



Family Planning and Huntington's Disease: Considering Options and Making Decisions

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Huntington's Disease Society of America

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Presenter Disclosures

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

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Huntington's Disease
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Overview

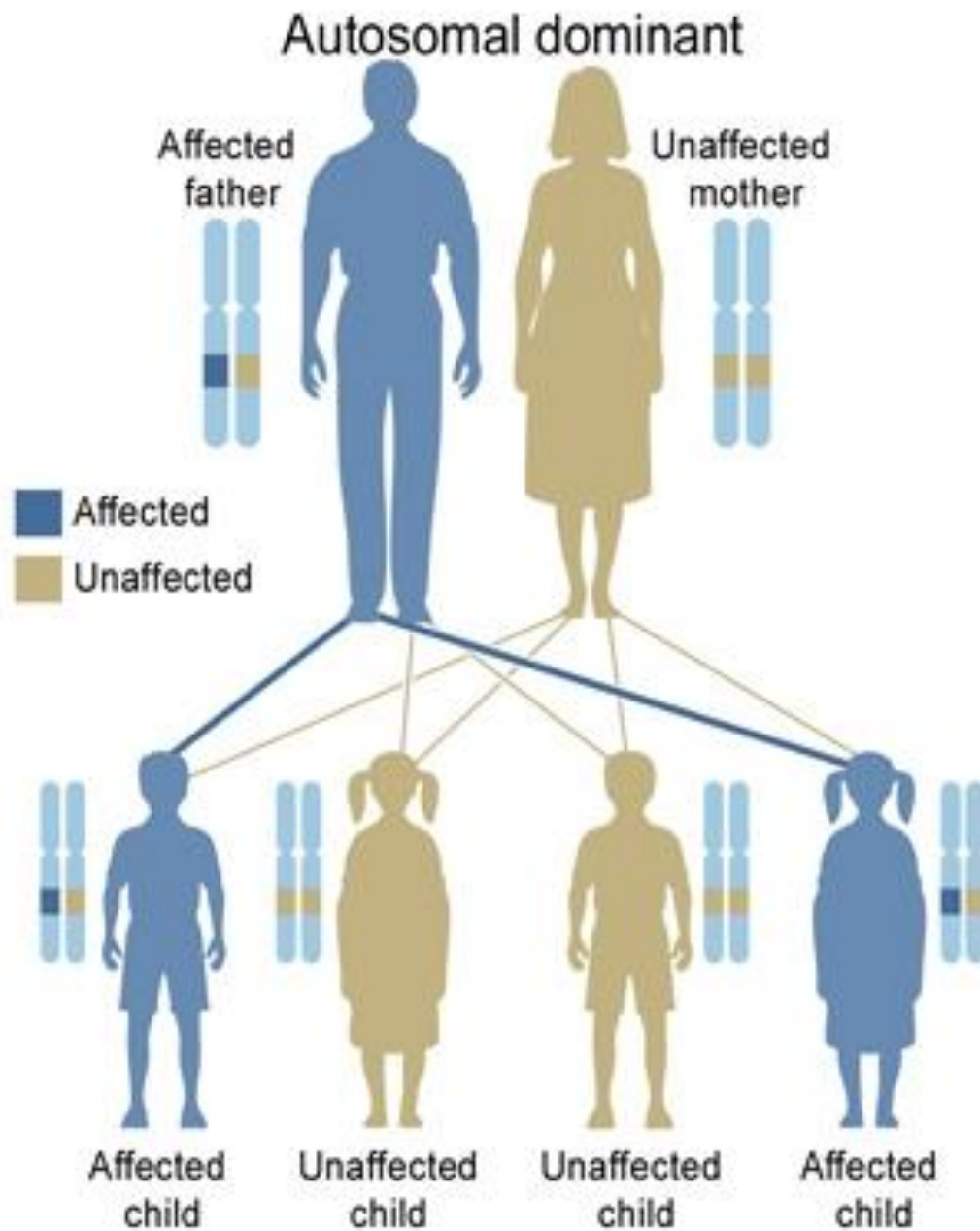
- Family planning and HD
- Brief Review of HD Genetics
- Approaching Family Planning Decisions
- Overview Family Planning Options
- Resources

Family Planning and HD

- Family planning: Planning if, how and when to have children
- Family planning and HD
 - Explores options for having children that modify the risk of passing HD onto children
 - Involves making personal decisions based on individual desires, beliefs, and circumstances

What are the genetic risks from HD and where do they come from?

