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Physician Knowledge, Attitude and Practice of Medical Genomics/Genetically Related Diseases in Bayelsa State, Nigeria

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A B S T R A C T

There is a paucity of data on the knowledge of genomic medicine and heritable diseases, attitudes and practice of genomic medicine among physicians in the Niger Delta region of Bayelsa State. This study was thus aimed at evaluating the knowledge of, attitude and practice of genomic medicine by physicians in a resource poor country in Africa as depicted by Bayelsa state, Nigeria. A structured questionnaire was distributed to a random sample of 200 physicians in public and private hospitals in Bayelsa State, Nigeria. The data was analyzed using SPSS. The results showed that 54.8% of the physicians had a fair knowledge of genetic medicine, 71.15% of respondents had a poor attitude and this was also reflected in their poor practice where most respondents, (83.33%) claimed that their practice was hampered by lack of diagnostic tools and only 33.04% had knowledge of basic genotyping techniques. In conclusion, the data suggests a fair knowledge of and a poor attitude towards genetic medicine amongst practicing physicians in Bayelsa state, Nigeria. This suggests that it is imperative to establish a perceived need to learn more about genetic medicine if the promise of genomics has to be established in Africa.

Keywords: Physician, Genomic Medicine/heritable disease, Knowledge, Attitude, Practice, Bayelsa state, Nigeria

Introduction

Genetically related diseases are caused by an abnormality in an individual's DNA. Abnormalities can range from a small mutation in a single gene to an entire chromosome or set of chromosomes been added or removed. Such diseases can be inherited from parents or other relatives. However, there are subtypes of genetic disorders called acquired ones, i.e. they originate due to mutations in a pre-existing gene or set of genes. Causes are usually complex and the environment has been implicated as an important possible cause. (NHGRI, 2022)

Medical genetics and genomics is concerned with the hereditary contribution to health and disease (The World Bank; 1999) and involves the application of genetic knowledge in clinical practice focusing on family history, detection of genetic risk factors, diagnosis, counselling about and providing treatment options if available. (The World Bank, 1999) The application of medical genomics is thought to have seen marked acceleration and the recent strides in development of its application will further widen the existing gap between the

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developed and the developing world (Ala' *et al.,* 2019) and it has become imperative that developing countries include genetic methods into their health care delivery systems. Though there are arguments on the affordability of the basic DNA diagnostic tools by the middle and low-income countries this is further worsened by the commercialization of genomics and DNA patenting. These, scientists believe will further within the gap between the developed and the developing countries. (Ala' *et al.,* 2019, The World Bank, 1999). The WHO has recognized some of these concerns and has made recommendations that includes training programs plus professional and public educations (Ala' *et al.,* 2019). Thus, the general consensus is that education should form part of a broader implementation strategy if the promise of genomic medicine in Africa will be realized.

The preparedness of medical specialties without specific genetic qualifications to play a vital role in this workforce has long been questioned. Adult learning theory indicates that for education to be effective, a perceived need to learn must first be established (Crellin*et al.,* 2019). Medical specialists in Africa have to perceive genomic medicine as relevant to their clinical practice (Mahmoud *et al.,* 2019)

Previous studies reveal that medical specialties views on clinical utility of genomic medicine are mixed. However, with the increasing demand for genomic investigations, medical specialists are beginning to practice genomic medicine (Mahmoud *et al.,* 2019).

METHODOLOGY

Study Area

The study was done amongst physicians attending an association meeting for doctors practicing in all private and government hospitals in Bayelsa State, Nigeria.

Subjects

The subjects in this study were all physicians (both male and female) who were attending an association meeting for doctors practicing in all private hospitals and government hospitals in all the local government areas in Bayelsa State. Participants had earned their MBBS or fellowship degree from Nigeria and were either specialist, interns and residents. 200 copies of a structured questionnaire consisting of 36 statements was distributed randomly to the physicians at the start of the meeting and the completed questionnaires were collected at the end of the meeting. The response rate was 52% (104/200).

