Care - drew 78 young people affected by HD for learning, fun and community-building in Calgary, AB, Toronto, ON and Halifax, NS.

Looking ahead, there are many exciting things on the horizon. Once we have a new CEO in place, we will be rolling out a new five-year strategic plan to guide us as we expand services for families, support clinical trials and advocate on key issues for the HD community.

Of course, amidst all these changes, some things stay the same. At our core, HSC continues to be a passionate group of Canadians working hard to help families affected by HD, which is why you will find Ralph Walker's favourite bench at the new office, reminding us of our roots as an organization.

MACK ERNO
Board Chair, Huntington Society of Canada

Message from the Interim CEO

Once the Huntington cause gets into your heart, it doesn't leave. So, when Mack called and asked if I could serve as interim CEO, saying "yes" was a very easy decision. I feel honoured to be invited back, and privileged to work with such a caring community and competent team once again.

While the Board seeks out a new CEO, we have lots to move forward with. That includes organizing the National Conference in Niagara Falls, ON. We are putting together an incredible lineup of speakers, and we expect this to be HSC's biggest conference ever. I warmly invite you to join us and hear about all the latest research developments. Stay tuned for more details but mark your calendars for Nov. 13 and 14, 2020 (with YPAHD Day on Nov. 12). I hope to see you there!

Bev Heim-Myers
Interim CEO, Huntington Society of Canada

Mack Erno in the Saddle as New Board Chair

By Josh Martin

Giddy-up! That about sums up go-getter Mack Erno, the new chair of the Huntington Society of Canada (HSC) national board of directors. In the morning, the rancher from Teepee Creek, AB could be roping cattle. By noon, you might find him working at his day job running an ATM business, or at his latest venture, a livestock feed operation. Come the weekend, he's shuttling his kids to 4-H club, piano lessons or football practice.

Then there's everything he does to support HSC. Mack's Huntington disease (HD) journey began in high school, when he started dating a girl named Amanda, whose mother had the disease. Eager to learn more, Mack did a science project about HD. And while we're not sure what grade he got, he ended up marrying Amanda.

Since then, his commitment to the cause has only grown stronger. In 2009, he and Amanda hopped in their truck and drove 15 hours to attend the *World Congress on Huntington Disease* in Vancouver, BC (where they first met Ariel Walker, HSC's co-founder). The event inspired them to take action. "We're not scientists. Far from it," he says. "But we asked, 'what can we do to get involved?'"

After returning home, Mack led a team of volunteers who founded HSC's Peace Country Chapter and organized a horseback trail ride as its first fundraiser. It poured rain on the big day, but Mack didn't let that stop him. "Hell or high water, we're going to ride," he told the folks who were calling him to see if it was still happening. Ten years on, the annual *HD Ride 4 a Cure* has raised more than $750,000 for HSC. "We're pretty proud of it," says Mack.

While out on one of those trail rides, HSC CEO, Bev Heim-Myers, suggested to Mack he should consider joining the national board of directors. In 2013, he did just that, bringing his business background and can-do attitude to the group.

Mack and Amanda divorced a few years ago, but his commitment to the HD cause is stronger than ever. Last September, Mack took on the role of chair of the board. With HSC's new five-year strategic plan ready to roll out later this year, he's keen to leverage the skills and connections of his fellow board members to serve the HD community. "We have a phenomenal set of people around the table," he says. "We want to do more of that good work and find ways to accomplish more. It's all about making that difference."

HSC's new board chair, Mack Erno, also runs a highly successful annual trail ride which has generated more than $750,000 for the organization in the last decade.

An Ariel View

Ariel Walker,
Co-Founder of HSC

Happy 2020! In honour of the new decade and the strategic vision the Huntington Society of Canada (HSC) will roll out in 2020, the *Horizon* editorial team suggested interviewing me, using some of Barbara Walter's favourite questions from the 20/20 television show as inspiration. So here goes…!

Do you have a philosophy of life?

I live each day as it comes. My coffee cup is always half full - I don't see the negative side of things. I just enjoy each minute and each hour of the day. I try to keep family and friends very close to me, and I try to help people. That's what life is all about.

What about Ralph?

He was very similar. His cup was always half full.

Complete this sentence: The Huntington Society of Canada is…

…the best little organization in the world!

When Ralph decided to step down from HSC, what did he miss the most?

That was a big decision. He missed the families and the scientists, but he knew if HSC was going to grow, it needed someone who could bring new ideas. If you have a dream, you must act on it. But you also have to know when to let that dream continue with someone else.

HSC's National Conference is going to be in Niagara Falls, ON this year. What kind of memories does that bring back?

