Family planning options for HD-free kids

By Shawna Feely, MS, CGC
UI HDSA COE Genetic Counselor, and
Carly Siskind, MS, LCGC
Stanford School of Medicine,
Neuromuscular Disorders Program

“Will my kids have what I have?”

This is a common question in genetic counseling. Sometimes this question comes after the kids are already grown, in which case we know the risk for Huntington disease is 50-50. But in other cases, this question comes before pregnancy, which leads to a conversation about reproductive options.

It is possible to make family planning decisions to prevent having a child that is at risk for HD. These are very personal decisions, and what is right for one person or family may not be right for others. Our job as genetic counselors is to present the available options, and to help the family determine what is best for them.

Prenatal options

If a woman is pregnant, the pregnancy can be tested through various methods to determine if it has the gene expansion that would cause HD.

Chorionic Villus Sampling (CVS):

CVS is performed between 10-12 weeks of pregnancy. Cells from a sample of the placenta are tested for the familial gene expansion. Complication rates vary by center and expertise of the person performing the test, but are usually quoted between 1-in-100 to 1-in-300. Complications can include miscarriage.

Amniocentesis:

This test is performed after 15 weeks of pregnancy, in the second trimester. A sample of the fluid that surrounds the pregnancy (amniotic fluid) is taken, and cells are analyzed for the familial gene expansion. The complication rates for amniocentesis are usually quoted lower than CVS, between 1-in-200 to 1-in-1,000.

What a family does with the information from this testing is ultimately up to them. However, typically this testing is offered to couples who plan to terminate a pregnancy if it is determined to have the HD gene expansion. It is not the right option for everyone, but this method can prevent future generations from having HD.

Preconception option

Preimplantation genetic diagnosis (PGD) is available if a couple does not want to have a child at risk for HD and does not want to have to make the decision about terminating a pregnancy. PGD is used in connection with in vitro fertilization (IVF), which is a procedure more commonly used for couples who have difficulty conceiving on their own. In IVF, egg and sperm are combined in a laboratory and grown into embryos. Different labs use different technologies, allowing a couple to have a baby without having to have a child at risk for HD.

Research ongoing at UI HDSA COE

Dr. Ed Wild of HDBuzz and University College London calls Huntington disease the most curable incurable brain disorder, in part because of the promise he and other researchers see in treating this disease that has a known genetic repeat as its cause. In order to continue moving closer toward treatments and an eventual cure, participants are needed to take part at all levels of research, from observational studies to clinical trials.

The following studies are currently ongoing at the UI HDSA COE.

Enroll-HD: The UI HDSA COE is...
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All in for HD Awareness Month

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Student excels at HD research


Little did she know it, but McKenzie Wallace was destined to study at the UI from when she was 11 years old.

That was when a close family friend with two daughters whom Wallace had played with for years passed away from Huntington disease.

“When she passed away, my dad told me, and I just broke down in tears,” Wallace recalled. “I couldn’t function that day. I was bawling my eyes out at the funeral. It was the first time that I had experienced death and just knowing what was going to happen to her daughters.”

The experience emboldened Wallace, a graduating senior majoring in nursing and international studies/global health. She chose the UI in large part due to its strength in HD research. Wallace approached Dr. Nancy Downing about helping out in her lab.

“Downing hired Wallace, and from that moment, Wallace was a co-leader of a project examining how diet and exercise affect the disease’s progression.

“She was a go-getter,” says Downing, an assistant professor of nursing. “She seemed to very quickly understand what was needed and not need much guidance. She took the initiative, which was exceptional (for a student) at her level.”

“Nancy just let me kind of take over as much as I wanted to,” related Wallace. “So, I just jumped in and tried to find out what would work.”

Wallace’s research included organizing the study, recruiting participants, conducting the research, analyzing data, and writing about the results. The payoff: first authorship on a paper that will be submitted to a peer-reviewed journal.

“Well, she earned it,” Downing says of Wallace’s lead authorship on the paper, a rarity for an undergraduate. “She was a very hard worker, and she didn’t just treat it like a job. She acted in the full capacity as a researcher, which is having intellectual conversations from the beginning.”

Loss of family friend to HD inspired Wallace to pursue research

but once an embryo has a certain number of cells (at least six), between one and ten are removed either by a biopsy, removed chemically or removed by a laser. The DNA from these cells is analyzed for the familial gene expansion.

The lab would choose the best embryos that are not affected with HD to be implanted back into the uterus of the mother to carry to term. PGD can be an amazing option for some families, but there are downsides, including the need for hormone shots and two surgeries. There is no guarantee that the embryo will implant when reentered into the uterus, so an additional cycle may be needed. Each cycle of IVF tends to cost $10,000 –$20,000, and not all insurances cover it. Furthermore, each PGD is often another $3,000–$5,000. Finally, many centers would suggest following up PGD with a CVS or amniocentesis to ensure that the embryo implanted really was free of the gene expansion causing HD.

All of these options are available for couples who know the affected parents’ gene status. They are also available if someone is at risk for HD, but does not want to know their own status. This is called non-disclosure testing, and the at-risk parents’ status will not be disclosed. Your local genetic counselor would be happy to review the specifics of these options in more detail. These are very personal decisions and only the family can know if one of these options is right for them.
The hot dish on spreading HD awareness

Excerpted from “Huntington's disease awareness: Spreading it, maximizing it and measuring success with a hot dish” by Sean Thompson in The Huffington Post on June 22, 2015.

