REVIEWING GENETICS AND GENOMICS RESOURCES FOR OB-GYN PROFESSIONALS

Abimbola M. Oladayo, BDS, MPH, MS
NIH T90 Fellow, College of Dentistry, University of Iowa.
2021-2023 ISCC-PEG Scholar: OB/GYN Genomics Curriculum Development Group.

Mentor: Louise E. Wilkins-Haug, MD, PhD
Division Director, Maternal Fetal Medicine and Reproductive Genetics at Brigham and Women's Hospital.

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

NIH National Human Genome Research Institute
Advancing human health through genomics research

OVERVIEW

- **Project goal**: Improving access to genetics/genomics education resources by creating a central resource point consisting up-to date information.

- **Target audience**: OB-GYN providers.

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

NIH
NHGRI

INCREASING RATE OF ADOPTION OF GENOMICS TECHNOLOGIES

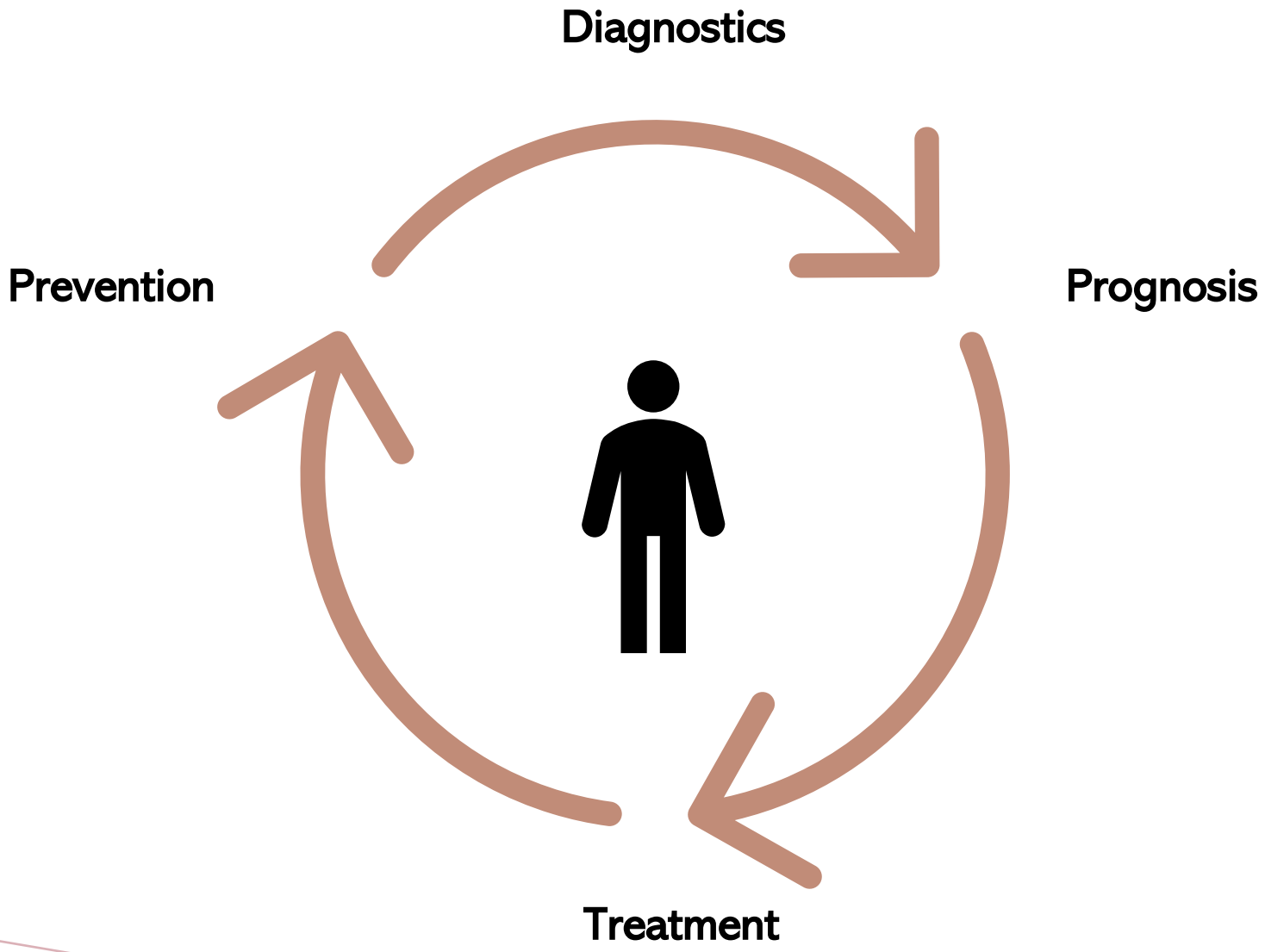


Image adapted from Franks et al., Technological readiness and implementation of genomic-driven precision medicine for complex diseases. J Intern Med 2021; 290: 602– 620.