We hosted our first international meeting there in 1983. There was such great camaraderie with people from all the different countries. I remember we were all having dinner in the revolving restaurant and a full moon came up over the falls. It was beautiful. Ralph spread his arms and said, "We arranged it all just for you!"

Then there was HSC's National Conference in Niagara Falls, ON the year after Ralph died. That was a real tear-jerker, but I'm very excited to be going back.

Did you know Ralph's old bench is welcoming visitors at HSC's new office?

Oh, good! I haven't seen the new space yet, but I've heard all about it. We've come a long way since Ralph shared an office with Mr. Blackwell up those steep stairs on Water Street.

Is the best yet to come?

For HSC, yes. The research that's going on right now is mind-blowing. For me personally, I don't know. Life is pretty good at the moment!

Q …I have a loved one with Huntington disease (HD) and we'll be taking a trip soon. What do I need to know?

A If travelling is new for your family, starting out with a shorter trip may be a good idea to get accustomed to travel before going on a long journey.

Because people with HD often have difficulty regulating their body temperature, it's important to consider the weather for the time of the year that you plan to travel. It's also advisable not to travel in high tourist season so as not to overwhelm the person with HD. If possible, request quiet and safe spaces for the person with HD to relax and re-group.

Before the trip, include everyone in the planning of where you will go and what you will see and do. Once the trip is planned, make an itinerary available so the trip details can be reviewed whenever necessary. Learn as much as you can about the place you are visiting, so you can anticipate what you will need for the trip. Consult with your health care team before your trip about your travel plans and ask about contingency plans and strategies that could be helpful in an emergency. Ask your doctor about safe medications to help with controlling nausea and other travel-related illness. If you are planning air travel, carry 4-5 days of medications in your carry-on baggage just in case your luggage is lost.

The best way to start is to contact your HSC Family Services worker (visit www.huntingtonsociety.ca/family-services-team-list/ for contact information). Together, you can discuss your specific needs and zero in on concrete options that are right for you.

Got a question you'd like to ask Dear HSC? Email it to us at communications@huntingtonsociety.ca

Helping Brain Cells Take Out the Trash

By Julie Stauffer

Dr. Dale Martin first became intrigued by the field of Huntington disease (HD) research as a PhD student at the University of Alberta. He was investigating how the normal huntingtin protein plays a role in something called autophagy – "just a fancy word that means self-eating," Dr. Martin explains. "The cell uses the process to remove toxic proteins and damaged parts of the cell and recycles them."

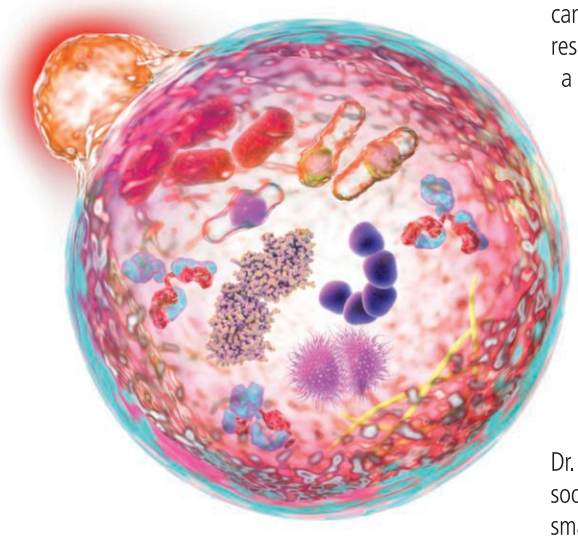
Normally, bubble-like structures called autophagosomes move around brain cells, collecting trash and taking it to lysosomes, which digest the garbage. The normal huntingtin protein seems to play a role in loading garbage into the autophagosomes and moving them around the cell.

It turns out that the mutant version of the huntingtin protein can't carry out this important junk-clearing job. When scientists look at the cells of people with HD, instead of seeing autophagosomes filled with garbage ready to be degraded, some of those autophagosomes are empty.

Dr. Martin describes this as a double whammy for the cell. Because mutant huntingtin can't clear away the toxic proteins, they start to build up and affect the brain cell. On top of that, the mutant huntingtin itself is toxic.

So, what's the solution?

Fortunately, cells have ways to trigger autophagy that don't require huntingtin. As a postdoctoral fellow in Dr. Michael Hayden's lab at the University of British Columbia (UBC), Dr. Martin was investigating how to switch on those alternative pathways in mice with HD and prevent toxic protein from building up in their brain cells.



Now he's continuing that work as a newly-minted professor at Ontario's University of Waterloo. One of the things he's doing is scrutinizing drugs that have already been approved for other diseases to see if they can promote autophagy.