The response of each participant was kept anonymous.

Survey format

The questionnaire was made up of four categories of questions; sociodemographic characteristics, knowledge of genetic medicine, attitude towards and practice of genetic medicine.

Knowledge of genetic medicine; participants were requested to define their understanding of what genetic medicine is, can genetic disorders arise from oil exploratory activities, are they treatable, are they preventable, how many diseases could they spontaneously write down.

Attitude to genetic medicine; The questionnaire requested the respondent to note what they will do if they make a tentative clinical diagnosis, will they encourage relatives of such patients to be screened and what heritable diseases in our environment should be on routine tests for relatives already diagnosed.

In their practice of medicine; Respondents were required to state how many genetically related diseases they have been able to identify in their practice, how were the diagnosis made and what were their treatment modalities.

Statistical Analysis

The data was analyzed using SPSS (Statistical package for social sciences). Non-parametric test was used to compare the means and P-value was considered significant at 95%confidence limit.

RESULTS

104 out of the 200 questionnaires were completed. Respondents were mostly male (83%). The majority were under the age of 35 years (Table 1) This was expected because this is the most active age of practice in the medical career.

60% were general practitioners and other subspecialties most represented were; surgeons (10.58%) gynaecologist/obstetricians (7.69%), paediatricians (5.77%)

Knowledge of genetic medicine; 63% could define genetic medicine in the context of practice of medicine. 15% thought genetic medicine involves research into the cause and inheritance of genetic diseases. 40% thought it is a specialty involving the diagnosis and management of heritable disorders, 20.19% thought it is a specialty of medicine and 16.35% thought it is a specialty of science in genetic research.

To the question of what the respondent thought genetic medicine will directly involve; 33.22% thought it involves research into the cause and inheritance of genetic diseases, 22.03% thought it directly involves genetic counsellors, 17.83% thought it directly involves clinical diagnostic laboratories, 16.78% thought it directly involves clinical practice of physicians and 10.14% thought it involves nutritionists.

Conditions that fall within the scope of genetic medicine; 16.17% of respondents acknowledged birth defects fall within the scope, 15.43% acknowledged cancers, 14.50% mitochondrial disorders, 13.75% teratogenesis, 12.64% mental retardation and 12.64% dysmorphology all fall within the scope of genetic medicine

To the question, are genetic diseases treatable; 58.65% confirmed it was treatable, 6.73% said it was not treatable and 18.27% had no knowledge.

Are genetic/heritable diseases preventable; 95.19% thought it was preventable, 1.92% thought it wasn't and 2.88% had no knowledge.

To the question of the genetically related/inheritable/heritable diseases known; Amongst all the genetically related diseases spontaneously mentioned by the respondent six had the highest frequency. First was sickle cell disease (SCD) with a frequency of 18.60%, Duchenne muscular dystrophy (10.99%), Haemophilia (7.69%,), Klinefelter's syndrome (4.40%), G6PD deficiency (4.18%) Cancer (3.08%,) diabetes mellitus (2.85%).

When respondents were asked if they do know any family with genetically related/heritable diseases running in the family**;** 54.81% knew families with genetically related disease running in the family while 45.19% knew non.

To the question of what type of heritable/genetically related diseases did they observe; 57.35% of the respondents reported sickle cell anaemia, 13.24% Downs Syndrome, 2.94% G6PD deficiency, 2.94% Haemophilia.

71.15% of the respondents has a poor attitude and 28.85% had a good attitude towards genetic medicine. 58.18% of the respondents accepted that referral of a patient to a genetic counselling unit was the most appropriate decision to take after making a tentative clinical diagnosis of a genetic disease. 23.48% thought referring to a physician was appropriate, 5.30% thought counselling and sending patients home was appropriate, while 3.03% thought referring to a surgeon was appropriate.