GENOMICS TECHNOLOGY IN THE CONTEXT OF OB-GYN PRACTICE

- DNA-based tests for preimplantation, prenatal and postnatal diagnosis.
  - Carrier testing
  - Aneuploidy screening
  - Gynecological malignancies – Breast, Endometrial, Peritoneal, Fallopian Tube and Ovarian.
- Impact treatment and prevention approach to fetal and maternal care.

LOW LEVELS OF GENOMIC LITERACY IN OB-GYN PROFESSIONALS

Low levels of genomic literacy in non-genetic HCPs, including OB-GYN professionals.

Shortage of genetic counselors means OB-GYN providers would have to be more involved in genetic testing.

Impact on delivery/incorporation of genomics medicine into care.

- Limited knowledge regarding testing options.
- Low confidence about using genetic test results in the patient care.
- Limited knowledge about how and when to make a referral to a specialist.
- Conducting risk assessment and using variant information in care decisions.

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

NIH NHGRI

BARRIERS TO PROVIDERS GENOMICS EDUCATION

- Time requirements for elective education.
- Commercial/for profit entities as major sources of information.
- Pace of advancement of genomics technology **vs**. update of available resources.
- Providers awareness of existing educational resources and ease of navigation.

These resources are for information purposes only and are not meant to be comprehensive. Referral to these resources does not imply the American College of Obstetricians and Gynecologists' endorsement of the organization, the organization's website, or the content of the resource. The resources may chan without notice.

This document reflects emerging clinical and scientific advances as of the date issued and is subject to change. The information should not be construed as dictating an exclusive course of treatment or procedure to be followed.

NIH NHGRI

Inter-Society Coordinating Committee for Practitioner Education in Genomics (ISCC-PEG)

AMERICAN JOURNAL OF OBSTETRICS & GYNECOLOGY- ABSTRACT

- Providers spent too little time on genetic counseling.
- More than half could not correctly identify ACOG recommendations.
- Barriers to counseling included time constraints, health literacy, lack of a visual aid, and language barriers.
- Future directions – need for the development and assessment of prenatal genetics educational tools to supplement counseling.

POSTER SESSION IV FRIDAY, FEBRUARY 10, 2023 • 3:30 PM - 5:00 PM | VOLUME 228, ISSUE 1, SUPPLEMENT , S694, JANUARY 2023

A cross-sectional survey of prenatal care providers' knowledge, barriers, and confidence in prenatal genetic counseling

Margaret Thorsen • Rose Mahoney • Franklin Enemuo • Huda B. Al-Kouatly • Melissa L. Russo

DOI: https://doi.org/10.1016/j.ajog.2022.11.1162 • Check for updates

METHODS

Literature review via Google Scholar & PUBMED.

Websites of scientific, clinical and professional organizations.

RESOURCES

PROFESSIONAL ORGANIZATIONS

- ACOG: American College of Obstetricians and Gynecologists.[1]
- American Medical Association
- ACMG: Genetics101 for Healthcare Providers
- ESHG - European Society of Human Genetics.[1]

PUBLIC DATABASES

MedGen – an online portal providing information on the genetic basis of human health and disease, including professional guidelines.

ClinGen – an NIH funded central resource providing information on the clinical relevance of genes and variants with summaries about ACMG recommended genes and conditions.

GeneReviews – a point of care resource for clinicians providing information on clinically relevant and medically actionable conditions including diagnosis, management and counselling.

ClinVar – a public archive of information about genomic variations and phenotypes.

OMIM – provides a compendium of human genes and genetic phenotypes.

NCI –presents physician data query (PDQ) summaries of the latest information on cancer detection, prevention and genetics.

PharmGKB – provides information on the impact of genetic variation on drug response.

GOVERNMENT SUPPORTED PROGRAMS

NHGRI: HCP Genomics Education Resources.

Genomics Education Resource Center (GenomeEd): https://www.genome.gov/GenomeEd

Global Genetics and Genomics Community (G3C): https://www.genomicscases.net/en

"Training Residents in Genomics" (TRIG): https://www.pathologylearning.org/trig

Healthcare Provider Genomics Education Resources: https://www.genome.gov/

The OBG project: Genome section

CDC - https://www.cdc.gov/genomics/resources/educational.htm

State Health Departments[1].

National Coordinating Center for the Regional Genetics Networks: https://nccrcg.org/#

NIH NHGRI

[1]: Provides both provider and patient educational resources.