HD Genetics



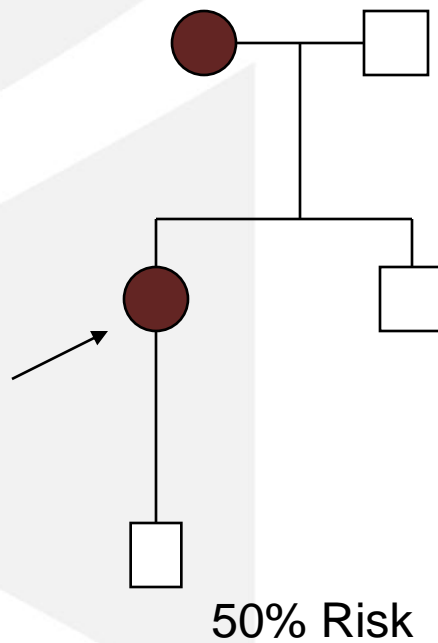
CAG Repeat Expansion

TAC--TTA--TAG--GAG--GTA--ATA—TAT--GCC--CCT--
GGT--CAG—TAC-TTA-TAG-GAG-GTA-ATA-TAT-GCC-
CCT-GGT-CAG--TTA--TAT—CAG—CAG—CAG—
CAG—CAG—CAG—CAG—CAG—CAG—CAG—
CAG—CAG—CAG—CAG—CAG--TAC-TTA-TAG-
GAG-GTA-ATA-TAT-GCC-CCT-GGT-CAG-TAG-CGT-
TAC-TTA-TAG-GAG-GTA-ATA-TAT-GCC-CCT-GGT-
CAG-TAG-CGT-TAC-TTA-TAG-GAG-GTA-ATA-TAT-
GCC-CCT-GGT-CAG-TAG-CGT

CAG Repeat Sizes and HD

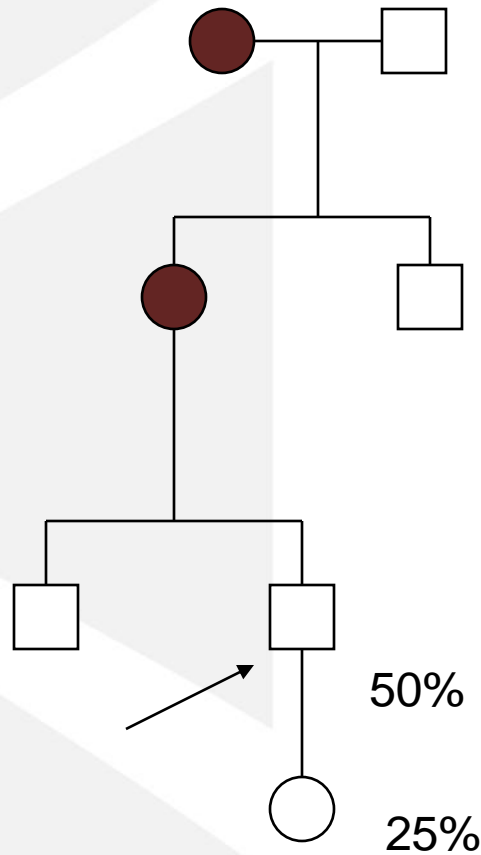
- 26 or Less CAG Repeats – No Risk for HD
- 27-35 CAG Repeats – Intermediate Repeats
- 36-39 CAG Repeats- Reduced Penetrance
- 40 or Higher CAG Repeats – HD

Risk Situations -Children at 50% Risk



● = HD Diagnosis
or + HD Testing

Risk Situations –Children at 25% Risk



● = HD Diagnosis
or + HD Testing

Family Planning Decisions: Factors to Consider

- Gene status
- Desire for biological vs. non-biological children
- Costs of each option
- Insurance: What does it cover? Do you have enough?
- Physical readiness and health

Family Planning Decisions: Factors to Consider

- Emotional readiness
- Moral/ethical issues
- Partner's wishes/concerns
- Support: family, community, medical
- Time frame

Adoption

Conceiving naturally

Deciding not to have children

Prenatal testing



Egg/Sperm/Embryo Donation

Preimplantation Genetic Diagnosis

Deciding Against Having Children: Factors to Consider

- Only the individual or couple can decide
- May consider other factors
 - Desire to have children
 - Lifestyle
 - Financial stability
 - Support from family/friends/community
 - Health including fertility issues
- Feelings about decision may evolve over time