Dr. Martin believes this research will also create insights into ALS, Alzheimer disease and Parkinson disease – all conditions where autophagy seems to be affected. "We're trying to use some of the results that we're finding in HD and see if we can apply them to some of the other diseases," he says. "I think there's a lot of promise there."

He is also continuing other exciting research he began at UBC: finding ways to deliver huntingtin-lowering drugs intravenously, so there is no need for lumbar punctures.

Injecting a drug into a patient's vein is a lot easier and less invasive than injecting it into their spine. However, the big challenge is finding a way to get the drugs across the blood-brain barrier that separates brain cells from the bloodstream.

This barrier acts like a heavy-duty security wall with checkpoints that allow only certain things to cross into the brain. Sugar, oxygen and a few other key molecules can pass through. Just about anything else, no.

That's why the current clinical trials for Roche's RG6042 drug and Wave Life Sciences' WVE 120101 and 120102 drugs involve injecting them into the cerebrospinal fluid next to the spine. This fluid surrounds the brain and the spinal cord. It naturally circulates, moving the drugs from the spine up to the brain where they can do their job.

HSC and Brain Canada are funding research into ways to package huntingtin-lowering drugs so they can slip across the blood-brain barrier instead – research that Dr. Martin was involved in. "There was a lot of progress there, and so this is something that I want to take forward," he says.

When Dr. Martin established his own lab in Waterloo, ON, he was inspired by Dr. Hayden's multidisciplinary approach, where neuroscientists, geneticists, cell biologists and bioinformaticians* work closely together. Currently, several undergrad students and a Master's student are busy working in his lab packed with fluorescent microscopes, cell cultures and gel imagers.

Dr. Martin plans to hire more graduate students soon, but he wants to keep his research group small enough that he can give his trainees plenty of attention. Meanwhile, his partner Dr. Shaun Sanders – a neuroscientist he met in Dr. Hayden's lab – will be taking up a position at the nearby University of Guelph this June, and the two plan to collaborate on HD projects.

Dr. Dale Martin of the University of Waterloo is scrutinizing drugs that have already been approved for other diseases to see if they can promote autophagy.

Since moving to the Kitchener-Waterloo area in 2018, Dr. Martin has been busy connecting with the HD community here. He has met staff at the national office and attended the Grand River chapter's Christmas party. He has also taken part in fundraising initiatives: a "Pins and Pizza" bowling event and the annual Grand River Chapter's Founders' Walk. Last fall, he also opened his lab for a tour and hosted the Community Education Forum for the region.

Meeting families is very inspiring, he says, and he is constantly impressed by their eagerness to understand the latest research. "The HD community is so well educated in what's going on," he adds. "Their questions were fantastic."

When he repeats the tour this Fall, Dr. Martin hopes to be able to show off a bigger team, a new cell culture area and more exciting research results.

**A bioinformatician combines research in biology, medicine, and health-related studies with information technology in order to collect and interpret data covering a range of fields, such as genetics or pharmaceutics.*

Community Education Forums Feed Appetite for Knowledge

By Josh Martin

Last year's Community Education Forums (CEFs) marked a big change for Huntington Society of Canada (HSC) outreach efforts. Instead of broadcasting a live symposium about the latest Huntington disease (HD) research, staff pre-recorded the keynote addresses and gave chapters the freedom to pick dates that worked best for them.

HSC set the bar high, aiming for 20 events and 300 participants with this new format. So, how did the organization do? "We blew that out of the water," says Annie Vanexem, HSC's Senior Manager, National Events and Chapter Development. All told, 23 communities from coast to coast hosted CEFs in 2019, attracting 490 participants.

The feedback shows that chapters loved the flexible timing. Meanwhile, the video presentations by Drs. Ed Wild and Michael Hayden provided plenty of "a-ha" moments about clinical trials and what to expect from them. The variety of local speakers at the events – including researchers, neurologists, youth mentors, nutritionists, self-care experts and more – were a big hit as well. Some people even attended multiple forums so they could hear from different speakers.

Recognizing that not everyone can physically attend a local forum, or may not have one near them, HSC also hosted a virtual CEF in early November. The virtual CEF featured Dr. George Yohrling, Senior Director of Mission and Scientific Affairs with the Huntington Disease Society of America. During his presentation, he provided insights into how clinical trials around the world are going and what's in the pipeline. Online polls and other interactive tools allowed participants to get involved and ask questions and it all went off without a hitch, thanks to tech support enlisted from the webinar platform provider.

The virtual event attracted viewers from across Canada, as well as the United States, Mexico, Argentina and Italy. "It's really great to see that a simple webinar initiative from HSC is having an international impact and drawing attention from around the world," says Annie.

The success of all the CEFs points to the HD community's appetite for research updates. Having access to that information is crucial, especially right now when there's so much happening with clinical trials. Moving ahead, HSC will continue looking for new and innovative ways to make it as readily available as possible – whether it's through webinars or events like the Community Education Forums.

