



FAQ about Genetic Testing and Huntington Disease

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Presenter Disclosures

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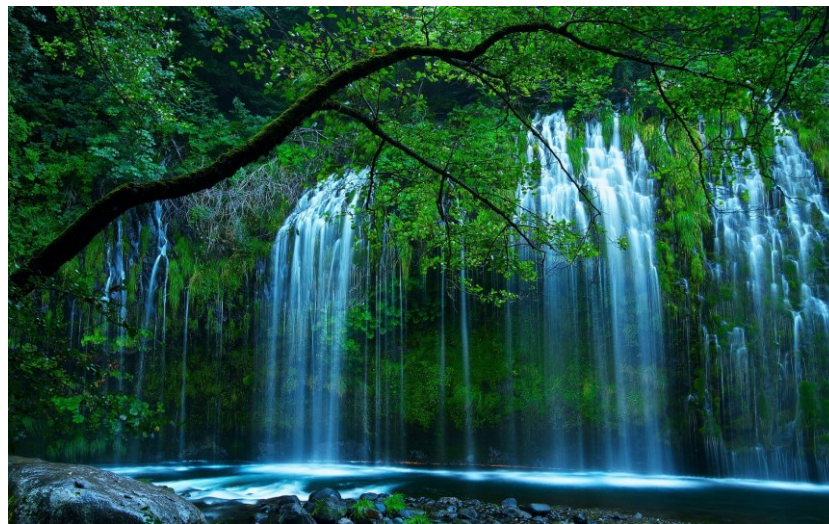
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Huntington's Disease
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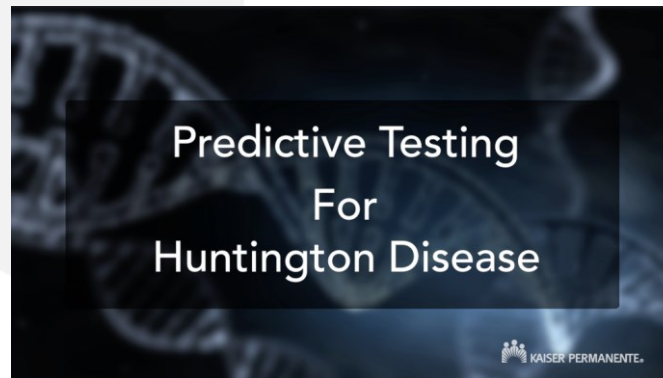
Overview of our time:

- Video about predictive testing
 - only 11 minutes!
- Presentation about some FAQ
- Q & A time
 - Privacy & Respect



Video about predictive testing

- ***Predictive Testing for Huntington Disease*** (11:39) - An 11 minute video about predictive testing created in partnership between Kaiser Permanente & UC Davis Huntington Disease Centers of Excellence. . <https://www.youtube.com/watch?v=4HW5YdgM4zs>



A Little History

The Past

- HD marker identified 1983, linkage analysis began in research studies in 1989 (Gusella et al, 1983)
- Gene identified 1993 (Huntington's Disease Collaborative Research Group, 1993) and CAG expansion found to be causative

The Present

- **2017:** DNA testing for HD is now faster, cheaper, more accurate, and more accessible.
 - **BUT.... testing for HD remains a process with the potential to have profound implications for the life of a person who chooses to be tested**
 - financial, emotional, & social issues that involve not only the person seeking testing, but also his or her immediate family - and often the extended family as well.

FAQ-Can my CAG # predict when I will get symptoms?

- CAG # correlates with age of onset but not the whole story.....
 - ~50-70% of the factors that contribute to age of onset
 - Other genes, general health
- **Normal:** 26 or fewer CAG repeats
- **Intermediate:** 27-35 CAG repeats
- **Reduced penetrance:** 36-39 CAG repeats.
- **Full-penetrance HD-causing:** 40 or more CAG repeats
 - Juvenile HD-causing alleles 60 or more

– Gene Reviews updated: December 11, 2014.

