



Family Planning in Huntington's Disease

Chelsea Chambers, MS, CGC
Certified Genetic Counselor
HDSA Center of Excellence at the University of Virginia



Huntington's Disease
Society of America

The information provided by speakers in workshops, forums, sharing/networking sessions and any other educational presentation made as part of the 2016 HDSA Convention program is for informational use only.

HDSA encourages all attendees to consult with their primary care provider, neurologist or other healthcare provider about any advice, exercise, medication, treatment, nutritional supplement or regimen that may have been mentioned as part of any presentation.

Presenter Disclosures

Chelsea Chambers, MS, CGC

The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

No relationships to disclose
or list

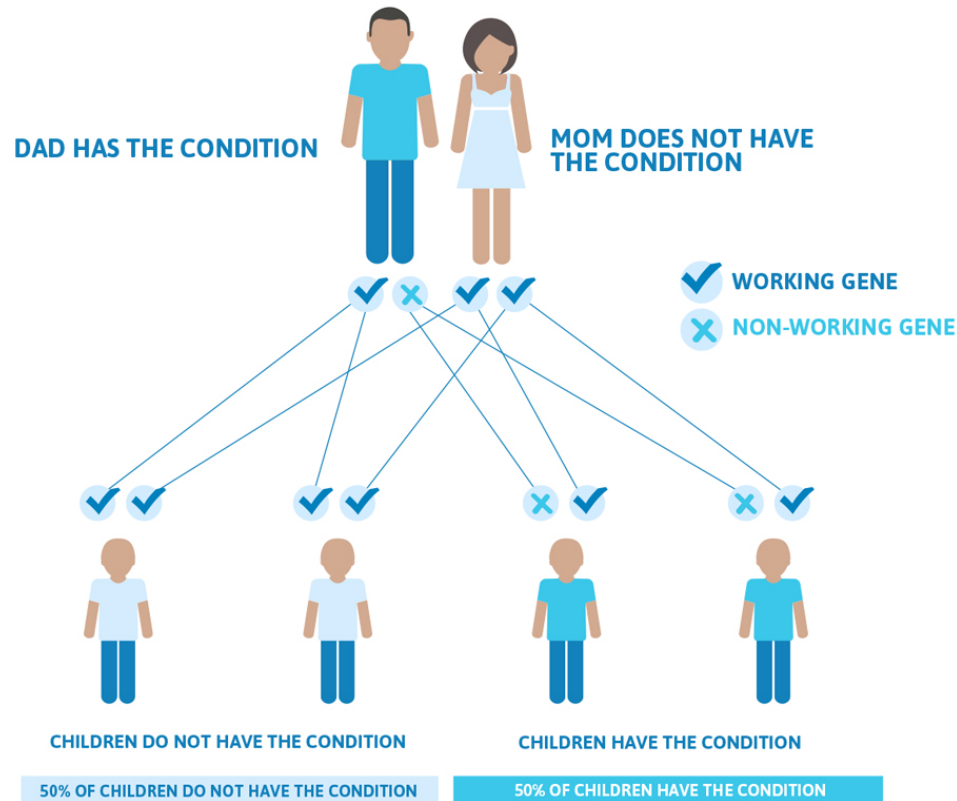


Overview

- Basics of HD Genetics
- Family Planning Options
- Issues to Consider
- Personal Experience: Emily's Story

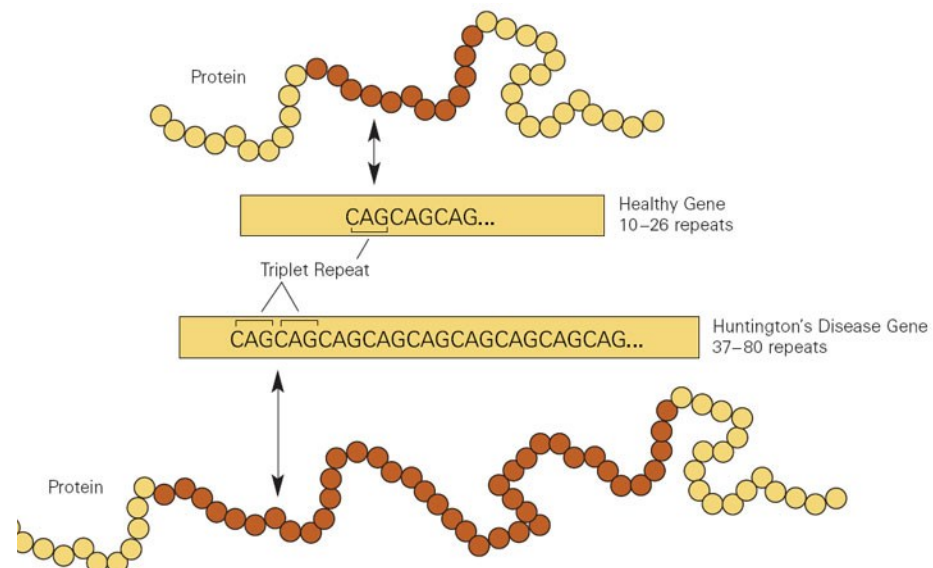
Huntington's Disease: Genetics Overview

Autosomal Dominant Inheritance Pattern



<https://www.geneticsupportfoundation.org/autosomal-dominant-inheritance>

Abnormal Trinucleotide Repeats:



