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BEYOND THE DOUBLE HELIX

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Medical genetics education in the midst of the COVID-19 pandemic: Shared resources

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Abstract

In the midst of the COVID-19 pandemic, it is appropriate that our focus is on patient care and preparation. However, the genetics community is well poised to fill in the educational gap created by medical students transitioning to limiting patient contact, creation of telemedicine patient care, and online learning modules. Our history of agility in learning and teaching is now only inhibited by the time constraints of current clinical demands on the genetics community. This publication is designed to offer ideas and resources for quickly transitioning our education to meet the current demands in the time of a pandemic. Not only will this allow us to continue our strong history of education, it will enhance our strong commitment to using modern educational techniques and tools to address the genetics workforce issues that have defined the recent past. We have the opportunity to aggressively educate for trainees that now have the capacity to learn, and to lead the way in showing how the genetics community rallies together no matter the challenge.

KEYWORDS

Genetics education, Telemedicine education, Communities of practice

1 | INTRODUCTION

In the midst of the COVID-19 pandemic, the role of agile medical education has become increasingly important. For the genetics community, there has been a growing passion to increase educational opportunities to increase our workforce (Bennett, Waggoner, & Blitzer, 2017). In this time of unprecedented changes in medical care and education, we must work as a community to create genetic education that is agile, caring, approachable, and innovative.

2 | MEDICAL EDUCATION THEORY

In this new educational environment, the importance of strong educational theory to guide our decision-making is paramount. Lave and Wenger described the Communities of Practice theory of education where direct interaction with a community of experts and mentorship is the basis of training (Wenger, 1998). Thus, the design of all of our educational endeavors needs to continue to bring trainees into our community.

Malcolm Knowles created andragogy, a learning theory that describes adult learners. This theory includes six major themes (Knowles, 1970; Knowles, 1985; Manning et al., 1987). First, self-concept describes that adults choose what they want to learn, when they want to learn it and how they want to learn. The second characteristic is experience, which explores the importance of the adult's past learning to share and grow in the active learning environment. Next, the adult learner needs to recognize and educational need. This is the one area that is most difficult for traditional genetics to lead to long term learning. Thus, we need to actively apply the educational content to current and important local and national events (i.e., genetic sequencing affecting COVID-19 outcomes, unique needs of rare disease patients, etc.). The fourth important concept of adult learning theory is to create problem centered learning. This style of problem-solving is innately a part of the diagnostic odyssey, which is a common finding in medical genetics. The role of internal motivation in adult learning creates an unknown variant. It is the common reason the most invested learners have personal experience with rare diseases. For this reason, the use of stories is important to increase

internal motivation. Finally, the most successful adult learners realize the importance of the knowledge and how it will be applied to problems that they will or are encountering.

3 | LEVERAGING CURRENT MEDICAL GENETICS EDUCATIONAL RESOURCES

Genetics education has historically been ahead of its time with the creation of online resources, modules, and even YouTube videos. As summarized in Table 1, the available online resources are vast. For example, by including patient interviews available on the National Organization of Rare Disorders web site to an online lecture could be an optional substitution for a patient panel session. Or the creation of a problem-based session with "real" newborn screen results and the Baby's First Test, ACMG ACT sheets, National Newborn Screening and Resource Center, and state lab resources take a dry lecture to an interactive case-based online session. Thus, the opportunity to learn while not in the hospital setting is available and robust. It is not a lack of educational resources, but instead a need to leverage the resources that exist to create a new generation of teaching materials that meet the unique needs of adult learners. In the current environment, where trainees are sequestered at home and tasked with independent learning, online modules, while potentially effective at educating, may lack the interactive quality trainee's desire. As described above, the opportunity to interact with the material, apply it to new and important situations, and bring in past knowledge in new ways, ensure optimal learning. Thus, in place of creating additional online modules, a group of genetics educators have worked to identify ways to create web-based AND interactive learning activities.