Domestic Adoption: Adoption with the United States

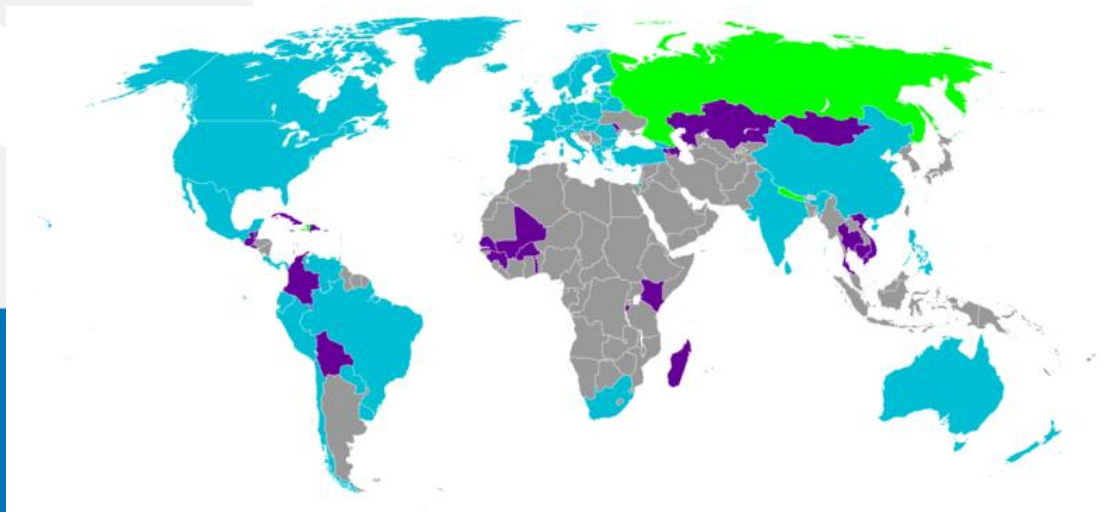
- Types of Domestic Adoption
 - Adoption of Relative
 - Public Agency/Foster Care System
 - Licensed Private Agency
 - Independent
 - Facilitated/Unlicensed

Domestic Adoption: Adoption with the United States

- May have access to child's history/medical background
- Openness – possible relationship with child's biological family
- Waiting times may be long
- Laws/options vary by state

International Adoption

- Children often raised in orphanages/institutional settings
- Limited access to history/medical information
- Placement process varies by country of origins
- Hague Adoption Convention: international convention dealing with international adoption



Range of Adoption Costs

- Public Agency (Foster Care) Adoptions \$0 - \$2,500
- Licensed Private Agency Adoptions \$5,000 - \$40,000+
- Independent Adoptions \$8,000 - \$40,000+
- Facilitated/Unlicensed Adoptions \$5,000 - \$40,000+
- Intercountry Adoptions \$15,000 - \$30,000

www.childwarefare.com

Adoption: Factors to Consider

- Providing a home to child in need
- No biological relationship to child
- Option for individuals/couples with infertility

Adoption: Factors to Consider

- Varying access to child's history/medical background
- Waiting time may be long
- Costs vary depending on type of adoption
- Agencies may consider family history of HD in assessment

Adoption: Where to Go for More Information

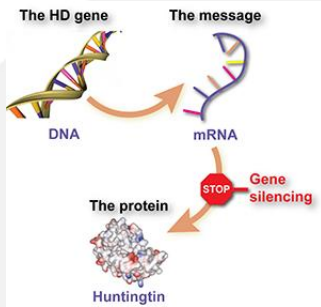
- US Department of Health and Human Services: Administration for Children and Families (www.childwelfare.gov)
- US State Department
- State Child Welfare Agencies
- Attorney specializing in adoption
- Other HD Families

Having Children without Genetic Testing

- Child biologically related to both parents
- Accepts the genetic risks
 - Hope that child will not inherit gene expansion
 - Hope that a cure will be found in child's lifetime
 - One can have a good life with HD
- Knowledge of parent's gene status not necessary

Having Children without Genetic Testing

- Avoids risks, costs, limitations of assisted reproductive technologies and prenatal testing
- Time – Can be achieved relatively quickly
- May result in anxiety/guilt over child's gene status



