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SPECIAL ARTICLE

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EuroGEMS.org: Guide and links to online genetic and genomic educational resources, valuable for all levels

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Abstract

There is a well-recognized growing need for improved access to online genetic and genomics education for professionals, students, teachers, and the public. Numerous individual online genetic and genomic educational resources have been developed, but many are difficult to identify or locate when required. Consequently, an easily navigated website, European Society of Human Genetics (ESHG) Genetic Educational Materials and Sources (https://www.EuroGEMS.org), has recently been created, on behalf of the ESHG, by the authors. It facilitates access by a wide variety of target audience types and levels to a broad range of 110 selected, free, high-quality educational online genetic and genomic resources around the world, including several in languages other than English. The website has been endorsed by the ESHG, directly linked from that society's web pages, and has now been used in over 105 countries.

KEYWORDS
education, ESHG, genetics, genomics, online, resources

1 | INTRODUCTION

A massive expansion has taken place in recent years in the development, discussion, and application of genetics and genomics, across all clinical specialties and in biomedical research. There has also been growing public exposure to genetics as a result of frequent media coverage of technological developments and ethical issues related to genetics and genomics, together with widening availability and marketing of direct-to-consumer testing. As a result, the perceived importance of genetics and genomics in the lives of the general public has markedly increased, despite its technological aspects often appearing daunting and even overwhelming (LaRue, McKernan, Bass, & Wray, 2018).

In fact, for several years, education has been recognized as a pivotal component of the successful introduction and utilization of genomics in the 21st century, and the provision of genomics education at all levels has been deemed critically important (Green, Guyer, & National Human Genome Research Institute, 2011). Now, more than ever, it is regarded as imperative that high-quality educational materials are made freely available, not only to genetics professionals but also to a range of target audiences at all levels, including school pupils, university students, patients, relatives, and non-genetics health professionals (Campion, Goldgar, Hopkin, Prows, & Dasgupta, 2019). Non-genetics specialists are, in fact, known to welcome the provision of educational support in this field (Crellin et al., 2019). This need has grown with the widespread adoption of diagnostic genetic and genomic testing in other clinical specialties (or "mainstreaming"). In addition, there may be a greater need for accessible genomic resources for patients and the public in countries or communities in which consanguinity is more common.

Responding to the resulting need for easily-accessible educational resources and the continuing exponential increase in the use of the internet for learning (McCutcheon, O'Halloran, & Lohan, 2018), there has been a remarkable growth in the number and scope of individual genetic and genomic educational resources available online. However, the intended audiences and types of content of these sources all vary considerably and are unfortunately often not apparent from the titles or from the internet addresses or uniform resource locators (URLs) of the resources. Moreover, many valuable resources are not those most highly ranked or most easily found by commonly used internet search engines. In addition, boosting genomics education in university courses can be particularly beneficial where it is otherwise underrepresented in medical or nursing curricula.

2 | DEVELOPMENT OF A GUIDE TO INTERNATIONAL ONLINE EDUCATIONAL GENETIC AND GENOMIC RESOURCES

2.1 | Aims

To assist a wide range of users in quickly identifying and accessing high-quality education resources that are up-to-date and most useful for their needs, a new educational website has therefore been created on behalf of the European Society for Human Genetics (ESHG). The aim of this website, ESHG Genetic Educational Materials and Sources (www.EuroGEMS.org) is to provide easily navigated guidance and secure, free, direct access to a wide range of types and sources of educational materials for a broad range of target audience types and levels (see Figure 1). They include healthcare professionals, university educators and students, primary (elementary) and secondary (high) school teachers, in addition to patients and their relatives. Multiple resources are linked from and described on each page. Links are also provided to several online resources (such as genome browsers, tools, and databases) that are primarily designed for genomic analysis by professionals but are also increasingly used in higher education.