Interpretation of genetic testing for CAG repeats

		CAG Repeats	Interpretation
Unaffected	Normal	26 or less	> No disease
	Intermediate	27 – 35	> Do not develop symptoms* but their children are at risk for developing Huntington's
Affected	Reduced penetrance	36 – 39	> May or may not develop symptoms at any age > May develop symptoms in old age
	Full penetrance	40 or more	> Have disease

* There are case reports of subjects within this range who develop mild symptoms of HD

Ref: Potter NT, Spector EB, Prior TW. Genet Med 2004;6:61-65



INNOVATE RESEARCH & DEVELOPMENT™

https://publications.nigms.nih.gov/findings/sept08/images/hunt_gene_big.jpg
<http://www.educatehealth.ca/media/369503/3-lightbox-hd-genetic%20testing-resources.png>

Anticipation: Earlier ages at onset in future generations

- Most often individuals inherit the exact same number of repeats as their parent
- Anticipation can occur when expansions occur in repeat length from one generation to the next
 - Mostly occur when inherited from affected father
 - Greater than 7 repeat increase is almost exclusively from a father
 - When inherited from mom, a child normally has the same number of repeats or a small increase of 1-3 repeats

Genetic Counseling

- Genetic Counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of hereditary disease
- Aids in informed decision making
- Pre-conception Genetic counseling is available to help a couple make family planning decisions together

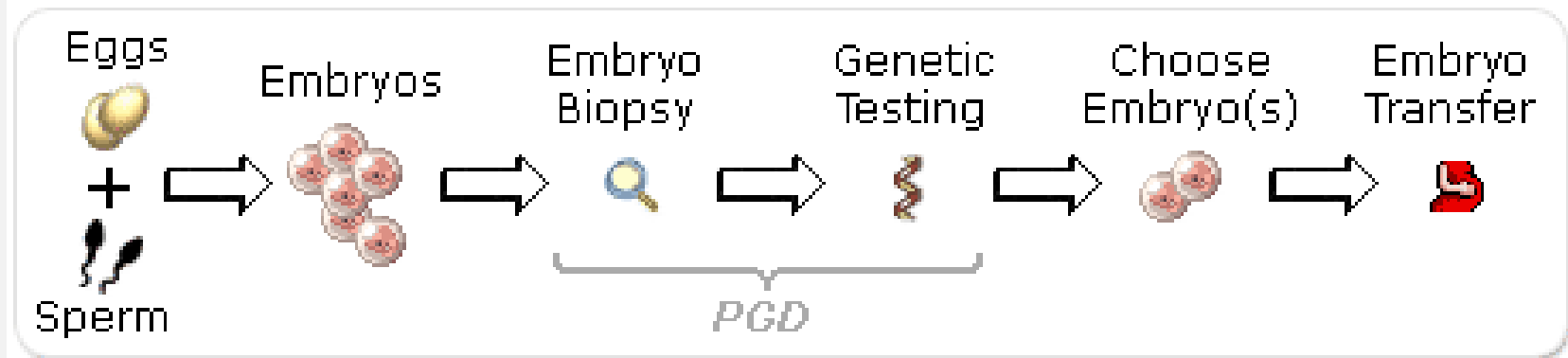
Family Planning Options Available

- Natural Conception
 - With or without knowing gene status
 - With or without prenatal testing
- Egg or Sperm Donation
- Embryo Donation
- Preimplantation Genetic Diagnosis with IVF
- Adoption

Concerns to consider with prenatal genetic testing

- Considered most challenging HD testing situation
- Simultaneous Pre-symptomatic and Prenatal testing
 - Challenging to prepare for emotional impact if both positive
 - Time constraint with prenatal testing
- If parents uncertain about termination:
 - Significant implications for unborn child
 - Emotional burden
 - Loss of ability to decide if want gene status information
 - Possible discrimination that is difficult to anticipate

Preimplantation Genetic Diagnosis (PGD) and In Vitro Fertilization (IVF)



- PGD: option available to couples seeking to have children who are known to be gene-negative and avoids ethical issues associated with termination a pregnancy
- IVF: assisted reproductive technology that is the process of fertilization by manually combining an egg and sperm in a laboratory dish, and then transferring the embryo to the uterus

<http://www.ingender.com/Gender-Selection/PGD/>

Cost & Insurance Coverage for PGD/IVF

- Average cost for IVF w/ PGD: \$17,000-\$25,000 in the US
 - Price may vary between centers and depends on amount of medication needed for IVF ovarian stimulation
- Some couples may have insurance coverage for IVF but it is rare for insurance to cover PGD
- Cost for PGD is ~\$5,000-\$10,000 (per embryo)
- Most patients will pay for PGD/IVF upfront and submit to insurance to try and get reimbursement

Success Rate for PGD/IVF

- Success rates is dependent on the doctors and IVF groups and can vary significantly between IVF centers
- Pregnancy success rates can vary between 40-70%
- PGD for single gene disorders is typically >90% accurate

Personal Experience:

Emily's Family Planning Story

Resources:

- HDSA Centers of Excellence
- HDSA Predictive Testing Centers
- National Society of Genetic Counselors