January-March 2019 Volume 6 Number 1
ISSN 2347-7946
eISSN 2347-7962

MGM Journal of MEDICAL SCIENCES

MGMJMS

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The Official Publication of
Mahatma Gandhi Mission Institute of Health Sciences
(Deemed to be University u/s 3 of UGC Act 1956)
Kamothe, Navi Mumbai, Maharashtra, India

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MGMJMS

MGM Journal of Medical Sciences

Volume 6 Issue 1 January-March 2019

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From the Editor's Desk

10.5005/mgm-6-1-iii

Currently, the proportion of the world's population above 60 years is estimated to be 12% and is likely to rise to 22% by 2050 according to the World Health Organization (WHO). With increasing age, physical and mental capacities are bound to decline, leading to various morbidities and death, but fortunately, these declines are neither linear nor consistent. An 80-year-old person may be in very good health, fully independent and enjoying life fully, whereas another 70-year-old person may be frail and dependent on the help from others. Besides elderly people are prone to, what have been named as geriatric syndromes. These are caused by multiple underlying factors like frailty, urinary incontinence, falls, delirium, pressure sores, etc, which further add to their health problems. Healthy aging is definitely achievable by living in a healthy environment, maintaining healthy behaviors throughout life (like taking balanced diet, abstaining from tobacco use, regular exercise, strength training to maintain good muscle mass), meditation, keeping good relations with family members and friends, etc. Communities must also provide supportive environments to their seniors like safe and accessible public buildings and transport, which will enable them to move about freely and safely.

The World Health Organization has formulated a global strategy and action plan on aging and health, in which 5 priority areas have been identified for action. These are commitment to healthy aging, aligning health systems with needs of older population, creating age-friendly environments and improving measurement, monitoring and understanding.

Current issue of MGM Journal of Medical Sciences (MGMJMS) carries an informative article about gaps and other inadequacies in awareness of primary care physicians about anemia in the elderly. As is well known, anemia significantly worsens preexisting morbidities in the old, like cardiovascular and neurological disorders, depression and cognitive decline. It also increases their susceptibility to falls and fractures.

We are pleased to present issue no 1, volume 6 of MGMJMS to our esteemed readers. Thank you for contributing your articles and papers for this journal and your whole-hearted support.

Shibban K Kaul MS, MCh, FIACS
Editor-in-Chief
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MGMJMS
MGM Journal of Medical Sciences
Volume 6 Issue 1 January-March 2019

Contents

ORIGINAL ARTICLE

Anemia in the Elderly: Knowledge, Attitudes, and Perception of Primary Care Physicians

Amrita Desai[1], Thomas Guerrero[2]

Abstract

Background: Anemia in the elderly can impair the quality of life as well as cognitive and physical functions and is even associated with an increased risk of death. Thus, in the elderly, anemia should not be considered as a consequence of aging and must be explored and appropriately treated by their primary care providers. There are no studies to assess the awareness of primary care physicians regarding anemia in the elderly. We designed a survey questionnaire to assess the knowledge, attitudes, and perception about anemia in elderly among primary care providers with the aim to design medical curriculum and academic teaching to maximize the current understanding of anemia in the elderly.

Materials and methods: A survey instrument, with 10 questions assessing the primary care physician's knowledge, attitudes, and perception, was prepared by two internal medicine (IM) residents and reviewed by a hematologist along with a primary care provider for appropriateness and reliability. The survey was then administered to the residents and primary care providers in the Departments of IM and Family Medicine (FM) at Memorial Hospital of Rhode Island.

Results: We obtained 69 surveys, out of which 55% of the responders were female and the median age was less than 30 years. There was equal representation of IM and FM doctors. On the knowledge component, 48% of the physician agreed that the hemoglobin threshold for the diagnosis of anemia was the same in young and elders, 52% agreed for routine anemia screening while 67% felt that it affects only morbidity but not mortality. Only 55% agreed that nursing home residents are at a higher risk of anemia than the elderly in the community and 23% did not think that anemia affects the physical performance in the elderly. There was no statistically significant difference based on the type of medical practice or the years of clinical experience for attitude and perception questions in the survey.

Conclusion: There are significant gaps in the knowledge and skills of primary care health providers which need to be addressed in the form of training sessions or periodic continuing medical education (CME) to ensure better management and care of the increasing elderly population.

Keywords: Anemia, Attitudes, Elderly, Hemoglobin, Knowledge, Survey.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1220

Introduction

Life expectancy in humans has gone up worldwide, leading to an increase in the elderly population. Older people (age >65 years) have different healthcare problems and needs. One of the common and often understudied medical problems in the elderly population is anemia. Systemic literature reviews estimated the prevalence of anemia up to 2.9–51% in elderly men and 3.3–41% in elderly women.[1] According to the National Health and Nutrition Examination Survey (NHANES) study in the USA, anemia of elderly (AIE) is of epidemic proportions with approximately 3 million seniors being anemic.[2] With the growing percentage of the aging population, these numbers could go up further. Given how prevalent this is in the community, the American Society of Hematology and National Institute of Aging had an expert panel to address this public health issue and came up with some key questions to further help understand and address this issue.[2]

Blood hemoglobin levels tend to decline with aging[3] gradually; however, currently, there is no separate definition and cutoff value of hemoglobin for anemia in elderly population, and we still use the conventional WHO definition for anemia, i.e., <13 g Hb/dL for men and <12 g Hb/dL for women.[4] In clinical practice, it is easy for us doctors to assume that slightly lower levels of hemoglobin in older people have no clinical significance[3] and consider it a normal variance. This could prove detrimental to further workup and treatment as numerous studies have shown that anemia in the elderly has a significant impact on morbidity and mortality.[3,5] Low hemoglobin levels have shown to increase the risk of falls, decrease physical mobility, increase weakness, worsen cognitive functions, increase depression, and reduce the quality of life.[3,5–7] Thus, treating anemia has been shown to be beneficial as this population, which already has many other comorbid conditions, showed improved quality of life.

It is imperative for us as physicians to understand the significance of anemia in the elderly if we aim for a healthier aging population. So, then the next question is why we as physicians are unable to identify these populations early in the disease course and make appropriate interventions? Do we as primary care physicians understand this disease entity, know how to screen, how to work up these patients, and how to treat them adequately and when to refer to a specialist? No studies are done to understand the perception

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How to cite this article: Desai A, Guerrero T. Anemia in the Elderly: Knowledge, Attitudes, and Perception of Primary Care Physicians. MGM J Med Sci 2019;6(1):1–5.

Source of support: Nil

Conflict of interest: None

and knowledge of primary care physicians about anemia in the elderly. It is essential for us to understand the knowledge gaps so that we can take necessary steps to increase awareness among primary healthcare professionals.

Hence, we decided to undertake this study to evaluate the knowledge and perception of primary care physicians about anemia in the elderly at our institute.

Materials and Methods

The study was conducted at Memorial Hospital of Rhode Island after institutional review board committee approval. The primary aim was to develop a validated survey tool for anemia in the elderly, based on our hypothesis that there is not enough awareness among primary care providers about anemia in the elderly. Our secondary aim was to identify the knowledge gaps, so that necessary teaching interventions can be undertaken in the future.

Two internal medicine (IM) residents prepared a survey instrument with 10 questions (Table 1A) assessing the primary care physician's knowledge, attitude, and perception about anemia in the elderly and reviewed by a hematologist and a primary care provider for appropriateness and reliability. The survey consists of four demographic questions and 10 questions assessing the knowledge, attitude, and perception of the physicians. Questions 1, 4, 7, 8, and 9 assessed the knowledge while 2, 3, 5, 6, and 10 assessed attitude and perception (Table 1B). A 7-point Likert scale was used for measurement of the items. Resident doctors and attendees working in the family medicine (FM) and IM departments at Memorial Hospital of Rhode Island were approached for the study. The providers were approached in their clinics. A two-page questionnaire with 10 questions about knowledge, attitude, and perception was given to each participant. The study was conducted over a period of 6 months. For analysis, scores of 1–3 were categorized as "agree," 5–7 as "disagree," and 4 as "neutral" for each question. Percentages were calculated and variability was assessed by Fisher's test.

Results

A total of 90 surveys were distributed among doctors in the IM and FM departments. We were able to obtain 69 completed surveys, with a survey response rate of 76.6%. Out of the 69 survey responders, 38 (55%) were females, 57 (82%) were between 20 and 39 years of age, and 35 (50%) were from the IM department. Fourteen (20%) were teaching faculty, 22 (32%) were postgraduate year-1 (PGY-1), 19 (28%) were PGY-2, and 14 (20%) were PGY-3 (Table 2).

Table 1A: Survey questions

1	The hemoglobin threshold for the diagnosis of anemia is the same for the elders and the young.
2	A hematology referral is useful for most cases.
3	I feel confident evaluating anemia in the elderly.
4	All patients over 70 years of age should be routinely screened for anemia.
5	I am confident in treating anemia in my elderly patients.
6	I feel confident teaching my patients about the risks of anemia.
7	Anemia in the elderly is more associated with decreased physical performance than anemia in young patients.
8	Elders in the nursing homes are at a higher risk of anemia than elders in the community.
9	Anemia in the elderly affects morbidity but not mortality.
10	I know when to refer my patients to a hematologist.

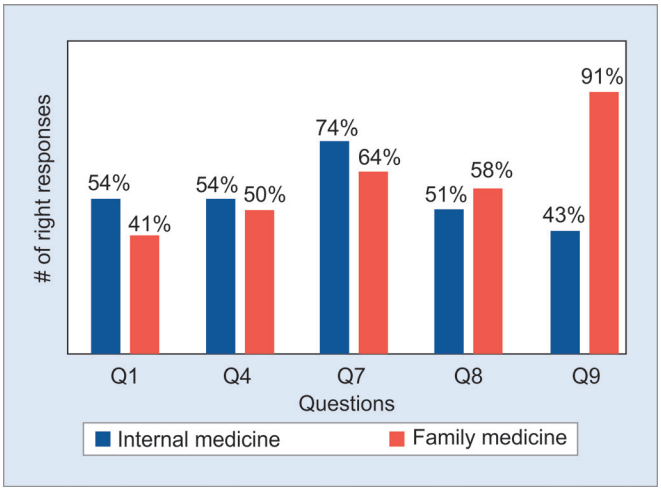

Fig. 1: Knowledge scores according to departments

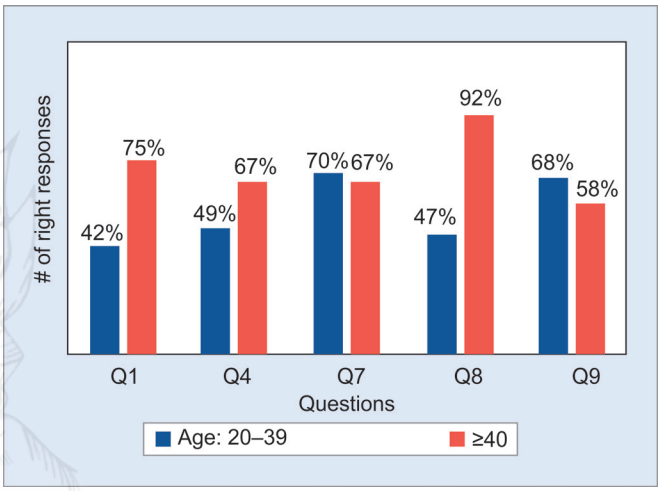

Fig. 2: Knowledge scores according to age groups

After evaluation of the survey responses for knowledge about anemia of elderly (AIE), we noted 33 (48%) as agreed that the hemoglobin threshold for the diagnosis of anemia is the same for the elders and the young, while 36 (52%) agreed to routine screening of adults >70 years of age for anemia. Forty-eight (70%) responders were associated with a more significant decrease in physical performance with AIE, 38 (55%) concurred that elders in the nursing homes are at a higher risk of anemia than elders in the community, and 46 (67%) felt that AIE affects not only morbidity but also mortality. Overall, FM providers scored more on the knowledge questions than IM providers with knowledge scores of 60.8 and 55.2, respectively (Fig. 1). Younger clinicians scored higher on the knowledge questionnaire than clinicians ≥40 years (Fig. 2).

