basic training

In Reproductive Medical Genetics

Preparing Professionals for Practice

Summary Report of the Inaugural Course

Academic Year 2020-2021

https://mfprecision.ucsf.edu/basic-training/

BASIC TRAINING
A COURSE IN REPRODUCTIVE MEDICAL GENETICS

A couple of years ago, we were discussing the increasing importance of genetics in perinatal medicine, and the challenges faced by many fellowship programs in providing comprehensive education and training in this complex and rapidly evolving field. We conceived the idea of an intensive short course in reproductive genetics and teamed up with two outstanding genetic counselors to produce a one-week program that took place in the New York Genome Center in July 2019. While the initial Reproductive Genetics "Bootcamp" was a great success, we had to pivot in 2020 when the pandemic altered our world.

We committed to developing an 8-week virtual course that could be a resource for the two month genetics rotation now required by the American Board of Obstetrics and Gynecology. The vision of "Basic Training" was to build knowledge and skills in genetics through a curriculum that started with the basics of genetics, evolved into implementation of clinical testing, and included content on important social and policy issues. The course included lectures, case presentations, interactive live workshops, and supplemental readings. Every week included quiz questions, office hours with the genetic counselors to answer questions, and ended with "rounds with the geneticists" to review concepts, quiz questions and interesting and complicated patients.

The 4-week fall curriculum covered basic genetics from underlying principles to gene structure and function, cytogenetics, and molecular laboratory techniques. In the 4-week spring curriculum, we built on these basics and delved into prenatal screening and diagnostics, maternal genetic disorders, ethical, legal and social issues, and adult and complex diseases. The attached report summarizes the curriculum, the pretest and posttest scores, and feedback from the fellows who participated.

The course would not have been possible without the support of our primary sponsor, Illumina, and other genetic laboratories including Integrated Genetics, Natera, Myriad Women's Health, Invitae, Fulgent Genetics and Eurofins. We are especially grateful for the administrative support of the Perinatal Quality Foundation, the tremendous faculty and genetic counselors who generously donated their time, and the support of the MFM program directors who allowed their fellows to participate.

We are very proud of the overall results, and believe the course was a tremendous success – we hope you agree. Moving forward we plan to offer Basic Training, A Course in Reproductive Medical Genetics for the 2021-2022 academic year and welcome the opportunity to share this education.

Basic Training 2021-2022 Weeks 1-4 October 4-29, 2021 Weeks 5-8 February 28-March 25, 2022

Mary Norton, MD and Ron Wapner, MD
Basic Training Course Directors
May 1, 2021

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Course Overview

Week 1 Principles of Genetics

Week 2 Gene Structure and Function

Week 3 Cytogenetic Laboratory Techniques

Week 4 Molecular Laboratory Techniques

Week 5 Prenatal Screening and Diagnostics I

Week 6 Prenatal Screening and Diagnostics II

Week 7 Genetics Puttanesca

Week 8 Adult and Complex Disease

A Sampling of Topics

Each week, the course built knowledge and skills by laying foundation through lecture, case presentations, supplemental reading, quiz questions, interactive LIVE workshops and rounds. Here's a sample of topics:

Genetic variants, mechanisms of disease, mitochondrial inheritance, germline and somatic mosaicism, trinucleotide repeats, complex inheritance, pedigree and risk analysis, imprinting, epigenetics, nondisjunction, structural rearrangements, microdeletions, microduplications, karyotypes, FISH, microarray, variant classification, dysmorphology, Sanger and next generation sequencing technologies, whole exome and whole genome sequencing, prioritizing variants by phenotype, noninvasive prenatal screening, expanded carrier screening, rare autosomal trisomies, confined placental mosaicism, twins, 22q11.2, chorionic villus sampling, amniocentesis, fetal anomalies, the prenatal exome, stillbirth and recurrent loss, preimplantation genetic diagnosis, maternal genetic disease, gene therapy, the role of industry in genetics, racial and ethnic health disparities in genetics, ethical legal and social issues, congenital heart disease, cardiovascular disease, neurogenetics, cancer genetics.

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Learning Outcomes

Weeks 1-4 had three targeted learning outcomes focused on clinical applications with the following reflecting the percentage of fellows that agreed or strongly agreed that the course achieved these goals:

1. Define genetic mechanisms of disease and inheritance as they apply to cytogenetics, molecular genetics and clinical genetics. --**93%**

2. Explain technologies underlying genetic tests and how those impact testing strategies and interpretation. --**87%**

3. Apply knowledge in case evaluation, genetic testing and genetic counseling. --**97%**

Weeks 5-8 had four targeting learning outcomes focused on clinical applications. The following percentage of fellows that agreed or strongly agreed that the course achieved these goals:

1. Describe techniques, interpretation, limitations and clinical applications of maternal serum screening, noninvasive prenatal testing, expanded carrier screening, and newborn screening. **–96%**

2. Compare and contrast diagnostic approaches: CVS, amniocentesis, ultrasound identified anomalies of specific organ systems, evaluation of the stillborn and newborn, preimplantation genetic diagnosis and the prenatal exome. **–92%**

3. Define the current and evolving status of these areas of genomic medicine: maternal genetic disease management, prenatal and postnatal gene therapies, racial issues in research and health disparities, secondary findings in genomic testing, the impact of industry on genetic testing, and ethical legal and social issues in reproductive medical genetics. **–96%**

