basic training

In Reproductive Medical Genetics

Preparing Professionals for Practice

Summary Report
the First Three Academic Years
2020-21, 2021-22, 2022-23

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

The importance of genetics is increasingly recognized in reproductive and perinatal medicine, yet fellowship programs are often 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, in 2020 we had to pivot to a virtual program due to the COVID-19 pandemic. We transitioned the program and developed "Basic Training," an 8-week virtual course meant to complement the two-month genetics rotation required by the American Board of Obstetrics and Gynecology.

The vision of "Basic Training" is to build knowledge and skills in genetics through a curriculum that begins with the basics of genetics, evolves into implementation of clinical testing, and includes content on important social and policy issues. The course includes lectures, case presentations, interactive live workshops, and supplemental readings; the outstanding faculty includes genetics experts from across the US. Every week we provide quiz questions, office hours with the genetic counselors to answer questions, and "rounds with the geneticists" to review concepts and discuss quiz questions as well as interesting and complicated patients the participants have seen. The overall goal is to integrate the course into clinical training, and we have sought to work with programs and fellows to allow them to take advantage of this educational opportunity.

The 4-week fall curriculum of "Basic Training" covers the basics of genetics from underlying principles to gene structure and function, cytogenetics, and molecular laboratory techniques. In the 4-week spring curriculum, we build on these basics and delve into prenatal screening and diagnostics, maternal genetic disorders, ethical, legal and social issues, and adult and complex diseases. After a very successful first year of the virtual course, we expanded the content and offered participation to Reproductive Endocrinology and Infertility (REI) programs and trainees. The basic course structure and most of the content was unchanged, while additional lectures, case discussions, and participating faculty were added. The REI content now 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 three years of the program, and feedback from many of the fellows who have participated. The course has overall been met with great enthusiasm and has become a useful component of reproductive genetics education for many fellowship training programs.

The course would not have been possible without the support of unrestricted educational grants from Illumina, Integrated Genetics, Natera, Invitae, Fulgent Therapeutics, and Juno Genetics. We are especially grateful for the administrative support of the Perinatal Quality Foundation, the tremendous faculty and many genetic counselors who have generously donated their time, and the support of the MFM and REI program directors who have allowed their fellows to participate.

We are very proud of the overall results and believe the course has been a tremendous success – we hope you agree. We plan to offer "Basic Training, A Course in Reproductive Medical Genetics" again for the 2023-2024 academic year and beyond.

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

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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

Each week, the course builds knowledge and skills by laying foundation through lectures, 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.

Please Join Us For
Basic Training 2023-24

Weeks 1-4: October 2-27, 2023
Weeks 5-8: March 4-29, 2024

Register online at:
Basic Training | Center for Maternal-Fetal Precision Medicine | UCSF

2022-2023 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:

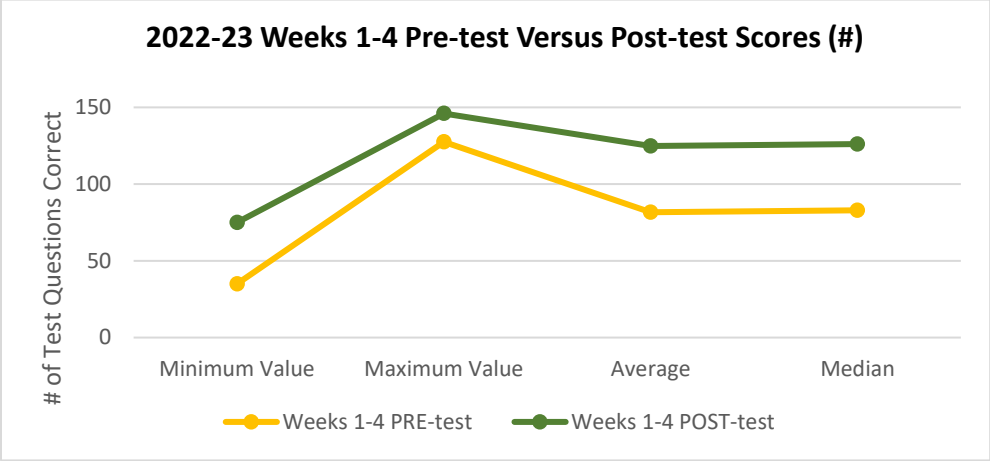
1. Define genetic mechanisms of disease and inheritance as they apply to cytogenetics, molecular genetics and clinical genetics. 100%
2. Explain technologies underlying genetic tests and how those impact testing strategies and interpretation. 96%
3. Apply knowledge in case evaluation, genetic testing, genetic counseling. 96%

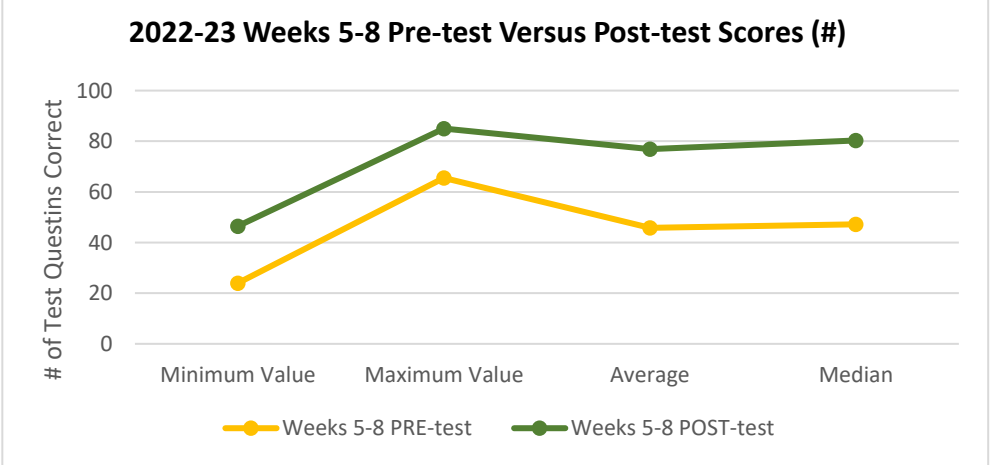
Weeks 5-8 had four targeted learning outcomes focused on clinical applications. The following percentage of fellows agreed or strongly agreed the course achieved these goals:

