Huntington's Disease Society of America

Family Planning in Huntington's Disease

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Presenter Disclosures

Carol Ludowese, 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

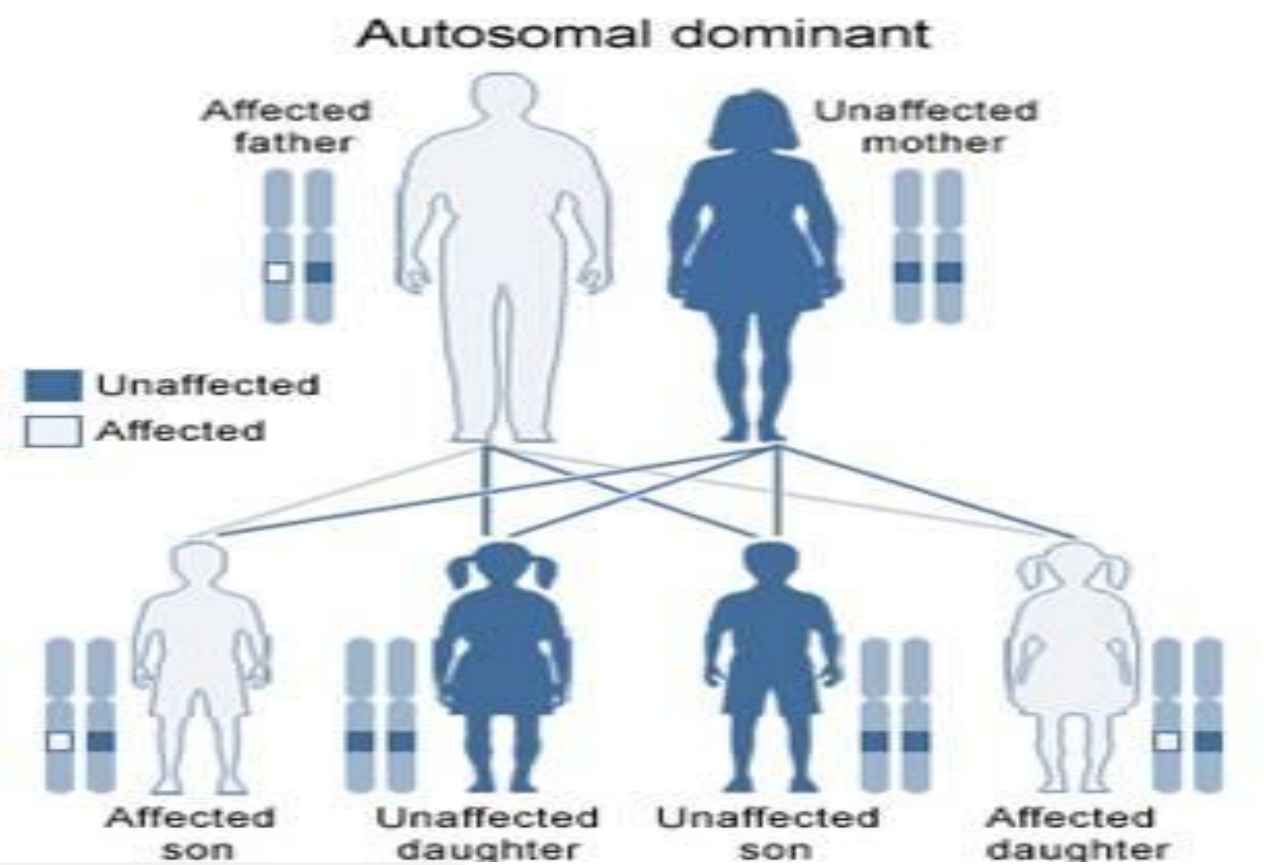
- Genetics of Huntington's disease
- Family planning options
- Things to consider

Huntington's Disease
Genetic Features

Autosomal Dominant Inheritance

- 50% risk to the children of an affected parent
- Males and females equally affected