93.2% strongly agreed that they would encourage screening individuals whose family relatives have a genetic disease, while 6.73% were indecisive

Four routine tests respondents thought should be part of a routine test list in every hospital and would be beneficial for relatives of patients diagnosed with genetic disease are; Sickle cell anaemia 61.59%, Downs syndrome 6.52%, Haemophilia 5.80%, Leukaemia 4.35%.

.**Practice of genetic medicine**; 71.15% of the respondents had made a diagnosis of heritable disease in their clinical practice career while 28.85 hadn't made a diagnosis. Of those who had made a diagnosis in their clinical practice, 79.73% had made a diagnosis of a genetic disease in less than 10 patients while 20.27% had made a diagnosis in more than 10 patients. Amongst the diagnosed patients, there were three most frequently diagnosed genetically related diseases. i.e. sickle cell anaemia (43.85%), Downs syndrome (20.00%,) G6PD deficiency (4.20%.).

The reason for the lack of cases was attributed by the respondents to the lack of diagnostic tools (83.33%), rare occurrences of genetic diseases (10.00%). Inability to clinically identify a heritable disease.

To the question of what basic genetic tests would you want to do; 33.15% of the respondents preferred karyotyping, 32.04%, genotyping, 32.04% preferred prenatal genetic diagnosis for common diseases like sickle cell diseases, 2.76% had no idea of what test is to be done. This is in agreement with previous works by Wonkam*et al* which also showed a poor awareness of DNA diagnosis amongst physicians and medical students in sub-Saharan Africa (Cameroon, a case study).

Table 1. Socio-demographic Data

Characteristics	Frequency n=104	Percentage (%)
Age		
Under 25 yrs.	4	3.85
26-35 yrs.	59	56.73
36-45 yrs.	15	14.42
46-55 yrs.	13	12.50
Over 55 years	13	12.50
Sex		
Male	83	79.81
Female	21	20.19
Religion		
Christianity	101	97.12
Islam	1	0.96
None	2	1.92
Educational Level		
Tertiary	91	87.50
Masters (post grad)	13	12.50
Area of Specialty		
General practice	60	57.69
Surgery	11	10.58
O & G	8	7.69
Paediatrics	6	5.77
Community medicine	6	5.77
Internal medicine	5	4.81
Family medicine	3	2.88
Mental health	2	1.92
Haematology	2	1.92
Ophthalmology	1	0.96
Designation		
Resident doctor	27	25.96
House officer	25	24.04
Medical officer	22	21.15
Consultant	14	13.46
GP	6	5.77
Registrar	4	3.85
Lecturer	3	2.88
PMO	3	2.88

Table 2. Knowledge score of Genetic Medicine

Characteristics	Frequency n=104	Percentage (%)
Knowledge scoring of 32 response		
<10 (Poor Knowledge)	9	8.65
10-20 (Fair Knowledge)	57	54.81
21-32 (Good Knowledge)	38	36.54

Table 3. Attitude scoring towards Genetic Medicine

Characteristics	Frequency n=104	Percentage (%)
Attitude scoring of 4 responses		
≤ 1 (Poor Attitude)	74	71.15
≥2 (Good Attitude)	30	28.85

Table 4. Practice of Genetic Medicine

Characteristics	Frequency n=104	Percentage (%)
If no, what do you think is the reason for the lack of cases (n=30)		
Lack of diagnostic tools	25	83.33
Genetic abnormalities are very rare	3	10.00
Inability to clinically identify a heritable disease	2	6.67
What basic genetic test would you want to do (n=181) (*Multiple Response*)		
Karyotyping	60	33.15
Genotyping	58	32.04
Prenatal genetic diagnosis for common diseases like SSA	58	32.04
I don't know	5	2.76

DISCUSSION

Bayelsa State is an oil-rich State located in the core Niger Delta region, in the Southern part of Nigeria. It has a land mass of 10,7731km^{2}and has a population of 1.705million (2006). It has a poverty rate of 93.2% (2010) and half of these are living below the international poverty line of <2 dollars per day and so can be described as a low-income society. The health care system comprises of the public and private sectors which are highly overburdened because of lack of manpower and adequate resources. The region, like most areas in the developing world is coming to terms with the gradual increasing load of non-communicable diseases with increasing genetic involvement an observation regional researcher has attributed to the inappropriate handling of oil exploratory waste materials. The importance of the knowledge of genetic disorders should therefore not be neglected and is imperative that practitioners pay considerable attention to prevent sudden development of diseases in families.