After evaluation of the survey responses for attitudes and perception about AIE, we noted that 52 (75%) knew when to refer to hematology, but only 13 (18%) thought that all AIE cases need hematology referral. Fifty-eight (84%) providers were confident in diagnosing anemia and 56 (81%) knew to treat it effectively. Fifty-five (80%) were confident in educating patients about the risks of anemia. Overall, IM providers were more confident in the

Table 1B: Survey questionnaire

General demographic information:								
1. Profession: IM: [ ] FM: [ ]								
2. Type: PGY-1: [ ] PGY-2 [ ] PGY-3 [ ] Attendee: [ ]								
3. Sex: M: [ ] F: [ ]								
4. Age in years: 20–29: [ ] 30–39: [ ] 40–49: [ ] 50–59: [ ] >60: [ ]								
Questions:								
1. The hemoglobin threshold for the diagnosis of anemia is the same for the elders and the young								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
2. A hematology referral is useful for most cases								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
3. I feel confident evaluating anemia in the elderly								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
4. All patients over 70 years of age should be routinely screened for anemia								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
5. I am confident in treating anemia in my elderly patients								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
6. I feel confident teaching my patients about the risks of anemia								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
7. Anemia in the elderly is more associated with decreased physical performance than anemia in young patients								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
8. Elders in the nursing homes are at a higher risk of anemia than elders in the community								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
9. Anemia in the elderly affects morbidity but not mortality								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly
10. I know when to refer my patients to a hematologist								
Disagree Strongly	1	2	3	4	5	6	7	Agree Strongly

diagnosis and management of AIE than their FM counterparts (Fig. 3), but this was not statistically significant. Clinicians ≥40 years of age were surer and more confident in the diagnosis and treatment of AIE than younger clinicians, but this also was not statistically significant (Fig. 4). There was a statistically significant ($p < 0.5$) difference in the knowledge between first-year postgraduate doctors vs second- and third-year postgraduate students, but no difference in their attitudes and perception about AIE.

Discussion

Anemia in the elderly is a significant healthcare problem with a prevalence in the community of 9.2–23.9% in elderly men and 8.1–24.7% in elderly women based on studies by Zakai et al.,[8] Guralnik et al.,[1] and Salive et al.[9] Studies have also shown that the prevalence of AIE rises with increasing age and the prevalence in the age group of 75–85 years is around 4.9–15.0% for men and 7.1–12.7% for women while for ages above 85 years is 29.6–30.7% for men and 16.5–17.7% for women, respectively.[9,10] Racial differences have been observed with higher prevalence in Hispanics and African-American populations compared to Caucasian whites.[1,11]

The most common causes of anemia in this population are chronic kidney disease, chronic inflammation, and nutritional deficiencies.[12] Retrospective studies like that of Artz et al.[12] have shown the prevalence to be higher in nursing home residents (48%). In our study, we noted that only 55% of the clinicians acknowledged that the occurrence of AIE was lower in the community vs nursing homes.

The hemoglobin threshold for anemia is the same for elderly and young adults as per the WHO definition. Majority of the patients have mild anemia, and only 3% have hemoglobin level <11 g/dL.[13] As per the NHANES study, the top three causes of anemia in elderly are nutritional deficiency/blood loss (34%), unexplained anemia

Table 2: Demographics

Baseline characteristics	*Total N (%)*
Survey response rate *N* (%)	69 (76.6)
Age (in years) *N* (%)	
20–39	57 (82)
≥40	12 (18)
Gender *N* (%)	
Female	38 (55)
Male	31 (45)
Department	
Internal medicine *N* (%)	35 (50)
Family medicine *N* (%)	34 (49)
Level of training	
PGY-1*	22 (32)
PGY-2	19 (28)
PGY-3	14 (20)
Faculty	14 (20)

*PGY: postgraduate year

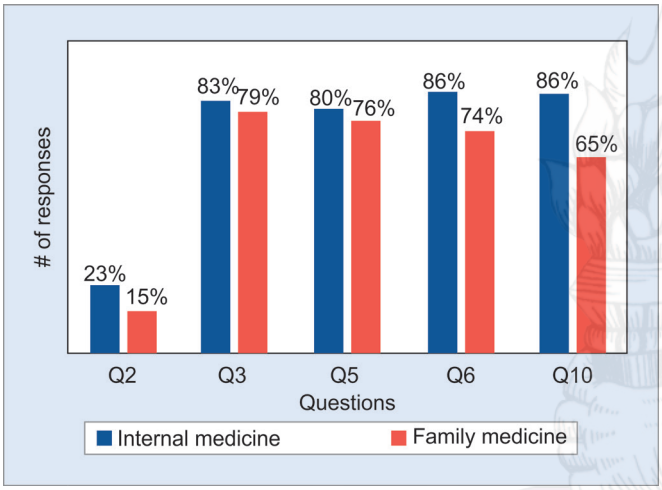

Fig. 3: Attitude and perception scores according to departments

(34%), and anemia due to chronic illness/inflammation or chronic renal failure (32%).[1] We noted in our study that only 48% of the providers were aware of the hemoglobin threshold for AIE and 48% felt that routine screening was necessary while 84% clinicians were confident about screening for AIE.

Several studies have shown that AIE leads to not only the overall decline in performance status but also worsening the preexisting conditions like cardiovascular disease, neurological conditions, increased fracture risk, depression, and cognitive function.[3,5–7,14–16] Studies like that of den Elzen et al.[16] have shown reduced functional capacity, mobility, increased fragility and falls, and decreased muscle mass and strength.[5–7] Lucca et al.[3] and Denny et al.[17] both have shown a decline in cognitive abilities with anemia while an Italian study by Onder et al.[18] showed the association between anemia and worsening depression. Hayashi et al.[14] showed that correcting anemia in elderly with chronic renal disease using erythropoietin improves left ventricular function. Thus, AIE is associated with increased hospitalizations and morbidity.[13,19] Many large cohort studies have shown anemia to increase the risk of mortality after controlling for other comorbid

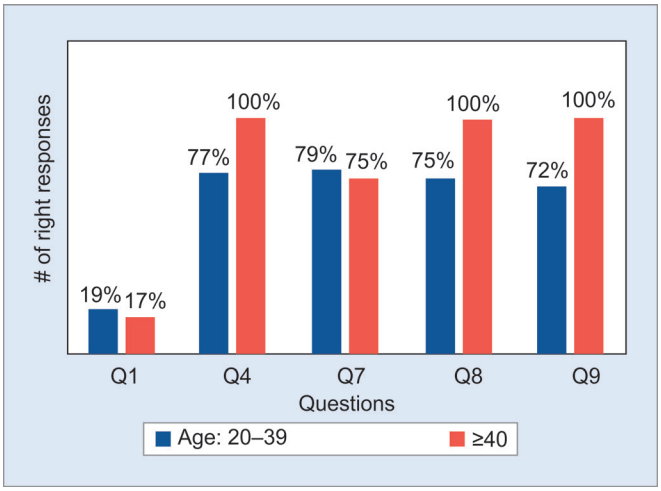

Fig. 4: Attitude and perception scores according to age groups

conditions.[20,21] AIE affects not only morbidity but also mortality, but in our survey, only 67% of doctors acknowledged that AIE affects both mortality and morbidity. Though the doctors scored low on the knowledge score, 80% of doctors were confident in treating it and educating their patients about its risks and effects on health.

AIE causes a significant increase in healthcare needs and contributes directly and indirectly to increased medical cost.[22–24] With an ever-increasing older population, the prevalence and burden of AIE will keep rising. This will have a substantial economic impact with an increase in healthcare costs and an increase in the use of limited medical resources. Thus, it is imperative for us physicians to correctly identify this population and treat them appropriately. In 2004, the American Society of Hematology and National Institute of Aging convened a panel of 20 experts to address this public health problem, and they came up with some key concepts and questions like what should be the hemoglobin threshold? Should everyone be screened, and who should diagnose and treat these patients to help further understand and address this issue?[2] They also came up with a practical approach to clinically evaluate AIE.[2] In our study, 52% clinicians agreed to routine screening, while only 13% felt that these patients need a hematology specialist, and 75% know when to refer to hematology.

In our survey, we found that the knowledge score was 50–60% for all the physicians; thus, there is a knowledge gap among providers. We further found that FM providers were more knowledgeable than their IM counterparts, but IM doctors were more confident in treating and diagnosing AIE. We also noted that though there was a statistically significant difference in the knowledge levels among junior and senior residents, they did not differ in their attitude and perception about AIE. Clinicians above >40 years were more confident in the treatment and diagnosis of AIE, and this could be because the majority of the clinicians <40 years of age were resident doctors in training. The knowledge gap could be due to the lack of educational training during medical school and residency, lack of awareness among primary care providers, and the absence of screening and treatment guidelines. The dearth of large prospective clinical studies on the epidemiology, morbidity, and mortality impacts of AIE also contributes to this knowledge gap.

The limitations of our study are that it was conducted at a single institute and did not include clinicians in private practice. We also

had many trainee doctors vs practicing doctors. However, we were able to create a survey tool to assess the awareness about AIE among primary care providers and test it in a small cohort for validation. The survey tool generated interesting results underscoring the fact that there is a lack of knowledge and awareness about AIE. The next step would be to test this questionnaire in a large cohort and also to conduct educational workshop for providers on AIE.

Conclusion

Although most primary care physicians feel confident in managing and treating anemia, this does not correlate with their low knowledge scores. Only 55% think that the threshold for making the diagnosis of anemia is the same in young and adults and only 67% are aware of its impact on mortality and morbidity. Our results reflect the need to revisit the training curriculum to fill out gaps that primary care providers have when facing elderly patients with anemia.

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ORIGINAL ARTICLE

Telemedicine: A Valuable, Epidemiological, and Therapeutic Tool for Cardiovascular Diseases in Rural Areas

Shridhar Dwivedi[1], Om P Yadava[2], Kalpeeta Roy[3]

Abstract

Introduction: Cardiovascular diseases (CVDs) are the leading cause of death in India. These diseases often go unnoticed in the rural heartland because of their geographical location and low socioeconomic status. By using telemedicine as a tool for providing health care in rural areas, we may be able to address their cardiovascular problems better.

Objectives: To study the incidence of noncommunicable diseases (NCDs) in a rural region adjacent to National Capital Territory and to observe the incidence of smoking and smokeless tobacco in rural subjects attending tele sessions.

Materials and methods: The present study was conducted at the National Heart Institute, New Delhi, in collaboration with Health Foundation of India (HFI) in Jewar, Greater Noida, where the majority of people were from low socioeconomic strata. We conducted biweekly telemedicine sessions over a period of 9 months (June 2018–February 2019) in which a total of 120 new patients were presented to us by HFI. After the first interaction, a tentative diagnosis was made and every patient was advised relevant lab investigations and lifestyle measures and a follow-up visit.

Results: We found out that 46% of the cohort was having diabetes, 25.83% had diabetes with other CVDs, 6% suffered from hypertension (HTN), 3.32% had HTN along with other comorbid conditions, 3.33% had hypothyroidism, and 2.5% had diabetes with hypothyroidism. Interestingly, only one case of 0.83% had isolated coronary artery disease (CAD). Notably, 62.5% of patients came for follow up in which 5.8% actually followed advice for quitting smoking and oral tobacco at the first instance.

Conclusion: Diabetes, hypertension and/or hypothyroidism have emerged in a big way in the rural National Capital Region. Reasonably optimum health care can be provided to the rural areas with the help of telemedicine. This tool can also be used effectively to propagate healthy life style, particularly tobacco habit and importance of exercise in rural population; thus preventing further spread of NCDs.

Keywords: Noncommunicable diseases, Rural scenario, Tele-ECG, Telemedicine.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1221

Introduction

Telemedicine is used for imparting health care and spreading health awareness from distance using telecommunication and information technology. It is also used to save lives in critical care and emergency situations. It has been used to overcome distance barriers and to improve access to medical services that would often not be consistently available in distant rural communities. As cardiovascular diseases (CVDs) have emerged as major causes of morbidity and mortality in urban as well as rural India, these need to be tackled at both places in an effective manner. However, adequate healthcare facilities are not yet available in most rural areas of the country. The National Heart Institute and Health Foundation of India undertook a joint venture using telemedicine to provide optimum health advice to patients near the Jewar village situated in Greater Noida. The aim is to provide health advice as well as study the epidemiological profile of these cases coming to the Jewar Health Centre. We present our initial observations of this innovative venture using telemedicine as a tool for providing health care to a village community.

Materials and Methods

Study Centre

National Heart Institute, Kailash Colony, New Delhi, in collaboration with Hospital Guide Foundation—NGO, eSanjeevni.in based at Jewar, Greater Noida.

Study duration: June 2018 to February 2019

Sample size: 120 patients

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How to cite this article: Dwivedi S, Yadava OP, *et al*. Telemedicine: A Valuable, Epidemiological, and Therapeutic Tool for Cardiovascular Diseases in Rural Area. MGM J Med Sci 2019;6(1):6–10.

