basic training

In Reproductive Medical Genetics

Preparing Professionals for Practice

Summary Report of the First Two Courses

Academic Years

2020-2021 and 2021-2022

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

BASIC TRAINING
A COURSE IN REPRODUCTIVE MEDICAL GENETICS

Genetics is increasingly important in reproductive and perinatal medicine while fellowship programs are increasingly challenged in providing comprehensive education and training in this complex and rapidly evolving field. Several years ago, 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 2019. While the initial Reproductive Genetics "Bootcamp" was a great success, we had to pivot in 2020 due to the pandemic.

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 would include lectures, case presentations, interactive live workshops, and supplemental readings. Every week we would also provide quiz questions, office hours with the genetic counselors to answer questions, and "rounds with the geneticists" on Friday to review concepts and discuss quiz questions as well as interesting and complicated patients.

After a very successful first year of the virtual course, we decided to expand the content and offer participation to Reproductive Endocrinology and Infertility programs and trainees in year two. The basic course structure and many of the lectures were pre-recorded and unchanged, while additional lectures, case discussions, and participating faculty were added. 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 new REI content includes embryology, genetics of infertility and recurrent pregnancy loss, and comprehensive discussion of preimplantation genetic testing. The attached report summarizes the curriculum, the pretest and posttest scores from the first two years, and feedback from the fellows who participated.

The course would not have been possible without the support of unrestricted educational grants from Illumina, Integrated Genetics, Natera, Invitae, and Fulgent Therapeutics. 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 first two offerings of the course have been a tremendous success – we hope you agree. Moving forward we plan to offer Basic Training, A Course in Reproductive Medical Genetics again for the 2022-2023 academic year and welcome the opportunity to share this education. Register your fellows online at [Basic Training | Center for Maternal-Fetal Precision Medicine | UCSF]

Basic Training 2022-2023 Weeks 1-4 October 3-28, 2022 Weeks 5-8 March 6-31, 2023

Mary Norton, MD and Ron Wapner, MD
Basic Training Course Directors
May 2022

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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 builds 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 the genetics of recurrent loss, preimplantation genetic testing, 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 and genetic conditions affecting fertility.

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2021-2022 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. **96.87%**

2. Explain technologies underlying genetic tests and how those impact testing strategies and interpretation. **93.55%**

3. Apply knowledge in case evaluation, genetic testing, genetic counseling. **96.78%**

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

1. Describe techniques, interpretation, limitations, and clinical applications of screening including noninvasive prenatal testing, expanded carrier screening, and newborn screening. **100%**

2. Compare and contrast diagnostic approaches: preimplantation genetic testing, CVS, amniocentesis, ultrasound identified anomalies of specific organ systems, evaluation of the stillborn/newborn, the prenatal exome and the genetics of recurrent loss. **100%**

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. **96.67%**

4. Recognize, assess risk, provide appropriate counseling and genetic evaluation for: genetic etiology in obstetric complications, common conditions in family histories, neurogenetic conditions, cancer genetic applications and genetic conditions that alter infertility. **100%**

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2021-2022: Weeks 1-4

Basic Training Pre-Test
October 2021
Statistics

124-point test (62 questions)

Count	102
Minimum Value	16.50
Maximum Value	106.00
Range	89.50
Average	68.51
Median	NaN
Standard Deviation	14.93
Variance	222.98

Grade Distribution
By Percentage Correct

90 - 100	0
80 - 89	2
70 - 79	5
60 - 69	28
50 - 59	39
40 - 49	21
30 - 39	1
20 - 29	5
10 - 19	1
0 - 9	0

Basic Training Post-Test
November 2021
Statistics

148-point test (74 questions)

Count	69
Minimum Value	0.00
Maximum Value	144.00
Range	144.00
Average	124.92
Median	128.00
Standard Deviation	20.61
Variance	424.77

Grade Distribution
By Percentage Correct

90 - 100	30
80 - 89	18
70 - 79	15
60 - 69	4
50 - 59	1
40 - 49	0
30 - 39	0
20 - 29	0
10 - 19	0
0 - 9	1