New HDSA Guidelines released 2016

GENETIC TESTING PROTOCOL FOR HUNTINGTON'S DISEASE

- HDSA first published “Guidelines for Predictive Testing for Huntington’s Disease” in 1989 and revised in 1994 and 2003
 - Collaborative effort written by HDSA w/input from clinicians, lab professionals, & people at risk
 - Recommendations are based on....
 - a review of the previous HDSA guidelines
 - the experiences of many who have been tested
 - experiences of providers who have offered the testing
 - and the growing body of knowledge about genetic testing for many other diseases.

New Recommendations Released 2016

- **Goals of new recommendations:**
 - Decrease barriers to testing while protecting the people who choose to be tested.
 - Allow flexibility in the way testing is offered while providing a framework for health care professionals
 - Allows for providers to use clinical judgment

IN GENERAL: Encourages centers to provide genetic counseling and psychological support services that allow predictive testing to be performed in a timely, sensitive, knowledgeable, and cost effective manner.

HDSA Provides Framework & Guidance, NOT Regulations

- HDSA does not enforce recommendations nor do/can they mandate that people use HDSA Centers of Excellence for testing
- HDSA does not monitor costs of testing
 - Experience has shown that some elements of the testing process should be considered **essential**:
 - **Pretest counseling, informed consent, and in-person results**
 - **A child should not be tested unless the child is exhibiting symptoms that cannot be attributed to any other condition.**

Why can't I have my child tested for HD?

- We don't test children unless they are showing signs of JHD without any other explanation.
- Children deserve the right to grow up and make their own decision about whether or not they want to know their gene status.
 - Remember only ~10% of the people at risk for HD want to know
- What if I learned their status and kept it from them?
 - This would be difficult
 - Secrets are often detrimental to healthy family relationships
 - You may have subconscious bias if you know their status
 - When and how would you tell them?

Why test at an HDSA Center of Excellence?

- Not all medical providers are familiar with HD and most don't understand the complex nature of this testing.
 - May not understand results, implications, or may misinterpret
- Most HDSA COE team members have many years of experience w/HD & systems are in place to support people through the process at COEs.
- People often underestimate the emotional nature of the testing process even when they truly believe they are ready to hear their results.
- Oversight by HDSA
- If you can't go to an HDSA COE, at least seek testing with a Neurologist or Geneticist.

Why do I need genetic counseling to be tested?

- Genetic Counselors are non-directive & non-judgmental.
 - We help people become better informed & support independent decision-making around emotional & personal testing decisions.
 - Discussing the issues w/caring professionals helps people make informed decisions about whether testing is right for them at this time in their lives.
 - Most people that didn't want counseling before testing tell us in retrospect.....
 - that they didn't really understand what was going to happen during the counseling session
 - Some feared GCs would tell them not to have children
 - felt counseling helped them clarify their thoughts/feelings
 - brought up some issues they hadn't thought about
 - overall found the process helpful

3 types of testing discussed in HD SA recommendations.....

- 1) Confirmatory
- 2) Predictive
- 3) Prenatal/PGD



FAQ: How do I get tested for HD if I am showing symptoms?

Also called
**CONFIRMATORY OR
SYMPTOMATIC TESTING**

CONFIRMATORY TESTING

- If someone is showing symptoms and has a family history of HD

OR....

- Has symptoms of HD but no family history
HD testing is sometimes part of the testing panel
- HDSA recommends that results should be given in person
 - Difficult to get these results even if expected
 - Taking away hope that it could be “something else”.

FAQ: How do I get tested for HD
before I am showing any symptoms?

Also called PREDICTIVE
OR PRE-SYMPTOMATIC
TESTING



Predictive Testing Protocol

Previously involved 2-4 appointments

- Geneticist, Genetic Counselor
- Neurologist
- Social Worker, Therapist
- In person results disclosure

NEW PROTOCOL RECOMMENDATIONS
DEVELOPED BY HDSA DEVELOPED 2016.