4 | CURRICULUM IMPLEMENTATION: THE LIVE TO REMOTE TRANSITION

4.1 | Example 1: Pediatric residents

Here we describe some ideas for how we have converted to a fully socially distanced learning system. In a pediatric residency program with over 100 residents, we transitioned in-person problem-based sessions to web-based problem sessions. The original lecture included having groups use a case as a basis for learning how to use online genetics resources (i.e., OMIM, PubMed, Genetics Home Reference, ACT sheets for Newborn screen, GeneReviews, summarized in Table 1) and create a picture-based memory mnemonic. Using the break-out room ability of Zoom (Zoom, 2020), we were able to transition this lecture to digital "break-out rooms." Participants had discussions with a guide (genetic counselors, genetics trainees, senior residents on a genetics rotation) to review the unknown case, "make the diagnosis," and create a memory mnemonic tool to share with the other participants. In one such lecture, residents were split into 5 groups and given a "common" rare disease including the following:

- Cystinosis: diagnosis made using OMIM. Required GeneReview and Clinicaltrials.gov for questions.
- 22q11 deletion syndrome: diagnosis made using OMIM. Required Genetics home reference, GeneReview to answer question.
- Tuberous sclerosis: diagnosis made using OMIM. Required GeneReview to answer question.
- Noonan syndrome: diagnosis made with Pubmed or OMIM. Required Genetics home reference, NORD, or GeneReview to answer questions.
- Phenylketonuria: required use of newborn screening and genetics guideline resources.

Following the disorder identification and answering the basic clinical questions, each group was assigned to make a "drawing" to help them remember the disorder. They were encouraged to be creative, use any tools they liked (i.e., one person drawing for the group, PowerPoint together, etc.). Following this, each group presented their case to the larger group. To manage the groups and time, the leader used the Zoom texting function to give time warnings and "dropped in" to each of the breakout rooms early in the time period when working through the case and later when working on the image to ensure each group was moving forward with the project.

4.2 | Example 2: Medical students

For medical students, the need to have meaningful, stimulating, and interactive learning opportunities became even more essential, as the AAMC published guidance to halt clinical rotations on March 17, 2020, which further were elucidated in publications March 23 and March 30 (summarized in AAMC, 2020) Their recommendation at that time was that medical students should not be involved in direct patient care unless there are dire health care provider needs in the immediate area (summarized in AAMC, 2020). Due to this, in our experience, students missed being a part of the hospital environment. While learning about COVID-19 has been an important component of their time, they continue to have sufficient bandwidth to expand learning to novel topics. Often, these discussions have been surrounding seminal articles in medical genetics (selected articles listed in Table 2). By creating a curriculum that directly relates to patient care (e.g., metabolic emergencies), offers education on expanding their examination skill knowledge (e.g., dysmorphology), and teaching counseling techniques have been well received.

For medical students, we created a picture-based Zoom lecture of dysmorphology cases using the chat function to "compete" for identifying abnormal features in the pictures. The anonymity of the chat function was even more effective than previous live lectures as it allowed for more inclusive participation. Thus, this method proved to be incredibly effective. A second institution created an in-depth case-based learning (CBL) curriculum that allowed for learners to have discussions with faculty over time (Table 3). By creating interactions with the faculty in the short and longer time period these programs allowed for interactions with medical genetics providers and expanded the understanding of our field to trainees at all levels.