<http://en.hdbuzz.net/122>



2CARE
Huntington Study Group



Research



PREDICT-HD
Huntington Study Group

CREST-EHDSTUDY



Huntington Study Group



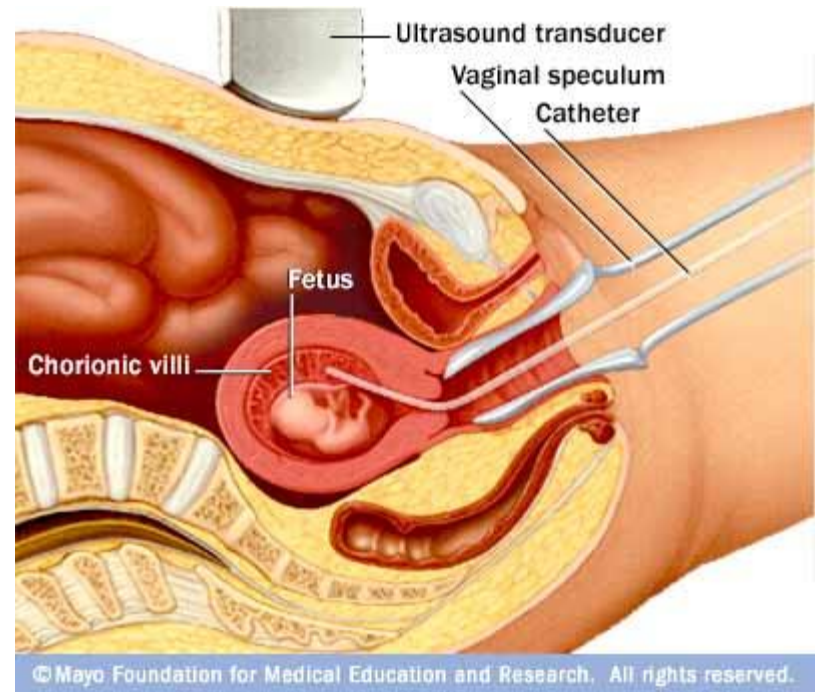
Huntington Study Group

Prenatal Testing for HD: What Is It?

- Genetically testing a fetus for a HD
- May require genetic samples from both parents/additional family members for optimal results
- Involves invasive procedures to obtain samples from fetus for testing:
 - Chorionic villus sampling (CVS)
 - Amniocentesis

Prenatal Testing: Chorionic Villus Sampling (CVS)

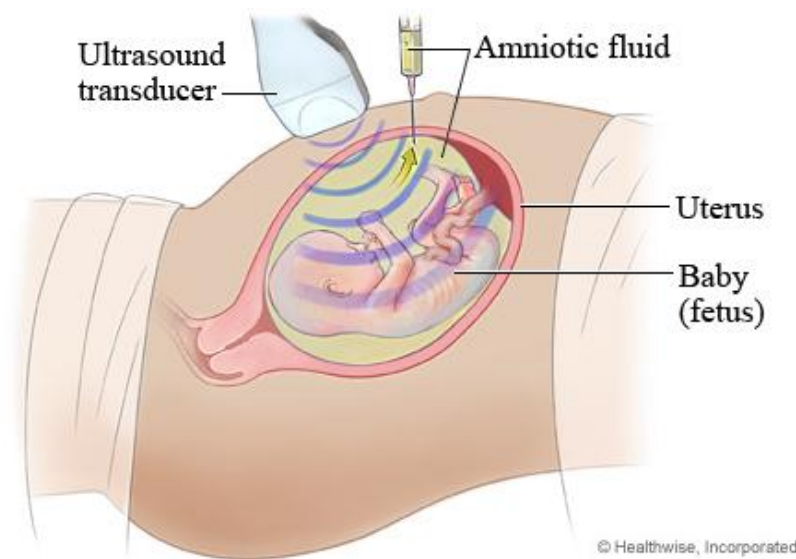
- 10th -13th week of pregnancy
- Testing on placenta
- Risk of Miscarriage: 1:100*



*US CDC MMWR Recommendations and Reports (1995).

Prenatal Testing: Amniocentesis

- Beginning in 15th week of pregnancy
- Testing performed on fetal cells from amniotic fluid
- Risk of miscarriage: 1:500-1:300*