2 visits and an in depth intake phone call and f/u phone call after results disclosure.

HDSA 2016 Recommended Components of HD Predictive Testing Process:

- **1. Telephone Contact** info re testing process, costs, & risks provided by a healthcare professional experienced in genetic counseling and testing for HD. Concerns re: sx and whether the individual may wish to include a neuro exam as part of the predictive testing process are also discussed.
 - Insurance, pedigree?, confirmation of dx, HD story, send information to review, video, discuss plan



- **2. Visit 1**
 - Genetic Counseling
 - Sign Informed Consent Document
 - Mental Health Assessment
 - Neurological Exam (if available/desired)
 - Draw Blood if all in agreement to proceed

- **3. Visit 2**
 - Disclosure of Results in Person
 - Arrange Post-result Follow-up

Encourage people to

bring a support person (preferably not an affected parent or an at-risk relative) for both visits.

clear their day/weekend/week as they can to allow for whatever reaction they have to the news.

think carefully about who they want to know that they are testing and when they are getting results.



- **4. Follow-Up**
 - Prearranged phone call or in-person visit

Neurological Evaluation (Exam)

- Neurological evaluation **may** be offered prior to the predictive test to any Individual who is or might be concerned about possible symptoms.
 - A normal exam can sometimes be enough reassurance that predictive testing is no longer desired.
 - However, refusal of a neurological examination cannot exclude the Individual from predictive testing.
 - If someone who is having symptoms (subtle or just unaware of their sx) declines the neuro exam they miss out on the opportunity to be better prepared for their results.

Additional care that may be indicated/provided for the Individual considering predictive testing

- *Neuropsychological testing*
- *personality inventory*
- *additional visits with the genetic counselor or mental health professional*
- *establishment of contact with a counselor outside of the testing program may be helpful/necessary.*
- *Additional support or counseling for the Individual's primary support person or family may sometimes be necessary*

Predictive Testing-some info from from guidelines

- *“HDSA recommends two in-person visits for an individual requesting predictive genetic testing unless concerns on the part of the testing team arise during the initial call or first in-person appointment that would necessitate additional services”.*
- *“Active psychiatric problems must be stabilized before an Individual undergoes predictive testing. Predictive testing cannot proceed if the responsible health professional believes it would be harmful to the Individual”.*
- *“An Individual who cannot or does not want to identify a testing companion cannot be excluded from testing”.*
- *“The decision to take a predictive test for HD must always be an informed, carefully considered, and freely-chosen personal decision. An Individual must not be pressured into testing by a spouse, another family member, a health care provider, an insurance company, or an employer”.*

Predictive Testing Counseling

- **Test for yourself**, not your spouse, children, other family members, employer, or friends
 - Talk to family and friends but ultimately your decision
- Think about **why you want to know your HD status**
 - Need to know, planning and decision making, clarify risks for children, family planning, education, retirement, etc
- **Once you know your HD status, you can't ever NOT know again**
 - Some people feel living with the ambiguity is easier than knowing they will eventually develop sx.

Issues Around Predictive Testing

- Life situation and mental stability to handle getting results at the time
- Support system
- How would results change your life?
- What would you do differently?
- Survivor guilt
- Risk for depression
- Suicide assessment
- Insurance concerns

Why does it take so long to go through this process?

- ***“Timing :*** *Predictive testing should ideally take place in a supportive environment during a time period when the Individual is not otherwise stressed. Testing must not be accompanied by a sense of urgency or emergency and should be considered in a cautious manner.*
- ***It is important to include enough time in the counseling process so that the Individual can fully consider the implications of the test and have a chance to reconsider his or her decision.”***

We usually ask for some type of confirmation of the diagnosis in your family before proceeding with predictive testing but.....