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. 100%
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%

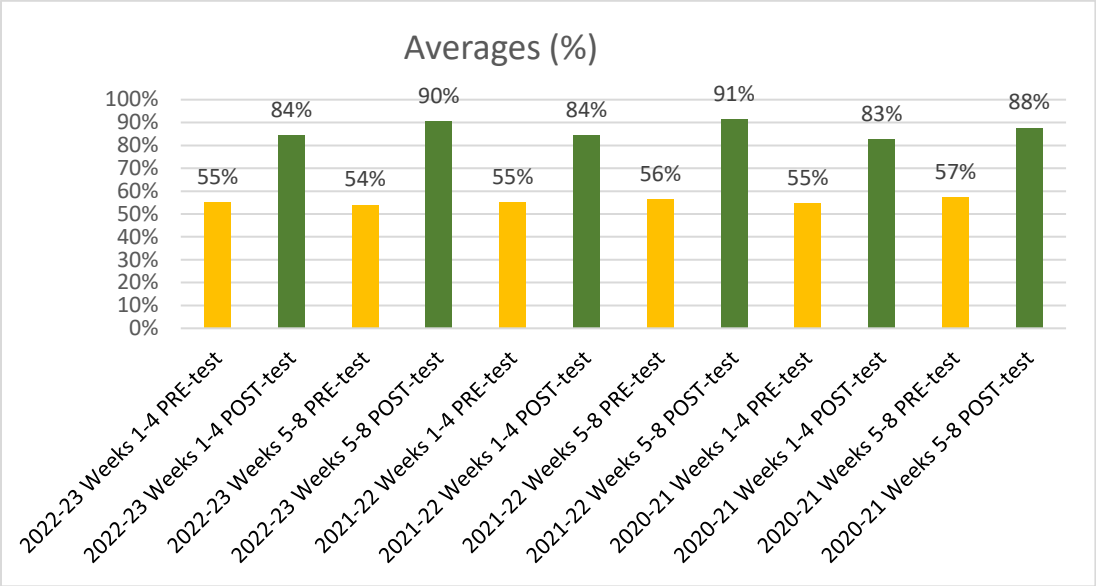
Basic Training 2022-23 Pre-Test Versus Post-Test Score Distribution

	Weeks 1-4 PRE-test 148 points	Weeks 1-4 POST-test 148 points	Weeks 5-8 PRE-test 85 points	Weeks 5-8 POST-test 85 points
Tests Submitted	119	78	82	61
Minimum Value	35	75	24	47
Maximum Value	128	146	66	85
Range	93	71	42	39
Average	82	125	46	77
Median	83	126	47	80
Standard Deviation	14	15	8	9
Variance	198	223	65	80





2020-2023 Cumulative Performance by Average



Course Registrants by Year	
Year	# Fellows
2020-21	119
2021-22	102
2022-23	119

Course Feedback from Fellows and Program Directors

From the 2022-2023 Fellows:

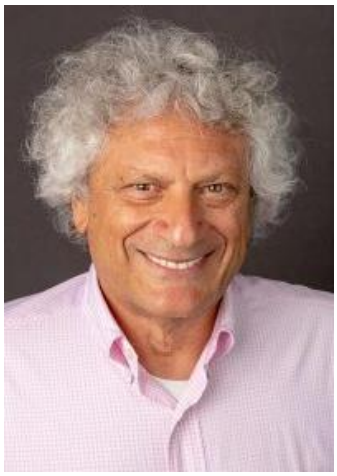
- *Thank you so much for the education and time/commitment/effort you all provided to make this very meaningful course. I wouldn't have had access to these kinds of resources in my fellowship. You all are such a friendly, approachable group!*
- *Thank you so much for this beautifully curated course, it has given me the scaffolding to better communicate with our geneticists, interpret results and plan work-ups.*
- *This was an excellent course. The material presented in Weeks 5-8 was especially relevant for clinical practice. The case-based learning format and interactive learning on Fridays were especially informative and enjoyable.*
- *This is a MUST for MFM fellows, especially before boards. I feel that our program has a strong genetic counseling program with fellow involvement, but this course has expanded my knowledge well beyond what I could have gotten from my own institution.*
- *This course was fantastic! The diverse modalities for learning (recordings, readings, cases, live rounds) and opinions from various experts in the field was very impactful. I wish I would of taken it in my 1st or 2nd year of fellowship to offer more context to everything I have seen thus far.*

From the 2021-2022 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.*
- *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.*
- *This course is required for genetic/genomic rotation in addition to rotating to genetic and molecular genetic labs and genetic counselors.*

Course Leadership

Course Directors



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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 have made Basic Training a reality.

Vimla Aggarwal, MBBS	Columbia University
Paul S. Appelbaum, MD	Columbia University
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Jessica Van Ziffle, PhD	University of California, San Francisco

Education Sponsors

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Natera

Invitae

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Juno Genetics

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