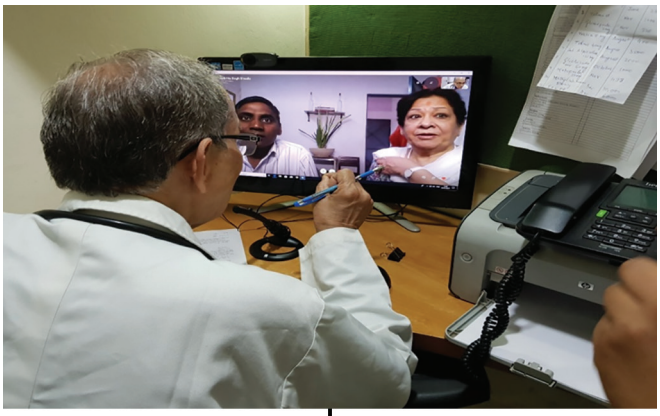
Source of support: Nil

Conflict of interest: None

Study Design

Telemedicine session was conducted at the National Heart Institute (NHI) on every Tuesday and Thursday between 12 and 1 pm. There were two separate teams: one at NHI and another at Jewar, Greater Noida. The NHI team included a doctor (SD), information technology technician, and a BSc. in Cardiac Lab Technologies (BCLT) trainee (KR). The Non-Governmental Organization (NGO) team included doctor, information technology technician, and coordinator of NGO. All subjects who presented themselves in the telemedicine session were included in this study. The basic details like name, age, sex, occupation, socioeconomic history, tobacco habit, height, weight, blood pressure, and blood sugar estimation of all cases were done

Flowchart 1: Operational mechanism of telemedicine session at NHI and Jewar. Real-time connection between the doctor at NHI and the patient at Jewar Health Centre



Store and forward data (investigations and lab reports) from Jewar health centre to NHI

Diagnosis is made, medicine prescribed and given and advised for a follow-up to evaluate the progress

by NGO and details beamed to telemedicine room at the NHI (flow chart real-time telemedicine session) (Flowchart 1).

Telecardiology basically involves the transmission of electrocardiograph (ECG) to the cardiologist *via* using the information and communication technology (ICT) platform. The ECG is essential for the screening diagnostic and monitoring for CVD[1] (Figs 1 and 2).

All relevant details like height, weight, blood pressure, blood sugar, etc., were maintained in separate computer files at Jewar and made available to the NHI team. Based on the history provided and interaction with the patient during the tele-session, a tentative diagnosis was made, investigations planned, treatment suggested, basic lifestyle measures like tobacco cessation, appropriate diet, proper exercise, and relevant investigations like total lymphocyte count (TLC), differential leukocyte count (DLC), hemoglobin (HB), erythrocyte sedimentation rate (ESR), routine urine examination, stool, liver function test (LFT), kidney function test (KFT), lipid profile, X-ray chest PA view, ECG, and ultrasound abdomen and pelvis were advised for all cardiovascular/diabetic cases. The grades of obesity are based on the criterion described by Lavie, Arena, Alpert, Milani, and Ventura.[2] The standard for determining high blood pressure in our study is based on JNC 8.[3] The diagnosis of diabetes in our study is based on the American Diabetes Association criterion 2018.[4] These tests were carried out at Jewar itself coordinated by the NGO. Reports including ECG, X-ray, and ultrasound were scanned and sent to the NHI information system well before the tele-session. The patients were reviewed along with all reports in the second visit. The final diagnosis based on history and lab reports was made during the second visit. The revised treatment schedule was started and compliance to lifestyle measures was also noted. Patients were then advised to come for follow up after 12 weeks. It may be worth mentioning that during all three visits, patients were counseled about the necessity for healthy lifestyle particularly the use of tobacco both smoking and oral. The medicines were provided by the NGO team free of cost.

Inclusion Criteria

All the patients registered with the hospital guide foundation coming to the Jewar center and presented to us *via* a telemedicine link were included in this communication.

Exclusion Criteria

The patients who did not come up for the follow-up visit were excluded in this study.

Statistical Analysis

Since the study is conducted on a very small number of populations, analysis of the data was done on the Microsoft office excel 2016.

Results

The present study includes 120 patients who presented to us *via* a telemedicine link. There were 62 (51.66%) males and 58 (48.33%) females. Ninety-eight (81.66%) were Hindus and 22 (18.33%) were Muslims. All patients belonged to low socioeconomic strata. About 28% of our study subjects were tobacco users, most being

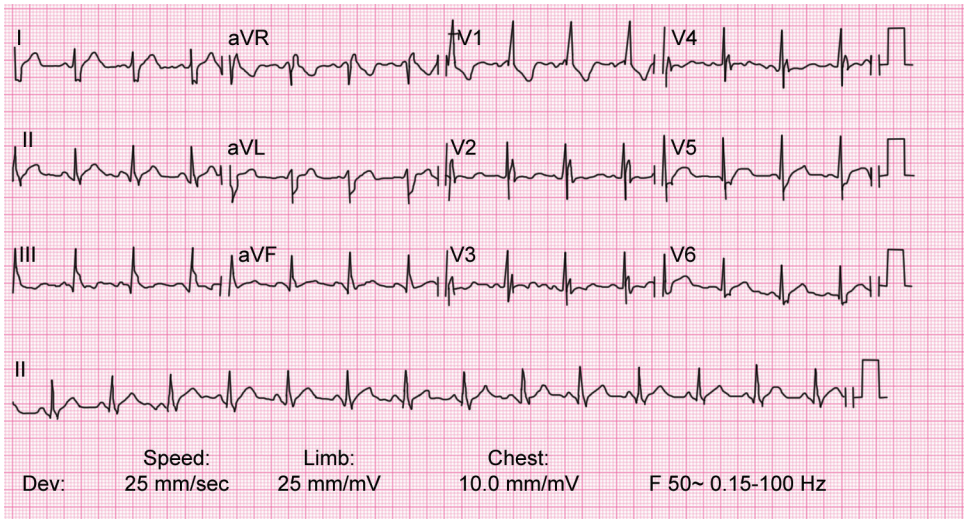


Fig. 1: Case 1: ECG report showing right bundle branch block (RBBB) sent by eSanjeevni.in during the telemedicine session

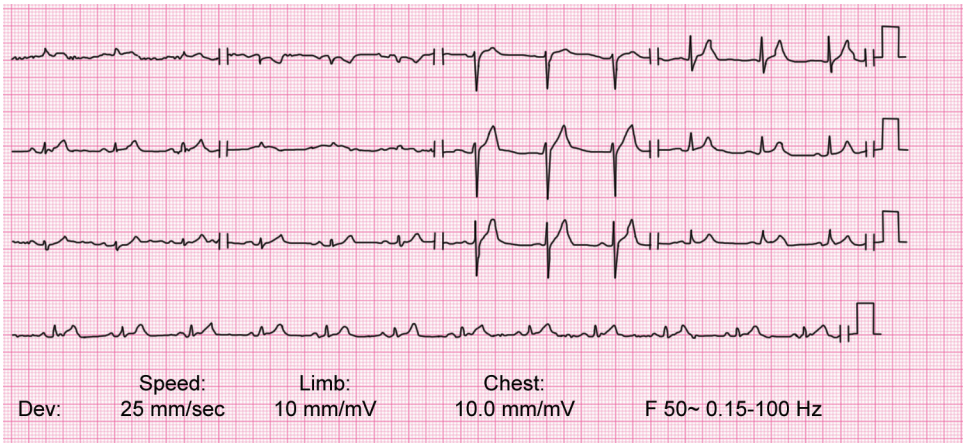


Fig. 2: Case 2: ECG sent during the tele-session. It shows ST elevation in L1, L2, V2–V5 possibly early repolarization

Table 1: SMO (smoker), SLT (smokeless tobacco), Ex-SLT (former smokeless tobacco user), Ex-smoker (former smoker) (*n* = 120). As regard the body mass index of the patients, it was observed that 56 (56.66%) of the study population had more than ideal weight (overweight, obese classes 1 and 2) (Table 2 and Fig. 3)

	Total (%)	*Male (%)*	*Female (%)*
Current smoker	19 (15.83)	18 (15)	1 (0.83)
SLT	4 (3.33)	3 (2.5)	1 (0.83)
SMO + SLT	3 (2.5)	2 (1.6)	1 (0.83)
SMO + Ex-SLT	2 (1.6)	2 (1.6)	0
Ex-SLT	1 (0.83)	1 (0.83)	0
Ex-smoker	5 (4.1)	5 (4.1)	0
Non-smoker + non-SLT	86 (71.66)	32 (26.66)	54 (45)

Table 2: Distribution of body mass index in study population

BMI	*Total (%)*	*Male (%)*	*Female (%)*
Underweight	6 (5)	3 (2.5)	3 (2.5)
Normal	46 (38.33)	25 (20.83)	21 (17.5)
Overweight	46 (38.33)	26 (21.66)	20 (16.66)
Obese class 1	19 (15.83)	8 (6.66)	11 (9.11)
Obese class 2	3 (2.5)	0 (0)	3 (2.5)

smokers (18.33%). Out of total tobacco users, only 2.49% were females (Table 1).

Disease Profile

More than 75% of the subjects had diabetes out of which 46% had diabetes alone and 29.99% had other comorbidities like hypertension, hypothyroidism and CAD. Most of them had uncontrolled blood sugar. Strikingly the prevalence of HTN alone was only 6%. These figures are eye openers for the health fraternity and health planners (Table 3 and Fig. 4). So, it is important to know that most of these diabetic patients were uncontrolled at the first visit because of noncompliance of medicine due to economic reasons. Even when the medicine is made available to them by the NGO team, there used to be a break in the continuity of taking medicine due to their unavailability to pick up medicine from the center in due time. Notably that many of the patients who were tobacco users particularly the smokers did not quit smoking and smokeless tobacco. These cases needed 3 to 5 tele-sessions to persuade them to stop smoking. Regrettably many of them did not stop tobacco even after persistent counseling.

Table 3: Disease profile: T2DM, HTN, CAD, and NCD (*n* = 120)

Disease	*Total (%)*	*Male (%)*	*Female (%)*
T2DM	56 (46)	27 (22.5)	29 (24.16)
HTN	8 (6)	6 (5)	2 (1.6)
CAD	1 (0.83)	1 (0.83)	0
Hypothyroidism	4 (3.33)	2 (1.66)	2 (1.66)
T2DM + other CVDs	31 (25.83)	21 (17.5)	10 (8.33)
T2DM + hypothyroidism	3 (2.5)	0	3 (2.5)
HTN + other CVDs	1 (0.83)	1 (0.83)	0
T2DM + HTN + hypothyroidism	2 (1.66)	0	2 (1.66)
Diseases other than NCD	14 (11.66)	4 (3.33)	10 (8.33)

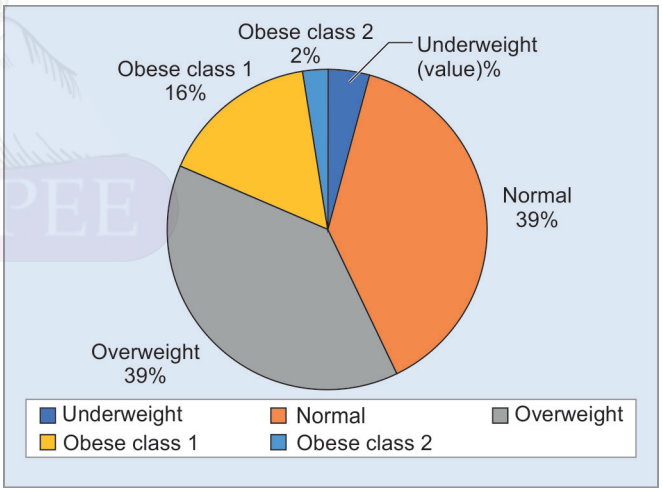


Fig. 3: Body mass index distribution (*n* = 120)

Discussion

According to the WHO, the goal of telemedicine is to provide good medical care to the people living remotely in a country like India where 75% of the population is poor and the number of health care professionals is not enough in remote areas.[5] Telemedicine is