What if you are the first person in your family to seek testing

- If the testing is confirmatory, your neurologist will order the test and give results & refer to a COE ?
- If it's predictive testing and we can't get confirmation of the diagnosis in your family we may still be able to offer testing but there are caveats about what a negative result really means.
- Sometimes neither parent apparently had/has HD:
 - parent may have died or “out of the picture” before symptoms of HD became obvious
 - Sometimes a parent has an “intermediate” or “reduced penetrance” sized repeat
 - Non-paternity

FAQ: Should I use my health insurance to pay for predictive testing?

- This is up to you but here's the take home message....
- If you use your health insurance your result will be accessible to your health care team (but protected under HIPAA & GINA) and anyone you give permission to view your medical records.
 - GINA is a federal law prohibiting your employer and health insurance from discriminating against people who test positive (who don't have any symptoms). BUT..... GINA doesn't apply to
 - Long Term Care Insurance
 - Disability Insurance
 - Life Insurance
- So it could be more difficult to get these types of insurance if your result is in your chart.
- Testing anonymously isn't available at all COE

Insurance Concerns: G.I.N.A.

- **Genetic Information Nondiscrimination Act**
- Health insurers and employers may not use or require genetic information to make enrollment, coverage, hiring or firing decisions
- Genetic test results will not be considered a “pre-existing condition” as long as person is **NOT** symptomatic

What doesn't G.I.N.A. cover?

- Health and Employment provisions do not apply to US military, veterans seeking care through VA or Indian Health Service
- Health provisions not in effect once there are symptoms of the condition/disease
- **Does not include protection from life, disability, and long-term care insurance genetic discrimination**

Electronic Medical Records

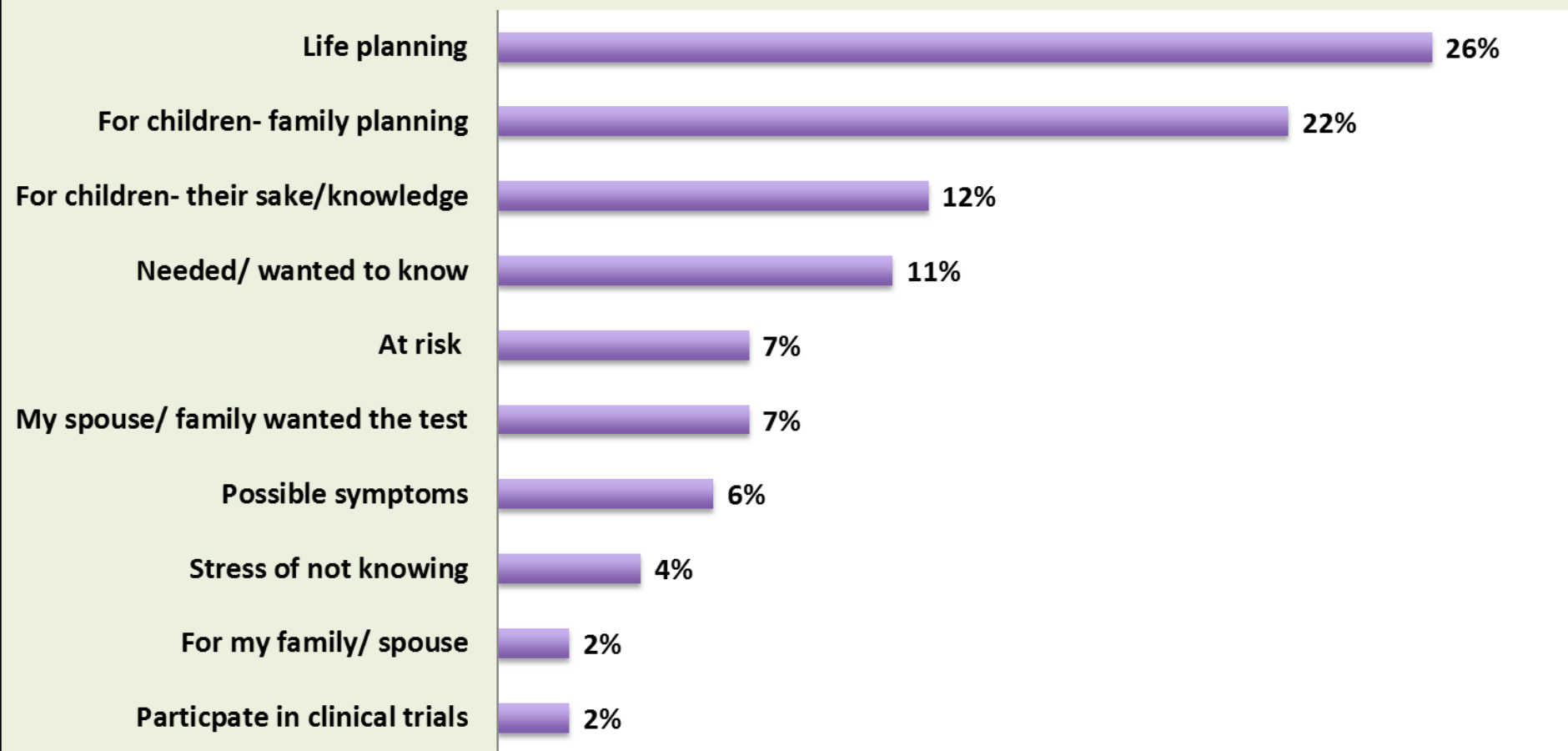
- Most health care organizations have electronic charts now
 - HIPAA, GINA
- Many organizations share electronic records (with your signed consent)
 - “CARE EVERYWHERE”
- How do we code people who are:
 - Asking about predictive testing?
 - VAGUELY!
 - “genetic counseling”
 - “family history of genetic condition”
 - E-mails to your providers become part of your record
 - Referrals may/may not be part of your record
- IF YOU ARE WORRIED ABOUT INFORMATION GETTING INTO YOUR MEDICAL RECORD AND TO INSURANCE COMPANIES (long term care, life, disability) IT MAY BE BEST FOR YOU TO TEST ANONYMOUSLY.