The predominantly male respondents totally agree with the observed male/female ratio amongst physicians at the level of the state and Christianity is the predominant religion practiced in the entire region.

The response rate is fairly poor though this could be expected taking into consideration the biased mindset of the average medical practitioner towards the field of genetics considering it to be a difficult and rare terrain for the average Nigerian-trained physician. The response is also comparable to that observed by researchers in Cameroon (Wonkam*et al.)* in their study of Knowledge and attitudes concerning medical genetics amongst physicians and medical students in Cameroon (sub-Saharan Africa). General practitioners were

most represented as against specialist doctors (60%:10.58%). The predominant respondent rate observed is a true reflection of the number of medical doctors who are actually interested in specialist training in the Niger Delta region of Bayelsa State. And this could also be an index that could be used to ascertain their would be interest in genomic medicine which is a relatively yet unexplored are of medicine considering that the first medical genetic services in Nigeria was established in the early 1970s in Ibadan (Adewale *et al.*, 2018). This could explain why 63% of physicians could appropriately define what genetic medicine is; however, the progress made in Nigeria has been very poor and currently the number of medical geneticists and counsellors are still very few as observed by Adewale et al and most patients with genetic disorders are seen by paediatricians while others are seen by specialists in internal medicine. In addition, medical diagnostic facilities are still very inadequate and generally unavailable in Nigeria. This thus means that training opportunities for medical genetics are not available within the country and interested trainees would have to travel out of the country. The study also showed (by the response of the respondents) that the genetic disorders with the highest frequencies in the region were sickle cell disease, Duchenne muscular dystrophy, haemophilia, Klinefelter's syndrome, and G6PD deficiency with sickle cell disease having the highest frequency, a fact that was further supported by the good number of respondents who had made a tentative diagnosis of SCD in the course of their clinical practice. This is in agreement with other data by Ade et al 2018 which places SCD as probably the single most common severe genetic disorder in Nigeria, occurring in up to 2-3% of new-borns with a steady prevalence rate of 22-25% for the sickle cell trait. From this study, most respondents (71.15%) had made a tentative clinical diagnosis of a heritable disease at one point or the other in their career, however, the number of cases identified per physician was less than 10. This may imply that the incidence of genetic/heritable diseases is low within Bayelsa State or physicians are not knowledgeable enough to make a tentative clinical diagnosis and thus many cases are undetected. The physicians also showed a poor awareness of basic DNA diagnostic tests which can greatly reduce misdiagnosis. Most respondents agreed screening family members of patients with genetic disease should be encouraged and in addition suggestions of four tests that should form part of a list of routine tests for relatives of patients diagnosed with genetic diseases was tests for SCD, Down syndrome, haemophilia and leukaemia. This could be a channel for preventive medicine which could be one promising area of focus for achieving prospects for genomic medicine in low-income(developing) countries such as Nigeria (Bayelsa State a case study)

CONCLUSION

The general knowledge of genetic medicine was considered to be fair (52.00%) while the attitude towards genetic medicine was poor (71.15%), and in their practice most physician expressed the opinion that they had very little tools to work with and therefore even when the physicians have the basic diagnostic tool to use to aid in making a diagnosis, such as karyotyping, this itself is not readily available. It is imperative that the average physician, though without specific genetic qualifications is sensitized about the importance of developing a workforce in Africa for genomic medicine and education should be part of a broader implementation strategy which could be achieved through organization of CMEs with genetics as a core content. It is only by establishing the perceive need to learn that the promise of genomic medicine will be realized in Bayelsa State and Nigeria.

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