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Developing educational iPhone, Android and Windows smartphone cross-platform apps to facilitate understanding of clinical genomics terminology

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1. Introduction

Clinicians are increasingly using genomic technologies to determine the molecular basis of presenting conditions, identify increased risk factors or guide treatment (Deciphering Developmental Disorders Study, 2015). Unfortunately, however, the bioinformatic terms and acronyms that are frequently used in the new field of Clinical Genomics (the study of all genes simultaneously, rather than of individual genes as in Clinical Genetics) can often be unfamiliar to many health professionals (Institute of Medicine Roundtable on Translating Genomic-Based Research for Health, 2015). These include clinicians who, increasingly, need to be able to accurately interpret clinical genomics-related laboratory reports and to understand genomics research papers and seminars. This is in addition to the requirement for these individuals to participate in discussions with genetics lab scientists and, on occasion, bioinformaticians. Unfortunately, online information can be unhelpful as the genomics definitions provided on the internet are often written for readers with much previous genomics-related knowledge. Excellent online summaries of genetic conditions and of standard genetics terms are available via GeneReviews (http://www.ncbi.nlm.nih.gov/books/NBK1116/), UpToDate (http://www.uptodate.com/home) and the National Human Genome Research Institute (https://www.genome.gov). These sources, however, currently do not provide explanations of many of the technical terms that relate to the field of genomics. Moreover, mobile internet access can be intermittent or slow, for instance, on public transport in the UK. A self-contained mobile app (application), in contrast, can provide a user-friendly, rapid and accessible alternative information source with its customized contained information being available offline. Furthermore, a high proportion of professionals and postgraduate students now use Apple (iOS), Android or Windows smartphones (Sandholzer et al., 2015; Payne et al., 2012).

1.1. Objectives

The objective was primarily to provide a concise bioinformatics dictionary for clinicians that is easy to access and to understand. Specifically, the aim was to create a smartphone app containing a small glossary of commonly used (but often unfamiliar) genomics terms (eg "FASTQ" and "PED file") that would be user-friendly, easily accessible, self-contained and accompanied by illustrated explanations. The app containing this information would, ideally, be capable of running on Android or iOS portable devices and on a PC. An additional objective was to create an interactive quiz as a self-assessment system to test the learning of this information, with a personal points score. Currently, no similar electronic mobile self-contained glossary exists that covers the highly specialized terms used in clinical genomics.

2. Methods

2.1. Software and hardware requirements

The software required for creating multi-platform apps included the latest Apple Xcode programme (version 6.3.2), together with both Apple iOS and Android software development kits (SDKs), versions 8.3 and 1.6.0, respectively, which were required in order to build device-specific apps. Creating apps for iOS devices also required an Apple iOS Developer Programme Licence.

The hardware required for app creation was an Apple Mac running the latest available operating system, OS X Yosemite (version 10.10.3).

2.2. The app creation process

A high-level app encoding language linked to iOS and Android SDKs, following installation on a MacBook computer, was used to build the apps. Apps were tested on iOS (iPod, iPhone and iPad) and Android (Samsung Galaxy smartphone) devices, as well as on Windows PCs. Apple's Xcode software was employed in order to transfer the app files directly onto testing devices running iOS operating systems.

2.3. Information sources

The text incorporated within the app was summarized from the information that is shortly to be provided in an award-winning Glasgow

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University course for postgraduate students and professionals (http://www.gla.ac.uk/postgraduate/taught/medicalgenetics/), in addition to the information in two internationally popular genetics textbooks (Tobias et al., 2011; Tobias & Connor, 2014) and an educational website (www.essentialmedgen.com) written by one of the authors, Edward Tobias.

2.4. Information covered

The genomics and bioinformatics terms covered within the app comprise, primarily, commonly-used sequence variant-related terminology (e.g. "SNV", "benign", "pathogenic", "somatic" and "germline"), frequently-encountered genomic file types (e.g. "FASTQ", "PED file", "BED file", "BCF" and "gVCF") and genomic bioinformatic software tools such as "BWA", "GATK" and "IGV".

2.5. Challenges encountered

An illustrated educational app plus an accompanying self-assessment quiz app were both created and then tested by the apps' developers and by several users, on Android, iOS and Windows devices. It was found that the creation of the educational text and images, as well as the code required for the apps to run, although a highly significant task in itself, only amounted to a fraction of the work involved in app creation. The many steps that required to be undertaken included obtaining and installing the latest software versions. For instance, while Apple's Xcode software (https://developer.apple.com/xcode/) contained the required SDK, installation of the Android SDK, in contrast, necessitated the installation of Android Studio software (http://developer.android.com/tools/studio/index.html) followed by the further installation of SDKs using the Android SDK Manager. The processes for App distribution (particularly on iOS devices) required the acquisition of an iOS Developer Programme licence (https://developer.apple.com/programs/) from Apple, the registration of individual app licence IDs and then the acquisition of Apple app "provisioning profiles". Furthermore, in order to provide optimized display and full compatibility with the latest devices such as the iPhone 6, it was necessary for each iOS app to be created with the appropriate range of app icons and splashscreens for display on the different devices themselves as well as multiple versions of screenshots for display on the App Store.