Attitudes and beliefs about predictive testing in HD

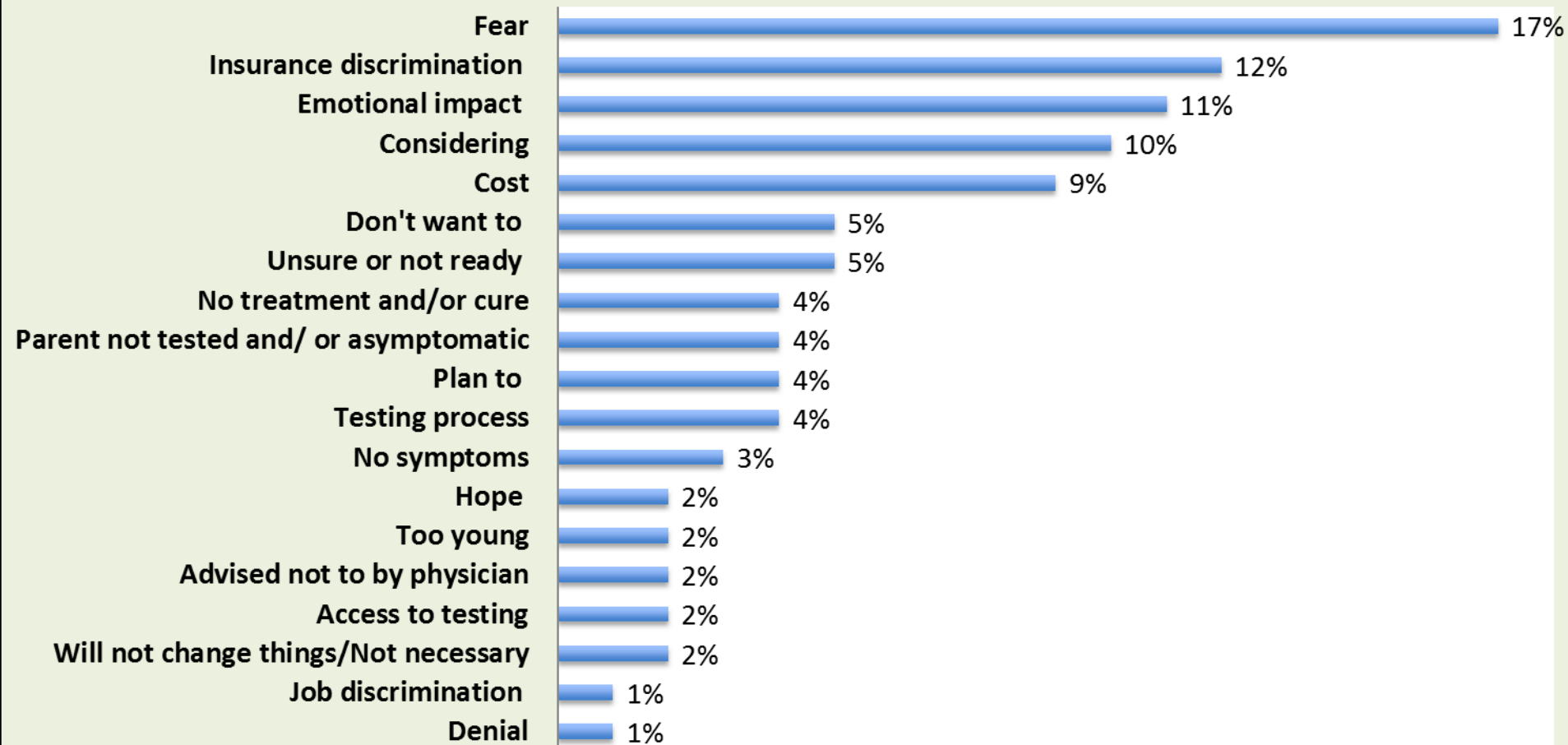
Survey conducted by neurologist Dr. Alexandra “Sasha” Duffy, 2014 & Cassandra Farrar, MS, LCGC (her thesis)

- Anonymous web-based survey, IRB approved
- Aims: To question a broad sample of individuals 18 and older who have previously undergone predictive testing *and* at-risk individuals who had not about their attitudes, beliefs, and concerns regarding predictive genetic testing in order to extend previous work and update current opinions.
- Methods: Multiple-choice demographic questions and open-ended responses regarding respondents’ experiences, beliefs, and attitudes
- Initially released in both paper and on-line versions at the 2014 HDSA national convention, shared with northern California HD support groups and then posted on HDSA website