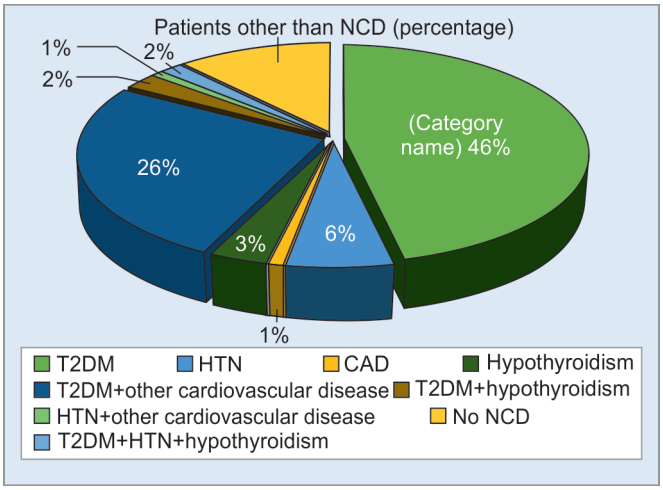


Fig. 4: Overall disease profile

an effective approach to deal with the problem.[1] It consists of five distinct domains:

1. Real time—this is the audiovisual ICT connection between the doctor and the patient.
2. Store and forward—in which the investigatory test reports are uploaded and transmitted through ICT to the doctor for examining.
3. Remote patient monitoring—this is a real-time transmission of data in which the patient is wearing wearable sensor capturing the vitals and transmitting them on the real-time basis.
4. Mobile health—various apps sending alerts to the patient by monitoring the vitals of the patient.
5. Health education delivered by ICT and online discussion with the health care professionals.[1]

Our study was primarily meant to give health advice to the rural population of Jewar. This also gave us an opportunity to study the epidemiological profile and prevalence risk factors for various cardiovascular disorders. Besides giving medical advice, we used the tele-sessions to impart healthy lifestyle education. In developing countries like India, the rising incidence of noncommunicable diseases (NCDs) is causing an increase in the morbidity and premature mortality. Smoking is one of the major risk factors in India. Consumption of tobacco causing mortality is huge. Smokeless tobacco and smoking of *bidi* are the most common types of tobacco used in India and the prevalence of tobacco consumption in rural areas of India is alarming. Young kids under 15 years consume tobacco.[6] Increasing tobacco use coupled with the rising incidence of diabetes and HTN are the major causes of concern for India. In the telemedicine, most of the diagnosis is done visually. The long-term effects of chronic smoking can be easily seen like darkening of lips, wrinkles, scarring pigmentation yellowing of teeth, and hair loss during visual sessions. The most noticeable immediate effects of smoking are stained teeth and tongue, gum discoloration (also called smoker's melanosis), and changes in the look of the roof of the mouth (palate). Our study revealed this in no less a manner wherein tobacco users among rural Greater Noida are as high as 28%. Also, an alarming prevalence of diabetes (75.32%) was seen. This is an eye opener for all of us. The irony is that most patients already knew that they have "sugar" (diabetes mellitus), "pressure" (HTN), and/or "heart attack" (CVD) but even then they failed to follow a healthy lifestyle. Most of them were not taking medicine for above ailments at the time of first reporting to tele-sessions. In spite of the fact that NGO provides medications and lab tests free of cost, they did not take medications regularly, which led to uncontrolled type 2 diabetes mellitus (T2DM) in many of the patients. About 38% of the cases do not visit for a follow up.

As we all know very well that diabetes is one of the major risk factors of CVDs burden in India, the prevalence of diabetes quadrupled in the rural areas of India because diabetes awareness is poor.[6] About 50% of people diagnosed with T2DM had at least one diabetes-related complication at the time of diagnosis.[7,8]

Using the telemedicine to impart health education and making people aware of the risk of faulty lifestyle along with providing medical advice and drugs can help in reducing the burden of CVDs in rural India and in remote locations. NCDs account for almost half the deaths among the low-income group people, leading to serious economic consequences.[9] Telemedicine assumes special relevance to reach such people and help them in reducing the prevalence of noncommunicable diseases like diabetes, HTN, CVDs, obesity, thyroid disorders, etc. It can be a very cost-effective tool.

Strengths and Limitations of the Study

The use of telemedicine for rural population, focusing on NCDs particularly CVDs and diabetes, has been done for the first time in India. The number being small is one of the lacunae which we hope to augment in the future. However, this study, though small, does give us useful information about the high prevalence of CVD and T2DM in a rural area of National Capital Region.

Conclusion

More than 76% of telemedicine patients had T2DM in the rural Jewar area. In most cases, blood sugar levels in follow-up visits were uncontrolled. People knew about diabetes and HTN but did not take it seriously as they failed to follow advice about lifestyle changes and did not take prescribed drugs properly. In most cases, urinalysis and other relevant workup were not done at the first instance. Uncontrolled T2DM had resulted in retinopathy, nephropathy, and neuropathy in some of our subjects. Telemedicine offers the opportunity of one-to-one interaction with visual images of both the patient and the doctor. In rural areas, people do not understand the severity of their condition. Telemedicine can be an effective tool for diagnostic care and prevention of CVD in rural areas.

Acknowledgement

The tremendous efforts put up by Mrs Indritta Singh for organizing "Telemedicine" session is gratefully acknowledged.

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ORIGINAL ARTICLE

Quality of Life in Patients with Acne Vulgaris

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Abstract

Background: Dermatological diseases have a significant impact on a patient's quality of life. The present study was to evaluate the impact of acne vulgaris on their quality of life.

Methods: Two hundred and fifty-six patients with acne vulgaris were considered for this study. Pre-tested personal interviews were done on all patients for the collection of data. This information was used for grading of acne vulgaris and its complications. Further evaluation of its impact on quality of life was done using the Dermatology Life Quality Index (DLQI) scale. The percentage and frequency of data were assessed by Chi-square test, and final analysis was done using Statistical Package for the Social Sciences (SPSS) version 20th.

Results: Small effect on DLQI was noted in 59 (45.7%) patients with mild severity acne, moderate effect in 42 (41.6%) patients with a moderate severity of acne and 14 (53.8%) patients with severe acne had a small effect on DLQI. There was a significant association between the outcome of the DLQI scale and grading of acne ($p < 0.0001$).

Conclusion: Depending on the severity, acne vulgaris has a considerable impact on the quality of life of patients. The study showed significant impairment of quality of life in acne patients. Proper treatment early in the course of the disease, along with counseling and assurance of the patient plays an important role in efficacious management and in reducing the psychosocial impact.

Keywords: Acne vulgaris, Dermatology life quality index, Quality of life.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1222

Introduction

Acne vulgaris is a common chronic skin disease in which there is inflammation of pilosebaceous unit. It has varying presentations, in the form of open and closed comedones, papules, pustules, and nodules in more severe cases. Face, upper chest and upper back are the most common sites affected.[1] Although it may occur in all age groups, it is primarily a disorder of adolescence. With an approximate prevalence of 85% in all individuals at one or the other time, peak incidence occurs during adolescence. Despite being a disease of youth, 12% of women and 3% of men will continue to have acne until the age of 44 years.[2] The pathophysiology of acne involves the complex interaction of multiple factors, both internal and external with the pilosebaceous unit. The scarring in acne occurs due to inflammation leading to the disfigurement of the face in eventually most of the cases associated with psychological comorbidity hampering the patient's quality of life.

Clinical assessment of acne alone cannot evaluate the impact of the disease adequately. Its impact on the health-related quality of life is thus a prime tool to assess the complete burden of the disease and treatment efficacy.[3] Quality of life (QoL), as defined by World Health Organization (WHO), is the "individual's perception of their position in the context of culture and value systems in which they live and in relation to their goals, expectations, standards, and concerns".[4] It is an important method to assess the general well being of an individual, focusing on both positive and negative aspects of life. It observes the functioning, well-being and life satisfaction of the individual, and indicates the individual's health status and quality of life. Skin diseases in general and acne in particular, greatly affect the quality of life of the patients. QoL thus serves as an important tool in analyzing the treatment effect at the patient's level.[5] The use of this entity helps us achieve a better understanding of the effect of acne on the life of the patient on a day-to-day basis and also the efficacy of therapy. This would ultimately enable us to use more targeted interventions for improved management of the disease. Dermatology Life Quality Index (DLQI), a questionnaire developed by Finlay and Khan, is a sensitive measure commonly used in clinical practice and research areas to assess changes in health-related QoL.[6,7] Studies on the impact of acne on QoL have been evaluated in the USA, UK, Spain, Brazil, Iran, Malaysia and Greece.[8] In India, fewer studies on this issue have been conducted.

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How to cite this article: Salve GN, Chavan SD, *et al*. Quality of Life in Patients with Acne Vulgaris. MGM J Med Sci 2019;6(1):11–14.

Source of support: MGMIHS

Conflict of interest: None

Materials and Methods

Our study was a cross-sectional, hospital-based, a prospective study done in the Outpatient Department of Skin and Vernal Diseases (VD) in a tertiary care teaching hospital, over a period of two years, from January 2015 to December 2016. It has been approved by the Institutional Research and Ethical Committee.

Patients diagnosed clinically with acne vulgaris, with age more than 12 years were included in the study. Informed consent of the patient and from guardian/ parent in case of minors, was taken in all cases. Patients with a history of any chronic medical/ surgical illness or known chronic mental disorders were excluded from the study.

A sample size of 256 was calculated using the prevalence of acne vulgaris and available literature on its a psychological impact. Patients of acne vulgaris meeting inclusion and exclusion criteria

were approached for participation in study till sample size was reached. Personal interviews of all patients were conducted using a pre-tested semi-structured schedule.

The collected data was analyzed using a Statistical Package for the Social Sciences (SPSS) version 20th. The quantitative data was represented in form of frequency and percentage. Chi-square test was applied to check the significant difference between the severity of acne and the DLQI, using *p* value at 0.05 level of significance. A detailed history of all patients was taken with respect to the presenting complaints, duration, and onset, the presence of any aggravating factors, seasonal variation, stress, use of any oral or topical medications or cosmetics, socio-demographic data, etc. Dietary history, menstrual history in females, associated symptoms like weight gain were noted. A single dermatologist conducted the cutaneous examination on all patients to avoid inter-observer errors. Grading of the severity of acne was done using Pillsbury, Shelley and Kligman Grading system.[9] Following grades were included:

- *Grade 1:* Comedone and occasionally small cysts confined to the face.
- *Grade 2:* Comedone with occasionally pustules and small cysts confined to the face.
- *Grade 3:* Many comedones and small and large inflammatory papules and pustules, more extensive but confined to the face.
- *Grade 4:* Many comedones and deep lesion tending to coalesce and canalize, and involving the face and upper aspect of trunk.[9]

The dermatology life quality index (DLQI) questionnaire was used as the study instrument.[5] After obtaining consent, all patients were asked to fill out the questionnaire without any assistance, since the questionnaire is self-explanatory. Domains covered in the DLQI questionnaire included: (a) physical symptoms and feelings (question 1 and 2), (b) daily activities (question 3 and 4), (c) leisure (question 5 and 6), (d) work/school (question 7), (e) personal relationships (question 8 and 9) and (f) treatment (question 10). Each question is scored as "very much" (score 3), "a lot" (score 2), "a little" (score 1), and "not at all", "not relevant", "question unanswered" (score 0). Final DLQI score is the sum of all scores (range 0–30). High scores indicate a poor quality of life. The DLQI can also be expressed as a percentage of the maximum possible score of 30. Interpretation of DLQI score: 0–1 no effect on patient's life, 2–5 small effect, 6–10 moderate effect, 11–20 very large and 21–30; extremely large effect.[6]

Interpretation of Incorrectly Completed Questionnaires

In the event of any mistakes by the patient in filling out the questionnaire, the interpretation was done using the following points:

- Any question left unanswered to be scored as 0 and the scores are summed and expressed as a usual maximum of 30.
- In the case of two or more unanswered questions, scoring is not done.
- If question 7 is answered yes this is scored 3. If question 7 is answered no or not relevant but then either a lot or a little is ticked this is then scored 2 or 1.
- If two or more response options are ticked, the response option within the highest score is recorded.
- If there is a response between two tick boxes, the lower of two score option is recorded.
- The DLQI can be analyzed by calculating the score for each of its six subscales. When using subscale, if the answer to one question in subscale is missing, that subscale should not be recorded".[6]

Results

The maximum patients were from age-group 21–30 years, i.e. 148 (57.8%) and whereas only 6 (2.4%) were from age-group 41–50 years. The mean age of the present study was 25.24 years (Table 1).

Out of 256 patients, 102 (39.8%) were male, and 154 (60.2%) were females (Table 1).

Out of 256 acne patients, the maximum patients 101 (39.4%) were from Grade 3 and minimum were observed in Grade 4, i.e. 26 (10.1%) whereas 44 (17.2%) and 85 (33.2%) patients were observed in Grade 1 and Grade 2 acne respectively (Table 2).