2.2 | Initial development

Strict criteria were applied when studying and selecting online resources for inclusion in EuroGEMS.org. The criteria by which the resources were selected were that they should be (a) free-to-access, (b) informative, (c) of high quality and free of obviously misleading information, (d) useful, and (e) containing up-to-date links, without broken URLs (or "404 errors") that would suggest a failure to maintain and update the website.

The EuroGEMS.org website's content and structure were discussed in detail with other members of the ESHG Education Committee, at the planning stage, during development, and upon completion. It was also discussed with many hospital colleagues (including consultant clinical geneticists, genetic counselors, clinical trainees, and laboratory scientists) and university educators; postgraduate and undergraduate university students (in medicine, dentistry, genetics, and biological science); school teachers, school pupils and the general public at outreach events; patients and patient-s'relatives; and with the ESHG Board and the Presidents of National Genetic Societies across Europe. Helpful suggestions with regard to additional content were invited and received from many of these individuals and from many professionals located around Europe and North America. As a result, further incremental improvements and additions were made sequentially. The few suggested sites that have been excluded have, for example, referred only to educational videos for which payment was required, strongly promoted commercial direct-to-consumer genetic tests, included misleading information, or contained many broken (expired) links.

2.3 | Structure and content

The website currently contains direct links to 110 free, high-quality, carefully selected, genetics and genomics educational sources located on servers distributed around the world. Each link is accompanied by a brief summary of the principal uses and content of the target resource.

To enable simple navigation, the many resources listed and described in EuroGEMS.org are grouped into audience-specific categories. Separate detailed annotated and illustrated pages of links are provided for: genetics professionals, university students and educators, high school teachers, primary school teachers, and patients and relatives, in addition to the home page. Each page contains links and descriptions for up to 36 online resources and also includes a section with links to localized initiatives in languages other than English. A page for educational sources specifically covering ethical issues, was also included. Very recently, a page of links selected for primary care and non-genetic specialists has been added, in view of the increasing need for the provision of access to genetic information for healthcare professionals other than geneticists, particularly with the growing "mainstreaming" of genetic testing.

To provide examples of the content, the online resources currently included within the frequently visited page entitled "For Genetics Professionals" are listed in Table 1. The linked resources include many useful sites for clinical professionals (e.g., HGVS for variant nomenclature as well as gnomAD, Locus Reference Genomic, and Leiden Open Variation Database databases) and also for non-clinical university academics. The resources also include numerous articles, videos, free genomics-terminology, and quiz smartphone apps in addition to many genetics-related animations, discussion topics, and pre-prepared educational activities for schools such as "DNA origami."

New resources are added regularly, including several of the latest websites for genetic variant interpretation (such as Ensembl's Variant Effect Predictor, Varsome, CanVar UK, and Variant Validator), as well as local initiatives in languages other than English (such as Huisarts en Genetica, based in the Netherlands) and links to Massive Open Online Courses.

A large number of highly impressive educational resources have already been developed and made available online, internationally. The purpose of the pages provided here is to assist educators and individual learners in identifying sources of educational materials, related to genetics and genomics, that they may find useful. In addition to the many sources to which these pages link directly, many of the linked websites themselves include many further links to substantial educational resources.

