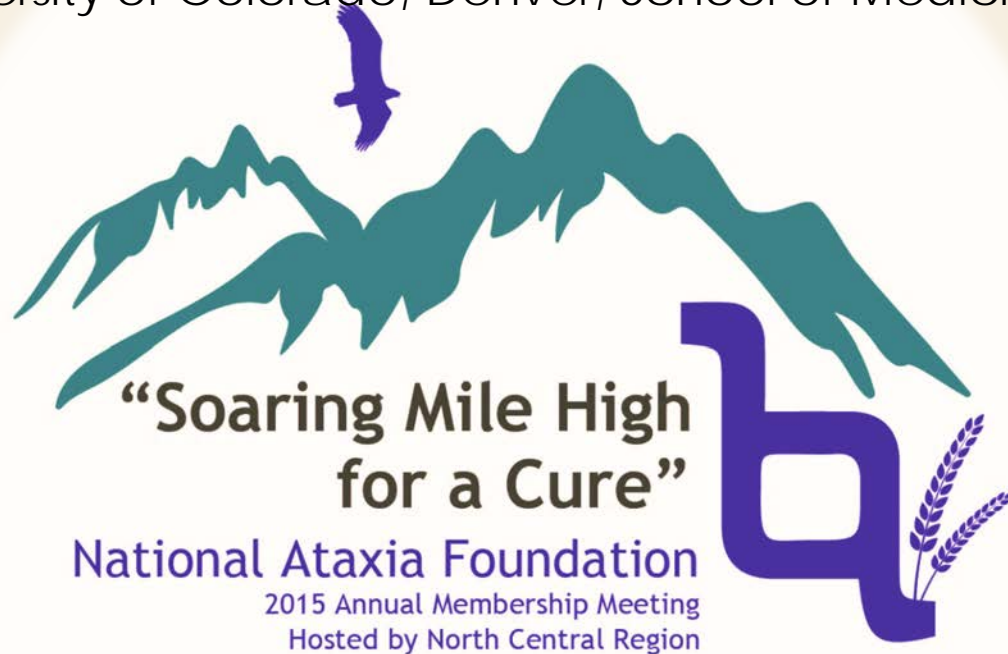


# Implications of Genetic Testing on Families with Ataxia

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# Disclaimer

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# Presenter Disclosures

- Melissa Gibbons
- No relationships to disclose or list



# Overview



- Who are genetic counselors
- Who should be offered genetic counseling
- Types of genetic testing
- To Test or Not Test
- Hurdles to Testing
- Discrimination?
- Now What? What Happens After the Results Arrive
- Talking Genetics With the Family



# Genetic Counselors

- Graduates of a Masters in Genetic Counseling Program
  - 2 year program
  - Clinical rotations in prenatal, pediatric, adult, and cancer
  - Coursework in both science and psychosocial
- Certified Genetic Counselors
  - Have graduated from an accredited program
  - Have completed supervised counseling sessions
  - Have passed a board examination and maintained their certification through continuing education
- What do Genetic Counselors do?
  - Genetic counselors help patients and families understand complex issues and support their emotional needs.



# Common reasons to see a Genetic Counselor

- ▶ I have a family history of a certain health condition, is there a genetic test I can take to find out if I'm at risk?
- ▶ My partner and I are planning a pregnancy, what types of testing are available to us?
- ▶ I am having symptoms should I get tested?
- ▶ I have a medical condition and want to learn whether I could pass it to my children.
- ▶ Can you help me share my genetic information with my relatives, or with my doctors?
- ▶ My child has a developmental or medical history that is concerning for a genetic condition



# The Right Test for the Right Patient at the Right Time



# Types of Genetic Testing

- **Diagnostic testing** involves identifying current disease states.
- **Carrier testing** determines whether an individual carries a certain genetic trait.
- **Predictive testing**, is used to determine whether a person has a genetic mutation that will lead to a late onset disorder or make them more susceptible to a conditions.





