NFXF MasterClass™ Genetics of Fragile X

Date: November 30, 2023
Time: 3:00 pm – 5:00pm ET
Location: Virtual Conference via Zoom
With recording for on-demand access
Focus:

This course is an overview of the genetics of Fragile X: the inheritance, indications for testing, the potential impact of the premutation and full mutation results, and resources to share with families.

3:00- 3:05 Hilary Rosselot
Introduction to the NFXF and NFXF MasterClass™

This presentation will provide an introduction to the NFXF, and its Mission, and share how this course is a part of our Strategic Priorities. Each presenter will receive a brief introduction.

1. Discuss the Mission of the NFXF
2. Define the role of the NFXF in the Fragile X community.
3. Introduce the course and how it relates to the NFXF Strategic Priorities.

3:05 – 3:35 Susan Howell
Introduction to Fragile X Genetics

This presentation will discuss the timeline of identifying the *FMR1* gene expansion. Discussing the recent nomenclature change. The talk will provide an overview of the complex inheritance, factors that influence the inter-generational expansions, indications for screening and diagnostic testing and briefly review the categorization of CGG repeat results.

Learning objectives

1. Review the history/timeline of identifying the *FMR1* gene expansion.
2. Recognize factors that influence the intergenerational expansion of the *FMR1* gene.
3. Summarize the categorization of the CGG repeat ranges.

3:35- 3:50 Susan Howell
FMR1-associated disorders: Fragile X syndrome (FXS)

This presentation will discuss the full mutation (>200 CGG repeats), methylation, mosaicism, and the relationship between FXS and autism spectrum disorder. This talk will also discuss common symptoms and the variability between males and females.

Learning objectives

1. Describe Fragile X syndrome and corresponding CGG repeat range.
2. Discuss the effect of genetic counseling on identifying inter-generational family members who may be appropriate for genetic testing.

3:50- 4:00 Break

4:00- 4:30 Matthew Walsh
FMR1-associated disorders – Fragile X premutation

This presentation will discuss the corresponding CGG repeat ranges, prevalence, and associated risks associated with individuals with a Fragile X premutation. This talk will also discuss currently identified disorders (Fragile X-associated Primary Ovarian Insufficiency (FXPOI) and Fragile X-associated tremor/ataxia syndrome (FXTAS)), prevalence, associated symptoms, treatment, and reproductive options.

Learning objectives

1. Review the identified associated conditions and disorders related to the Fragile X premutation: Fragile X-associated primary ovarian insufficiency (FXPOI) and Fragile X-associated tremor/ataxia syndrome (FXTAS) and corresponding CGG repeat ranges.
2. Discuss the effect of genetic counseling on identifying inter-generational family members who may be appropriate for genetic testing.
3. Explain the clinical and reproductive implications of *FMR1* expansions.

4:30- 5:00 Susan Howell and Matthew Walsh
Genetic Counseling Considerations and Challenges
This presentation will discuss advancements in testing and identification for an *FMR1*-associated condition or disorder. This talk aims to discuss considerations, recommendations for cascade testing, and appropriate referrals and resources. The presentation will be followed by a 15-minute Q&A.

Learning objectives

1. Identify barriers and challenges in providing genetic counseling to newly diagnosed patients
2. Describe psychosocial and ethical considerations related to testing for FMR1 expansions.
3. Review and answer attendee questions.

5:00- 5:10 Hilary Rosselot

Resources and Research for FMR1-associated Conditions and Disorders
This presentation will discuss currently available resources for families and professionals. This talk will also discuss the role of research in developing new treatments, interventions, and methods for diagnosing or detecting disease.

Learning objectives

1. Summarize the resources and referrals available to patients impacted by an *FMR1* expansion.
2. Discuss the ongoing research opportunities and programs available for families and professionals.

This course was presented by:

NATIONAL **FRAGILE X** FOUNDATION