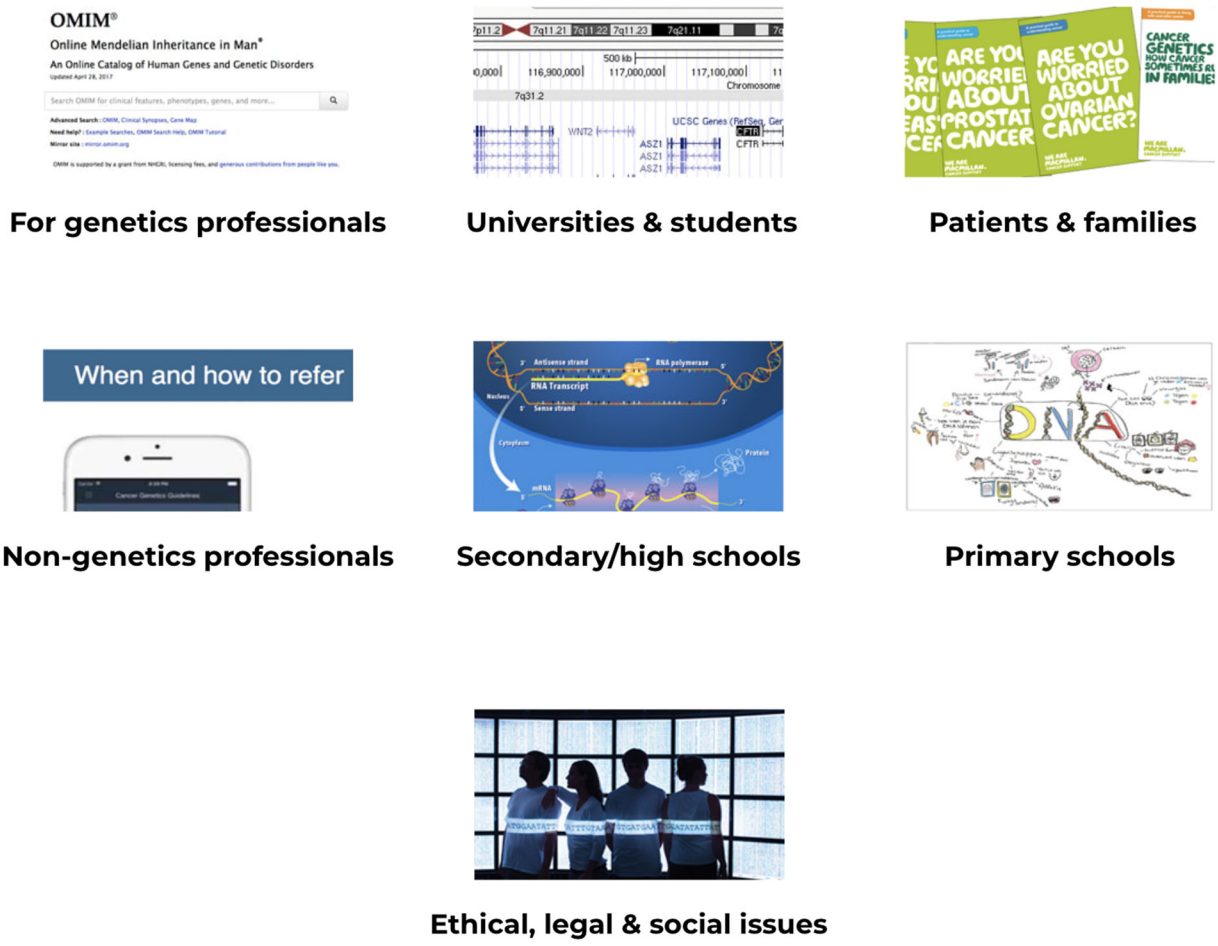

FIGURE 1 Home page at www.EuroGEMS.org showing the seven contained sections for a range of audiences, including the most recently added section (for non-genetics professionals)

2.4 | Technical aspects

A protected domain name was purchased, in addition to high-speed professional hosting with a high bandwidth (to permit a very large number of simultaneous users). Secure Sockets Layer cryptography was incorporated to ensure the security of transmitted data, resulting in the use of the https prefix in the website's full URL (https://www.EuroGEMS.org). Password-limited access was used to facilitate access and review by experts during the development phase of the website, before publishing. The website was also designed to be

TABLE 1 The online resources to which annotated links are currently provided within its most popular section (the page entitled "For Genetics Professionals")