Thank you again to the CEF sponsors – Roche, uniQure, Vaccinex and Wave Life Sciences – for making these events possible!

Missed the CEFs? Look for the video presentations and the entire virtual forum on our YouTube channel at www.youtube.com/HuntSocCanada.

At HSC, we are always working to ensure that our programs, services and resources are continuing to benefit our community. Times change and sometimes the way things are done needs to as well!

Currently, two surveys are open and we'd like your feedback.

The first is on *Horizon* and our Community Newsletters with the aim to provide feedback to our communications team about areas for improvement going forward. The second survey will enable the chapter development/events team to learn more about the motivations of our community and how our Chapters can serve our community better.

Links to these surveys on SurveyMonkey can be accessed at https://www.huntingtonsociety.ca/tell-us-what-you-think.

Both surveys will be open until Friday February 28, 2020 at 5 p.m.

We look forward to hearing from you!

2020 Winter Events Calendar: Coming to Your Neighbourhood Soon!

For a full listing of events near you, visit www.huntingtonsociety.ca/events.

NATIONAL EVENTS

Throughout 2020

RaceHD
Canada-Wide

COMMUNITY EVENTS

Sun. Dec. 1, 2019 – Fri. Feb. 28, 2020

Toronto Sports Getaway Raffle
Ontario-Wide
www.hscevents.ca/SportsGetawayRaffle

Thurs. Jan. 30

Strumbellas Concert Awareness Event
Kitchener, ON
events@huntingtonsociety.ca

Sat. Feb. 22 – Sun. Feb. 23

HD500
Lockport, MB
https://p2p.onecause.com/hd500

Sat. Feb. 29

4th Annual Niagara Benefit Concert
Niagara Falls, ON
https://tockify.com/senecaqueentheatre/detail/187/1583024400000

Sat. Mar. 7

Okanagan Dance
Okanagan, BC
Email events@huntingtonsociety.ca for information.

Sat. Mar. 14

Sarnia Pot O'Bowl
Sarnia, ON
https://p2p.onecause.com/sarniabowl

Sun. Mar. 15

Achilles St. Patrick's Day 5k Run
Toronto, ON
http://p2p.onecause.com/raceHD

Thurs. Mar.26

Toronto Comedy Night
Toronto, ON
Email events@huntingtonsociety.ca for information.

Fri. Apr. 24

Wawota Walk
Wawota, SK
Email events@huntingtonsociety.ca for information.

Equipping Alberta Families with Tools for Resilience

By Josh Martin

As a resource centre director, Bernie Modrovsky supports families throughout northern Alberta. Her counterpart, Shelley Thiele, does the same in the southern part of the province. However, the people they serve rarely get to interact with each other.

To bring Chapters and families across Alberta together, Bernie and Shelley organized "Tools for Resilience." Held in September at the public library in Red Deer, AB - between Calgary, AB and Edmonton, AB - the event drew 30 participants, including individuals at risk for HD, those at various stages of HD, family members and carers.

The afternoon provided attendees with great opportunities to socialize, share strategies and resources, learn what other Chapters were up to and find comfort in knowing they are not alone. "There was warmth, there was laughter, there was just a sense of belonging," says Shelley.

A diverse panel of speakers discussed personal directives, trusteeship, homecare and long-term care options, mentorship services for youth and more. Meanwhile, keynote speaker, Kim Anderson, reminded participants about the importance of self-care, taking control of your life and not letting your disease define you.

A model of resilience, the world champion para-triathlete recounted how she has persevered through several crises – including suffering injuries when she was hit by a drunk driver and being diagnosed with HD.

In the end, the participants left feeling better informed, connected and uplifted. "We wanted to really take a positive look at things," says Bernie. "We wanted to be inspirational."

A big thanks to Tara Johnson-Ouellette and Stan Weber, whose generous donations made this event possible!

The agenda at the "Tools for Resilience" event in Red Deer, AB last September was jam packed!

YPAHD Column

By Doug Mallock, YPAHD President

Young People Affected by Huntington Disease (YPAHD) Day 2019 is in the books as our biggest get-together ever! Last November, 78 young people from British Columbia to Newfoundland came out to our three events across the country. We had everyone from teens to "old-timers" like me, including a lot of new people.

The weekend kicked off with a Friday night social activity (just saying, the dragonflies from the Halifax Paint Nite were much better than Toronto's).

The next day, we had workshops on everything from genetic testing to fundraising to family planning. As always, people loved connecting with each other and left really energized. Events like these certainly refuel my batteries.

