

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).

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(4-6). However level of physicians knowledge remains a barrier for delivering benefits of genomic medicine(7).

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 practice(8). Single nucleotide polymorphism (SNP) microarray is another versatile diagnostic technique which has many benefits other than diagnosis (9).

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 (10). 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 (11,12).

Direct- to- consumer genetic testing (DTC-GT) is also becoming popular due to increased consumer access via internet and greater autonomy and privacy(13). 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(14).

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 (15,16). 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 (17).

In Sri Lanka availability of clinical genetic services dates back to 1981 and genomic medicine was implemented in 2010(18). However there is a paucity of scientific evidence about physicians' knowledge regarding different genetic and genomic diagnostic methods and their applications.

Main Text

This cross sectional descriptive study was carried out among Sri Lankan doctors using an on-line questionnaire. Convenient sampling technique was used and email addresses were obtained from official websites and upon requests from individuals and institutions. Questionnaire was created using Google forms and settings were adjusted to allow a single response per participant. Email containing a link to the questionnaire and consent was sent to individuals. Informed consent was taken in the form of a dialog box. Anonymous responses were collected to an online database.

The questionnaire 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. Level of knowledge was categorized as adequate, some and poor. Third component was used to identify the attitudes and opinions regarding the limitations of their knowledge and clinical genetics in practice. In this section participants either agreed, disagreed or remained neutral to different statements. The questionnaire was pre tested among 5 medical graduates and relevant changes were introduced depending on feedbacks.

Results

: A total of 123 (12.64%) individuals out of 973 contacted doctors completed the questionnaire. 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 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%). Intern house officers, consultants and university academics accounted for 14.9%,10.7% and 8.3% respectively. 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.

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 received an adequate knowledge on genetic diagnostics as an undergraduate	as	11.4%	36.6%	52%
Need a continuous medical education program to fill this gap in knowledge	to fill this	86.2%	12.2%	1.6%

Table 3: Opinion on adequacy of genetic education

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 comes in to 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 (17,19).

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(20). The need of expanding genetic services in the country has been emphasized before(18).

Inadequacy of genetic knowledge among physicians is 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 (11,12,17,21).

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 and a more generalizable conclusions. Furthermore broadening of the scope to include ethical, legal and social implications associated with genomic medicine would have strengthen the study.

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- Next Generation Sequencing, PCR- Polymerase Chain Reaction, SNP- Single Nucleotide Polymorphism

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.

Competing interests: Author declares that he has no competing interests.

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