COVID-19 links
COVID-19 resources for healthcare professionals
Clinical information
GeneReviews
OMIM
Orphanet
Genome browsers
Decipher
Ensembl Genome Browser
UCSC Genome Browser
Variant analysis and interpretation
Decipher
Leiden Open Variation Database (LOVD)
ClinVar
Broad Institute: gnomAD
Human Genome Variation Society (HGVS)
Human Gene Mutation Database (HGMD)
Varsome DNA variant analysis
Cancer predisposition (CanVar-UK)
Variant Effect Predictor (VEP)
Variant Validator
General information
Locus Reference Genomic (LRG)
Clinical Genomics Glossary and Quiz apps
European Reference Networks
Localized/National initiatives
Health Education England: Genomics Education Programme
PHG Foundation
NHGRI Genome TV
France Genomique: Training
Mediathek Humangenetik (Germany)
First German Academy for Further Medical Training on Rare Diseases
Dutch Guidelines for Cancer genetics referral
Scottish Genetics Education Network
Scottish Genomes Partnership
Massive Open Online Courses (MOOCs)
Clinical Bioinformatics: Unlocking Genomics in Healthcare
Cancer in the 21st Century: The Genomic Revolution
Genomic Technologies in Clinical Diagnostics: Next-Generation Sequencing
The Genomics Era: The Future of Genetics in Medicine
Genomic Medicine: Transforming Patient Care in Diabetes
Genome-wide association studies
NHGRI-EBI Catalog of Human Genome-Wide Association Studies
CDC Public Health Genomics and Precision Health Knowledge Base

displayed clearly using any browser, on any platform, including tablets and smartphones. Search engine optimization was achieved by analysis of the site using Google Webmaster Tools to verify the absence of any errors (whether loaded on computers or mobile devices) and a General Data Protection Regulation-compliant online service (Statcounter) was used to count the number of visitors as far as possible.

2.5 | Further development

Frequent additions and updates continue to be made, incorporating, where appropriate, links to carefully selected additional resources, including many links to local initiatives in languages other than English. Moreover, as mentioned above, an additional section has now been incorporated that has been specifically designed for non-genetic healthcare professionals, in view of the increasing use of genetics and genomics testing by non-geneticists. Furthermore, ongoing maintenance of the site is being undertaken using regular checking of all links to detect any non-functional URLs, with updating or removal of links carried out as required.

Additional sources will continue to be added. To this end, the footer of every page on the website requests that users get in touch, using the provided contact form, with any comments or suggestions for amendments, including the addition of up-to-date, high-quality, free educational sources. The corresponding author (Prof. Edward S. Tobias) also learns of additional resources from colleagues and through his various educational (and other professional) activities: attending conferences, running educational courses and sessions for postgraduate and undergraduate students, undertaking clinical genetics work, carrying out genetic and genomic research, meeting other clinical specialists and laboratory staff in regular multidisciplinary team meetings and as an active member of several international committees (including the ESHG Education Committee and the European Board of Medical Genetics).

In addition, the translation of the website into languages other than English, as suggested by the ESHG Education Committee, has now been commenced, firstly into Spanish (by a bilingual genetics professional). Subsequent translations into additional languages are planned. Future plans also include the addition of a search facility within the website, as suggested by a user of the website.

3 | INTERNATIONAL USE AND POPULARITY OF EUROGEMS.ORG

At the time of writing, the total number of page-views at the EuroGEMS.org website (excluding all those that could not be counted as a result of cookies being blocked or rejected) is 10,915 (using data received from Statcounter.com) on February 7, 2020. EuroGEMS has, by this date, been viewed from over 105 countries (see Figure 2 and Table S1), with approximately 20% of visits originating from