Autosomal Dominant Inheritance



Huntington's disease
Molecular Genetic Features

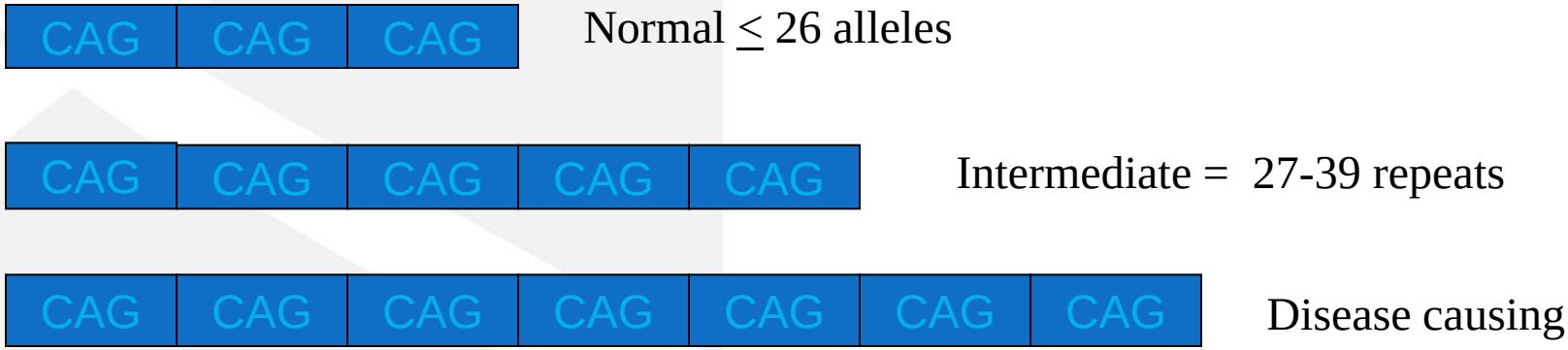
Trinucleotide repeat disorder

- CAG repeat expansion
- CAG-CAG-CAG-CAG

- Repeat count in a gene must exceed threshold to become disease causing

Huntington's Disease
Molecular Genetic Features

The genetic test will reveal the CAG count of a person's huntingtin genes



Huntington's Disease
Family Planning Options

- Decide not to have children
- The at-risk parent is tested, have children only if the test is negative
- Natural conception, no testing during pregnancy
- Prenatal testing
- Pre-implantation genetic diagnosis
- Sperm/egg donation
- Adoption

Huntington's Disease
Family Planning Options

Natural conception, no testing

- If the parent is a carrier of the gene:

  The pregnancy would have a 50% chance of inheriting the HD gene
- If the parent's gene status is unknown:

  The pregnancy would be at 25% chance of inheriting the HD gene

Prenatal testing
Direct gene testing

- Test the baby's genes for number of CAG repeats
- Highly accurate
- Can be done whether or not the at-risk parent has been tested

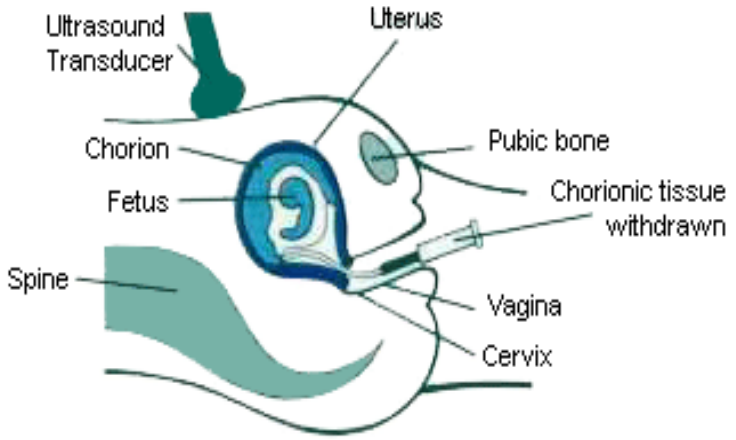
Family Planning Options
Prenatal Testing