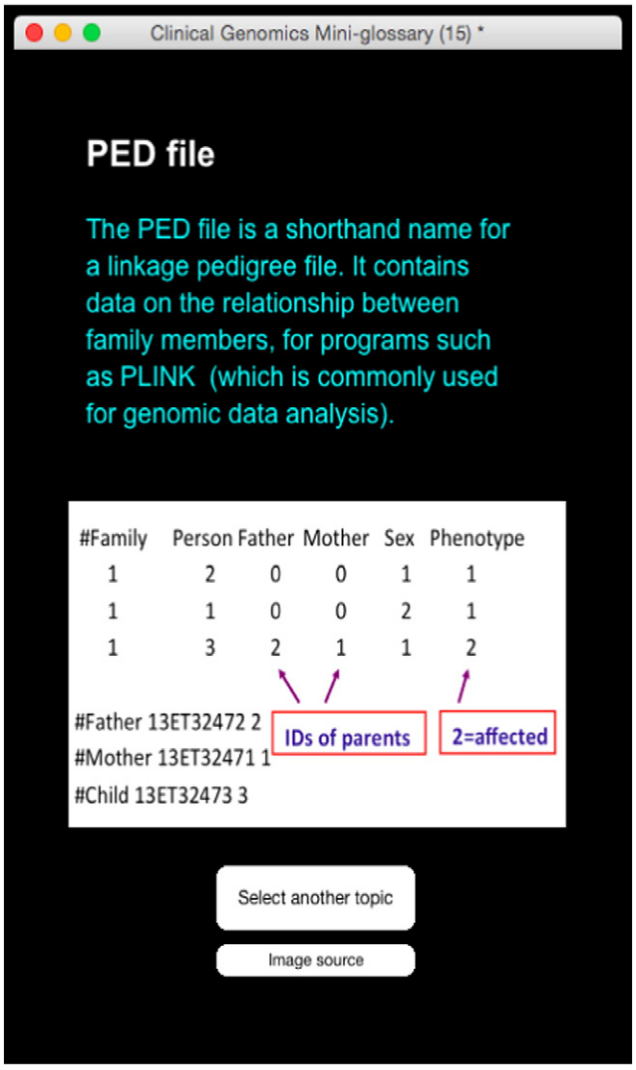

Fig. 1. Screenshot of app information screen providing a synopsis and illustration describing the PED file format.

3. Results

The authors created two educational apps related to clinical genomics terminology, providing straightforward definitions of terms relating to several genomic data file types, computer programmes and variant types. The first such app (Clinical Genomics Mini-glossary (Tobias & Tobias, 2015a)) was designed specifically to provide rapidly and easily accessible concise illustrated definitions of several of the most frequently used terms, including a PED file (Fig. 1), a FASTQ file (Fig. 2) and a BED file. The app also illustrated the principal steps involved in the generation of variant call format (VCF) files from DNA, including read alignment and variant calling and showing the types of data files that are involved at the various stages, such as FASTQ and BAM files.

The second app generated (Clinical Genomics Quiz (Tobias & Tobias, 2015b)) was a self-assessment quiz app, designed to test the user's knowledge of the various terms, in which the closest genomics-related acronym or term must be selected from a list of possible answers, for each description displayed. The questions are presented in a random order, with a single mark awarded for any correct answer given on first attempt.

End-users, including postgraduate (Masters-level) students and clinical professionals (at trainee and consultant level), were consulted during the design process in order to ascertain which genomics terms were most frequently encountered and also which terms were generally unfamiliar.

The completed apps were demonstrated on Apple iOS devices (iPod, iPhone and iPad) and Android smartphone at the European Society of Human Genetics conference held in Glasgow, Scotland in June 2015. The apps were tested there by 25 professionals, including clinical geneticists, senior laboratory scientists, genetic counsellors and leaders of the major educational programme of Genomics England. Extremely positive feedback was received when demonstrating the apps to these conference attendees, visiting from many different countries around the world, including England, Scotland, US, Canada, Mexico, Australia, Estonia, Latvia, Spain, Portugal, Netherlands and South Africa. In fact, many of them requested full use of the apps "as soon as possible". Feedback from all of the many individuals who used the apps was highly positive, with the apps being described for example, by the director of the diagnostic genetic laboratory of South-west England as "a great learning tool".