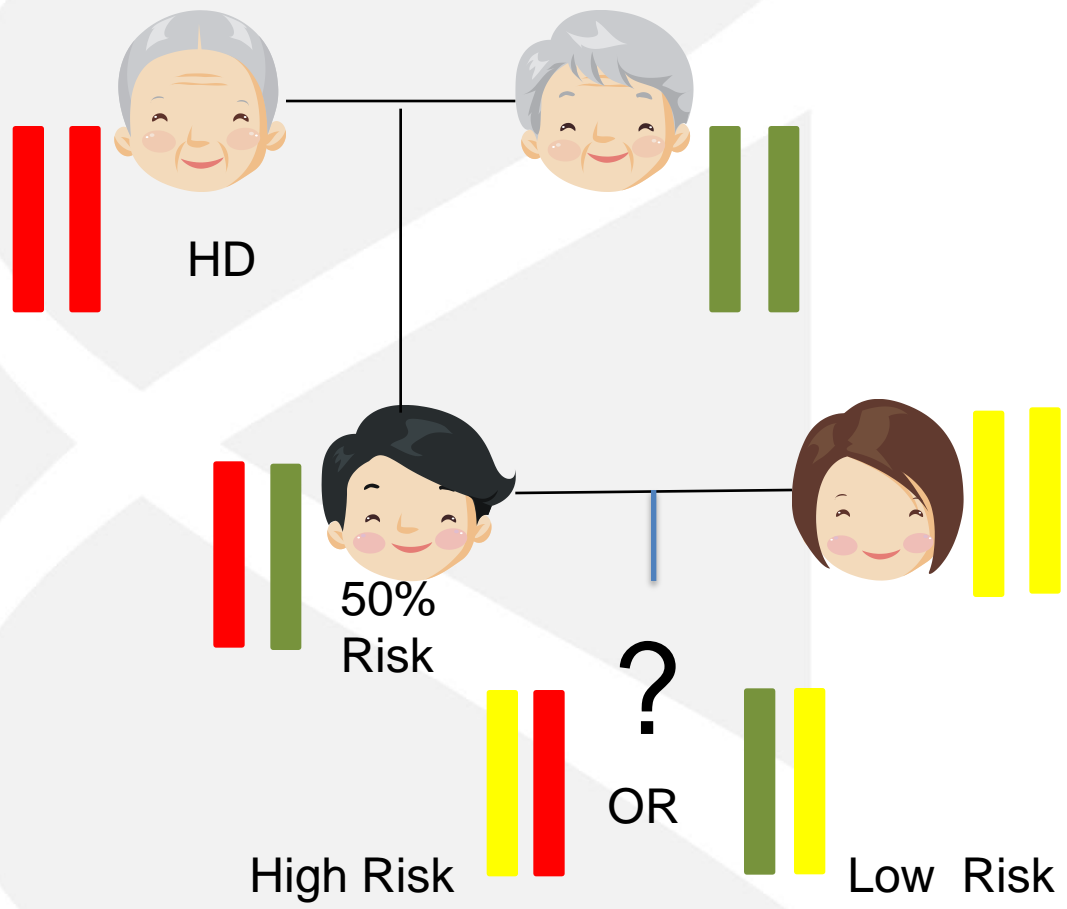




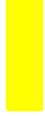
*US CDC MMWR Recommendations and Reports (1995).

Prenatal Testing

- Genetic testing performed on cells from CVS or amniocentesis
 - Direct DNA testing
 - Indirect testing (Exclusion testing)

Exclusion Testing



-  =Possible HD Gene
-  =Normal Gene
-  =Normal Gene

Prenatal Testing: Direct DNA vs. Exclusion Testing

Direct DNA Testing

- Determines gene status of pregnancy
- May reveal gene status of at-risk parent
- Decisions about terminating gene positive pregnancy

Exclusion Testing

- Determines risk status of pregnancy
- Hides gene status of at-risk parent
- Decisions about terminating a potentially gene negative pregnancy

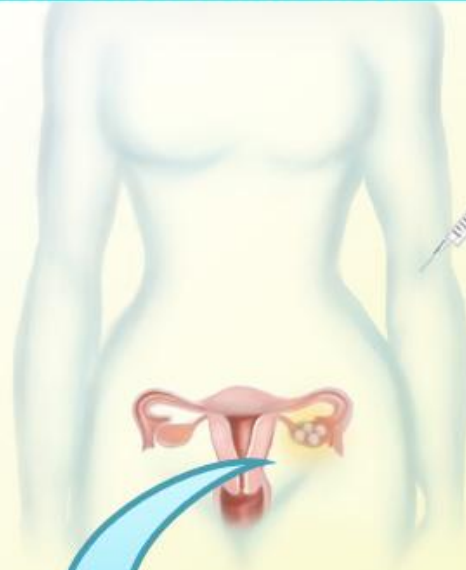
Prenatal Testing: Factors to Consider

- Option for unplanned pregnancy
- May not require testing of at-risk parent (exclusion testing only)
- Insurance may not cover the cost
- Testing is time sensitive – decisions must be made quickly