TABLE 1 Online resources for medical genetics education

ACMG	Multiple resources vetted by ACMG	https://www.acmgeducation.net/Public/Catalog/Home.aspx?tab=2
ASHG	Multiple resources vetted by ASHG	https://www.ashg.org/discover-genetics/
Baby's First Test	Newborn screen maps and ideas	https://www.babysfirsttest.org/
Children's National Genetics Education Site	Free warehouse of learning ideas and modules for general use and re-purposing. Your submissions are welcome!	LearnRD.com→ CNMC Genetics Courses→ Educational Resources in the Time of COVID 19
Clinical and Translational Sciences Institute	Each CTSI has educational resources through their translational workforce development module for learners at all levels.	https://ncats.nih.gov/ctsa (check for a CTSA near you!)
Cold Spring Harbor	While aimed at high school students, might be a fun way to get some ideas for your lectures!	https://dnalc.cshl.edu/
Elements of Morphology	Open access publications on the key elements for physical exams.	https://elementsofmorphology.nih.gov/index.cgi and/or https://onlinelibrary.wiley.com/toc/15524833/2009/149A/1
The Gene: An intimate history	This PBS Ken Burns special weaves together science, history, and the personal story of the human genome. Also included are short 2-min films on the basics of genetics.	https://www.pbs.org/show/gene/ (Premier April 7, 14)
GeneReviews Glossary	Basic GeneReview for direct to consumer testing, current approaches, how to order, etc.	https://www.ncbi.nlm.nih.gov/books/NBK481802/
Genetic Metabolic Learning	Many free with registration. Developed by Mark Korsen and Georgianne Arnold.	http://gmce.thinkific.com/
Genetics Home Reference	Try the Classroom link for lots of ideas of hands-on ideas	https://ghr.nlm.nih.gov/resources
Genetics Science Learning Center	University of Utah's education curriculum with all age examples	https://learn.genetics.utah.edu/
National Human Genome Research Institute	A website of educational resources for all levels of learners	https://www.genome.gov/about-genomics
National Newborn Screening and Resource Center	All the information about NBS you could want for a lecture or for a trainee! This also has the ACT sheets and screening by state maps.	https://genes-r-us.uthscsa.edu/
National Organization for Rare Disorders	NORD has a stellar website with webinars, resources, and disease pages.	https://rarediseases.org/
New England Regional Genetics Network	Designed for schools supporting patients with a diagnosis, this resource is a great quick description of diseases.	https://www.negenetics.org/content/genetic-education-support-system
The New England Consortium of Metabolic Programs	Video library, including a video of the first PKU treatment experiment	https://newenglandconsortium.org/[newenglandconsortium.org]
North American Metabolic Academy (NAMA)	A comprehensive set of ppt slides of metabolic pathways. Free with SIMD membership (contact the authors if you need assistance!)	https://www.simd.org/NAMA/index.asp
Pathways of Human Metabolism	Stanford's metabolic map to allow learners to have an overview map at their home site that can be "mapped" to content during a discussion. Free email with registration.	https://metabolicpathways.stanford.edu/
Rare Disease Clinical Research Network	This NCATS-funded group of consortia have amazing websites and educational materials on multiple rare diseases.	https://ncats.nih.gov/rdcrn/consortia
Consider checking the web site for any pharmaceutical and product sites for education content. We have found these to be very helpful in modeling effective curriculum.		

Note: This table contains organization names, a short description of their resources, and their website for readers to use as a reference.

TABLE 2 A list of interesting, landmark and important articles in the history of medical genetics. Learning questions will be hosted on Children's National Genetics Education Site (see Table 1)