Thanks to TD Securities, the SickKids Foundation and Kindred Home Care, we were able to give out a record amount of travel funding. YPAHD Day makes an impact in a lot of young people's lives, and it wouldn't be possible without these phenomenal sponsors.

Now, our executive team is busy pulling together all the details for YPAHD Day 2020, which will happen on November 12, 2020 just before HSC's National Conference (November 13-14, 2020) . We can't wait to see everyone in Niagara Falls, ON – it's going to be our biggest and best event to date!

However, YPAHD Day isn't the only way to connect. Last summer, I volunteered at the Huntington's Disease Youth Organization's (HDYO) Camp in California, USA. It was my fifth one, and every summer, I come away amazed by the strength of the young people throughout North America.

Meanwhile, the HDYO first-ever International Young Adults Congress is happening in Scotland this May. Picture YPAHD Day, but on a global scale. It's open to anyone 18 – 35 who is impacted by HD. Check it out at www.hdyocongress.org.

So, pick a way to get involved. Once you've made that first step, you'll never look back. Email us at ypahd@huntingtonsociety.ca, and let's connect!

Again, thank you to our 2019 YPAHD Day Sponsors:

kindred home care

SickKids FOUNDATION

TD

Youth Mentorship Comes Full Circle

By Josh Martin

When Erin Stephen welcomed the seven new recruits to the Huntington Society of Canada (HSC) Youth Mentorship training session last September, she was delighted to see that two of them were former mentees. "They got the support, and now they are in such a good place that they want to give that support," the Saskatchewan resource centre director and youth mentorship coordinator says.

To learn more about the Youth Mentorship Program, visit www.huntingtonsociety.ca/youth-mentorship-program.

Since HSC launched the program in 2013, the organization has conducted five rounds of training and now has mentors in almost every province. Today, 32 mentors are helping their younger peers face the day-to-day challenges of growing up in a family affected by Huntington disease (HD). Sometimes that means checking in with a friendly text. Other times it's lending a listening ear over the phone or connecting face-to-face at a Young People Affected by Huntington Disease (YPAHD) event.

Interest in the program continues to grow, with 30 mentees signed up across the country. However, some parents still hesitate to get their children involved. "[Parents] want them just to be kids and not have to worry about this stuff," says Corey Janke, HSC's national social worker, who co-led the training with Erin. "However, they see and hear the changes happening in the family and therefore they are worried. Let's give them a place to talk about that worry."

Erin and Corey work hard to match mentees with older peers who share similar life experiences. If someone is interested in genetic testing, they'll look for a mentor who's been through that process. If the applicant is a young carer, they'll find a match who knows what that's like. And it's not just for teens. Young people in their 20s grapple with lots of issues, so Erin and Corey will match them with mentors in their 30s.

The steady growth of this internationally-recognized program points to its success, as do the testimonials we receive from participants and parents. But the biggest sign, according to Erin, is seeing former mentees step up to organize fundraisers, contribute to local chapters, get involved in YPAHD – or become a mentors themselves. "It's full circle," she says.

YPAHD Day Another Huge Success!

By Kelsey Laidlaw, HSC Community Events Assistant

Huntington disease (HD) comes with many challenges for those who are affected by it, especially youth. That's why the Huntington Society of Canada (HSC) created a youth chapter a few years ago to provide a community and support network for Young People Affected by HD (YPAHD).

"The biggest issue youth face, is feelings of isolation. They feel as though their peers do not understand what they are going through and they feel very alone," says Erin Stephen, HSC's Saskatchewan resource director and coordinator of the HSC Youth Mentorship Program. "YPAHD Day provides an opportunity for youth to connect with others who have similar experiences and provides them with the information and support they crave."

YPAHD Day is a one-day conference that brings youth together at three sites across the country to help them connect with their peers.

"The atmosphere was very supportive," says Josh, a first time YPAHD Day attendee from Toronto, ON. "I was able to hear stories from others who understand what I'm going through and I feel like I'm less alone in facing HD."

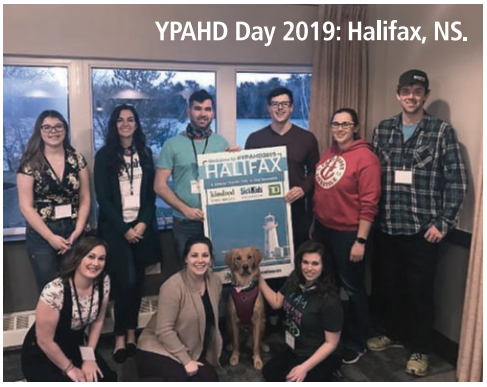
YPAHD Day 2019: Toronto, ON

YPAHD Day 2019: Calgary, AB

Not only does YPAHD Day provide a chance for youth to engage with each other, it also provides an opportunity to learn about HD research and coping strategies to combat the challenges that come along with HD. Youth can participate in a variety of sessions with topics including genetic testing, clinical trials, family planning, guilt, care strategies and more.