Direct Gene Testing

- Chorionic villus sampling (CVS)
- Genetic Amniocentesis

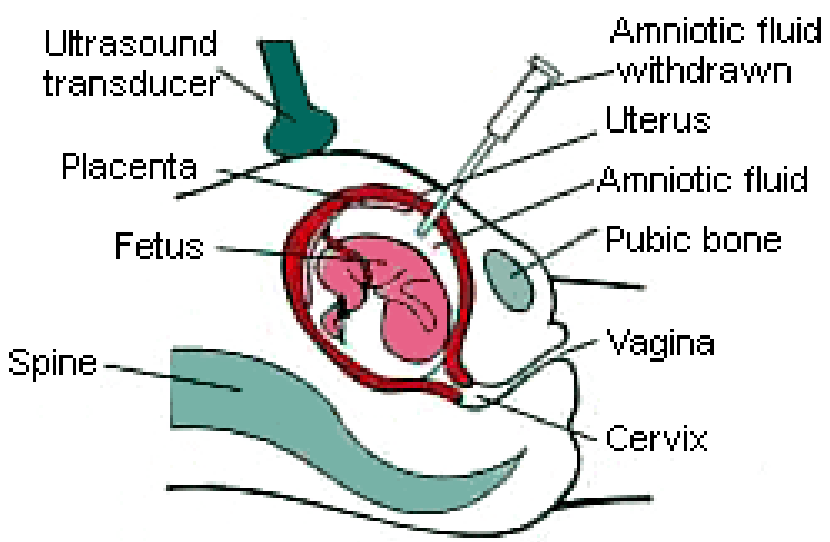
Chorionic Villus Sampling (CVS)

- 10-12 weeks of pregnancy
- Up to a 1% risk of pregnancy complications
- Results: ~ 2–3 weeks
- 98% accurate
- Cost ~$2500



Genetic Amniocentesis

- After 15 weeks of pregnancy
- Up to a 0.5% risk for pregnancy complications
- Results: ~ 2-3 weeks
- >99% accurate
- Cost ~$2000



Prenatal testing
Direct gene testing

Things to consider

- What are you going to do with the information?
- Would abortion be an option if the pregnancy is affected?
- Can you accept the risk of miscarriage associated the procedure?
- If your HD status is unknown and the pregnancy is affected then your status will be disclosed

Family Planning Options

Assisted Reproductive Technology

Pre-implantation Genetic Diagnosis (PGD)

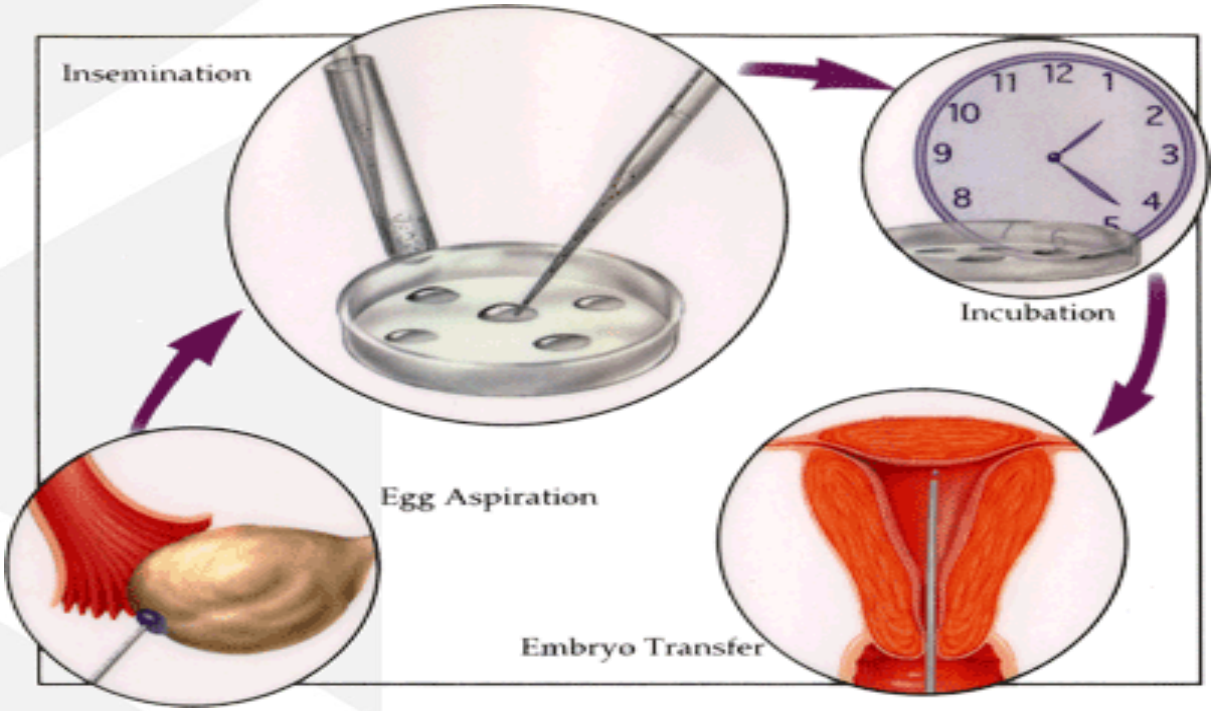
- Tests the embryos for genetic disorders before transferring them into the uterus
- Is performed in combination with in-vitro fertilization
- Offers an alternative to traditional methods of prenatal diagnosis (CVS or genetic amniocentesis)
- Can be done whether or not the at-risk parent has been tested