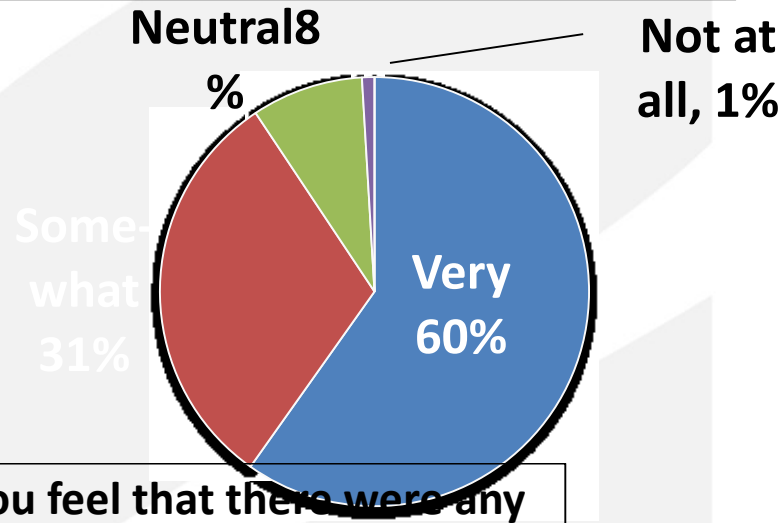
Reasons given FOR testing



Reasons given for NOT testing

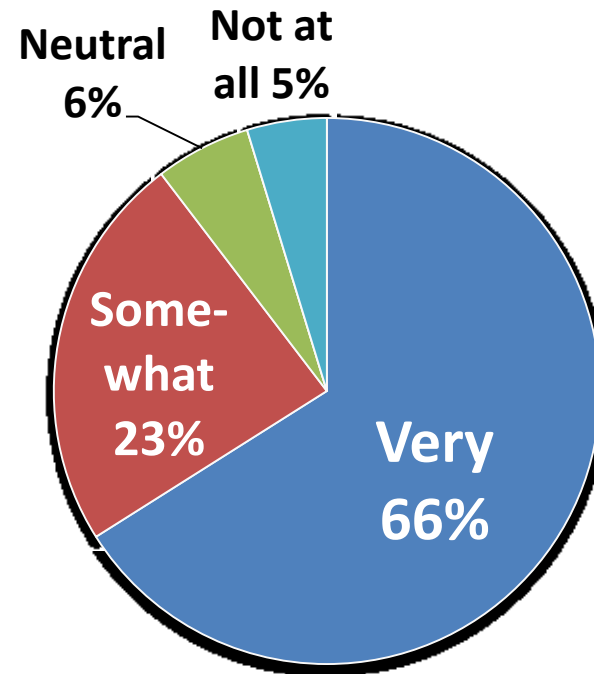


How satisfied were you in the predictive testing process?

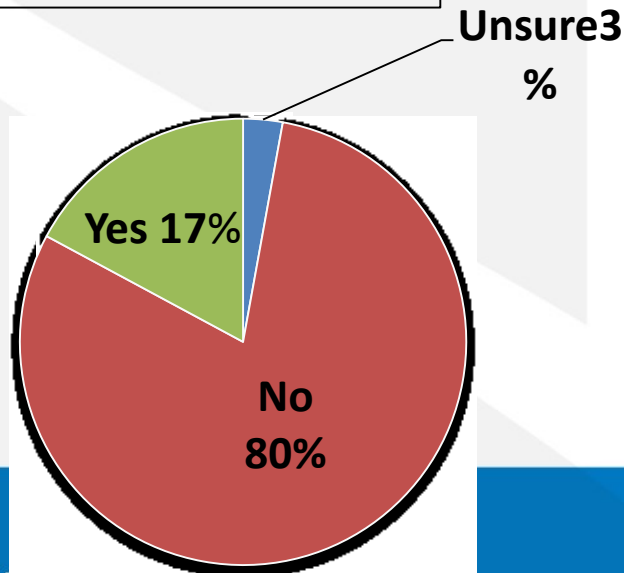


For those who had undergone predictive testing

How confident are you that your privacy was maintained during the process?

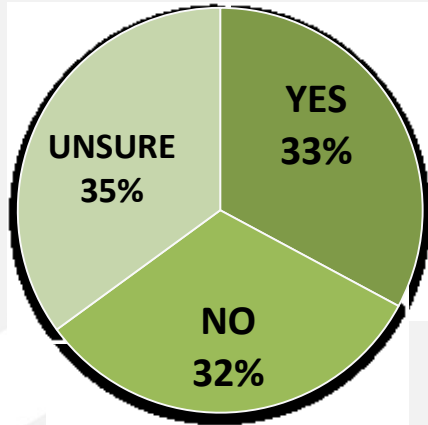


Do you feel that there were any inadvertent disclosures about your identity?