"YPAHD Day gave a face to HD which can sometimes feel cold and clinical," adds Emily from Calgary, AB. "I feel more knowledgeable and empowered to use the resources available to me."

The need for YPAHD Day is evident as it continues to grow year after year and reaches new individuals to provide them with opportunities for learning and connection. In 2019, 78 people attended, a 12 per cent increase from 2017. Of these attendees, 35 per cent were attending for the very first time. None of this would be possible if it were not for generous sponsors: Kindred Home Care, SickKids Foundation and TD Securities. The support of these partners helped



YPAHD Day 2019: Halifax, NS.

to fund more than 85 per cent of registrants, who would not have been able to attend otherwise. Their contribution is critical in keeping YPAHD Day accessible for youth.

YPAHD Day is much more than a single day conference. It is an opportunity to learn important strategies and create lifelong friendships that demonstrate to youth that they are not alone. Though they cannot always be together physically, YPAHD day reminds youth that they are supporting each other in spirit.

Making Families the Old-Fashioned Way

The Choice to Conceive Naturally

By Julie Stauffer

When Mack and Amanda started dating, Amanda's Mom had been diagnosed with Huntington disease (HD). They knew Amanda had a 50/50 chance of carrying the same gene mutation that was slowly taking her Mom. So, after they got married and were ready to start a family, Amanda took the genetic test. It came back positive.

It was a serious blow, but it didn't stop her from pursuing her dreams – including her dreams of having children.

Initially, Mack and Amanda tried in vitro fertilization with pre-implantation genetic diagnosis (IVF with PGD). The first round produced five embryos that carried the HD gene mutation and only one without. They implanted that one, but the pregnancy wasn't successful.

In the second round, they ended up with only two viable embryos. One had the HD gene mutation. The other was inconclusive. Today, PGD involves removing several cells from an embryo and testing them for the HD gene mutation. However, when Mack and Amanda went through the process in 2011, clinicians took only a single cell, which meant they weren't always able to get clear genetic results.

The news threw Mack and Amanda for a loop. "Inconclusive" wasn't a result they had prepared for. Sitting in a hotel room near the clinic, 1,400 kilometres from home, they had only a few hours to make a choice. Should they try implanting the inconclusive one and hope it didn't carry the HD gene mutation? Or should they invest more time and money on another physically and emotionally exhausting round with the same chance of failure? As the clocked ticked down, they did a lot of soul searching and made a lot of calls to family.

What helped them come to a decision was a conversation with Amanda's father. Amanda's mother was in long-term care at that point, and she and her sister had both inherited the HD gene mutation. "You know," he told them, "I have two beautiful daughters and I wouldn't change a thing."

"In the end, we did decide to implant the embryo," Mack says. "We made that decision to roll the dice and have that family." Once again, however, the pregnancy wasn't successful.

At that point, it wasn't a big leap to decide to take that same 50/50 chance, but this time without the intervention of doctors and labs.

They did consider other options, Mack says. They discussed the possibility of adoption, but (because this was before the Genetic Non-Discrimination Act became law) Amanda's genetic status would have weighed against them. They talked about conceiving naturally and then doing in-utero genetic testing, terminating the pregnancy if the fetus had the HD gene mutation. However, as Amanda pointed out, "I'm here, and I've had a great life and I'm sure glad that nobody terminated me."

In the end, they chose to take their chances and conceive naturally. Claire was born in 2011, a few short months before Amanda's mom passed away. Claire's brother, Tyson, came along 19 months later. Today, Claire is a busy little girl who loves to read and write, tell stories and draw pictures. Tyson is a rough-and-tumble, happy-go-lucky little guy. And neither Mack nor Amanda can imagine life without them.

In recent years, Amanda began showing HD symptoms. Of course, they wonder if Claire and Tyson carry the HD gene mutation. But when Mack looks at his kids, he's optimistic about what their future holds.

"I believe quite strongly that by the time our kids have to worry about whether they have the HD gene mutation or not, there will be a treatment," he says. With each new clinical trial launched, his conviction grows. And, as Amanda has pointed out many times, having the HD gene mutation doesn't stop you from having a good life.

Mack and his former wife, Amanda, conceived Claire and Tyson naturally after much consideration of their options.

"We don't regret going through the IVF with PGD process. I mean, we wished the results would have been different, but we don't regret it," Mack says. Nor does he regret conceiving Claire and Tyson the natural way.

When the *Globe and Mail* posted a video about their journey, some viewers criticized the choices Mack and Amanda made. Mack shrugs that off. "It's always easier when you're not in someone's shoes," he says.

