Families At Risk: Long-term Impact of Huntingtons Presymptomatic Genetic Testi

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Country

USA

Title of project or programme

Families At Risk: Long-term Impact of Huntingtons Presymptomatic Genetic Testi

Source of funding information

NIH (NINDS)

Total sum awarded (Euro)

€ 1,219,644.04

Start date of award

15/08/2014

Total duration of award in years

3

The project/programme is most relevant to:

Huntington's disease

Keywords

Genetic screening method, Huntington Disease, presymptomatic testing, Test Result, Family

Research Abstract
DESCRIPTION (provided by applicant): Huntington’s disease (HD) has for decades served as a model for how we think about genetic testing, and its benefits and risks for tested individuals

and their families. In 1983, the gene for HD was mapped to chromosome 4, allowing linkage tests to be developed for use in presymptomatic genetic testing for HD. In 1986, Johns Hopkins launched one of the first two such testing programs in the United States. This work influenced guidelines for the provision of HD genetic test results, which have subsequently influenced contemporary guidance for other adult-onset neurodegenerative diseases. This early experience also arguably influenced our collective thinking about many issues related to genetic testing and the provision of genetic test results. Almost 30 years later, we are still in contact wth many members of this early cohort. As increasing numbers of genetic tests are being used to predict adult-onset neurodegenerative disease, and as large-scale genetic testing is increasingly integrated into clinical care, it is critical that we understand not only the implicatons of presymptomatic testing for the at-risk individual over several years, but also for the at-risk individual and her/his family over the course of their lives. Here, we have a unique opportunity to take a retrospective look over decades at how at-risk individuals and their families communicate about and cope with test results not months or years following testing, but decades following testing. We will do this through in-depth interviews with a subset of those at-risk individuals who were tested between 1986 and 1996, focus groups with a subset of family members of tested individuals, and a survey sent to all at-risk individuals who were tested from 1986 to 1996 and their family members. The outcomes of this research will improve our understanding of how the results of genetic testing for serious, currently incurable disease are communicated in and through families, how this information influences choices (e.g., career, participation in research, having children) and trajectories of at-risk individuals and their families over decades, and will help inform policies and procedures for communicating such results. Under Specific Aim 1, we will explore the impact of presymptomatic genetic testing for Huntington’s disease (HD) on the lives and choices of at-risk individuals, 18-28 years after testing. Under Specific Aim 2, we will build on prior work with this cohort to determine longitudinal changes in individuals’ feelings about and understanding of their testing experience as they have progressed through life’s stages. Under Specific Aim 3, we will explore the impact of presymptomatic genetic testing for HD on family members (spouses, partners, children) of tested individuals, 18-28 years after testing. Under Specific Aim 4, we will build on the results of Aims 1-3 to explore how the views of individuals and their families comport with the current paradigms and policy for the conduct of clinical genetic and genomic testing.

Lay Summary

PUBLIC HEALTH RELEVANCE: This project is designed to improve our understanding of the impact of presymptomatic genetic testing for Huntington’s disease on families over time. We will talk with individuals who received testing 15-25 years ago, along with spouses, partners, and children of those individuals to learn about the impact of the testing and the resulting informatio on the family. We hope this information will help inform current thinking and policy governing the return of results from genetic testing.

Further information available at:

Types:
Investments > €500k

Member States:
United States of America

Diseases:
Huntington's disease

Years:
2016

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N/A

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