In the present study, 129 (50.4%) patients were having mild acne, 26(10.1%) were having severe acne, and 101 (39.5%) were having moderate acne (Table 3).

In this study, maximum 96 (37.5%) acne patients were having a small effect and 88 (34.4%) patients were having a moderate effect. Only 5 (2%) patients showed an extremely large effect (Table 4).

In mild severity of acne, 59 (45.7%) patients were having a small effect of DLQI. In moderate severity of acne, 42 (41.6%) patients were having a moderate effect of DLQI. In severe acne patients, 14 (53.8%) patients were found to have a small effect of DLQI (Table 5). There was a significant association between the severity of acne and DLQI scale outcome (p <0.0001).

Discussion

This is a cross-sectional observational study conducted in dermatological patients, and its results may not generalize to other population. In this study, we evaluated the prevalence of psychiatric morbidity in patients with acne vulgaris and to what extent DLQI affected them.[10] The study group included patients above 12 years of age.

In this study, about 39.8% of the patients were males, whereas 60.20% were females. The study by Schafer et al.[11] showed that acne was more prevalent in males 29.9% than females 23.7% which is in variance to this study, in which females predominated. In the

Table 1: Distribution of patients according to age-group and gender

S. No.	*Variables*		*Frequency*	*Percentage (%)*
1	Age	<20	60	23.4
		21–30	148	57.8
		31–40	42	16.4
		41–50	06	2.4
		Total	256	100
		Mean SD	25.24+/–6.49 years	
2	Sex	Male	102	39.8
		Female	154	60.2
		Total	256	100

Table 2: Distribution of patients according to grading of acne

Grading	*No. of patients*	*Percentage (%)*
Grade 1	44	17.2
Grade 2	85	33.2
Grade 3	101	39.4
Grade 4	26	10.1
Total	256	100

Table 3: Distribution of patients according to the severity of acne

Severity of acne	*No. of patients*	*Percentage (%)*
Mild	129	50.4
Moderate	101	39.5
Severe	26	10.1
Total	256	100%

Table 4: Distribution of patients according to dermatological life quality index

DLQI	*No. of patients*	*Percentage (%)*
No effect	16	6.3%
Small effect	96	37.5%
Moderate effect	88	34.4%
Large effect	51	19.9%
Extremely large effect	5	2.0%

Table 5: Association between severity of acne and DLQI score:

	DLQI					
Severity of acne	*No effect*	*Small effect*	*Moderate effect*	*Large effect*	*Extremely large effect*	*Chi-square value*
Mild	12	59	39	17	2	Chi-square value $s = 25.26\%$
Moderate	4	23	42	30	2	$p < 0.0001$
Severe	0	14	7	4	1	Statistically significant
Total	16	96	88	51	5	

distribution of patients according to age group maximum patients (57.8%) were from the age group 21–30 years. Mean age was 25.24 years. Schafer et al.[11] and Collier et al.[12] found a predominance of acne in the age group of 14 to 29 years which is similar to our study.

A maximum number of patients in our study (39.4%) had Grade 3 acne and least (10.1%) had Grade 4. Severity-wise, 50.4% had mild acne whereas severe acne and moderate acne percentages were 10.1% and 39.5% respectively in our study. DLQI score grading in our patients showed: 6.3% patients no effect, 37.5% had a small effect, 34.4% had a moderate effect, and only 2.0% of patients had an extremely large effect. While checking for the association between severity of acne and DLQI score we found that in mild severity acne, 45.7% patients had a mild effect on their DLQI whereas percentage of no effect, moderate effect, very large effect, extremely large effect were at 9.3%, 30.2%, 13.2%, 1.6% respectively. In moderate severity acne, maximum patients were found in moderately affected DLQI, i.e. 41.6% whereas the percentage of patients in no effect, small effect, very large effect, and extremely large effect groups were 4%, 23%, 30%, and 2% respectively. In severe acne, maximum patients (53.8 %) had a small effect on DLQI, whereas no effect, moderate effect, very large effect, and extremely large effect percentages were 0%, 26.4%, 15.4%, and 3.8% respectively. These figures show a significant association between the severity of acne and DLQI score ($p < 0.0001$). Martin et al.[5] observed that quality of life (QoL) score correlated with the severity of acne. They also observed that QoL in the facial acne correlated with patient's psychiatric disease severity. Pawin et al.[13] reported in their study that quality of life was affected by 48% of the patients with acne. These findings are similar to those of our study in which 189 patients out of 256 (73.8%) showed impaired QoL. However, Yazici et al.[14] found no correlation between the severity of acne and DLQI which is at variance with our study.

Saker et al.[10] found that in most of the patients (42%) the effect of the disease on their lives was minor, in 33.6% it was very large effect and in only 6.4% there was no effect of the disease on their lives. The relation between DLQI and severity of acne was statistically significant. In patients having mild severity of acne, there was a small effect on DLQI, whereas, in patients with severe acne, the effect was relatively large. On comparing our study with that of Saker et al., we found corroborative results viz. effect of acne on DLQI was small in 45.7% patients and moderate in 41.6%.

Conclusion

The visibility of skin diseases increases the likelihood of stigmatization. Skin diseases should be measured by physical, psychological and social parameters along with symptoms. Understanding of mind and body interaction and interventions can help to improve patient's skin conditions and ultimately their quality of life. Physicians concerned with patients' mental wellbeing should also consider referral to properly trained specialists in cosmetic camouflage to diminish or disguise facial or other disfigurements. Acne vulgaris, especially those involving the face, is a common skin disorder that can cause disfigurement of appearance. It affects many young people and is usually considered to be a cosmetic issue only.[15] Dermatologists need to be aware of psychological issues caused by acne, which can have serious consequences. It may be prudent to provide a basic standardized quality of life index assessment upon clinical evaluation of new and current acne patients. Consistent utilization of this tool would allow clinicians to better identify coexisting psychiatric comorbidity in their patients and tailor a regimen to each individual patient that would help to optimize treatment outcomes.[15] Management of skin disorders may be optimized by nonpharmacological or pharmacological-cum-psychological interventions.

Limitation

Following are the limitations of this study:

- This is a cross-sectional study with a limited sample size, and its result may not be generalized to other populations.
- The finding of assessment of psychiatric scales, i.e. DLQI of this study was based on information provided by acne vulgaris patients on above mentioned psychiatric scale questionnaire which are subject to some informant bias.

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ORIGINAL ARTICLE

Distensibility in a Small Size Vein as a Factor for Brachiocephalic Arteriovenous Fistula Maturity: A Single-centre Experience

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Abstract

Introduction: National Kidney Foundation Kidney Disease Outcomes Quality Initiative (NKF-KDOQI) guidelines require an arterial diameter of more than 2 mm and a venous diameter of more than 2.5 mm for arteriovenous fistula (AVF) creation. The failure rate in vein diameters of less than 2 mm is as high as 40%. An audit done in Hospital Kuala Lumpur demonstrated that up to 67% of patients who are referred for AVF creation have veins smaller than 2.5 mm. So as per the NFK-KDOQI guidelines, creation of AVF is not suitable for them.

Objective: The objective of this study is to demonstrate venous distensibility as a reliable assessment tool prior to fistula creation in small size veins (1.6–2.4 mm).

Materials and methods: A single-center prospective study was conducted at the Vascular Unit, Department of Surgery, Hospital Kuala Lumpur (HKL). Sixty patients with cephalic veins in cubital fossa 1.5–2.4 mm were assessed per the standard care for fistula creation. A tourniquet was applied using a sphygmomanometer cuff and inflated to occlude superficial venous return. Pre- and post-compression readings were taken at cubital fossa at 3 cm and at 6 cm above the cubital fossa. Patients were planned for surgery per the standard care. Follow-up was done at 2 weeks for clinical assessment, at 6 weeks to determine the maturity for early access, at 3 months to identify mature fistula, and at 6 months to identify any failure of fistula maturity if any, after the intervention.

Results: The mean vein diameter of selected veins was 2.1 mm. The mean percentage of vein distention preoperatively for fistula maturity with a tourniquet was 30%. Five patients did not turn up for follow-up. One fistula thrombosed, one fistula was ligated due to the steal syndrome, and three patients passed away from myocardial infarction. The total number of patients recruited was 60, five patients dropped out for reason as stated. Hence, 55 patients developed mature fistulas.

Conclusion: Venous distensibility as a preoperative assessment tool in patients with smaller veins can be used to select a larger number of patients for the creation of AVF without compromising fistula patency and maturation rate.

Keywords: Arteriovenous fistula, Brachiocephalic fistula (BCF), Dialysis, Fistula, Tourniquet, Venous distensibility.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1223

Introduction

Primary arteriovenous fistula (AVF) is considered as the first choice for vascular access due to better primary and secondary patency rates along with fewer infections and thrombotic complications as compared to prosthetic arteriovenous grafts.[1]

The National Kidney Foundation Kidney Disease Outcomes Quality Initiative (NKF-KDOQI) guidelines recommend that at least 50% of new hemodialysis patients have a primary AVF as these have better patency rates along with lower access-related costs.[2] Per these guidelines, patients planned for fistula creation should be assessed clinically and vein mapping done using a duplex scan, to assess suitability of vessels for fistula creation. The arterial diameter should be more than 2 mm and the venous diameter should be more than 2.5 mm.[3] The failure rate in vein diameters less than 2 mm has been reported to be as high as 40% in recent literature.[4–8] There are no guidelines that advocate venous distensibility as a predictor for fistula maturity.

A local audit conducted in Hospital Kuala Lumpur (HKL) demonstrated that up to 67% of patients do not fulfill the criteria of vein diameter for the creation of AVF. Lockhart et al. described that prior to the creation of a radiocephalic fistula, the use of a venous tourniquet increased the number of patients suitable for forearm fistulas without compromising the patency significantly and suggested that a tourniquet be routinely used in patients with small cephalic veins.[9] An article in the Annals of Vascular Surgery also identified venous distensibility as an indicator of successful radiocephalic fistula creation and demonstrated that distensible veins had a four-fold higher success rate.[10] van der Linden et al. using forearm venous distensibility measured the venous diameter using the ultrasound duplex with strain-gauge plethysmography prior to surgery and demonstrated that venous distensibility and not the luminal diameter is a predictor for AVF success.[1]

The aim of this study is to demonstrate venous distensibility of small-sized veins (1.6–2.4 mm) as an evaluation tool for creating an AVF. Post-distention increase in the diameter by 40% or an increase

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How to cite this article: Arvind M, Idris MA, *et al.* Distensibility in a Small Size Vein as a Factor for Brachiocephalic Arteriovenous Fistula Maturity: A Single-centre Experience. MGM J Med Sci 2019;6(1):15–18.

Source of support: Nil

Conflict of interest: None

in the diameter to 2.5 mm or more after distention was considered suitable criteria to use the veins for fistula creation.

Materials and Methods

This study is registered under the National Medical Research Register (NMRR), Malaysia, and has been approved by the Medical Research and Ethics Committee, Ministry of Health, Malaysia along with the Research Ethics Committee, National University Malaysia. It is a prospective cohort single-center study conducted at the Vascular Unit, Department of Surgery, Hospital Kuala Lumpur from May 2017 till January 2018. Chronic kidney disease (CKD) stage IV and above patients planned for native brachiocephalic AVF creation at the Department of Surgery, HKL, with 18 years and above of age, having cephalic vein luminal diameter between 1.6 mm and 2.4 mm which distended by more than 40% or achieved a diameter of more than 2.5 mm after distention, as measured on a duplex scan, were included in this study.