# Types of Genetic Testing

- **Prenatal Testing** offered during pregnancy to help identify fetuses with certain diseases.
- **Preimplantation Genetic Testing** is a screening test used to determine if a genetic disorder is present in an embryo
- **Newborn Screening** used to test babies soon after birth for treatable conditions that require immediate intervention

# Why Get Testing?

Might explain the symptoms I am having

*Family planning*

A diagnosis may lead to better treatment

Information for other members of the family

I just want to know

*Financial Planning*

**Control**

The results could alter my Retirement/Travel Plans

*Access to Services*

**Answers**

Research Opportunities

# Why Not To Get Testing?

Might raises more questions than answers

*Family planning*

A diagnosis may lead to discrimination

other family members do not want to know

I am not sure I want to know

*Financial Planning*

**Lack of Control**

The results could alter my Retirement/Travel Plans

*Guilt over passing on a faulty gene*

# What About Discrimination

- ▶ The Genetic Information Nondiscrimination Act of 2008 (GINA)- a federal law that protects people from genetic discrimination in health insurance and employment
  - ▶ The genetic information protected by the law includes family health history, the results of genetic tests, the use of genetic counseling and other genetic services, and participation in genetic research.
  - ▶ GINA makes it against the law for health insurers to request, require or use genetic information to make decisions about:
    - ▶ Your eligibility for health insurance
    - ▶ Your health insurance premium, contribution amounts or coverage terms
    - ▶ Consider family health history or a genetic test result as a pre-existing condition
    - ▶ Ask or require that you have a genetic test
    - ▶ Use any genetic information they do have to discriminate against you, even if they did not mean to collect it
- ▶ GINA does not cover life insurance, long-term care or disability



# What About Discrimination

- ▶ The Genetic Information Nondiscrimination Act of 2008 (GINA)- a federal law that protects people from genetic discrimination in health insurance and employment
  - ▶ GINA makes it against the law for employers to use your genetic information in the following ways:
    - ▶ To make decisions about hiring, firing, promotion, pay, privileges or terms
    - ▶ To limit, segregate, classify, or otherwise mistreat an employee
  - ▶ It is also against the law for an employer to request, require or purchase the genetic information of a potential, current employee or his/her family member
  - ▶ GINA applies to all employers with 15 or more employees, regardless if it is a not-for-profit organization or a corporation.

# Hurdles On the Path to Genetic Testing



1

2

3

# Now What?

*“While the testing is simple, dealing with the results has been traumatic”*

*“It gave us a chance to learn about the syndrome, connect with people like us, and gain an increased ability to deal with day to day issues”*

*“Knowing this has allowed me to take control of my life”*

*“The problem is that at present , there is no cure...so we have to face the problem without having a chance to solve it completely”*



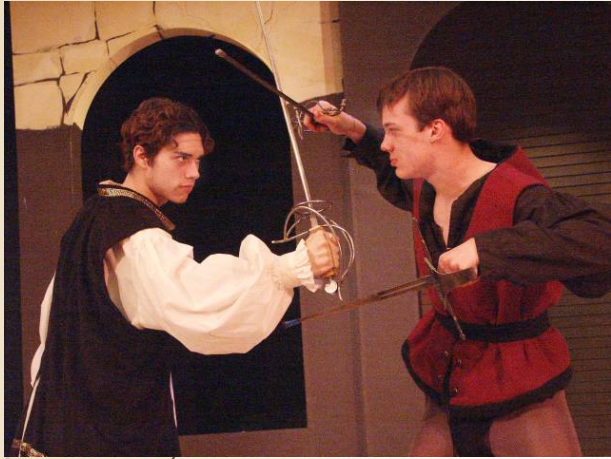
Excerpts from Genetic Testing Stories- Genetic Alliance