Assisted Reproductive Technology
Pre-implantation Genetic Diagnosis (PGD)

The Process

- Ovarian Hyperstimulation
- Egg Retrieval
- Fertilization
- Pre-implantation Genetic Diagnosis (embryo biopsy and testing)
- Embryo Transfer
- Establishment of Pregnancy

In vitro fertilization



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http://novaivf.com/images/ivf.gif

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Assisted Reproductive Technology
Non-disclosing PGD

- Have biological children that have not inherited the HD gene while not revealing the status of the at-risk parent
- The couple may not be informed of certain details of their IVF cycle

Assisted Reproductive Technology
Pre-implantation Genetic Diagnosis (PGD)

Advantages

- Opportunity to test an embryo for HD or other genetic condition prior to implantation
- Avoid abortion issue
- Increase likelihood for the baby to be disease-free (95-98%)
- Allows couples to have a biologic child without HD

Assisted Reproductive Technology
Pre-implantation Genetic Diagnosis (PGD)

Disadvantages

- Cost:
  - In vitro fertilization: $12,000 – 20,000
  - PGD: $3,500 - $5,000
- Success rate of pregnancy
  ~50% (under maternal age of 35)
- Increased chance for multiple gestation
- Prenatal testing still suggested for confirmation

Family Planning Options
Assisted Reproductive Technology (ART)

- Egg donation (at-risk or gene positive woman)
- Sperm donation (at-risk or gene positive man)

Can be done whether or not the at-risk parent has been tested

Assisted Reproductive Technology
Egg donation

- The donor undergoes ovarian hyperstimulation therapy to produce numerous eggs/cycle
- Eggs are retrieved and are fertilized with the man's sperm
- The woman is on hormone therapy to prepare for a pregnancy
- The embryos are put into the woman's womb
- Pregnancy is established
- The baby will be biologically related to it's father

Assisted Reproductive Technology
Sperm donation

- May be done either through
  - intrauterine insemination
  - in conjunction with IVF
- May need to go through a number of cycles (5 – 10) before becoming pregnant
- The baby will be biologically related to it's mother

Assisted Reproductive Technology
Sperm or egg donation

Advantages

- Eliminates the risk of HD
- Biological child to the unaffected parent
- Avoids abortion issue

Assisted Reproductive Technology
Sperm or egg donation

Disadvantages

- Uncertainty regarding donor's family history
- In vitro fertilization required for egg donation
- Success rate: < 50% with donor eggs
  10 – 20% with donor sperm (each cycle)
- Cost
  - Egg Donation - $15,000 – 25,000 per cycle
  - Sperm donation/intrauterine insemination - $300 - $500/cycle (total $1,500 – 4,000)

Family Planning Options
Adoption

- International
  - Health history is considered
  - Cost $25,000 – 45,000
  - Take about 2 years to complete adoption
- Domestic
  - Health history is considered
  - Cost is ~$20,000
  - Time to complete adoption varies

Family Planning Options
Important Questions to Consider

- What is the goal of the family? Is the couple equally motivated?
- What decision will be made if the baby is determined to have a the HD causing gene?
- Why pursue prenatal diagnosis if abortion is not an option?
- How important is the promise of treatment or cure?
- Adequate family support available?

Family Planning Options
Genetic Counseling

- Opportunity to learn more about the family planning options that are available
- Chance to discuss in detail the pros and cons of each option
- Help clarify which decision may be best for you and your family

For More Information

- For more information on HD, genetic testing and to find an approved HD testing center :
  Huntington' s Disease Society of America www.hdsa.org

- To find a local genetic counselor :
  National Society of Genetic Counselors www.nsgc.org

- To learn more about assisted reproductive techniques including PGD, sperm and egg donation:
  American Society for Reproductive Medicine www.asrm.org

- For more information about IVF, donor egg and sperm, PGD and to find providers:
  Infertility Resources for Consumers www.ihr.com