Prenatal Testing: Factors to Consider

- Risk of procedures
- Involves decisions regarding termination
- May results in the unintentional presymptomatic testing of the baby
- Can have a high emotional impact

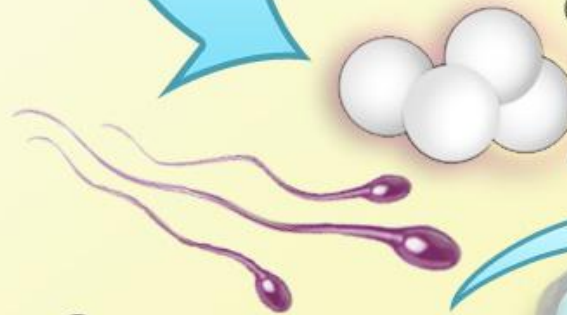
IN-VITRO FERTILIZATION



1 The doctors give the women hormones, causing her to superovulate - that is, to develop and release many eggs at once.



2 Once the hormone-treated woman has many mature eggs, the doctors surgically remove them from her ovary (the organ where eggs mature).

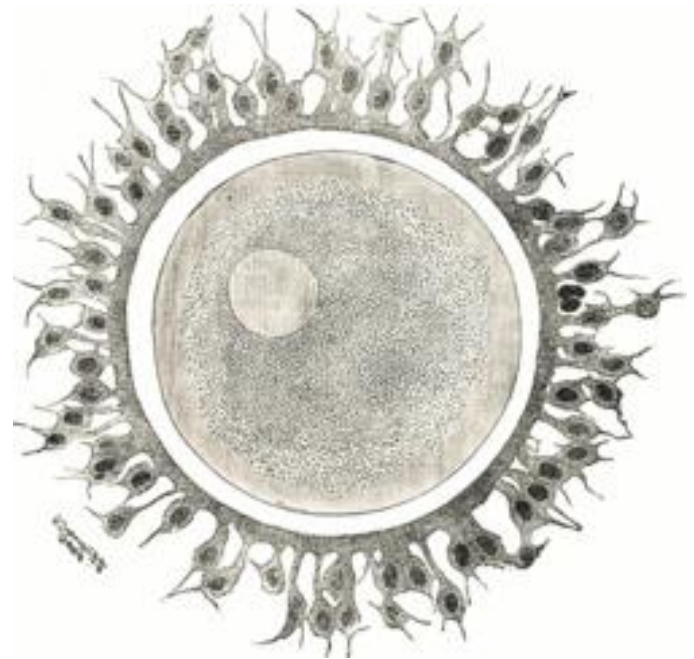


3 Once removed, the eggs can be combined with the man's sperm in a laboratory dish (this is the in-vitro part). The process of a sperm joining an egg is called fertilization.



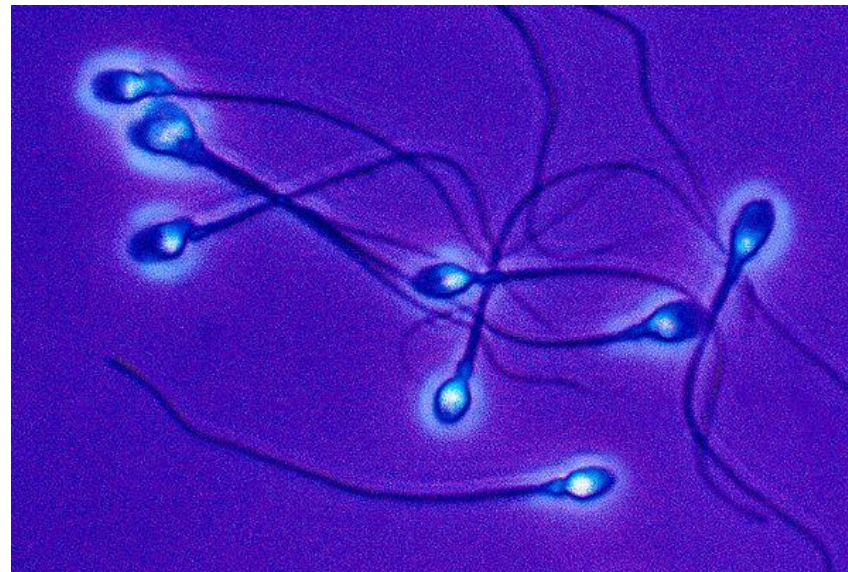
Egg, Sperm & Embryo Donation

- Egg Donation
 - Donated egg can be used when mother has or is at risk for HD
 - Egg is donated anonymously or by friend/or relative



