Family Planning in Huntington’s Disease
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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

DAD HAS THE CONDITION

MOM DOES NOT HAVE THE CONDITION

WORKING GENE

NON-WORKING GENE

CHILDREN DO NOT HAVE THE CONDITION

CHILDREN HAVE THE CONDITION

50% OF CHILDREN DO NOT HAVE THE CONDITION

50% OF CHILDREN HAVE THE CONDITION

https://www.geneticsupportfoundation.org/autosomal-dominant-inheritance
Abnormal Trinucleotide Repeats:

<table>
<thead>
<tr>
<th>CAG Repeats</th>
<th>Interpretation</th>
</tr>
</thead>
<tbody>
<tr>
<td>26 or less</td>
<td>No disease</td>
</tr>
<tr>
<td>27 – 35</td>
<td>Do not develop symptoms* but their children are at risk for developing Huntington’s</td>
</tr>
<tr>
<td>36 – 39</td>
<td>May or may not develop symptoms at any age</td>
</tr>
<tr>
<td>40 or more</td>
<td>Have disease</td>
</tr>
</tbody>
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* There are case reports of subjects within this range who develop mild symptoms of HD

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