EDUCATION PORTALS

Not for Profit Organizations

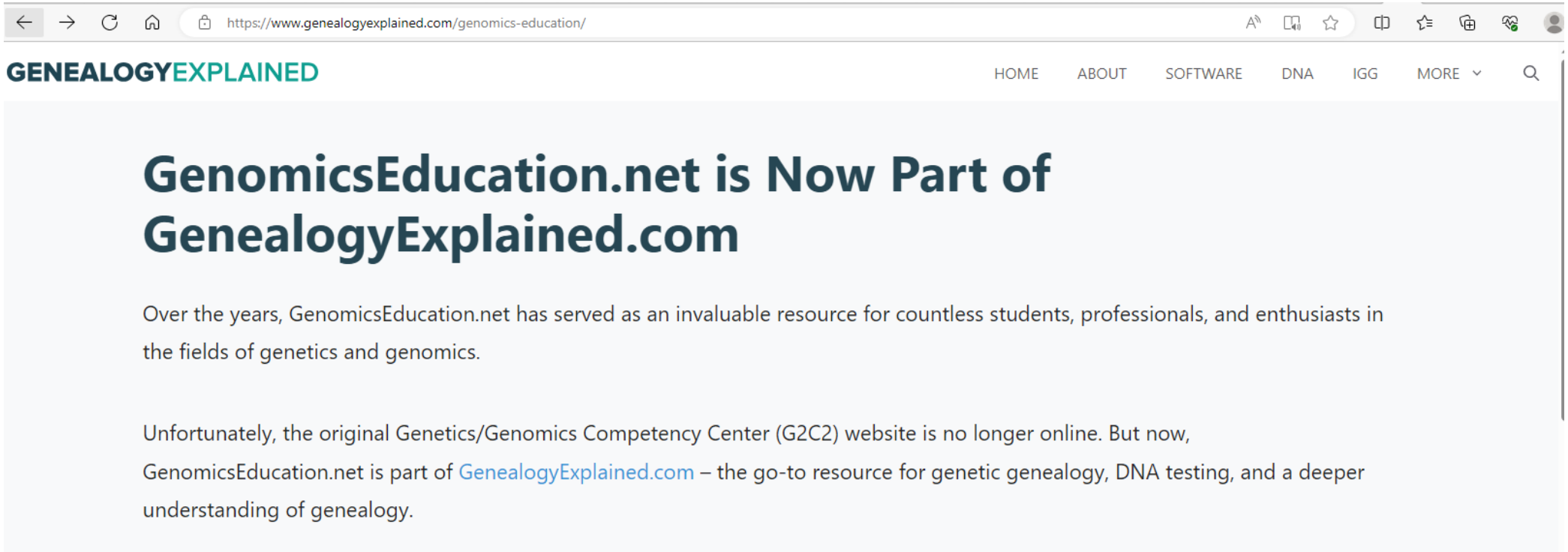
- Perinatal Quality Foundation: Online Genetic Education Modules developed by the PQF seeks to educate patients and providers about prenatal screening and diagnostic testing options.
- Jackson Laboratory.
- Genetics Education Canada.

MOOCs at Center for the Integration of Research, Teaching, and Learning.

Genomics Education Program - Health Education England*[1].

[1]: Provides both provider and patient educational resources

GENETICS AND GENOMICS COMPETENCY CENTER (G2C2)



ARE WEB-BASED GENOME ED APPROACHES EFFECTIVE?

- Web-based genetics education was highly effective in increasing health care providers' confidence about using genetics.

ELSEVIER

Genetics in Medicine

Volume 24, Issue 1, January 2022, Pages 214-224

Education Report

Improved provider preparedness through an 8-part genetics and genomic education program

Catherine Hajek [1, 2], Allison M. Hutchinson [1], Lauren N. Galbraith [3], Robert C. Green [4, 5, 6, 7], Michael F. Murray [8], Natasha Petry [9, 10], Charlene L. Preys [4, 11], Carrie L.B. Zawatsky [4, 7], Emilie S. Zoltick [3], Kurt D. Christensen [3, 5, 12]

Imagenetics METRICS Team

DISCUSSION AND RECOMMENDATIONS

Organize resources based on “keywords”/ “common categories”.

Improve providers awareness about education resources and a clear plan for dissemination.

Review the available education resources.

DISCUSSION AND RECOMMENDATIONS

Understanding and supporting patients post testing – referral to genetic counsellors.

Familiarity with practical and ethical issues.

Propose an alliance consisting of multiple organizations to develop a standard and central educational approach –ISCC-PEG???

ACKNOWLEDGMENT

Mentor: Louise E. Wilkins-Haug, M.D., Ph.D.

ISCC-PEG Obstetrics and Gynecology (OB/GYN) Genetics Curriculum co-chair- Barbara O'Brien.

ISCC-PEG Co-chairs: Rich Haspel, M.D., Ph.D. and Donna Messersmith, Ph.D.

National Human Genome Research Institute.

Butali Craniofacial Genetics Lab, COD, Ulowa: Azeez Butali, DDS, Ph.D, FICD.

NIH R01 DE028300 and NIDCR T90 Training Grant- DE023520.

NIH NHGRI

THE UNIVERSITY OF IOWA

NIDCR® National Institute of Dental and Craniofacial Research

Inter-Society Coordinating Committee for Practitioner Education in Genomics

NIH NHGRI

REFERENCES

- **ACMG** – www.acmg.net.
- **NIH** – www.genome.gov/11510197.
- **MedGen** – www.ncbi.nlm.nih.gov/medgen/.
- **ClinGen** – https://clinicalgenome.org/.
- **GeneReviews** –www.ncbi.nlm.nih.gov/books/NBK1116/.
- **ClinVar** – ClinVar www.ncbi.nlm.nih.gov/clinvar/.
- **OMIM** – www.ncbi.nlm.nih.gov/omim.
- **NCI** – www.cancer.gov/about-cancer/causes-prevention/genetics/overview-pdq.
- **PharmGKB** – www.pharmgkb.org/.