Feeling supported and understood by friends and the general public, and not feeling utterly alone anymore. This is how so many in the HD community would like to feel. Making inroads toward this level of support involves achieving one of those nebulous activism buzzwords: awareness.

Whether talking about HD or any other cause or human condition, we know awareness is a good thing. But beyond that, the whole idea of awareness can be murky. How exactly is awareness beneficial for those who need it? And how can awareness of HD be achieved?

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The purpose of Enroll-HD is to accelerate discovery of effective treatments, and improve clinical care for HD patients by identifying best practices. Visits typically take no more than two-and-a-half hours and can often be completed on the same day as other research or clinical care. Travel compensation is provided.

PREDICT-HD: The UI HDSA COE’s flagship study has continued to see new participants in our center, and current participation in the study will conclude in August.

Recent participants have been individuals who have been willing to undergo a spinal tap procedure in order to collect cerebral spinal fluid (CSF). Study Coordinator Bella De Soriano says an emphasis has been placed on collecting CSF samples because researchers think examination of the proteins and molecules in CSF may lead to further understanding of HD, and could potentially lead to new methods to diagnose and treat HD.

Thanks to study participants and their continued willingness to give their time and energy toward to contribute to this 14-year study, De Soriano says recruitment goals have been met and no new visits are being scheduled until further notice.

Clinical trials coming soon

The UI HDSA COE has long been a site for multiple ongoing clinical trials to test the latest potential treatments for HD. Currently, the center is nearing the final stages of approval to begin recruiting participants for at least two clinical trials. UI HDSA COE Social Worker Amanda Miller says announcements will be coming soon with more information about opportunities to take part in these studies featuring promising new therapies for HD.

For advocate Sara Dean, “casserole awareness” would mean knowing how to react to HD.

“Awareness is our bridge to the world,” Dean says, “so that when someone says “I have Huntington’s disease,” people will respond with ‘We’re in this together – here’s a casserole!’”

It’s funny (humor can be a huge awareness technique as well), but Dean’s hot dish example is a goal to strive for. With dedicated advocates like Dean, Palka, Walker and many others, the HD community is on its way to achieving a new wonderful term coined by Dean: “casserole awareness.”

For more information or to participate in any of these studies, contact us at hdcenter@uiowa.edu or 319-353-4307.

HDSA National Youth Alliance 2015 Ultimate Youth Researcher Jolene Luther (middle) is joined by NYA members Amanda Butterworth (left) and Angela Mabry (right).

Luther wins research award at convention

Jolene Luther, was named the HDSA National Youth Alliance's 2015 Ultimate Youth Researcher at this year's HDSA National Convention for her accomplishments at the UI HDSA COE.

During her year as a research associate, Luther coordinated four studies and cowrote a manuscript on psychiatric symptoms in early HD.

Luther is continuing her career in HD research this fall as a graduate student at the Medical College of Wisconsin.
Could lifestyle explain why women live longer?


Women tend to live longer than men. But it wasn’t always that way, a new study says.

In the study published in the Proceedings of the National Academy of Sciences, researchers began by examining common causes of death of people going back more than 200 years.

The researchers focused on five major causes of death: cardiovascular disease; stroke; cancer; influenza and pneumonia; and smoking-attributable deaths, which they estimated from rates of lung cancer and other smoking-related diseases.

In general, men and women born in the 19th century had much shorter lives than people born in the 20th century. That’s because people who lived in the 1800s had less access to clean water and food, and they didn’t have antibiotics to help them fight infectious disease, said Hiram Beltran-Sanchez, a demographer at the University of Wisconsin in Madison and the lead author of the study.

But once antibiotics, safer water and more nutritious food became available, the odds of dying in any given year between the ages of 40 and 90 fell by 0.29% for women, compared with 0.17% for men.

While these numbers are small, they add up over time – for people born between 1900 and 1935, men were two to three times more likely to die in their 50s and 60s than were their female counterparts, the researchers wrote. This gender imbalance is caused primarily by cardiovascular disease and smoking-related deaths, he said.

By looking at data from the United Nations Food and Agriculture Organization, the team saw that the gender gap in life expectancy really opened up when people started eating more animal fats, a shift that began in adults born at the end of the 19th century.

These data mean the difference in life expectancy between men and women probably isn’t due to biological factors alone, Beltran-Sanchez said. And that would be good news.

“If this is something that has been triggered by lifestyle, or diet, we could potentially ameliorate it or diminish it,” he said.
Join the fun and volunteer to help with the Iowa City/ Coralville Team Hope Walk!

Saturday, November 7, 2015
Coral Ridge Mall | Coralville, IA

- Raise awareness about HD
- Meet new friends impacted by HD
- Increase funds to support HDSA

Volunteers are needed to help with all aspects of planning the walk including Team Captains, securing silent auction items, identifying potential walk sponsors, and helping the day of the event.

For more information or to get involved, contact

**Ingrid Wensel**
Walk Coordinator
319-431-7823
iowacity@walkforhd.org

*The Team Hope Walk program is HDSA’s signature grassroots fundraising campaign designed to provide hope and support for those touched by this devastating disease. All proceeds support HDSA’s mission to improve the lives of people affected by HD and their families. Since their inception in 2007, Team Hope Walks have occurred in over 100 different cities and have raised over $7 million for the cause.*