All patients fulfilling the inclusion criteria were counseled and, if agreeable, were asked to give consent to enroll in this study. Subsequently, a tourniquet (sphygmomanometer cuff) was applied to the arm of the patient and inflated to a pressure of 40 mm Hg and kept for 1 minute to occlude superficial venous return. Then a repeat duplex scan was carried out to assess the distensibility of the cephalic vein. The total number of patients meeting these criteria was 60. All these patients underwent brachiocephalic AVF creation. After surgery, all patients were followed up as follows:

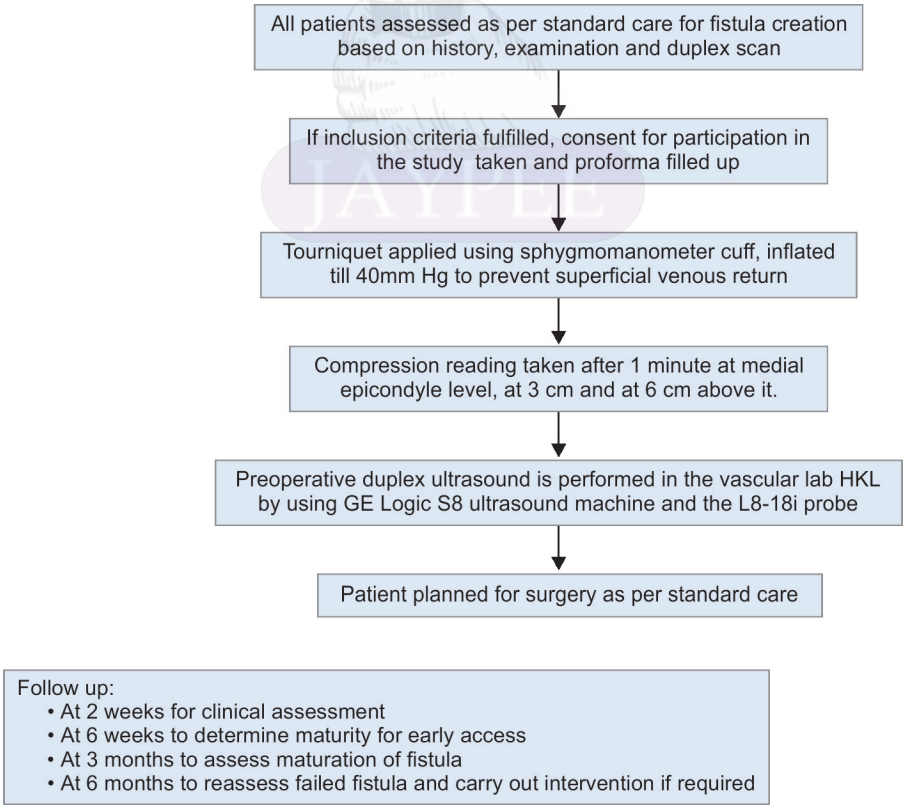
- At 2 weeks for clinical assessment (wound healing, presence of thrill)
- At 6 weeks to assess if the fistula is ready for cannulation, using duplex sonography, measuring flow, depth from the skin, and diameter of the vessel
- At 3 months to assess fistula maturity
- At 6 months, if fistula fails to mature and/or requires any intervention
- At any point during the follow-up period should the patient require primary assist procedures, i.e., percutaneous transluminal angioplasty, ligation of accessory veins, creation of a more proximal neo-anastomosis, interposition of a short segment of vein graft or thrombectomy, these procedures are carried out.

The statistical analysis was performed with Statistical Package for the Social Sciences (SPSS) Statistics V21 (IBM Corp., Armonk, NY). Categorical variables are presented in frequency and percentage, while the numerical variables are presented in mean and standard deviation. The usable segment of the fistula was taken between the 3-cm and the 6-cm point corresponding to the inflow and outflow points. This measurement was used due to the practicality where the fistula could be punctured during dialysis.

A mature fistula is defined as a fistula suitable for cannulation with a flow of 600 mL per minute, a luminal diameter of 6 mm, and 6 mm depth from skin.[2] A non-maturing fistula is defined as a fistula that fails to mature after 6 weeks. The primary failure is defined as fistulas that are not usable for dialysis or that fail within 3 months of use.[11] The late failure was defined as fistulas that failed to mature at 6 months, even after primary intervention.

All the ultrasonography assessment of fistula pre- and postoperatively was performed by two trained medical attendants with more than 2 years of experience in fistula assessment using ultrasound and the primary author (Flowchart 1).

Flowchart 1: Flowchart of methodology



Results

A total of 60 patients who fulfilled the inclusion criteria were recruited to participate. Their baseline characteristics are listed in Table 1.

Table 2 shows distensibility of veins less than 2.5 mm in diameter. Twenty-three percent of veins were distended by 40% and remaining distended by less than 40%. However, the veins which distended less than 40% fulfilled the inclusion criteria by achieving an increase in the diameter of 2.5 mm or more.

Table 3 shows that the mean comparison along with the standard deviation between pre- and post-distention was examined that demonstrated a statistically significant *p* value. The *p* value was generated using the paired *t* test analysis. Therefore, on an average, venous distention led to an increase in the diameter of the vein allowing it to fulfill NKF-KDOQI guidelines.

Table 4 shows that anteroposterior (AP) diameter of veins along with three fixed points distended by an average of 30% with a 95% confidence interval.

Five patients were dropped out of the study. One fistula thrombosed after 1 month, one fistula was ligated due to the steal syndrome 5 days after surgery, and three patients passed away from myocardial infarction before the 6 weeks follow-up review. Remaining 55 patients were available for follow-up.

Table 5 shows the size of fistula at three levels (cubital fossa, 3 cm, and 6 cm from the cubital fossa) during the follow-up at 6 weeks, 3 months, and 6 months. All 55 fistulae matured in terms of size mostly within 6 weeks. Only in two patients, the fistula took longer time to mature but their maturation was also complete at 6 months without any primary intervention.

Table 1: Baseline characteristics of patients with small veins (*n* = 60)

Characteristics	*n (%)*
Gender	
Male	23 (38.33)
Female	37 (61.67)
Comorbidities	
Diabetes	45 (75.00)
Hypertension	49 (81.67)

Table 2: Percentage of venous distensibility post-tourniquet

Anterior–posterior diameter (pre-distention)	*Increase in diameter post-distention n (%)*	
	≥40%	*<40%*
<2.5 mm (*n* = 60)	14 (23%)	46 (77%)

Table 3: Comparisons of pre-distention and post-distention vein diameters (*n* = 60)

	Mean (standard deviation)		
Vein diameter	*Pre-distention*	*Post-distension*	*p value*[a]
Cubital fossa	2.32 (0.50)	3.00 (0.77)	<0.001
3 cm	2.15 (0.48)	2.77 (0.60)	<0.001
6 cm	2.10 (0.42)	2.84 (0.61)	<0.001

[a]Paired *t* test

Table 4: Mean percentage of vein distensibility (*n* = 60)

Vein diameter	*Mean percentage (95% confidence interval)*
Cubital fossa	30.46 (24.62, 36.29)
3 cm	30.40 (25.94, 34.85)
6 cm	36.90 (30.94, 42.86)

Table 5: Fistula size during follow-up visits (*n* = 55)

	Mean (standard deviation)		
Anterior–posterior diameter	*Week 6*	*Month 3*	*Month 6*
Cubital fossa	6.51 (1.11)	6.30 (1.10)	5.90 (0.14)
3 cm	6.61 (1.12)	6.37 (0.88)	6.20 (0.00)
6 cm	6.63 (1.07)	6.37 (0.88)	6.20 (0.28)

Discussion

In 2014, there were 34,767 patients in Malaysia receiving dialysis, which shows a two-and-half-fold increase from 13,356 patients in 2005. While the new dialysis patients included in 2005 were 3,167, the number had increased to 7,055 in 2014.[12] Among the dialysis patients, 13.4% were on peritoneal dialysis. There was also significant demographic change in dialysis population. Patients above 55 years made up 58% of all new dialysis patients in 2014 vs 52% in 2005. In 2014, the death rate was 12% among hemodialysis patients and 16% in the peritoneal dialysis group.[12] A majority of dialysis patients died due to cardiovascular disease.[12]

Compared to AVFs, tunneled central venous catheters are associated with a 15-fold greater risk for bacteremia.[13] Infection-related hospitalizations were higher in patients on peritoneal dialysis as compared to those on hemodialysis.[14] This leads to the increase in healthcare costs in patients undergoing peritoneal dialysis as compared with hemodialysis. Hemodialysis, using native access of AV fistula, remains the renal replacement therapy of choice.

Satisfactory maturation of AV fistulae for use in hemodialysis is largely dependent on the size of veins. The failure rate of veins less than 2.5 mm size is as high as 40%. But, in practice, we do have many patients with small caliber cephalic veins (less than 2.5 mm) in cubital fossa. According to NKF-KDOQI guidelines, such patients would not qualify for brachiocephalic fistula creation. So, we carried out this prospective study to identify if patients with small caliber cephalic veins (less than 2.5 mm) can undergo the creation of brachiocephalic fistula successfully. We hypothesized that if we can use distensibility of veins prior to surgery as an assessment tool and accept those veins which distend by 40% or more or achieve a post-distention diameter of 2.5 mm or more, we can create a fistula in such patients. Our results show encouraging outcomes. There are many other modifiable and non-modifiable factors influencing fistula maturity which have been summarized by Smith et al.[16] These include smoking, obesity (body mass index >35), and cannulation before 14 days, which lowered patency rates.

Complications, which may arise from small vein AVF creation, are similar to those of normal sized fistula creation, namely infection, pseudoaneurysm formation due to repeated puncture at the same site, ischemic neuropathy, thrombosis of fistula, and the possibility of central vein stenosis due to the presence of the previous indwelling catheter.[15] In our study, thrombosis of the fistula was seen only in one patient that may be because the patient was returning early to his labor-intensive work. The steal syndrome was seen in one patient which is most likely due to the surgical technique as it was seen within 3 days from surgery.

This study was limited to only brachiocephalic fistulas. We believe that this finding can be extrapolated for all types of native fistula creation. If possible, a future randomized trial with a larger number of patients and accepting venous distensibility of even less than 40%, as in this study, will provide more useful data in success rates of creating AV fistulae in small caliber veins.

Conclusion

Hemodialysis using native access AV fistulae remains the best option for renal replacement therapy of the end-stage renal disease patients. To create a successful AV fistula, a minimum vein size of 2.5 mm is recommended. However, in practice, especially in the Asian population, many patients have cephalic veins less than 2.5 mm in size. This study demonstrated that venous distensibility can be used as a reliable assessment tool prior to operation to create AV fistulae in patients with small-sized veins. If veins are distensible by 40% or if they achieve a diameter of 2.5 mm or more post-distention, they can be used with a predicted maturity rate of 90%. This will enable us to offer construction of brachiocephalic AV fistula in a larger number of the end-stage renal disease patients requiring hemodialysis.

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ORIGINAL ARTICLE

Antibiotic Prophylaxis with Episiotomy—Is it Necessary?

Arina Chandrababu[1], Sushil Kumar[2] and Ajita Goli[3]

Abstract

Background: A hospital-based prospective randomized controlled trial was conducted at a tertiary care hospital. The aim of the study is to determine if a single dose of intravenous antibiotic was superior to a course of oral antibiotics post-episiotomy or no antibiotics at all.

Materials and methods: A total of 300 cases undergoing normal vaginal delivery with episiotomy were included in this study and were randomly divided into three groups.

Group A received a single prophylactic injectable dose during episiotomy, group B received oral antibiotics for 3 days after the delivery, and group C received no antibiotics. The groups were compared per day 3 total leukocyte count (TLC), presence of foul-smelling lochia, episiotomy gaping, and mean redness, edema, ecchymosis, discharge, approximation (REEDA) score at the time of discharge.

Results: The mean age of the study group was 25.09 years with no difference between the study groups ($p = 0.356$). No difference was seen among IV antibiotic, oral antibiotic, and no antibiotic groups with respect to the mean total leukocyte count on the third postoperative day ($p = 0.69$). The prevalence of episiotomy gaping was seen in 2%, 3%, and 1% cases among IV antibiotic, oral antibiotic, and no antibiotic groups, respectively ($p = 0.60$). The mean REEDA score in subjects of IV antibiotic, oral antibiotic, and no antibiotic groups was 3.91, 4.11, and 4.04, respectively ($p = 0.49$). None of the patients in any of the groups showed long-term complications like keloid.

Conclusion: The present study showed that there is no additional benefit of prophylactic antibiotics, given before or after episiotomy.

Keywords: Episiotomy, Gaped episiotomy, Prophylactic antibiotics, REEDA score.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1224

Introduction

The episiotomy is defined as deliberate incision on perineum to enlarge the vaginal opening. This surgical incision is given when the presenting part is distending the vulva, almost at the end of the second stage of labor. After delivery of the baby and placenta, the episiotomy wound is sutured with an absorbable suture material.[1] Episiotomy was introduced in obstetric practice to facilitate vaginal births. Episiotomy wound is a clean wound but because of proximity to the anal canal, vaginal and bowel flora may contaminate the area. Therefore, episiotomy wound is named as "clean-contaminated" wound.[2] Antibiotic prophylaxis is given to avoid infection or contamination by surgical incision. It is generally given in the case of complete perineal tear, manual removal of placenta, or some cases of cesarean section.[3] Episiotomies are anatomically similar to a second-degree perineal laceration, involving the vaginal mucosa, connective tissue, and underlying muscles, and might not warrant the routine use of prophylactic antibiotics.[3–6] However, the use of prophylactic antibiotics for episiotomy seems to vary widely. In high-income countries, the use of prophylactic antibiotics for episiotomy is not practiced without any evidence of infection.[4,5] However, in low-income countries like India, administering antibiotics with episiotomy is almost a norm.[6] Antibiotic prophylaxis is a low-cost, accessible intervention that may prevent considerable maternal morbidity. It is, therefore, important to establish the benefits of prophylactic antibiotics and also to assess whether there are any adverse effects on the mother or the infant. However, the current evidence regarding the routine use of prophylactic antibiotic with episiotomy is limited. With this background, the present study was planned to determine whether antibiotic prophylaxis used before or after episiotomy reduces the incidence of wound infection.