4. Recognize, assess risk, provide appropriate counseling and genetic evaluation for: genetic etiology in obstetric complications, common conditions in family histories, neurogenetic conditions, and cancer genetic applications. **–96%**

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Assessment: Weeks 1-4

Basic Training Pre-Test
October 2020

Statistics

100 point test (50 questions)

Count	119
Minimum Points	0.00
Maximum Points	84.00
Range	84.00
Average	54.79831
Median	55.50
Standard Deviation	12.12716
Variance	147.0681

Grade Distribution
By Percentage Correct

Greater than 100	0
90 - 100	0
80 - 89	2
70 - 79	8
60 - 69	25
50 - 59	52
40 - 49	25
30 - 39	4
20 - 29	0
10 - 19	2
0 - 9	1
Less than 0	0

Basic Training Post-Test
November 2020

Statistics

100 point test (50 questions)

Count	77
Minimum Points	36.00
Maximum Points	100.00
Range	64.00
Average	82.82467
Median	86.00
Standard Deviation	12.47068
Variance	155.51796

Grade Distribution
By Percentage Correct

Greater than 100	0
90 - 100	29
80 - 89	26
70 - 79	14
60 - 69	4
50 - 59	1
40 - 49	2
30 - 39	1
20 - 29	0
10 - 19	0
0 - 9	0
Less than 0	0

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Assessment: Weeks 5-8

Basic Training Pre-Test
Weeks 5-8
March 2021

Statistics
81 point test (81 questions)

Count	101
Minimum Points	15.00
Maximum Points	62.50
Range	47.50
Average	46.52722
Median	47.25
Standard Deviation	8.13309
Variance	66.14715

Grade Distribution
By Percentage Correct

Greater than 100	0
90 - 100	0
80 - 89	0
70 - 79	7
60 - 69	33
50 - 59	43
40 - 49	13
30 - 39	2
20 - 29	2
10 - 19	1
0-9	0

Basic Training Post-Test
Weeks 5-8
April 2021

Statistics
81 point test (81 questions)

Count	82
Minimum Points	27.00
Maximum Points	81.00
Range	54.00
Average	71.79268
Median	74.00
Standard Deviation	8.78822
Variance	77.23293

Grade Distribution
By Percentage Correct

Greater than 100	0
90 - 100	47
80 - 89	23
70 - 79	6
60 - 69	5
50 - 59	0
40 - 49	0
30 - 39	1
20 - 29	0
10 - 19	0
0 - 9	0

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From the Fellows

- *I thoroughly enjoyed this rotation. I loved the variety in speakers as well as the multi-media format (including LIVE and recorded lectures, supplemental readings, case presentations and quizzes), this truly reinforced the information. I also enjoyed this second course with the powerpoints before hand, it helped me to be able to follow the lectures and take better notes!*

- *I was initially only casually interested in genetics. Now, I wish I had become an MFM-geneticist! I feel that the course has cemented knowledge I had developed throughout fellowship and added so much more information that I had never learned. I feel much more confident heading into my attending position this summer. Thank you for an incredible learning opportunity.*

- *This course has drastically improved my genetics knowledge and approach to genetics and genomics. It is a course which should be considered by all obstetricians. I have honestly never done a better course and being constantly taught by such experts in the field is a once in a career opportunity.*

- *This has been a truly incredible opportunity to be part of this pilot and I am SO thrilled that this will continue to be offered. The course design was well thought out and I appreciated the foundational recall lectures (Oh yeah! Centromeres!) that kicked off Part I. Please keep those types of "basic" lectures in the curriculum!*

- *This course was extremely valuable! Genetics can be an intimidating part of MFM, and it was very helpful to have a structured course to guide me through the core topics. I feel much more comfortable approaching genetics topics/cases as they arise now during my fellowship. I do feel that the second portion of the course (weeks 5-8) was more valuable than the first, as it was more clinically-focused and clinically-relevant.*

- *The courses were segregated into basic science and clinical. As I was taking the second portion of the course, I was realizing there were portions of the first course I didn't understand. I also felt the same in the first course. In the first part, some of the lab concepts were very abstract so I couldn't completely grasp them without some clinical correlation. I learned so much from both courses and have a huge increase in my knowledge. I hope I can continue learning more about the field of genetics in the future.*

- *Overall, dramatically improved my genetics knowledge. Loved the cases and talks from genetic counselors and Britton Rink. Norton and Wapner were incredible during the workshops. I am recommending to all my co-fellows to take!*

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Course Leadership

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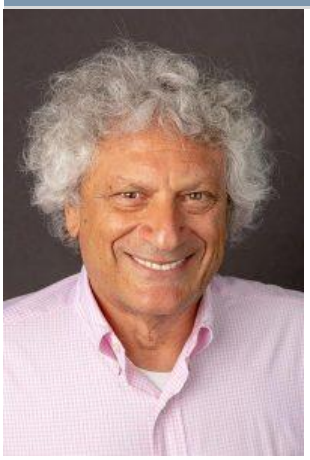

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The Faculty

These faculty donated their time and expertise in LIVE and recorded content. We are grateful for these experts, who each contributed to the vision of an outstanding online medical genetic education resource for fellowship programs. These faculty made Basic Training a reality.

Vimla Aggarwal, MBBS	Columbia University
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Education Sponsors

The following companies provided unrestricted educational grants that made this course possible. We are grateful for their support of physician education in reproductive medical genetics.

Illumina
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and

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Fulgent Genetics

Eurofins

With in-kind support from

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