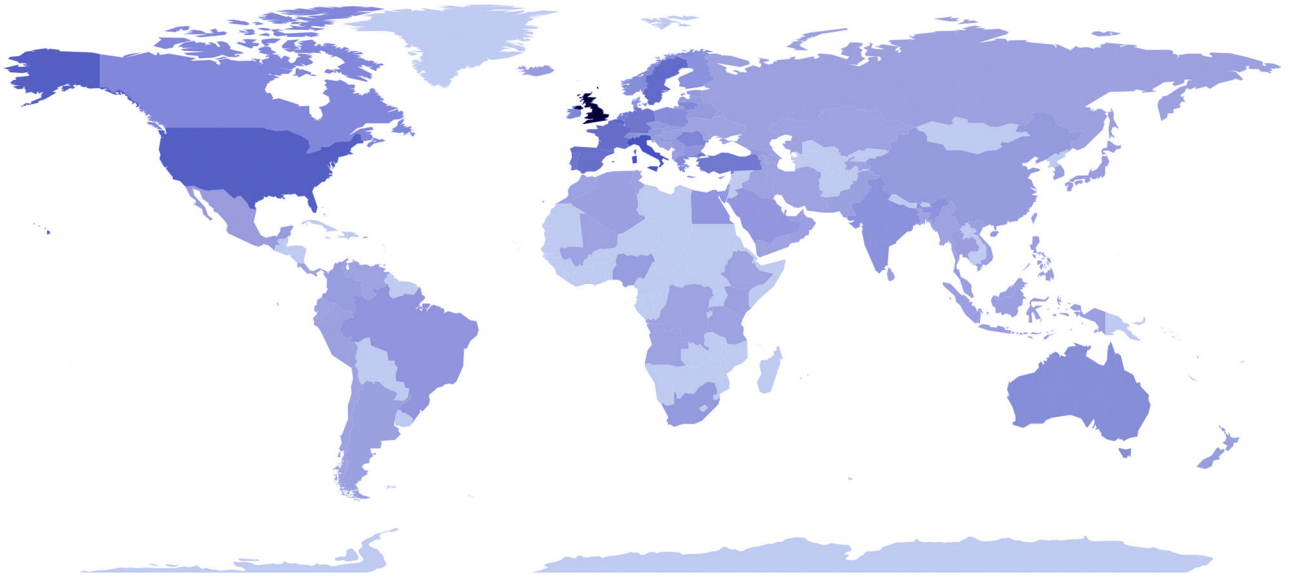

FIGURE 2 Countries of origin of visitors to EuroGEMS.org with 108 countries represented (with the intensity of shading according to the number of users). Analytics data from Statcounter.com

outside Europe (particularly from North America). Since the launch of the website, there has been a steady increase in the frequency of visits from returning visitors (see Figure 3). Returning visitors are important as they are an indication of the degree to which a website's initial visitors perceive the website to be sufficiently useful to be worthy of a further visit.

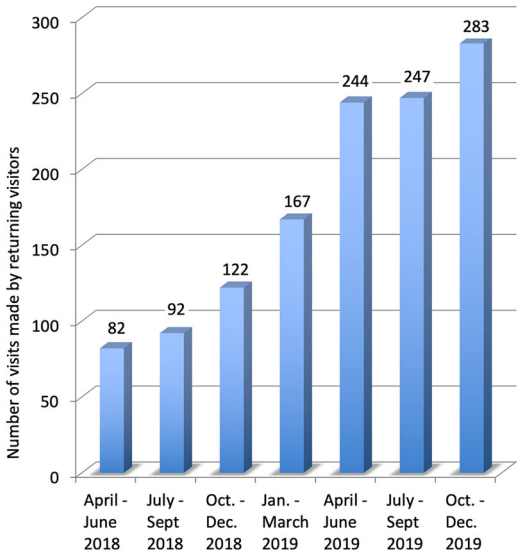

FIGURE 3 The use of the website since its launch in 2018, showing the total number of visits by returning visitors in each quarter-year, for the first seven quarters. The total in the seventh quarter was approximately 3.5-fold greater than the total in the first quarter. The greater increase in April to June 2019 may have been a result of repeated uses of the website by attendees of the ESHG Conference (in June 2019, in Gothenburg). Analytics data from Statcounter.com

