Knowledge on Genetic and Genomic Diagnostics among Sri Lankan Medical Practitioners

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Research note

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Abstract

Objectives : Up-to-date knowledge on changing genetic and genomic diagnostic approaches is essential for a contemporary medical practitioner. This study was carried out to describe Sri Lankan Medical Practitioners' current knowledge and opinion on modern genetic and genomic diagnostics techniques.

Results : Data was collected to an electronic database using an online self-administered questionnaire. A total of 123 respondents completed the questionnaire. Majority had either some or adequate knowledge on applications of basic genetic diagnostic methods such as karyotyping and polymerase chain reaction. Similarly most had either some or adequate knowledge on the use of predictive genetic tests in cancer. However the level of knowledge on the applications of modern diagnostic techniques like Fluorescent in-Situ Hybridization and Next Generation Sequencing remained poor. Majority had inadequate knowledge on newer concepts such as direct-to-consumer genetic testing and personalized medicine. Most agreed upon the inadequacy of undergraduate genetic education and the need of a continuous medical education program to fill this knowledge gap. Both undergraduate and continuous medical education approaches should be modified to improve the knowledge on increasingly complicated technological and ethical aspects associated with modern genetic diagnostics.

Introduction

Lack of knowledge in genetics and genetic testing among non-geneticist doctors has been identified as a global problem in medical education. Studies have shown that there is a significant weakness among clinicians particularly in interpreting genetic tests(1,2). This would be a limiting factor in integrating genomics in to the medical practice(3).

A study in five European countries describes significant deficiencies regarding medical genetics particularly among primary care physicians. It also states the need of a coordinated European effort to improve the background knowledge on medical genetics among primary care physicians(4). Another study carried out among primary care physicians in Italy shows that the majority had inadequate knowledge regarding proper use of genetic testing(5).

With the development of the Next generation sequencing (NGS), the cost for DNA sequencing has been reduced dramatically and it has enabled us to use sequencing data in a wide range of clinical practice. It has particularly revolutionized use of genetic data in the management of Mendelian, and complex diseases including cancer as well(6–8). However level of physicians knowledge remains a barrier for delivering benefits of genomic medicine(9).

Diagnostic methods such as Array Comparative Genomic Hybridization (array CGH) and Fluorescence in situ hybridization (FISH) are being used increasingly in today's clinical genetics practice¹⁰. Array CGH is a method of high resolution genome wide screening of segmental Copy Number Variations (CNV) used to detect unbalanced chromosomal abnormalities(11). FISH is a technique used to identify numerical and

structural chromosomal abnormalities and to determine the chromosomal location of specific sequences(12). Single nucleotide polymorphism (SNP) microarray is another versatile diagnostic technique which has many benefits other than diagnosis (13).

Awareness regarding common cancer predicting genetic tests like BRCA1/2 is generally high among physicians(14). With the advancement of our knowledge in cancer biology, use of genetic diagnostic goes beyond predicting the risk. Cancer treatments are also becoming more and more targeted and tumor specific. Gene expression data has become more important in predicting outcome of modern cancer treatments(15). Oncologists and other physicians have indicated the need of increasing genomic literacy and the need for more education about genetic testing for susceptibility of cancer (16,17).

Direct- to- consumer genetic testing (DTC-GT) is also becoming popular due to increased consumer access via internet and greater autonomy and privacy. These tests can be ordered online without a physician's referral(18). Studies have identified the need of improving the level of awareness among general public as well as health professionals regarding DTC-GT due to potential disadvantages associated with it(19).

Genetic profiling rather than single gene testing would help to identify individuals with high risk for common complex diseases. It would be beneficial since appropriate preventive measures can be taken well before the development of the diseases (20,21). Studies have shown that there is a significant knowledge gap regarding pharmacogenomics and the need of clinically oriented educational resources especially electronic resources to fill this gap (22).

In Sri Lanka availability of clinical genetic services dates back to 1981 and genomic medicine was implemented in 2010(23). However there is a paucity of scientific evidence about physicians' knowledge regarding different genetic diagnostic methods and their application in the clinical practice.

Main Text

This cross sectional descriptive study was carried out using an on-line questionnaire created using Google forms. Google form settings were adjusted allowing only single response per participant. Convenient sampling technique was used and an email requesting participation of this online study was sent to 973 individuals. Email contained a link to the questionnaire and informed consent was taken in the form of a dialog box. Anonymous responses were collected to an online database. Email addresses were obtained using official websites and through personal contacts. The questionnaire was pre tested among 5 medical graduates and relevant changes were done accordingly. It consisted of three components. First component was used to gather demographical data. Second component was used to assess the level of knowledge regarding different genetic and genomic diagnostic techniques. Third component was used to identify the attitudes and opinions regarding the limitations of their knowledge and clinical genetics in practice.

Results

Out of 973 individuals contacted via emails only 123 (12.64%) replied. Their age ranged from 26 to 48 with the mean age of 33.87 years (SD=5.02). All but three participants were graduates from local universities. Sixty individuals (48%) either had either finished post graduate studies or involved in post graduate studies at present.

