ISCC Genetics Education Curriculum Summary Presentation

Barbara M. O’Brien, MD
Louise Wilkins-Haug, MD, PhD
Richard Haspel, MD

The need

- Knowledge of human genetics is increasing dramatically and OB/GYNs are often called upon to incorporate genomics and genetic testing into medical practice

- Data has shown that OB/GYNs are undertrained in their residency and even subspecialty rotations in genetics and genomics

Background - Existing Resource Challenges

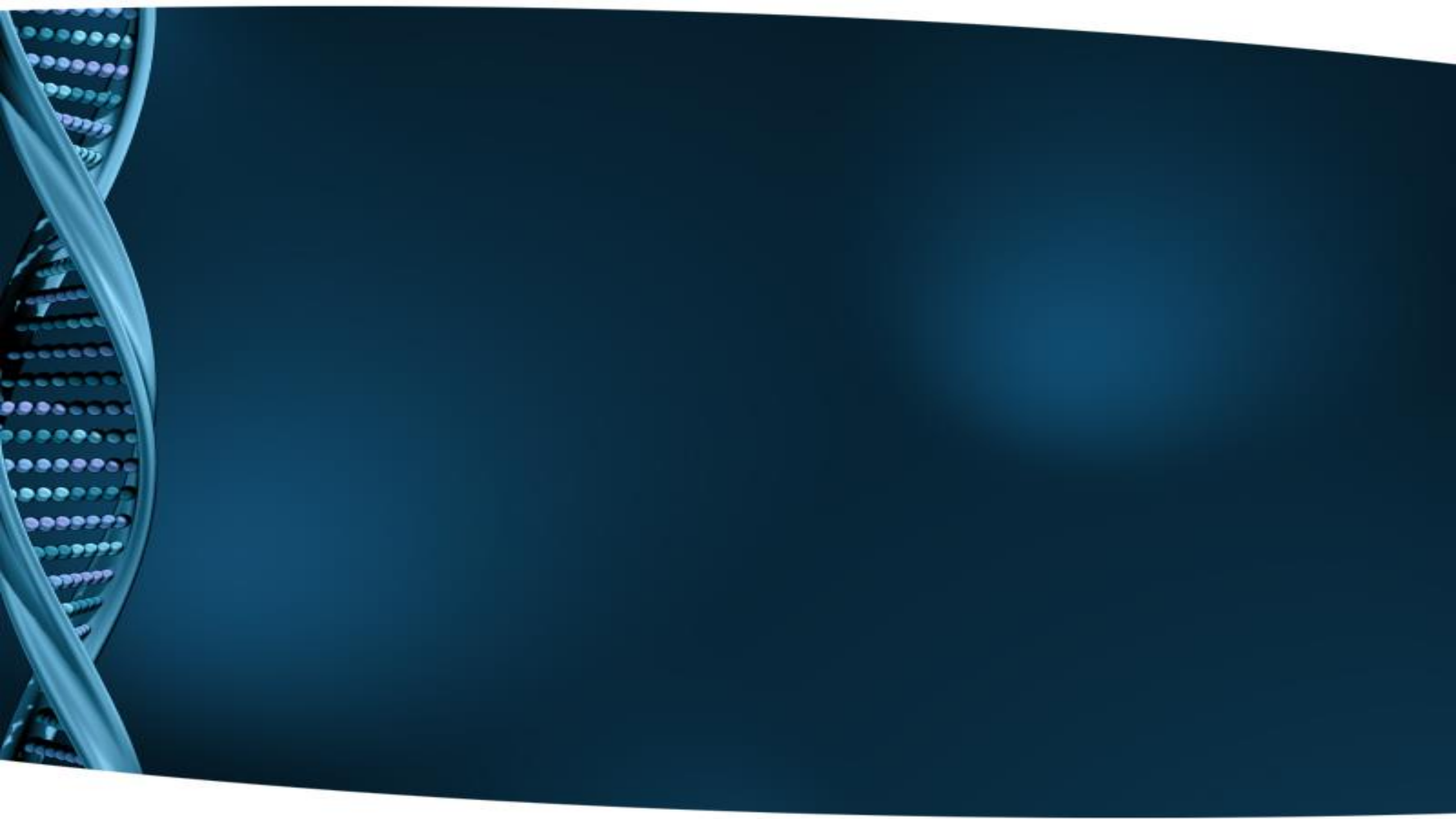
- Curriculum development based on single specialty (CREOG guidelines)
- Pre and post test available but lacking validation
- Commitment to update and alter format and content not systematic

Will this be adequate for the OBGYN residency graduates in 2020 ?