TABLE 2 The 20 most frequently used exit links from the website

% of total	Exit link
6.22	https://www.yourgenome.org/
6.11	https://www.ensembl.org/index.html
4.44	https://www2.le.ac.uk/projects/vgec/schoolsandcolleges
4.33	http://www.scotgen.org.uk/information/leaflets/?page=1
3.33	https://www.genome.gov/10005911/genetic-education-resources-for-teachers/
3.33	https://genome.ucsc.edu/cgi-bin/hgGateway
3.11	http://www.ru.nl/wetenschapsknooppunt/english/
2.89	https://www.ncbi.nlm.nih.gov/books/NBK1116/
2.89	http://www.genecards.org/
2.56	https://ghr.nlm.nih.gov/resources
2.44	https://www.contact.org.uk/
2.00	https://vimeo.com/60747882
2.00	http://www.kumc.edu/GEC/
2.00	http://www.eurogentest.org/index.php?id=226
2.00	http://learn.genetics.utah.edu/content/basics/
1.89	https://www.primarycaregenetics.org/?page_id=109&lang=en
1.78	https://www.omim.org/
1.78	https://www.genome.gov/education/
1.67	https://www.youtube.com/playlist?list=PLHdemSStztKa79voJGhTkk7sinywEG9qz
1.67	https://www.jeansforgenesday.org/educational-resources

Note: The number of hits shown is displayed as a percentage of the total that was recordable, within a period of 1 year.

The page most often visited to-date (other than the Home page) is the page "For Genetics Professionals," closely followed by the page for Universities and Students, then the pages for Secondary Schools; Patients and Families; Primary Schools; Ethical, Legal, and Social Implications; and finally, the page for Non-genetics Professionals (which was only recently added).

The majority (52.0%) of visitors to EuroGEMS.org have reached the website directly by using the URL, whereas another 41.3% have reached it via the ESHG website. Smaller proportions (6.0% and 0.7%) of visitors have arrived via a web search or Facebook, respectively.

The most commonly used exit links from the website (in order of decreasing frequency) are listed in Table 2, with the most frequently used link being www.yourgenome.org, perhaps as a result of the quality, usefulness, and range of concise educational information that it contains.

4 | TESTIMONIALS AND ENDORSEMENT FOR EUROGEMS.ORG

Feedback received from university students and international professionals has consistently been highly positive and is detailed in Table 3.

TABLE 3 Written feedback comments received from university students (anonymously), professionals in genetics and genomics across Europe, and a secondary (high) school head teacher

From university students (undergraduate and postgraduate):

- "The website was wonderful for accessing genetics databases and looking up genetic conditions."
- "The EuroGEMS website was particularly useful."
- "We were all very grateful for all of the resources."
- "Thank you for igniting my passion for genetics."
- "Excellent. All links have been really useful."
- "If more topics were taught like this then it would make a big difference."
- "It has made me consider pursuing further learning opportunities in this area of medicine."
- "Informative, amazing … sparked my interest in genetics."

Professionals' written comments and endorsements:

- "I was amazed! …How much we could profit from your amazing site."
- "I am really impressed by your terrific work."
- "Congratulations on making EuroGEMS.org truly beautiful and comprehensive. I can clearly see how much effort has gone into this."
- "Simply wonderful….A suggestion: a searchable database by subject, to be placed at the home page, could be very helpful…."
- "Thank you once again for the excellent web resource overview you have made!"
- "EuroGEMS.org is great! I think this is absolutely brilliant."
- "Congratulations on creating an excellent, comprehensive, and highly useful resource for education on the ESHG web page!"

From a head biology teacher at a secondary school:

- "Your website looks terrific… I will certainly share the website within the department and I am confident that it will be very useful indeed."

The details of the aims, structure, and content of the website were presented, upon invitation, to the ESHG Board and also to the committee of the National Human Genetics Society (NHGS) presidents, at the annual ESHG Conference in Gothenburg in June 2019. A slightly updated version of the presentation can currently be downloaded from an ESHG web page (https://www.eshg.org/index.php?id=84). That presentation provides additional details, particularly regarding the links and annotated content within each of the pages. Direct links to EuroGEMS.org have been created from the ESHG website, on which it has been described as "the excellent website dedicated to educational resources" and as "the dedicated ESHG website containing a wide range of educational resources for genetic and genomic education."

