

# The Need for Development of Medical Genetics and Genomics Curriculum for Medical Schools in Saudi Arabia

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Received 11 February 2019;

Accepted 25 February 2019;

Published 01 March 2019

## Abstract

Next generation genomic tools including dense SNP arrays and high throughput sequencing technology have led to the exponential increase in delineating genetic defects in rare as well as common genetic disorders. Advanced genetic tools have found clinical applications in almost all areas of medicine specifically in clinical diagnostic setting. Genomic testing has entered clinical practice and physicians are using genomic information to guide treatment options. Medical schools throughout the world have integrated medical genetics and genomics courses in the curriculum to prepare medical students and future physicians to understand the role of genetic factors in human health and disease. However, in Saudi Arabia, the existing undergraduate curriculum in most of the medical schools is insufficient to prepare medical students to cope with rapidly advancing genomic science. Moreover, the majority of medical schools in Saudi Arabia have no geneticist as an academic staff, and where there are such academician available relatively less teaching time is allocated for them and negligible use is made of their specialized knowledge. Overall in Saudi Arabian medical schools, little genetics teaching is given to medical students. An objective oriented genetic and genomic course need to be integrated in medical school curricula which would subsequently prepare generation of the student who can develop a critical understanding and knowledge of core concepts in medical genetics and genomics.

**Keywords:** Medical Education, Genetics and Genomics, Medical Schools, Saudi Arabia, Next generation Sequencing Tools

## Introduction

Medical practice has been transformed by new information in genetics and increased understanding of molecular basis of human disease. In the past decade, technological advancement in molecular biology has led to the rapid progress in biotechnology which subsequently directed countless discoveries in medical genetics. Next generation sequencing technology has achieved enormous improvement in generating sequencing reads by employing massively parallel sequencing strategy. Whole exome and even whole genome can be sequenced in few days and in a cost effective way. The technological advancement played important role in the determining disease, interpretation of clinical laboratory data, proper use of therapeutic agents, disease treatment and patient counseling (Feero *et al.*, 2010; Mayer *et al.*, 2011). Throughout the world, medical curriculum development committees recognized that clinicians entering into practice need to understand human genetics in more detail. Healthcare authorities and medical educators agreed on strong need of advance genetics and genomics curricula in medical schools so that medical students and future physicians get enough training to be able to understand basic principles of genomics and are capable of interpreting personal genome testing results (Wiener *et al.*, 2010)

## Why a Physician Need to Understand Genetics and Genomics?

Standard molecular tests usually involves analysis of gene rearrangements or particular gene variants while genomic test refers to the ability to analyze large portions of the genome with a single "test." Genomic test may involve sequencing of all nucleotide bases (*whole genome sequencing*), or just the protein coding part (*whole exome sequencing*) or only those genes that are expressed (*whole transcriptome sequencing*). Next-generation sequencing (NGS) methods are cost effective and time efficient. The cost per test is rapidly decreasing and the turnaround time has reduced from years to weeks. Gene rearrangements, large insertions and deletions and global expression analysis can be measured by microarray technology where millions of single nucleotide polymorphisms (SNPs) are genotyped in a single test.

The field of genomic medicine is advancing at an unmatched pace, and many primary care and specialty physicians routinely use genomic test results to guide and individualize patient management. Incorporating next generation genomic technologies in a medical setting require clinicians to enhance their understanding of genetics and genomics and the role of genomics

in disease pathology. Disease diagnosis based on genomics, such as association of single nucleotide polymorphisms, mRNA expression profiling, and protein analysis, has the potential to improve healthcare through disease prevention, predictive diagnoses, improved prognoses, and personalized treatment regimens. In order to use such next generation genomic tools effectively, physicians need to know their benefits, risks, utilities, and limitations. Moreover, based on patients' genomic profiles, physician must understand the patient management and counseling.

### **Genetic testing in disease diagnosis and need for advanced genetic curriculum**

In order to diagnose monogenic disorders and chromosomal aberrations, routine genetic testing is used. Moreover, genetic testing has also been used to calculate the risk of single-gene disorders in unaffected and carrier individuals. In developed countries, increased used of genetic testing has been observed for common and multifactorial disorders where the outcome of genetic test is used to tailor therapy and predict disease prognosis. Next generation genomic tools including dense SNP (single nucleotide polymorphism) microarrays and massively parallel sequencing instruments have the ability to investigate thousands of variants in a genome in a cost effective and timely manner (Bentley *et al.*, 2008; Drmanac *et al.*, 2011). Physicians from different hospitals in Saudi Arabia including King Khalid University Hospital (KKUH), King Fahad Specialist Hospital (KFSH), King Faisal Specialist Hospital and Research Center (KFS&RC), King Abdullah Medical City are recommending patient samples for gene panels testing to guide treatment options or submitting samples for clinical whole exome sequencing to test maternal blood for fetal risk of Down syndrome and inherited disorders. Reports have revealed that most clinicians failed to understand and interpret simple genetic diagnostic tests and reports (Guttmacher *et al.*, 2007; Plon *et al.*, 2011). Regrettably, the existing undergraduate curriculum in most of the medical schools is insufficient to prepare medical students to cope with rapidly advancing genomic science (Thurston *et al.*, 2007). Incorporating cytogenetics along with molecular genetics and genomics in the curriculum of medical school and exercises related to genomic annotation and communication of results to patients would help in preparing the generation of clinicians who can use genetic and genomic data for patient management and treatment (Nelson and McGuire, 2010; Korf, 2011). Practicing clinicians need to consider advanced genomic tools and must be able to interpret the results obtained from genomic DNA of patient. So that, they can make decision regarding patient management and disease treatment.