KEY GENETIC CONCEPTS	
Sex chromosome aneuploidy	Ford CE, Jones KW, Polani PE, et al. A sex-chromosome anomaly in a case of gonadal dysgenesis (Turner's syndrome). Lancet. 1959; 1(7075):711-3.
Imprinting	Nicholls RD, Knoll JH, Butler MG, *et al*. Genetic imprinting suggested by maternal heterodisomy in nondeletion Prader-Willi syndrome. *Nature*. 1989; 342 (6247):281–285.
Genetic Dominance	Wilkie AO. The molecular basis of genetic dominance. *J Med Genet*. 1994; 31 (2):89–98. Goldstein JL, Brown MS. Familial hypercholesterolemia: identification of a defect in the regulation of 3-hydroxy-3-methylglutaryl coenzyme A reductase activity associated with overproduction of cholesterol. *Proc Natl Acad Sci U S A*. 1973 Oct; 70(10):2804-8.
X-linked Dominant	Amir, R., Van den Veyver, I., Wan, M. *et al*. Rett syndrome is caused by mutations in X-linked *MECP2*, encoding methyl-CpG-binding protein 2. *Nat Genet* 1999; 23, 185–188.
X-inactivation	Lyon MF. X-chromosome inactivation: a repeat hypothesis. *Cytogenet Cell Genet*. 1998; 80(1-4):133–137.
The two-hit phenomenon	Knudson AG Jr. Mutation and cancer: statistical study of retinoblastoma. *Proc Natl Acad Sci U S A*. 1971; 68(4):820–823.
Gene Dosage	Patel, P., Roa, B., Welcher, A. *et al*. The gene for the peripheral myelin protein PMP–22 is a candidate for Charcot–Marie–Tooth disease type 1A. *Nat Genet* 1992; 1, 159–165. P.F. Chance, M.K. Alderson, K.A. Leppig, *et al*. DNA deletion associated with hereditary neuropathy with liability to pressure palsies. Cell, 1993; 72:143-151.
Trinucleotide repeats	Budworth H, McMurray CT. A brief history of triplet repeat diseases. *Methods Mol Biol*. 2013; 1010:3–17. MacDonald *et al*. A novel gene containing a trinucleotide repeat that is expanded and unstable on Huntington's disease chromosomes. *Cell* 1993; 72, 971–983.
Mapping a gene to a chromosome	Gusella, J. F., *et al*. A polymorphic DNA marker genetically linked to Huntington's disease. *Nature* 1983; 306, 234–238. King MC. "The race" to clone BRCA1. Science. 2014; 343(6178):1462-5.
Mosaicism	Lindhurst MJ, Sapp JC, Teer JK et al. A Mosaic Activating Mutation in *AKT1* Associated with the Proteus Syndrome. *N Engl J Med* 2011; 365:611-619.
Sickle cell disease	Pauling L, Itano HA, Singer SJ, Wells IC. Sickle Cell Anemia, A Molecular Disease. Science 1949; 110(2865): 543-548. A. C. Allison. Protection afforded by sickle-cell trait against subtertian malaria infection, *Br. Med. J*. 1954; 1, 290–294.
CONCEPTS OF TRIPLET REPEAT DISORDERS	
Anticipation	P S Harper, H G Harley, W Reardon, and D J Shaw. Anticipation in myotonic dystrophy: new light on an old problem. Am J Hum Genet. 1992 Jul; 51(1): 10–16.
Number of repeats influence expression	Kuhl PA, Pizzuti A, Pieretti M *et al*. Variation of the CGG repeat at the fragile X site results in genetic instability: Resolution of the Sherman paradox. Cell. 1991; 67; 6: 1047-1058.
PRENATAL GENETICS	
Teratology	Jones KL, Smith DW, Ulleland CN, Streissguth P. Pattern of malformation in offspring of chronic alcoholic mothers. Lancet. 1973 Jun 9; 1(7815):1267-71.
Prevention of Neural Tube Defects	Smithells RW, Sheppard S, Schorah CJ, et al. Apparent prevention of neural tube defects by periconceptional vitamin supplementation. *Arch Dis Child*. 1981; 56 (12):911–918.
Paternal age	Orioli IM, Castilla EE, Scarano G, Mastroiacovo P. Effect of paternal age in achondroplasia, thanatophoric dysplasia, and osteogenesis imperfecta. Am J Med Genet. 1995; 59:209–17.
Circulating cell-free DNA	Lo YM, Corbetta N, Chamberlain PF, Rai V, et al. Presence of fetal DNA in maternal plasma and serum. *Lancet*. 1997; 350(9076):485-7.