Friends and family were supportive, and Mack and Amanda don't have to look too far to see the benefits of their decision. "We're extremely happy with our family and love them very much," he says. "We wouldn't change a thing."

Ultimately, he says, every couple must make their own choices – and he acknowledges IVF with PGD has likely improved since their experience ten years ago. "Just explore all the options available to you, and go in with realistic expectations," he suggests.

This is the final installment in a three-part series about family planning and HD. For more family planning information, contact your local Family Services team member (visit www.huntingtonsociety.ca/familyservices-team-list/ for contact information).

HealthPartners Launches E-Newsletter

By Eileen Dooley
CEO, HealthPartners Canada

At some point in their lives, 87 per cent of Canadians are likely to be affected by a chronic disease. Donations to HealthPartners directly help 16 of Canada's most trusted health charities – including the Huntington Society of Canada (HSC) – which all work to prevent chronic disease and to care for those affected by it.

Together with our member charities and partners, we are building healthier workplaces, healthier Canadians and healthier charities. One way we do this is by researching and packaging health information and sharing it in workplaces, on our website (www.healthpartners.ca), on our social media accounts – and now, we are excited to announce the launch of our new email newsletter, the HealthPartners e-News!

Our goal with the HealthPartners e-News is to create a trustworthy and informative newsletter that will give you news about healthy living, disease prevention and the latest research developments on the treatment and cure of chronic disease. We hope you'll subscribe at http://eepurl.com/gJzT3j and find something that will be of interest to you and help you improve your health and well-being.

Last year was a very eventful year for everyone within the Huntington disease community with several promising drugs currently undergoing clinical trial, and even more being planned in 2020. HealthPartners is proud to have contributed to the success of this research through our over 30-year partnership with HSC. We are very much looking forward to seeing what advances may arrive in coming months – and, in the meantime, we wish you a wonderful year ahead.

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Charities At Work

Genetic Non Discrimination Act Update

By Bev Heim-Myers
Chair, Canadian Coalition for Genetic Fairness

The Canadian Coalition for Genetic Fairness (CCGF) comprises a group of organizations dedicated to preventing genetic discrimination against individuals, based on their genetic test information. CCGF advocates within governments (at the federal, provincial and territorial levels) to create positive changes for the Huntington disease (HD) community and all Canadians. Huntington Society of Canada (HSC) and the HD community have been strong supporters of this issue.

In May 2017, the Genetic Non-Discrimination Act (GNDA) received Royal Assent and was passed into law. Prior to that date, genetic test information of Canadians was not protected. Parliament enacted the GNDA pursuant to its criminal law power as a response to expert evidence that showed some Canadians avoided taking genetic tests, despite knowing about the potential health benefits. The GNDA empowers all people living in Canada with the chance to make informed decisions regarding health and reproduction, without fear of genetic discrimination.

In December 2018, the Cour d'appel du Québec (Québec Court of Appeal) gave its opinion that the GNDA does not constitute a valid exercise of Parliament's criminal law power. The Québec opinion did not overturn the GNDA, but did put the GNDA at risk.

In response to the opinion of the Québec Court of Appeal, CCGF filed a notice of appeal, referring the case to the Supreme Court of Canada (SCC). The appeal was accepted in January, 2019 and this officially made CCGF an appellant at such time when the case would be heard at the SCC.

The SCC hearing took place on Oct. 10, 2019 in Ottawa, ON. I was fortunate enough to attend and was joined by Senator Jim Cowan (ret.), and his former Senior Policy Advisor, Barbara Kagedan, who continue to remain very engaged with this file and whose historical knowledge and guidance is critical moving forward. Of course, the CCGF legal team also attended and, as you would expect, our lawyers did an excellent job in presenting CCGF's perspective.

The GNDA still remains as federal law. Genetic test information remains protected for people living in Canada. As soon as we are advised of the opinion of the SCC (sometime in 2020), we will be sure to update you.

CCGF thanks our team of lawyers, Senator Cowan and Barbara Kagedan for their continuing hard work on this file, on behalf of all people living in Canada.

Champion of Hope: Sharon Haig

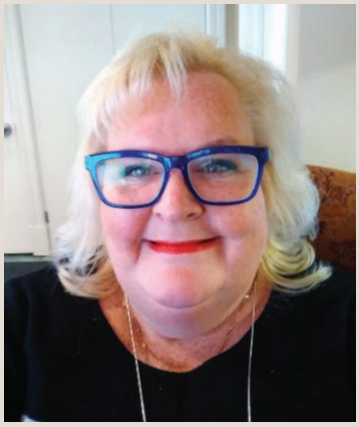
By Josh Martin

Sharon Haig is a passionate advocate for families with Huntington disease (HD) and their carers. Through her time as a volunteer for the Huntington Society of Canada (HSC), and as president of HSC's Ottawa Chapter over the past three years, she has delivered presentations to hospital CEOs, senior medical staff of medical organizations throughout the Champlain Local Health Integration Network (LHIN), Members of Parliament and other community stakeholders. She serves on the Champlain LHIN Patient and Family Advisory Council, where she regularly conveys the complex medical and psychosocial needs and long medical journey of people affected by HD to further educate those working with HD patients and carers alike.