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How to cite this article: Chandrababu A, Kumar S, *et al*. Antibiotic Prophylaxis with Episiotomy—Is it Necessary? MGM J Med Sci 2019; 6(1):19–21.

Source of support: MGMIHS

Conflict of interest: None

Materials and Methods

The prospective randomized control study was conducted at the Department of Obstetrics and Gynecology, MGM Hospital, Navi Mumbai, Maharashtra. The study population included all the patients undergoing vaginal delivery with episiotomy or having the first- or second-degree perineal tears during the study period of November 2015 to October 2017. A total of 300 cases undergoing normal vaginal delivery with episiotomy were included in the study. The patients were randomly divided into three groups (100 each) using computer-generated random numbers.

1. Group A: Received a single prophylactic IV dose of augmentin 1.2 g (clavulanic acid 200 mg + amoxicillin 1000 mg) just before an episiotomy.
2. Group B: Received tab augmentin 625 mg (clavulanic acid 125 mg + amoxicillin 500 mg) twice daily for 3 days after the delivery.
3. Group C: Received no antibiotics.

Inclusion Criteria

- Patients undergoing full-term normal delivery.
- Patients with no associated comorbidities like anemia, gestational diabetes mellitus (GDM), pregnancy induced hypertension (PIH), or premature rupture of membranes (PROM).
- Patients not allergic to the above-mentioned antibiotics.
- Patients having leukocyte counts below 25,000.

Outcome Measures

The following outcome measures were compared between the three groups:

- Day 3 total leukocyte counts
- Foul-smelling lochia
- Wound gape
- Keloid (long-term outcome)

The significance threshold of the *p* value was set at <0.05.

Results

The mean age of the study group was 25.09 years with no difference between the study groups ($p = 0.356$). Out of the total 300 cases, 60.3% were registered cases, while 39.7% were unregistered deliveries (Table 1). Most of the patients were primipara (64.7%), while the remaining 35.3% were multipara. No difference was observed between the study groups with respect to parity ($p = 0.356$). Most of the deliveries in all three groups were term deliveries (37–42 weeks) with no difference between the study groups in terms of gestation age ($p = 0.412$). Normal episiotomy wound was achieved in 92.7% cases overall, while first-degree perineal tears (Table 2) were seen in 5%, 7%, and 6% cases of IV antibiotic, oral antibiotic, and no antibiotic groups, respectively. Second-degree perineal tears were inflicted in 2%, 1%, and 1% cases of IV, oral, and no antibiotic groups, respectively ($p = 0.93$). No difference was seen among IV antibiotic, oral antibiotic, and no antibiotic groups with respect to the mean total leukocyte count (Table 3) at admission and on the third post-op day ($p > 0.05$). The prevalence of foul-smelling lochia (Table 4) was seen in 3%, 5%, and 5% cases among IV antibiotic, oral antibiotic, and no antibiotic groups, respectively ($p = 0.72$). The prevalence of episiotomy gaping (Table 5) was seen in 2%, 3%, and 1% cases among IV antibiotic, oral antibiotic, and no antibiotic groups, respectively ($p = 0.60$).

Table 1: Distribution of cases as per registration of pregnancy

Registration of pregnancy	*Group*			
	IV antibiotics	*Oral antibiotics*	*No antibiotics*	*Total*
Registered	59	60	62	181
	59.0%	60.0%	62.0%	60.3%
Unregistered	41	40	38	119
	41.0%	40.0%	38.0%	39.7%
Total	100	100	100	300
	100.0%	100.0%	100.0%	100.0%
p value: 0.772				

Out of the total 300 cases, 60.3% were registered cases, while 39.7% were unregistered deliveries

Table 2: Distribution of cases as per status of episiotomy wound

Episiotomy wound	*Group*			
	IV antibiotics	*Oral antibiotics*	*No antibiotics*	*Total*
Normal	93	92	93	278
	93.0%	92.0%	93.0%	92.7%
First-degree perineal tear	5	7	6	18
	5.0%	7.0%	6.0%	6.0%
Second-degree perineal tear	2	1	1	4
	2.0%	1.0%	1.0%	1.3%
Total	100	100	100	300
	100.0%	100.0%	100.0%	100.0%
p value: 0.93				

Normal episiotomy wound was achieved in 92.7% cases overall, while the first-degree perineal tears were present in 5%, 7%, and 6% cases of IV antibiotic, oral antibiotic, and no antibiotic groups, respectively. The second-degree perineal tears were inflicted in 2%, 1%, and 1% cases of IV, oral, and no antibiotic groups, respectively ($p = 0.93$)

Table 3: Distribution of cases as per mean leukocyte count on day 3

Variables	*Group*	*N*	*Mean*	*SD*	*p value*
TLC (day 1)	IV	100	9987.40	971.80	0.77
	Oral	100	10034.20	673.40	
	Placebo	100	10923.00	781.20	
TLC (day 3)	IV	100	13369.21	971.80	0.69
	Oral	100	14043.11	673.40	
	Placebo	100	13999.65	781.20	

No difference was seen among IV antibiotic, oral antibiotic, and no antibiotic groups with respect to the mean total leukocyte count at the admission and on the third postoperative day ($p > 0.05$)

Table 4: Distribution of cases as per presence of foul-smelling lochia

Foul-smelling lochia	*Group*			
	IV antibiotics	*Oral antibiotics*	*No antibiotics*	*Total*
No	97	95	95	287
	97.0%	95.0%	95.0%	95.7%
Yes	3	5	5	13
	3.0%	5.0%	5.0%	4.3%
Total	100	100	100	300
	100.0%	100.0%	100.0%	100.0%
p value: 0.72				

The prevalence of foul-smelling lochia was seen in 3%, 5%, and 5% cases among IV antibiotic, oral antibiotic, and no antibiotic groups, respectively ($p = 0.72$)

Table 5: Distribution of cases as per presence of gaping of episiotomy wound

Episiotomy gape	*Group*			
	IV antibiotics	*Oral antibiotics*	*No antibiotics*	*Total*
No	98	97	99	294
	98.0%	97.0%	99.0%	98.0%
Yes	2	3	1	6
	2.0%	3.0%	1.0%	2.0%
Total	100	100	100	300
	100.0%	100.0%	100.0%	100.0%
p value: 0.6				

Prevalence of episiotomy gaping was seen in 2%, 3% and 1% cases among IV antibiotic, oral antibiotic and no antibiotic groups, respectively ($p = 0.60$)

Discussion

Episiotomy was introduced in obstetric practice to facilitate normal births and prevent maternal and fetal complications. Though there is a very high risk of contamination due to proximity with the vagina, urinary meatus, and the anal canal, the incidence of episiotomy infection is only between 0.3% and 5%.[7] The real aim of using antibiotic with episiotomy is to prevent infection by reaching therapeutic antibiotic levels at the time when microbial contamination is most likely to occur.[8] Prophylaxis is characterized by the use of broad-spectrum antibiotics (e.g., ampicillin, amoxicilline, cephalosporin, or combination of antibiotics) effective against the microorganism most likely to cause infections. The routine use of antibiotic prophylaxis varies greatly around the world.[7] As episiotomy is anatomically similar to a second-degree perineal laceration, the routine use of prophylactic antibiotics is not warranted in the case of the first- and second-degree perineal tears.[3–5] More importantly, the routine use of antibiotics increases the risk of antibiotic resistance among microorganisms. In the present study, we, thus, aimed to compare the utility of routine use of antibiotics after episiotomy and also to compare the efficacy of single dose of intravenous antibiotic with a full course of oral antibiotics. A total of 300 cases undergoing normal vaginal delivery with episiotomy were included in the study and they were randomly divided into three groups: Group A received a single prophylactic injectable dose just before the episiotomy, Group B received oral antibiotics for 3 days after the delivery, and Group C no antibiotics. The three groups were then compared for the development of symptoms and signs of infections. The mean age of the study group was 25.09 years with most of the females being primipara (64.7%). No difference was observed among the study groups with respect to age, parity, gestation age, and type of episiotomy wound. Normal episiotomy wound was achieved in 92.7% cases overall. The second- or third-degree perineal tears were inflicted in 2%, 1%, and 1% cases of IV, oral, and no antibiotic groups, respectively (p 0.93). On the follow-up examination during the hospital stay, no difference was observed among IV antibiotic, oral antibiotic, and no antibiotic groups with respect to the mean total leukocyte count on the third post-op day (p 0.69). Similarly, the prevalence of foul-smelling lochia and episiotomy gaping was seen in 3%, 5%, and 5% and 2%, 3%, and 1% cases each among IV antibiotic, oral antibiotic, and no antibiotic groups, respectively (p 0.72, 0.60).

Thus, in the present study, we observed that the universal infection control measures like wearing a mask, hand washing, use of sterile gown and gloves, disinfection of perineal area, and sterilization of instruments which were followed at our institute minimized the risk of episiotomy infection. Therefore, the prophylactic antibiotic has no additional role in the prevention of infection after episiotomy. To the best of our knowledge, no randomized control trial has compared the outcome after routine antibiotic prophylaxis in episiotomy cases as compared to the non-usage of antibiotics. Most of the available literature is on the efficacy of antibiotic prophylaxis for the third- and fourth-degree vaginal tears, which is less frequently encountered complication of episiotomy (when performed correctly). Buppasiri et al.[9] did not find any difference in episiotomy wound complications at the time of discharge of the patient and at 6 weeks after delivery. Liabsuetrakul et al.[10] also did not find a reduced infection rate with the use of prophylactic antibiotics. Bonet et al.[7] in another recent review concluded that evidence is not strong enough to recommend prophylactic antibiotics for the third- or fourth-degree perineal tears after delivery.

Conclusion

We, thus, conclude that the use of routine prophylactic antibiotics is not recommended after episiotomy as it gives no additional benefits; instead, it may have adverse effects on the mother and the baby. Side effects include drug resistance, elimination of beneficial microbes from mother, and the baby and drug reactions. It also increases the cost of healthcare which is not affordable by a large number of our countrymen.

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ORIGINAL ARTICLE

Plasmodium vivax: Chloroquine Drug Resistance in Strains Isolated from Navi Mumbai, Maharashtra, India

Gurjeet Singh[1], Raksha[2], Anant D Urhekar[3]

Abstract

Aim: Malaria imposes a significant public health burden worldwide. Chloroquine (CQ) resistance has been shown to be associated with point mutations in *Plasmodium vivax* chloroquine resistance transporter (Pvcrt) and *Plasmodium vivax* multidrug resistance transporter (Pvmdr1). The present study was carried out to study the association of Pvcrt-o K10 (lysine) insertion and Pvmdr1 Y976 mutations with CQ resistance in Northeast Indian *Plasmodium vivax* isolates.

Materials and methods: The study was conducted in the Parasitology Laboratory at the Department of Microbiology, Mahatma Gandhi Mission Medical College and Hospital, Kamothe, Navi Mumbai, Maharashtra. A total of 22 *Plasmodium vivax* isolates were subjected to the *in vitro* CQ-sensitivity test and the polymerase chain reaction (PCR) test for the Pvmdr1 Y976 and Pvcrt-o K10 (lysine) insertion mutations.

Result: Five isolates of *Plasmodium vivax* were found to be resistant to CQ by the *in vitro* antimalarial drug-sensitivity test, while 17 were found to be CQ sensitive. All the CQ-resistant isolates showed the presence of Pvmdr1 and Pvcrt mutations. CQ-sensitive isolates were negative for these mutations. Strong linkage disequilibrium was observed between the alleles at these two loci [Pvmdr1 Y976 and Pvcrt-o K10 (lysine) insertion].

Conclusion: Our study supports the use of molecular methods for the detection of Pvmdr1 Y976 and Pvcrt-o K10 (lysine) insertion mutations to identify CQ drug resistance in *Plasmodium vivax* and to provide early and proper treatment to patients suffering from vivax malaria.