(Continues)

TABLE 2 (Continued)

KEY GENETIC CONCEPTS	
Concept of gonadal mosaicism	Pyott, S., Pepin, M., Schwarze, U. *et al*. Recurrence of perinatal lethal osteogenesis imperfecta in sibships: Parsing the risk between parental mosaicism for dominant mutations and autosomal recessive inheritance. *Genet Med* 2011; 13, 125–130.
Twinning	Souter VL, Kapur RP, Nyholt DR, *et al*. A Report of Dizygous Monochorionic Twins. N Engl J Med 2003; 349:154-158.
IN THE ERA OF GENOMIC ASSESSMENT & NEXT GENERATION SEQUENCING:	
Genome-wide association studies	Visscher PM, Wray NR, Zhang Q, et al. 10 Years of GWAS Discovery: Biology, Function, and Translation. *Am J Hum Genet*. 2017; 101(1):5–22.
Chromosomal microarray	Miller DT, Adam MP, Aradhya S, *et al*. Consensus statement: chromosomal microarray is a first-tier clinical diagnostic test for individuals with developmental disabilities or congenital anomalies. Am J Hum Genet. 2010; 86(5):749-64.
The role of exome sequencing in molecular discovery	Bamshad MJ, Ng SB, Bigham AW, et al. Exome sequencing as a tool for Mendelian disease gene discovery. Nat Rev Genet. 2011; 12(11):745-55.
Assessing a genetically & phenotypically heterogenous condition	Shen Y, Dies KA, Holm IA, et al. Clinical Genetic Testing for Patients With Autism Spectrum Disorders. Pediatrics 2010, 125 (4) e727-e735.
Understanding how molecular variants are interpreted in a molecular laboratory	Richards S, Aziz N, Bale S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med*. 2015; 17(5):405–424.
Exome sequencing as a first-tier test for infants with complex disease suggestive of an underlying genetic etiology	Stark, Z., Tan, T., Chong, B. *et al*. A prospective evaluation of whole-exome sequencing as a first-tier molecular test in infants with suspected monogenic disorders. *Genet Med* 2016; 18, 1090–1096.
The power of segregation analysis	Jarvik GP, Browning BL. Consideration of Cosegregation in the Pathogenicity Classification of Genomic Variants. *Am J Hum Genet*. 2016; 98(6):1077–1081.
ETHICAL CONCEPTS	
Terminology & treatment needs to evolve with patient needs	Lee PA, Houk CP, Ahmed SF, et al. Consensus Statement on Management of Intersex Disorders. Pediatrics Aug 2006, 118 (2) e488-e500.
History of Gene Patents	Kenneth Offit, Angela Bradbury, Courtney Storm, *et al*. Gene Patents and Personalized Cancer Care: Impact of the Myriad Case on Clinical Oncology. Journal of Clinical Oncology 2013; 31:21:2743-2748.
In consideration of population based genetic testing	King M, Levy-Lahad E, Lahad A. Population-Based Screening for *BRCA1* and *BRCA2*: 2014 Lasker Award. *JAMA*. 2014; 312(11):1091–1092.
Reinterpreting genetic information	Appelbaum PS, Parens E, Berger SM, *et al*. Is there a duty to reinterpret genetic data? The ethical dimensions. Genet Med. 2020 Mar; 22(3):633-639.
TREATMENT	
Gene Therapy	Maguire AM, Simonelli F, Pierce EA, *et al*. Safety and efficacy of gene transfer for Leber's congenital amaurosis. N Engl J Med. 2008; 358(21):2240-8. Finkel RS, Mercuri E, Darras BT, *et al*. Nusinersen versus Sham Control in Infantile-Onset Spinal Muscular Atrophy. *N Engl J Med*. 2017 Nov 2; 377(18):1723-1732.
Mutation-specific treatment	Ramsey BW, Davies J, McElvaney NG, *et al*. A CFTR potentiator in patients with cystic fibrosis and the G551D mutation. N Engl J Med. 2011; 365(18):1663-72.
CRISPR/Cas9	Jinek M, Chylinski K, Fonfara I, Hauer M, Doudna JA, Charpentier E. A programmable dual-RNA-guided DNA endonuclease in adaptive bacterial immunity. *Science*. 2012; 337(6096):816–821.
Single-cell sequencing	Yin Y, Jiang Y, Lam KWG et al. High-Throughput Single-Cell Sequencing with Linear Amplification. Molecular Cell. 2019; 76(4):676-690.