# A Family Affair



# Sharing Genetic Results



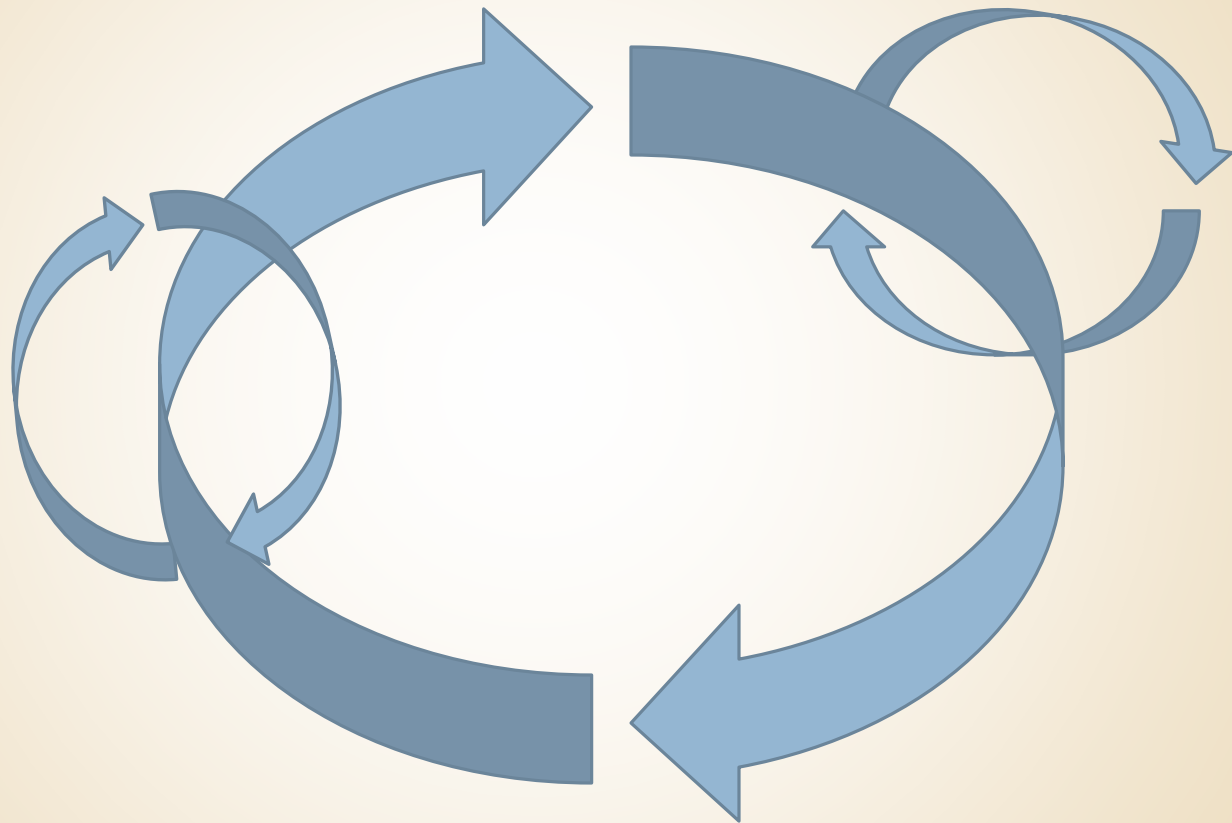
**Family Dynamics**



**Family Member(s) do not want to know**



# Impact on Family Members



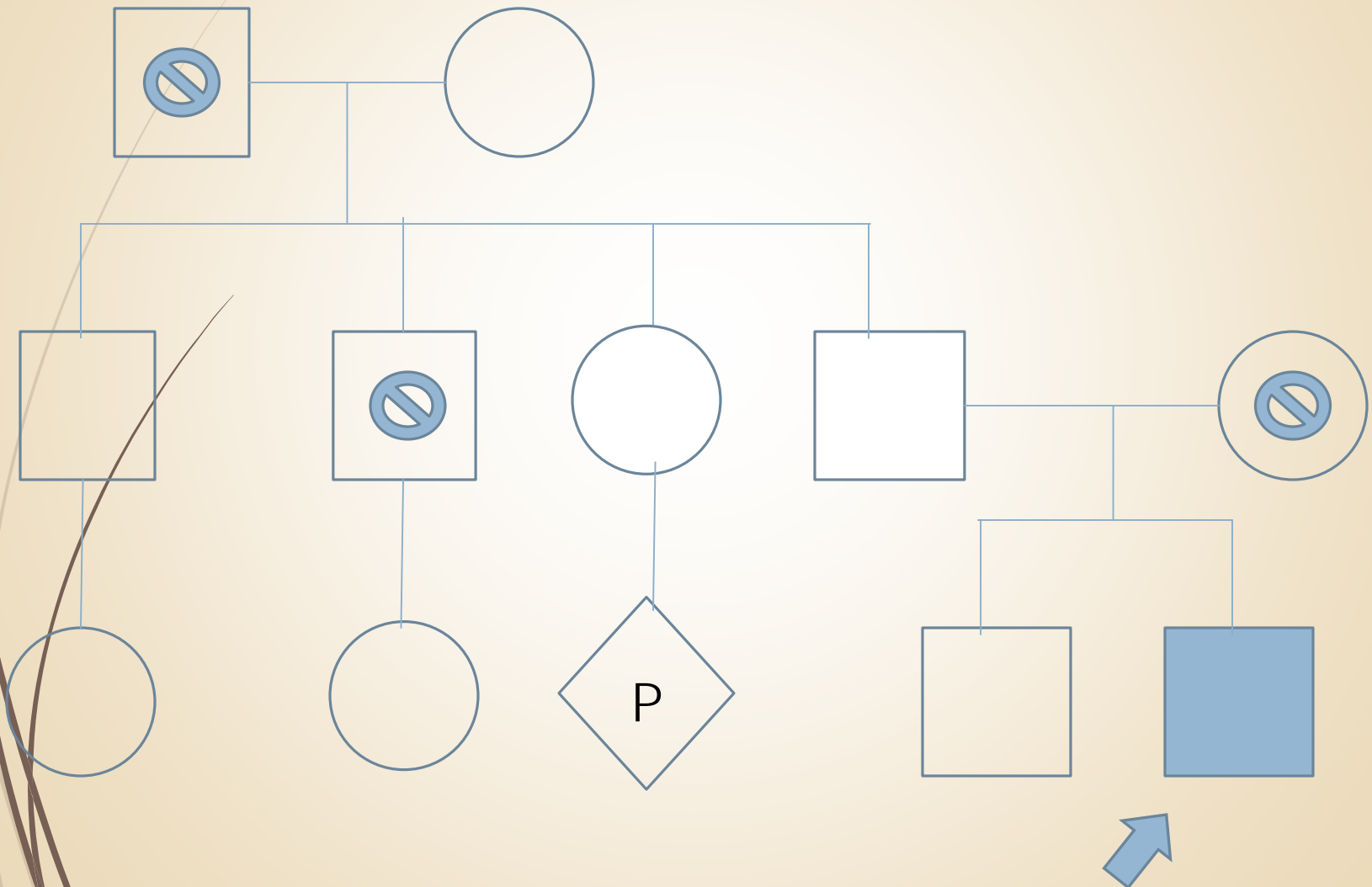


# Impact of Family Planning

## Reproductive Options

- ▶ Natural Conception
- ▶ Gamete (Sperm or Egg Donation)
- ▶ Preimplantation Genetic Diagnosis with IVF
- ▶ Prenatal Diagnosis
  - ▶ Chorionic Villus Sampling (CVS)
  - ▶ Amniocentesis
- ▶ Adoption

# Impact on Family Dynamics





# Resources

- ▶ Find a Genetic Counselor at [NSGC.ORG](http://NSGC.ORG)
- ▶ Find out more about GINA at [GINAHELP.ORG](http://GINAHELP.ORG)
- ▶ Does it Run in My Family Toolkit, Understanding Genetic and How To Talk to My Family about My Genetic Condition - [GENETICALLIANCE.ORG](http://GENETICALLIANCE.ORG)
- ▶ To learn more about all the ways that genetic is part of your life visit [GENESINLIFE.ORG](http://GENESINLIFE.ORG)