Sharon understands that clinical research, support groups and other important services for families affected by HD depend on fundraising. Over the years, she's completed four 5K walks for HSC, as well as a challenging 10K version. "I was very proud of myself," she says, adding that walking on behalf of those who are not able is a powerful motivator.

Meanwhile, since getting involved with the Chapter's premiere event, the Race for the Cure, Sharon has helped revenues grow from $13,000 the first year to more than $42,000 eight years later. She has also encouraged more team members and walkers for the Tamarack Ottawa Race Weekend, raising many thousands of dollars, and supporting Amaryllis sales. "It's not just me," she's quick to insist, referring to her "small but mighty" team of volunteers at the Ottawa Chapter.

Sharon digs into her own pocket regularly to support HSC. To ensure she doesn't forget to make those contributions, she decided to become a monthly donor in early 2019. As a Champion of Hope, Sharon's automatic donations help provide a stable funding base that HSC can count on.



"It's a fast and easy way to provide HSC with very important donor dollars," she says. "You just put in your credit card number and away you go."

Find out more about how you could become a monthly donor at www.huntingtonsociety.ca/monthly-giving.

Beryl Pitfield: A Spirit of Generosity

By Julie Stauffer

When Beryl Pitfield died in 2018 at the age of 95, the Huntington disease (HD) community lost a generous donor, while friends and family lost a lovely soul.

Photo: Maddalena Costa

Through friendship with Marguerite Evans (L), Beryl Pitfield (R) became a stout supporter of HSC.

Beryl's life didn't get off to an easy start. When Beryl was two, her mother had a stroke and died a few years later. Beryl was sent to live with two maiden aunts, while her oldest sister left school to look after her other siblings.

However, Beryl always focused on her blessings: her aunties, the weekly visits from her father, the opportunity to take piano lessons and summers spent at the cottage with her cousins. "She really didn't have a lot but was grateful for whatever she had," says Maddalena Costa, a long-time neighbour and friend.

Church was a cornerstone of her life. She attended St. Paul's Anglican Church in Toronto, ON until her 90s, donning her best hat for Sunday service, taking Bible study during the week and putting her sewing and knitting talents to work for church causes.

Beryl never married, continuing to live with her aunties. When they got older and needed support, she quit her job at an insurance company to care for them. That spirit of generosity defined Beryl. "Sweet and kind," is how her niece, Chris Kovac, remembers her.

Beryl's connection to HSC came through Marguerite Evans, who married one of Beryl's relatives. Marguerite's husband died from HD and five of their children developed the disease. Whenever Marguerite raised money for HSC, Beryl was always one of her biggest contributors.

When Beryl passed away in 2018, she left a large gift to HSC in her will – just one of several causes that benefited from her generous spirit. "She didn't do it for any glory," says friend Rosemary Broos. "She was just a very giving person."

Nephew Randall Masales agrees. "It was important to her that she share what she had," he says.

For that, HSC is incredibly grateful.

If you'd like to leave a legacy to the Huntington Society of Canada, we'd be happy to discuss the options. Call Ian Foss, national director of development at the HSC office: 1-800-998-7398, ext. 125 or ifoss@huntingtonsociety.ca.

Thank you

On behalf of families living with HD, thank you for your continued partnership and generous support. Our community makes the difference as we reach out to families who are not yet connected to HSC, continue to support and advocate for families from coast to coast, invest in world-class research, and play a leadership role in the international Huntington disease community. With your help, we are continuing to improve the quality of life for people with HD, cultivating strength and resilience in the Huntington disease community and providing substantive reasons for hope. If you have questions, story ideas or comments about *Horizon* or the Huntington Society of Canada, please contact us at info@huntingtonsociety.ca or call us at 1-800-998-7398.

The Huntington Society of Canada is committed to reaching out to as many Canadians as possible. Should you wish to explore the French side of our website, select the Français option at the top right hand corner of our website www.huntingtonsociety.ca.

La Société Huntington du Canada a pour mission d'éduquer et d'aider autant de Canadiens que possible. Si vous souhaitez explorer la partie française de notre site Web, veuillez cliquer sur l'option française en haut à droite de la page suivante : www.huntingtonsociety.ca.