4.3 | Example 3: Medical subspecialists

At the request of the fellowship faculty from multiple other divisions, we have offered our faculty as a learning resource during this time. For example, neonatology asked if we could do a virtual "review" session with fellows on the carbohydrate metabolism disorders, based on a chapter in their review textbook. I am certain that many reading this article are similarly cringing as the author did with this request. What could have been a very dry lecture was actually very interactive and fun when each person was given a case to "present" to the group after taking a few minutes to research what they thought the child had (based on pictures or a single people of additional information to a low glucose level). By giving them a

TABLE 3 Using case-based learning (CBL) in clinical genetics

Goals of teaching	CBL prompts to student and teacher
Identify key clinical features	• Are features suggestive of a malformation, deformation, dysplasia, interruption • Are features primary or secondary?
Developing a clinical suspicion for underlying single-gene disorder	• Recognizing pathognomonic features of single gene disorders • Assessing a medical condition in the context of age of onset, clinical course, sex
Assessing a complex patient	• Develop skill to incorporate clinical confounders into clinical assessment • Considering associations for additional phenotypic assessment
Develop a differential diagnosis	• Note features that support and lean against conditions considered on differential diagnosis
Incorporate family history into assessment	• Recognize patterns of inheritance • Note how the following impact risk assessment: age of diagnosis, age of death, sex of affected, number of affected, and unaffected individuals
Consider genetic testing strategies	• Clinical utility of various testing strategies • Consensus statements and best practices • Strengths and limitations of each testing modality

baby of a diabetic mother (radiograph of sacral agenesis and a large baby), picture of an infant with Beckwith–Wiedemann syndrome, a newborn screen showing elevated leucine (Maple syrup urine disease), a baby with *Escherichia coli* sepsis and liver failure (galactosemia), rash (sepsis) and elevated C3 level on newborn screen (propionic vs. methyl malonic aciduria), each person had a "story" to share about a child with hypoglycemia and the reasons for this diagnosis.

4.4 | Example 4: Expanding the medical genetics community: learners of all kinds!

At the request of medical students, pediatric residents, and medical genetics residents sequestered in their homes, but wanting to continue to be involved in patient care, we expanded our rotation size to allow each attending to have a "learner" dedicated to their telemedicine visit day. By assigning each learner to a mentor, they were able to "join" the telemedicine visits with the attending physician. This allowed them to not only learn about genetics or metabolism, but also about the intricacies of telemedicine. From learning how to help a family log-on to the telehealth portal, to eliciting an exam over a camera, to determining what you can tell about a child from their running around their home, to building rapport over an internet line, these interactions were useful to both the learner and the mentor. The technical requirement was only that the telehealth portal was able to allow entry to more than one individual to the visit. On our review of the available telehealth platforms, this was a common ability and in the future, this should be a requirement for any platform to be used in the genetics setting. Initially, it was though that this would be a "better than nothing" experience. Instead, it became a "this could be an amazing resource that I have learned to use for the rest of my career."

Due to the remote learning functions of the learners on our rotation, we were concerned that their experiences would not be consistent between different providers. To help address this issue, we began a seminar series for all learners on any of our rotations. From dysmorphology cases to metabolic 101 to identifying the case management needs of a family, the short educational series allowed the whole team to come together, share joys, share hurdles, and created a subcommunity within the greater community. The participation in each of these sessions varied based on the subject content and the number of faculty and staff joining in for the educational session. For session only for earlier learners, they were often with the attending and only a few learners (i.e., six participants) and for the larger activities of dysmorphology cases available to all clinicians, staff, and learners about 40 individuals joined to learn from each other.

We realize that every community, hospital, genetics program, and care team will have unique abilities to educate and expand the genetics community during this time period. It is our hope that this article will serve as a source of ideas, resources, and encouragement to find ways to expand the Genetics Community of Practice in the middle of the COVID-19 pandemic.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

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