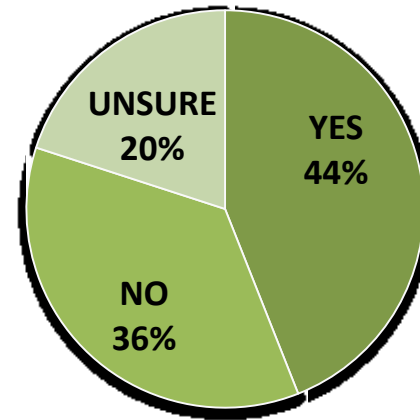


Would you change your decision to test if anonymous testing were an option for you?

Respondents who have NOT TESTED

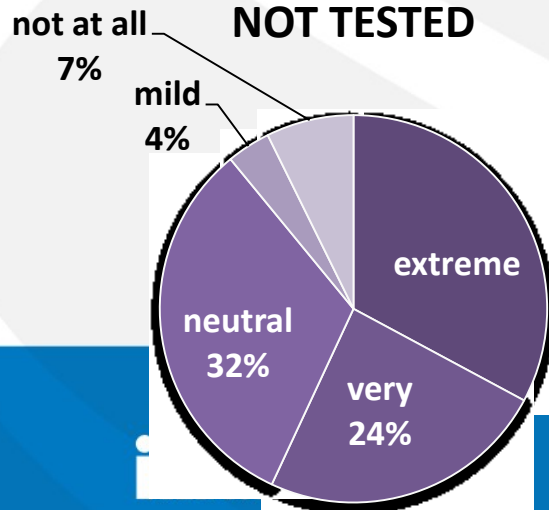


Respondents who have NOT DECIDED

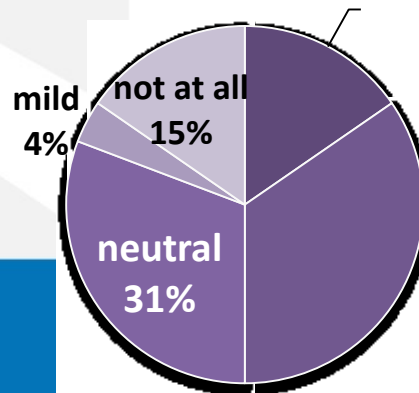


How important would the option of testing anonymously be to you if you were given the option to test anonymously?

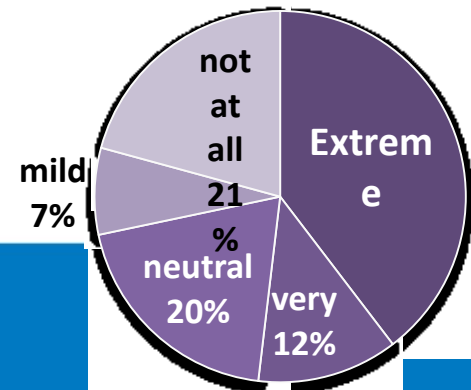
NOT TESTED



UNDECIDED



TESTED



HDSA Centers of Excellence are here for you....

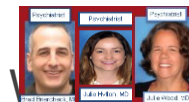
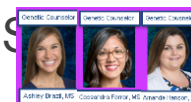
- We want to help make your journey with HD a little bit easier so please feel free to reach out to us when questions or issues arise.



Kaiser Permanente Genetic Movement Disorder Clinic-Sacramento HDSA Partner Center of Excellence



- Movement Disorder Neurologist-Dr. Khandhar
- Geneticists-Drs. Lipson, Tezcan, and Moghaddar
- Pediatric Neurologists-Drs. Friederich X2, Nelson & Masselink
- Genetic Counselors-Ashley Brazil, MS, Cassie Farrar, MS, Amanda Hanson, MS
- Social Services-Elle Tadina-Siau, MSW, Lori Penery, MSW
- Palliative Care, Janet Britton, RN
- Physical Therapy- Jeanine Perry, PT
- Psychiatry-Drs. Brad Briercheck, Julie Hyltunen & Julie V
- Clinic Coordinator-Mara Sifry-Platt, MS, LCC



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Questions?