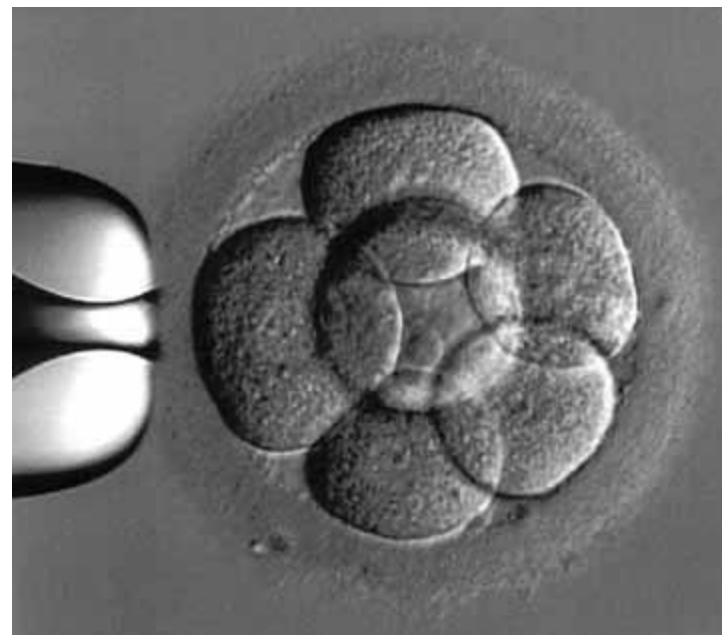
Egg, Sperm & Embryo Donation

- Sperm Donation
 - Donated sperm can be used when father has or is at risk for HD
 - Sperm is donated anonymously or by friend/relative



Egg, Sperm & Embryo Donation

- Embryo Donation
 - Donated embryo can be used with either parent is at risk for or has HD
 - Embryo donated from couple with embryos remaining after completing IVF



Egg, Sperm, Embryo Donation: Factors to Consider

- Eliminates risk of HD in child by not using genetic material from at risk parent
- Embryo donation puts potentially unused embryos to use
- One or both parents will not be biologically related to child
- Cost (~\$15, 000 to \$20, 000 for use of donor egg)

Egg, Sperm, Embryo Donation: Factors to Consider

- May result in birth or multiples (twins, triplets etc)
- Success rates may vary (up to 55% of embryo transfers using donor egg may result in birth of child*)

*Society for Reproductive Technology www.sart.org

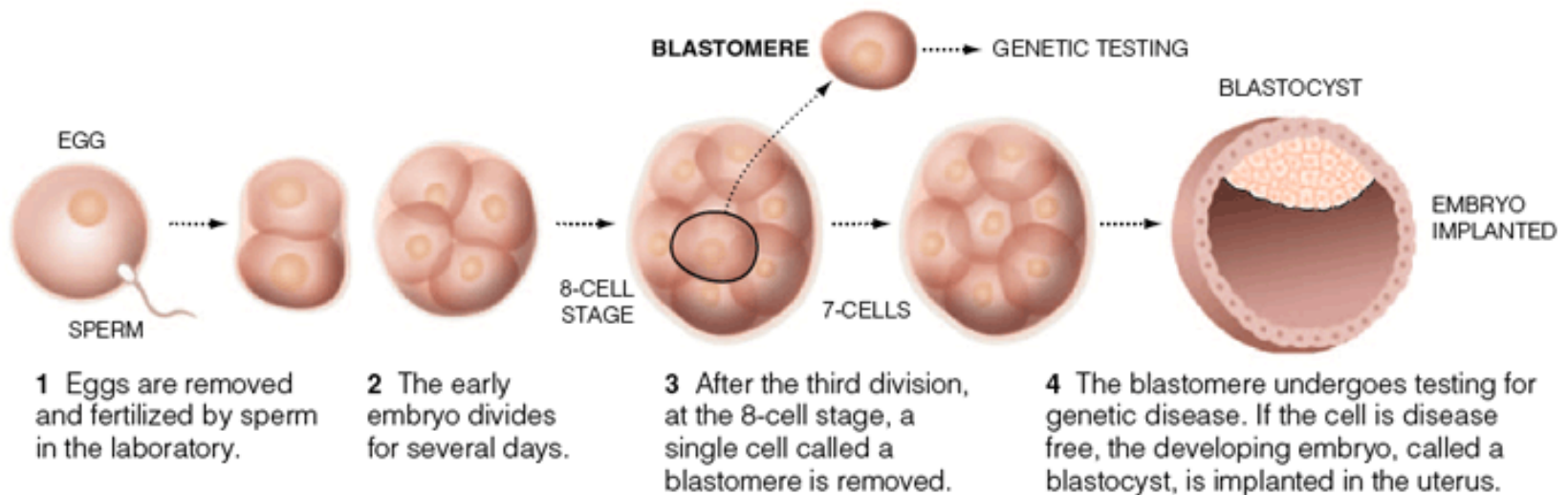
Preimplantation Genetic Diagnosis: What Is It?