In addition, one consultant clinical geneticist commented that the genomics glossary app provided concise helpful explanations of the precise terms ("FASTQ", "BED file" and "gVCF") that she had just heard mentioned, unexplained, during a genomics lecture. She commented that possessing such an app on one's smartphone would enable a listener to quickly determine the meaning of unfamiliar terms without having

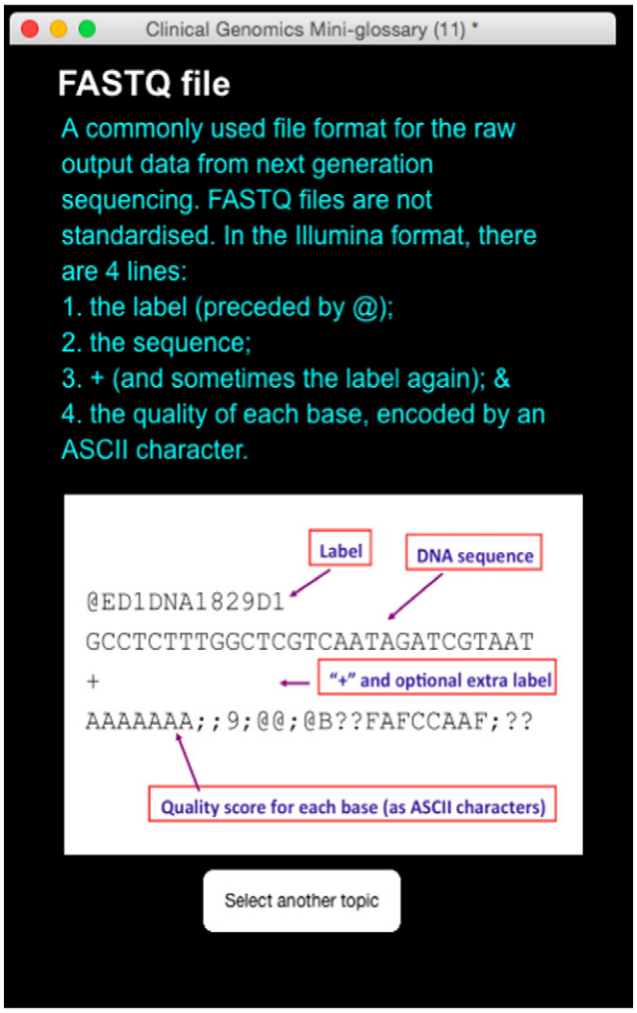

Fig. 2. Screenshot of app information screen providing a synopsis and illustration describing the FASTQ file format.

to ask the speaker for clarification or to attempt to browse the internet during lectures.

No-one who used the apps could identify any important genomics-related term or acronym that was absent, except for one bioinformaticist who commented that a definition of "SAMtools" should be added. This is now underway and the apps are shortly to be submitted, initially to Apple's App Store.

3.1. Student feedback

Prototype versions of the authors' apps were offered as downloads from a university virtual learning environment (Moodle 2) system, to all 273 of the year-3 undergraduate medical students. These students had received teaching related to medical genetics on topics such as Mendelian inheritance mechanisms, mitochondrial inheritance and cancer genetics. The apps proved to be highly popular, with 978 total downloads (mean: 3.58 downloads per student). Anonymous feedback, collected electronically from the students, regarding the supplementary learning sources provided, revealed that 100% of responders felt that these were helpful.

4. Conclusions

The creation and distribution of Apps can be challenging, involving many steps. Several obstacles had to be overcome by the authors, including the fulfilment of all the requirements for the App Store (for example for multiple icon and screenshot resolutions). Nevertheless, useful instructive and self-assessment iPhone, Android & Windows multiplatform smartphone apps were created that explain (and test knowledge of) commonly used genomics and bioinformatics terms and acronyms.

Smartphone apps represent a convenient, rapid and popular means of obtaining information and enhancing learning. Apps can provide an important modern component of technology-enhanced learning and teaching. These apps provide an important direct source of genomics-related information that is not otherwise easily accessible.

Based on many discussions with physicians, genetic counsellors, genetic laboratory scientists and students it is anticipated that these clinical genomics apps will be used in the future most frequently by clinical professionals who receive genetic laboratory reports that they must interpret for the benefit of patients, as well as by genetics professionals and postgraduate students during genomics or bioinformatics lectures and when reading genomics articles.

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