### **High burden of genetic diseases in Saudi Arabia**

Several reports have shown that the rate of cousin marriages is very high in Saudi Arabia (KSA). Cousin marriages led to increased inbreeding which subsequently results in a high rate of autosomal recessive genetic disorders in human populations (El-Mouzan *et al.*, 2008 Bittles, 2008; Al-Owain *et al.*, 2012; Hannan *et al.*, 2015). Incidence of genetic disorders in Saudi population is one of the highest in the world (Hannan *et al.*, 2015; Sulaiman and Al-Owain, 2018). The burden of genetic diseases, in particular, autosomal recessive disorders can be reduced by the effective planning of culturally sensitive, genetic prevention services of high ethical standards at national level. Education of both medical students and the public is needed concerning the prevalence of genetic diseases in the population.

### **Department of Medical Genetics in Saudi Medical Schools**

Department of Medical genetics of Al Faisal University has initiated a genetic counseling program. The aim of this program is reduce the burden of genetic diseases by educating and screening families, pre-marriage counseling and looking for preventive reproductive choices. In order to look for genetic department in medical school, I searched websites of eleven Saudi universities. I found that only two universities has department with the term "genetics/genomics". King AbdulAziz University in Jeddah has department of Genetic Medicine and King Faisal University Riyadh has department of Anatomy and Genetics. I also found a Genetics Division in the Department of Pediatrics in King Saud Bin AbdulAziz University of Health Sciences.

### **Involvement of teaching faculty in Genetic Research and Vice Versa**

Medical schools should encourage and involve teaching faculty both basic and clinical genetics research. There are two funding agencies, deanship of scientific research (DSR) for intramural grants, and King Abdul Aziz City for Science and Technology (KACST) and Research Development Office (RDO) of the ministry of education, at the national level.

Extensive search of faculty members of the Taibah University (Author is employed in Taibah University) revealed that faculty working in medical school and other colleges of Taibah University have completed several research projects related to genetic and genomics. Research publications have shown that experts in emerging genetics and genomic technologies are part of the Taibah University, however, they are involved only in research work and their role in academic teaching and curriculum development is minimal (Alharbi *et al.*, 2008; Basit *et al.*, 2018; Alharby *et al.*, 2017). These studies would be helpful in starting a future graduate-research programs (Msc, PhD) in genetics and thus can provide a genetic laboratory for the practical sessions of students from medical school.

### **Practical Sessions Positively Impact Learning Process**

Laboratory courses should be structured largely. These courses must reflect the material presented in the theory. Wet laboratory experiments related to basic genetics failed to produce much excitement. However, experimentation of clinical relevance attract student attention. A comprehensive and updated medical genetics experimental manual need to be compiled. Laboratory experiment in which student is a subject may enhance learning. Students can be offered a personal genome testing as an elective course. This course must consists of interactive lectures and hands-on analysis of genomic data. They can be involved in analyzing their own personal genomic data or data from publicly available databases (Salari *et al.*, 2011). Study have shown that using personal genomic data can enhance the learning ability of students in those courses which teaches concepts of genomics and genetic testing (Salari *et al.*, 2013).

### **Curriculum design with integration of molecular pathology with genomics**

A curriculum can be designed keeping in mind the current technological advancement and community need. For example, the summary of approaches used for quantifying DNA, lecture related

to Sanger sequencing and an overview of next generation sequencing technology. Further lectures on NGS methods and SNP based genotyping as well as topics on applications of molecular pathology and genomics in hematology, pharmacogenomics, solid tumors and inherited disorders.

Moreover, lectures discussing institutional ethical approval, regulatory bodies and commercial issues related to personal genomics. Furthermore, a brief overview of genomic data analysis tools in research and clinical settings, annotation of NGS data and American College of Medical Genetics (ACMG) guidelines of variant interpretation.

## Conclusion and Recommendations

Author searched medical school curriculum of medical schools located in technologically advanced countries. I found that Baylor College of Medicine has implemented a new syllabus that improves genomic understanding of medical students. The curriculum is designed with an ultimate goal of enabling medical students to provide more effective and personalized medical care. Authors believe that the genetics and genomics syllabus at Baylor College of Medicine can serve as a prototype for Saudi medical schools. It is recommended that recent development in genetics and genomics must be incorporated in curriculum of medical schools. Moreover, interactive and participatory approach of using genomic testing data has the potential to increase student's knowledge and awareness of genetic testing. Author recommends a genetics and genomic course which may include DNA/RNA structure and function, traditional and non-traditional inheritance patterns, population diversity and genomic variations, methods used in diagnostic laboratory, clinical bioinformatics approaches, interpretation of results and assessment of genetic risk. Additional lectures related to ethical issues in genetic testing, social and cultural practices, psychological issues in genomic medicine/healthcare and population and public health aspects of genomics would result in further benefits.

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