- Genetically testing embryo for HD before it is implanted in uterus
- In vitro fertilization required to create embryos that are then tested for HD
- HD testing for at risk parent is not necessary: PGD can be performed so that gene status of an at-risk parent remains hidden

Preimplantation Genetic Diagnosis

Genetic Testing of Embryos

Doctors can now test embryos for genetic disorders and gender before implantation in the uterus.



New York Times 2005

Preimplantation Diagnosis: HD Testing of Parent Not Required

- Exclusion testing
- Direct testing with non-disclosure
 - Embryos tested directly for CAG repeat expansion but results not revealed
 - Additional information may be kept from parents in order to hide HD gene status (ex. # of viable embryos created etc)
 - Test results will be known to select staff at fertility clinic and laboratory

Preimplantation Diagnosis: Factors To Consider

- Avoid HD risk to children by testing embryos before pregnancy
- Children will be biologically related to both parents
- PGD may be an option for couples with infertility
- Cost (~\$15,000) not often covered by insurance

Preimplantation Diagnosis: Factors To Consider

- Time – several months
- Success rates (birth of baby) similar to those with IVF
 - Per egg retrieval ~22%*
 - Per embryo transfer ~29%*
- Requires IVF procedures and all the associated risks
- Concern about what is done with unused embryos

*Harper et al (2012)

Family Planning: Next Steps

- Seek genetic counseling in your area
- Get input from spouse/family/friends/HD community members
- Talk with your personal physician
- Assess your health/emotional readiness
- Contact a fertility clinic if you are considering PGD or other assisted reproductive option
- Financial planning

Planning for the Care of Child When a Parent Develops HD

- Build a support network: family members, community members, health care providers, mental health care providers
- Discuss plans for care of child with your partner or spouse
- Seek advice on talking to children about HD
- Financial Planning
 - Talk to an expert about estate planning
 - Locate/review your resources for outside care
 - Assess your insurance coverage

Resources

- General HD Information
 - Huntington's Disease Society of America
 - hdsa.org
 - HD Buzz
 - en.hdbuzz.net
 - Testing for Huntington's Disease: Making An Informed Choice
 - <http://depts.washington.edu/neurogen/downloads/hungtinton.pdf>
 - Huntington Study Group
 - www.huntington-study-group.org

Resources

- Adoption - Domestic
 - US Dept of Health and Human Services/Child Welfare Information Gateway
 - www.childwelfare.gov
 - AdoptUsKids
 - www.adoptuskids.org
 - National Foster Care & Adoption Directory
 - www.childwelfare.gov/nfcad
 - State Child Welfare Agencies

Resources

- Adoption - International
 - American Academy of Pediatrics (Pediatricians with special interest in adoption)
 - www.aap.org/sections/adoption/SOAFCAAdoptionDirectory2.pdf
 - The US State Department
 - adoption.state.gov
- Egg/Sperm/Embryo Donation
 - Society for Assisted Reproductive Technology
 - www.sart.org

Resources

- Genetic Counseling
 - National Society of Genetic Counselors
 - nsgc.org
- Prenatal Testing
 - March of Dimes
 - www.marchofdimes.com
 - Mayo Clinic
 - Amniocentesis (www.mayoclinic.com/health/amniocentesis/MY00155)
 - CVS (www.mayoclinic.com/health/chorionic-villus-sampling/MY00154)

Resources

- Preimplantation Genetic Diagnosis
 - Genesis Genetics - <http://www.genesisgenetics.org/>
 - Centre for Genetics Information. Fact Sheet: Preimplantation Genetic Diagnosis. www.genetics.edu.au/FS18.pdf
- Children and HD
 - Hennig, Bonnie. Talking to Kids About HD. 2004.
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<http://onlinelibrary.wiley.com/doi/10.1111/cge.12089/full> June 15, 2013.
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