Medical officers/ senior house officers were the largest (35.5%%) group followed by Postgraduate trainees(30.6%). Rest of the sample was composed with intern house officers (14.9%), consultants(10.7%) and university academics (8.3%).

Table 1 illustrates the responses given for different knowledge components.

Knowledge component	Adequate	Some	Poor
Applications of Karyotyping and Polymerase Chain Reaction (PCR)	24.4%	65.8%	9.8%
Applications of FISH	3.2%	35%	61.8%
Applications of SNP arrays	-	22%	78%
Applications of Next Generation Sequencing(NGS)	3.2%	18.8%	78%
Applications of array CGH	-	9.8%	90.2%
Applications of predictive genetic testing in cancer(PCG)	11.4%	74%	14.6%
Direct to Consumer Genetic Testing(DTC-GC)	-	22.8%	77.2%

Table 01: Knowledge on applications of different genetic/ genomic diagnostic approaches

Table 2 illustrates the respondents' opinion on availability of genetic diagnostic services. in Sri Lanka.

Opinion on limitations of using genetic diagnostic service	Agree	Neutral	Disagree
Cost of genetic tests are beyond the reach of public	88.6%	9.8%	1.6%
Latest genetic tests are not readily available	74%	20.3%	5.7%
Genetic diagnostic facilities should be widely made available island wide	82.1%	8.1%	9.8%

Table 2: Opinion on availability of genetic diagnostic services in the country.

Table 3 illustrates the respondents' opinion on adequacy of genetic education.

Opinion on genetic education	Agree	neutral	Disagree
I satisfy with the adequacy of knowledge on genetic diagnostics during undergraduate period	11.4%	36.6%	52%
Need a continuous medical education program to fill this gap in knowledge	86.2%	12.2%	1.6%

Table 3: Opinion on adequacy of genetic education

Undergraduate education was the only source of knowledge in medical genetics in 53.7% while 34.2% had gained knowledge from multiple additional sources such including PG training, CME, peer learning, etc.

Discussion

Majority of participants had either adequate (24%) or some (65.9%) knowledge on applications of basic genetic diagnostic methods such as karyotyping and PCR. Level of knowledge on the applications modern diagnostic techniques like Fluorescent in-Situ Hybridization (61.8%), SNP arrays (78%), Next generation sequencing (78%), and array-CGH (90.2%) was generally poor. Most had either some (74%) or adequate (11.4%) knowledge on the use of predictive genetic tests in cancer. Thus except for conventional genetic diagnostic tests and predictive cancer genetic tests, overall knowledge on genetic diagnostics remains insufficient.

Majority (77.2%) had poor knowledge on direct-to-consumer genetic testing. This is a new frontier where advanced genetic diagnostics come into clinical practice. Interpretation of DTC genetic test results has been identified as a challenge for future physicians(2). Many studies have found that the knowledge on concepts such as personalized medicine, pharmacogenomics remain low (22,24).

Majority accepted high cost beyond the reach of general public (88.6%) and unavailability in most areas of the country (74.6%) as barriers for access to the benefits of modern diagnostic techniques. Majority (82.1%) agreed the need for expanding of genetic diagnostic facilities. A study done in Canada describes difficulty in accessing resources, cost, distance and poor patient engagement as key issues in the practice of clinical genetics particularly in rural areas(25). This need of expanding genetic services in the country have been highlighted before(23).

Inadequacy of genetic knowledge among physicians has been identified as an area of concern around the world (1,2,4,5). In this sample undergraduate education is the only source of knowledge regarding genetic diagnostics for a majority (53.7%). Furthermore only 11.4% agreed upon the adequacy of undergraduate genetic education to apply in the practice while 86.2% agreed the need of a continuous medical education program to update their knowledge. Slow adaptation of genetic technology to clinical care has been identified as a challenge to achieve genetic literacy among physicians(3). The need for a CME program particularly an online strategy to improve the genetic literacy has been highlighted in previous studies (16,17,22,26).

Conclusions

Sri Lankan doctors' knowledge on applications of modern genetic and genomic diagnostic techniques is inadequate. Both undergraduate and post graduate curricula should be revised and continuous medical education is the way to keep them up to date regarding ever changing frontiers of genetic and genomic diagnostics.

Limitations

The methodology used to contact participants did not provide expected results. Use of conventional paper based questionnaire would have yielded a larger sample size. Furthermore using a stratified cluster sampling method with randomization and adequate sample size for each strata would have given results that can be inferred to the general study population. It would have also helped to identify subgroups with inadequate knowledge.

Abbreviations

CGH- Comparative Genomic Hybridization, CME- Continuous Medical Education, CNV- Copy Number Variation, DTC-GT – Direct To Consumer Genetic Testing, FISH- Fluorescent In Situ Hybridization, , NGS-

Declarations

Ethics approval and consent to participate: Ethical approval was obtained from Ethics Review Committee, Faculty of Medicine and Allied Sciences, Rajarata University of Sri Lanka (ERC/2016/023).

Consent to publish: Not applicable

Availability of data and materials: Raw data set is attached as a supplementary spread sheet document.

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