

Young writer earns lab coat

Perth Modern student Emma Congear, who received a prize for an insightful essay about Huntington's disease, won the opportunity to swap her school uniform for a lab coat at the Western Australian Institute for Medical Research for a week.

The year 10 student from South Perth first wrote her article as part of a health assignment, but it was entered into a competition, with work experience at WAIMR as the prize.

The 15 year old, who is trying to decide what she'd like to do after leaving school, made the most of her opportunity to learn about genetics at the WAIMR laboratories.

"I really want be able to help people," she said, "and that's shaping what I want to do as a career."

Scientists at WAIMR are involved in a number of exciting projects, investigating the genetic and environmental causes of diseases, including nerve and muscular disorders.

Emma's Article follows...

Media contact: Carolyn Monaghan cmonaghan@waimr.uwa.edu.au 0448 021 932.

HUNTING FOR SURVIVAL - LIFE WITH HUNTINGTON'S DISEASE

Emma Congear reports:

"In my head, I'm not different.

In my heart, I'm not different."

- James Birdsall, age 30.
Diagnosed with HD in 2008

Trapped in a body that won't respond, watching your entire life – your career, friends, family, and body – slowly slip away before your very eyes. This is just part of the torment and dejection which rules over the cold harsh reality of a life with Huntington's Disease (HD).

Huntington's Disease is a terminal progressive neurodegenerative genetic disorder which affects approximately 7 out of 100 000 Australians (however experts say that this number could be much greater due to people keeping their condition a secret because of fears of discrimination). Affecting both men and women, HD gradually impairs a person's body movements, speech, and mental ability; but worst of all, HD is hereditary. This means that if a person with HD wanted to have children there would be a 50% chance that the child will be born with the disorder. However the symptoms of HD do not usually appear straight away. Although the age at which HD first shows itself is different for each individual, HD often appears during middle age.

So how do people find out if they have HD?

Due to the discovery of the HD gene in 1993 by Nancy Wexler, there is now a direct test for HD, by taking a person's blood sample and counting how many CAG repeats are in the HD gene region. This is called a pre-symptomatic test. Before, during and after undertaking the

test, a person is required to undergo counselling sessions, as taking the test is an extremely important and personal decision, which can have detrimental consequences.

As there is currently no known cure for HD, taking a pre-symptomatic test that returns positive for HD can be devastating and, to many, the equivalent of a death sentence. Many implications follow this result, such as discrimination at work, social alienation, complications with childbearing, and an increased chance of committing suicide. Knowing that one day you're going to wake up and notice the first symptoms of HD appearing, and see yourself begin to progressively deteriorate is a horrifying reality. You will never be able to have children without a 50% chance of dooming them to the same grim daunting fate as yourself. Not only does this affect your self-esteem, it also affects romantically finding someone and whether marriage is a good idea.

Nancy Wexler, discoverer of the HD gene, has had a history of HD in her family. However after years of research and now finally having the pre-symptomatic test available, both Nancy and her sister have decided not to take the test. The reason, she says, is that there's no effective treatment for Huntington's Disease, nothing she could do if the test showed she'd get the disease. But there is one thing, Wexler said, that would change her mind:

Excellent
Emma!

Children. If Nancy was to have children she said that she would want to be certain that they would have no chance of inheriting HD.

A terrible and unjust result of HD for both the person carrying it and their entire family is discrimination. Many insurance companies tend to refuse covering people with a family history of HD. What is surprising is that discrimination is no worse for those who test positive for the Huntington's gene than for those who merely have a family history of the illness. Fortunately, most European countries and now the United States, have protections and provisions made to protect its citizens from genetic discrimination.

James Birdsall is a 30 year old American who lives with his mother, Melissa Billiard. As a child he enjoyed a typical childhood such as riding his bike around the local streets and hitting the surf whenever the weather permitted. However, now with Huntington's Disease, Birdsall struggles to come to terms with his disorder, getting out of the house is a major effort, let alone riding the waves or a bike.

"I call it a closeted disease because people who have it don't talk about it. They're ashamed. But there's nothing to be ashamed of. It's not their fault," Biliardi said. "When you have an illness or disease like this, people need to realize it's not contagious."

With the costs of professional care for James exceeding his mother's own income, Melissa quit her job so that she could provide James with full time care. When asked to comment on her financial

position, Billiard had this to say: "If you do have it and test, what happens then? What happens with your work? They're not supposed to discriminate, but they do. And what happens to your insurance? Everything is affected by the outcome of the test...On the other hand, social services aren't in place." Whilst there are social services to help with the medical expenses of Huntington's disease, there is an agonizing waiting period of two years before any cover is received. "In that time, people lose their jobs, they lose their families and they lose their homes because they lose their mental capacity. They need immediate help, not to be put on a waiting list while their lives, as they knew them, slip away," Biliardi said. Although it took a lot of effort to compose his thoughts, James was able to comment on his life with HD: "In my head, I'm not different. In my heart, I'm not different. But sometimes I'll be talking to somebody and I won't be able to finish the conversation because I've forgotten what we're talking about, or I'll stutter and keep stuttering," Birdsall said. "Sometimes I'll be sitting quietly just like this and my leg will shoot into nowhere. I hate that."

Living a life with HD is a horrible burden. However there is a vast amount of research for HD in place, trying to slow the progression of HD as well as finding that miraculous cure which many have been wishing for. To help the research for HD as well as those who are suffering please make a donation today and spread awareness about HD.

To make a donation, please contact Huntington's WA on 9346 7599 or visit their website at <http://www.huntingtonswa.org.au/>

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