- NO
  - ACOG has supplemental genetics training available with an excellent list of lectures
    - Requires an 8 hr onsite course, plus a concluding 3 hr on site course
    - Requires additional attendance at annual meeting session
    - Completion of on-line modules
    - Cost

Genetics training OBGYN residency does not align with real–life OBGYN practice

- Primary care provider across women's reproductive lifespan
  - Preconception, prenatal and cancer (not just breast) genetic screening
- New genetic tests are continually being developed
  - Inability to assess test marketing claims
  - Inability to present reports to patients
  - Inability to communicate with other specialties (pathology, pediatrics, internal medicine)
- Genetic counselors are a limited resource



Progress Thu Far

- Genetic learning objectives were made
- Survey developed

- Survey results…..

Objective	Rating					
Basic Genetics and Genomics Principles	1	2	3	4	5	6
Distinguish genotype vs. phenotype (Q1_1)	0 (0.0)	2 (28.6)	0 (0.0)	0 (0.0)	2 (28.6)	3 (42.9)
Delineate the components of the human genome including exons, introns, splice sites and repeat regions (Q1_2)	1 (14.3)	1 (14.3)	2 (28.6)	1 (14.3)	1 (14.3)	1 (14.3)
Define the variant annotation (benign, pathogenic, uncertain significance) (Q1_3)	0 (0.0)	0 (0.0)	0 (0.0)	1 (14.3)	2 (28.6)	4 (57.1)
Define point mutations, deletions, and structural chromosomal abnormalities (Q1_4)	0 (0.0)	0 (0.0)	1 (14.3)	1 (14.3)	2 (28.6)	3 (42.9)
Define silent, missense, nonsense, frameshift, and splicing defect variants (Q1_5)	0 (0.0)	1 (14.3)	1 (14.3)	1 (14.3)	2 (28.6)	2 (28.6)
Apply concepts within a clinical framework of epigenetic regulation, including methylation and the role of imprinted genes (Q1_6)	0 (0.0)	1 (14.3)	1 (14.3)	2 (28.6)	2 (28.6)	1 (14.3)

Resource	Rating					
	Have not heard of this	1	2	3	4	5
ClinVar (Q18_1)	1 (14.3)	0 (0.0)	0 (0.0)	2 (28.6)	2 (28.6)	2 (28.6)
PolyPhen (Q18_2)	3 (42.9)	0 (0.0)	2 (28.6)	1 (14.3)	0 (0.0)	0 (0.0)
Stanford Decision Tool for Women with BRCA Mutations (Q18_3)	2 (28.6)	0 (0.0)	1 (14.3)	1 (14.3)	1 (14.3)	1 (14.3)
KM Plotter for breast cancer (Q18_4)	5 (71.4)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	1 (14.3)
Mycancergenome (Q18_5)	3 (42.9)	0 (0.0)	0 (0.0)	1 (14.3)	1 (14.3)	1 (14.3)
CIVic (Q18_6)	5 (71.4)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)
OncoKB (Q18_7)	5 (71.4)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)
Jackson Laboratory CKB (Q18_8)	4 (57.1)	0 (0.0)	0 (0.0)	1 (14.3)	1 (14.3)	0 (0.0)
COSMIC (Q18_9)	5 (71.4)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)
ClinicalTrials.gov (Q18_10)	0 (0.0)	0 (0.0)	0 (0.0)	1 (14.3)	2 (28.6)	4 (57.1)
ClinGen (Q18_11)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	2 (28.6)	3 (42.9)
OMIM (Q18_12)	1 (14.3)	0 (0.0)	0 (0.0)	2 (28.6)	1 (14.3)	3 (42.9)
cBioPortal (Q18_13)	5 (71.4)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)
Integrative genomics viewer (IGV) (Q18_14)	5 (71.4)	0 (0.0)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)
Genetic Testing registry (Q18_15)	2 (28.6)	0 (0.0)	0 (0.0)	3 (42.9)	0 (0.0)	2 (28.6)
Gene Reviews (Q18_16)	1 (14.3)	0 (0.0)	0 (0.0)	2 (28.6)	0 (0.0)	4 (57.1)
Genetics home reference (Q18_17)	1 (14.3)	0 (0.0)	0 (0.0)	2 (28.6)	1 (14.3)	3 (42.9)
genome AD (Q18_18)	4 (57.1)	0 (0.0)	1 (14.3)	0 (0.0)	0 (0.0)	2 (28.6)

Project Group Discussion