5 | CONCLUSIONS

A website has been developed that enables a wide range of individuals to easily access a broad range of high quality educational genetic and genomic resources around the world. Many of these valuable resources, all of which have been carefully selected and are free to access, would otherwise be considerably more difficult and time-consuming to locate. The opinions of many potential users from a range of target audiences have been taken into account in the initial and ongoing development of this website. In combination with the authors' custom-designed free educational smartphone apps (www.genomicsapps.org; Tobias & Tobias, 2015), it has already been found invaluable in teaching postgraduate and undergraduate students, who, in turn, have expressed their considerable appreciation in anonymous written feedback. Much highly positive feedback has also been received from clinical professionals and from university and high-school staff (see Table 3).

The EuroGEMS.org website has been enthusiastically welcomed and endorsed by the ESHG, and there is a steady growth in the number of its returning visitors. Although over half of the visitors now arrive directly (presumably by entering the name or URL of the website into a browser), it is suspected that many potential users worldwide are still unaware of the website's existence. It is hoped that readers will inform others of the website and that other organizations and authors will consider providing a link to it for their readership. In fact, it is understood that the authors of the forthcoming new edition of a class-leading genetics textbook are strongly considering incorporating it as the core internet resource.

6 | INVITATION FOR COMMENTS

The website's authors would welcome any comments and also further suggestions for any additional online educational source (particularly in languages other than English) that is free-to-access, useful, and of high quality, together with a brief summary of

the source's target audience, general content, and specific advantages. Please address correspondence to edward.tobias@glasgow.ac.uk

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CONFLICT OF INTERESTS

Adam P. Tobias and Edward S. Tobias are the authors of the www.EuroGEMS.org website; the new COVID-19 educational web-pages for professionals and patients at www.ScotGEN.org.uk; and also, the free (and not-for-profit) educational genomic apps at www.genomicsapps.org. Adam P. Tobias created the www.uems-ecmgg.org website for the European Certificate in Medical Genetics and Genomics (ECMGG) professional examination. Edward S. Tobias is the Clinical Director of the MSc in Medical Genetics and Genomics at the University of Glasgow, United Kingdom (www.gla.ac.uk/postgraduate/taught/medicalgeneticsandgenomics).

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REFERENCES

Campion, M., Goldgar, C., Hopkin, R. J., Prows, C. A., & Dasgupta, S. (2019). Genomic education for the next generation of health-care providers. *Genetics in Medicine*, *21*(11), 2422–2430.

Crellin, E., McClaren, B., Nisselle, A., Best, S., Gaff, C., & Metcalfe, S. (2019). Preparing medical specialists to practice genomic medicine: Education an essential part of a broader strategy. *Frontiers in Genetics*, *10*, 789.

Green, E. D., Guyer, M. S., & National Human Genome Research Institute. (2011). Charting a course for genomic medicine from base pairs to bedside. *Nature*, *470*(7333), 204–213.

LaRue, K. M., McKernan, M. P., Bass, K. M., & Wray, C. G. (2018). Teaching the genome generation: Bringing modern human genetics into the classroom through teacher professional development. *Journal of STEM Outreach*, *1*(2), 48–60.

McCutcheon, K., O'Halloran, P., & Lohan, M. (2018). Online learning versus blended learning of clinical supervisee skills with pre-registration nursing students: A randomised controlled trial. *International Journal of Nursing Studies*, *82*, 30–39.

Tobias, A. P., & Tobias, E. S. (2015). Developing educational iPhone, Android and Windows smartphone cross-platform apps to facilitate understanding of clinical genomics terminology. *Applied and Translational Genomics*, *6*, 15–17.

SUPPORTING INFORMATION

Additional Supporting Information may be found online in the supporting information tab for this article.

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