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2021-2022 Weeks 5-8

Basic Training Pre-Test
Weeks 5-8
March 2022

Statistics
85 point test (85 questions)

Count	72
Minimum Value	11.25
Maximum Value	77.75
Range	66.50
Average	48.03
Median	NaN
Standard Deviation	9.70
Variance	94.10

Grade Distribution
By Percentage Correct

90 - 100	1
80 - 89	3
70 - 79	2
60 - 69	15
50 - 59	38
40 - 49	9
30 - 39	2
20 - 29	1
10 - 19	1
0 - 9	0

Basic Training Post-Test
Weeks 5-8
April 2022

Statistics
85 point test (85 questions)

Count	46
Minimum Value	51.50
Maximum Value	85.00
Range	33.50
Average	77.61
Median	NaN
Standard Deviation	6.74
Variance	45.44

Grade Distribution
By Percentage Correct

90 - 100	32
80 - 89	11
70 - 79	1
60 - 69	2
50 - 59	0
40 - 49	0
30 - 39	0
20 - 29	0
10 - 19	0
0 - 9	0

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2020-2021 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	54.79
Median	55.50
Standard Deviation	12.12
Variance	147.06

Grade Distribution
By Percentage Correct

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

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	82.82
Median	86.00
Standard Deviation	12.47
Variance	155.51

Grade Distribution
By Percentage Correct

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

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2020-2021 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	46.52
Median	47.25
Standard Deviation	8.13
Variance	66.14

Grade Distribution
By Percentage Correct

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	71.79
Median	74.00
Standard Deviation	8.78
Variance	77.23

Grade Distribution
By Percentage Correct

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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From the Program Directors

- *I just wanted to reiterate that our 4 fellows who have participated so far all have found the course very useful. I looked at the qualifying board exam scores of the 2021 grads and they were through the roof in genetics.*
- *This is an amazing resource to have for fellows. The REI fellows loved the first half of the course and said it was amazing.*
- *Genetic counseling support is available for our program. Going forward, we are planning to rebuild our genetics curriculum with integration of this course- that means fellows would be working with genetic counselors during the course and that would facilitate the necessary collaboration*
- *The fellow participates in sessions with a pediatric geneticist and genetic counselor for one of the months. This has worked out very well in our program.*
- *We assign our fellows to spend 1/2 day per week with our GCs during these 2 genetics months.*
- *We used this program to provide most of the didactic lectures on genetics but continued our own curriculum on application to fetal disorders. We used the two months to dedicate one month to an outpatient prenatal genetics month where they also worked with our genetic counselor and our MFM/genetics faculty. The other month was a pediatric genetics elective that allowed them to also work with our genetics physicians and the counselors.*
- *The course has been rolled into our fellowship curriculum as a required 2-months in the 1st year, before they spent dedicated time in our Prenatal Diagnosis clinic in their 2nd year.*
- *During the first month, the course is paired with a prenatal diagnosis/ultrasound rotation. During the second month, the fellow rotates with a pediatric geneticist.*
- *Our first-year fellow participated in the course. Next year, he will be seeing patients in the newly established MFM/Genetics clinic one day per week.*
- *This course is required for genetic/genomic rotation in addition to rotating to genetic and molecular genetic labs and genetic counselors*
- *Outstanding foundational knowledge. Relevant clinical vignettes*
- *I am very interested in incorporating this course into our fellowship training and would also like to know if there is a refresher available for faculty members as well. I have some faculty members who are also interested.*
- *Thank you so much for offering this course! We are so excited. Enrolling fellow now!*

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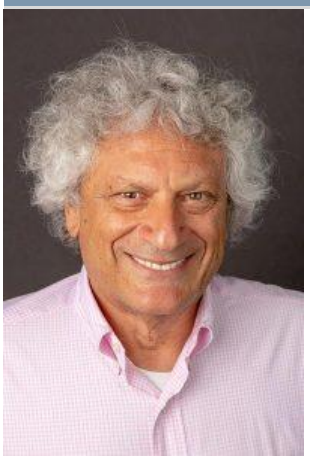

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The Faculty

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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Illumina

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Natera

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Columbia University

University of California, San Francisco

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