Keywords: Chloroquine, Polymerase chain reaction, Vivax malaria.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1225

Introduction

Malaria remains one of the leading causes of morbidity and mortality in the world. About 2 million deaths are attributed to malaria each year globally. Approximately 2.48 million malarial cases are reported annually from South Asia, of which 75% of cases are from India alone. Drug resistance is one of the major factors contributing to the resurgence of malaria, especially resistance to the most affordable drugs, such as chloroquine (CQ). Prior to the emergence of resistance, CQ was considered a very effective, safe, and inexpensive antimalarial drug.[1] However, resistance to CQ developed in the early 1960s at two loci, one in Southeast Asia and another in Latin America, and has spread to all areas where malaria is present. It has been estimated that mortality from falciparum malaria increases up to five-fold in areas where resistance to the antimalarial CQ is established.[1]

To reduce the mortality rate, understanding the mechanisms of such resistance and the development of new treatments, including new drugs, are urgently required. Great progress has been made recently in studying the mechanisms of drug action and drug resistance in malaria parasites. These efforts are highlighted by the demonstration of mutations in multiple drug-resistant genes.[2]

Malaria transmission depends on two primary factors, i.e., location of mosquito breeding sites and clustering of human habitations where people serve as reservoirs of parasites for mosquito infection.[3] The resistance to CQ antimalarial drug in *Plasmodium falciparum* is one of the main issues contributing to the global increase in morbidity and mortality. Various methods have been developed to detect the antimalarial drug-resistant pattern of malarial parasites. CQ resistance in *Plasmodium vivax* has also been observed in the Pacific and developing countries including India and nearby the countries but has not yet been reported in Pakistan and Afghanistan. CQ remains effective against *Plasmodium vivax* in India.[4–14] Therefore, the present study was undertaken to detect the CQ antimalarial drug sensitivity in *Plasmodium vivax*.

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How to cite this article: Singh G, Raksha, *et al*. *Plasmodium vivax*: Chloroquine Drug Resistance in Strains Isolated from Navi Mumbai, Maharashtra, India. MGM J Med Sci 2019;6(1):22–24.

Source of support: Nil

Conflict of interest: None

Materials and Methods

Study type: Prospective and analytical study.

Study period: January 2014 to December 2014.

Place of study: The study was conducted in the Parasitology Laboratory at the Department of Microbiology and Central Research Laboratory, Mahatma Gandhi Mission Medical College and Hospital, Kamothe, Navi Mumbai, Maharashtra, India, and Eurofins Genomics, Bengaluru, Karnataka, India.

Ethical committee approval was obtained from MGM Institute of Health Sciences, Navi Mumbai, before conducting the study.

Study participants: A total of 22 patients having confirmed *Plasmodium vivax* malaria were included in this study.

Sample size: Twenty-two *Plasmodium vivax* malaria-positive samples.

Informed written consent was obtained from the patients. For antimalarial drug-sensitivity and molecular analyses, approximately 3 to 5 mL of the blood sample was collected from the patients who tested positive for *Plasmodium vivax*. Blood samples with proper

identification number were stored in cryovials at –20 °C. A total of 22 blood samples positive for *Plasmodium vivax* malaria were included in the study. The DNA extraction of the samples was done by using the DNA extraction kit (Invitrogen, USA) spin column method. Primers were procured from Eurofins Genomics, Bengaluru, Karnataka, India. Primers for nested polymerase chain reaction (PCR) for the detection of a drug-resistant gene in *Plasmodium vivax* were selected from articles.[15,16] For the *Plasmodium vivax* pvmdr1, the forward primer was GCGAACTCGAATAAGT ACTCCCTCTA and the reverse primer was GGCGTA GCTTCCCGTAAATAAA, and for pvcrt-o, the forward primer was CGCTGTCGAAGAGCC and the reverse primer was AGTTTCCCTCTACAC CCG. DNA was extracted from 200 µL of *Plasmodium vivax*-positive blood with the DNA extraction kit (Invitrogen, USA) per the instructions given in the manual and stored at 4 °C until PCR could be completed. Nested PCR amplifications were done using a standard procedure. Known *Plasmodium vivax*-positive and -negative samples were used as controls. DNA bands were visualized and documented by using the gel documentation system (BioEra, India).

The amplified products of nested PCR containing genes pvcrt-o and pvmdr1 were directly subjected to sequencing in both directions using a 3730XL DNA sequencer (Sanger method, big dye terminator chemistry, and Pop 7 polymer gel) from Eurofins IT Solutions India Pvt Ltd. (Bengaluru, India). We found that all isolates showed the mutant allele F976 of codon 976 in pvmdr1 genes and K10 (lysine) insertion in pvcrt-o genes.

Results

Out of 22 samples, five isolates showed resistance to CQ, whereas 17 isolates were sensitive to CQ. All 22 isolates were subjected to the nested PCR test, of these, five showed pvmdr1 and pvcrt-o genes and the rest 17 isolates showed none of the genes. The results showed the same sensitivity using both methods. All 22 amplified products of nested PCR were subjected to purification on gel to proceed for gene sequencing and confirm the mutations. Out of 22 amplified products, only five amplified products showed band on gel and rest 17 showed no band on gel that means only five isolates had pvmdr1 and pvcrt-o genes.

Sequencing analysis of 22 *Plasmodium vivax*-positive strains (five CQ resistance and 17 CQ sensitive) showed that the mutant allele F976 of codon 976 was detected in five samples, whereas the normal allele Y976 of codon 976 of Pvmdr1 was seen in 17 samples. For Pvcrt-o K10 codon, five samples showed K10 (lysine) insertion, whereas 17 samples did not show lysine insertion. Sequencing studies of pvmdr1 and pvcrt-o genes in our study revealed that the mutant F976 gene in codon 976 of pvmdr1 was found in 22.73% samples of CQ resistance (Table 1).

Discussion

The treatment of *Plasmodium vivax* malaria has changed a little in the past 60 years. In most areas, CQ plus primaquine is the first-line treatment, but this status quo is increasingly threatened by the emergence and spread of CQ-resistant *Plasmodium vivax*.[17,18] The extent of this threat is unclear because primaquine has intrinsic blood-stage activity, which could mask low-level CQ resistance, and modest reductions in therapeutic efficacy can be either masked or accentuated by various methodological issues inherent in the study designs applied.

Table 1: Drug-resistant pattern of *Plasmodium vivax*

In vitro sensitivity status CQ resistance (5) and CQ sensitive (17)		
Codon 976 of Pvmdr1 (*n* = 22)	Allele Y976	17
	Allele F976	05
Codon K10 of Pvcrt-o (*n* = 22)	Without K10 insertion	17
	K10 insertion	05

Decreasing antimalarial efficacy is shown by the ability of malaria parasites to grow in the presence of adequate bloodstream drug concentrations. At low levels of resistance, an initial clinical response occurs, often followed by a return of illness caused by recrudescent parasitemia (a late treatment failure or late parasitological failure). The length of the interval from the start of treatment to parasite recrudescence depends on the pharmacology of the initial treatment regimen, the degree of drug resistance, and the level of host immunity.[19] Increasing drug resistance enables parasite growth in high drug concentrations, which slows parasite clearance and shortens the interval to the first recurrence. In studies with a greater risk of recurrence by day 28, illness tends to recur sooner (rs = –0.58). Highly resistant parasites continue to grow despite high blood concentrations of the drug, which results in early treatment failure.

The epicenter for CQ-resistant *Plasmodium vivax* studies has consistently shown high-grade resistance manifested by early clinical deterioration requiring hospitalization, by delayed parasite clearance, and by early recurrent parasitemia.[17,18,20] Several reports of severe and fatal vivax malaria have been published in the past few years.[21,22]

In vitro antimalarial drug-sensitivity testing of CQ was done for 22 isolates of *Plasmodium vivax* using a method similar to the WHO III plate method according to Singh et al.[2] Out of 22 isolates, only five were resistant to CQ, whereas 17 were sensitive.

All 22 isolates were subjected to the nested PCR test and there were the same numbers, i.e., five showed pvmdr1 and pvcrt-o genes and the rest 17 isolates showed no genes. The results showed the same sensitivity using both methods.

All 22 amplified products of nested PCR were subjected to purification on gel to proceed for gene sequencing and confirm the mutations. Out of 22 amplified products, only five amplified products showed band on gel and rest 17 showed no band on gel that means only five isolates had pvmdr1 and pvcrt-o genes. Sequencing analysis of 22 *Plasmodium vivax*-positive strains (five CQ resistance and 17 CQ sensitive) showed that the mutant allele F976 of codon 976 was detected in five samples, whereas the normal allele Y976 of codon 976 of pvmdr1 was seen in 17 samples (Table 1 and Fig. 1). For the pvcrt-o K10 codon, five samples showed K10 (lysine) insertion, whereas 17 samples did not show lysine insertion (Table 1 and Fig. 2). Sequencing studies of pvmdr1 and pvcrt-o genes in our study revealed

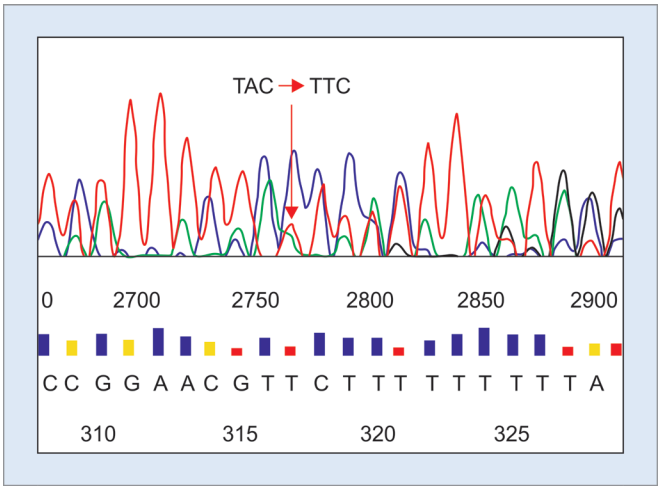

Fig. 1: Mutant allele F976 mutation in pvmdr1 gene

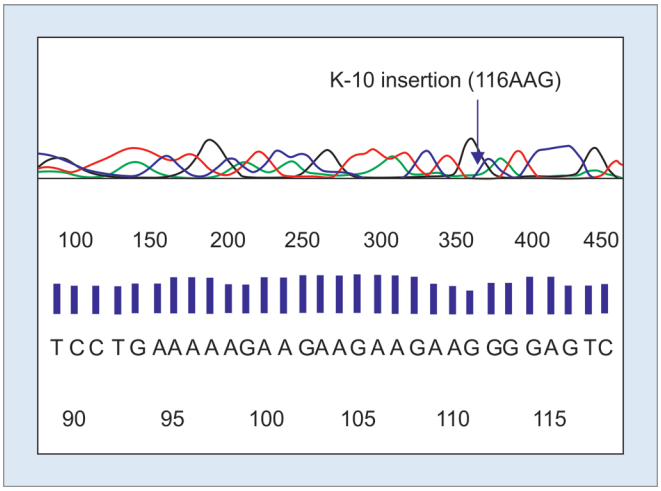

Fig. 2: Mutant allele K10 insertion in pvcrt-o gene

that the mutant F976 gene in the codon 976 of pvmdr1 was found in 22.73% samples of CQ resistance. Mint et al.[15] reported that the majority of the isolates with successful PCR amplification (76/86, i.e., 88%) were characterized to be of the wild-type pvdhfr genotype, while the remaining 10 isolates carried the S58R and S117N double mutations. In their study, all isolates had the wild-type pvdhps genotype SAKAV. For the pvmdr1, 75 of 103 (73%) had the wild-type Y976, and 28 (27%) carried the mutant F976. Most (98%) carried the mutant L1076 codon. Of 105 isolates, 102 (97%) had one copy and 3 (3%) had two copies of the pvmdr1 gene.[15] Chehuan et al.[16] reported that 12 out of 112 isolates were considered resistant to CQ, resulting in 10.7% (IC95%, 5.0–16.4), while 3 out of 47 (6.4%; IC95%, 0.0–12.8) were resistant to mefloquine (MQ). A discrete correlation was observed between IC50s of CQ and MQ (Spearman = 0.294; $p = 0.045$). For the pvdhps gene, a non-synonymous mutation was found at the codon 382 (S C) in 5/8 CQ-sensitive samples and 1/9 CQ-resistant samples ($p = 0.027$). The other molecular markers were not associated with CQ susceptibility.[16]

Conclusion

The gene sequencing study in our work revealed the presence of the mutant allele F976 of the codon 976 of pvmdr1 (*Plasmodium vivax* multidrug-resistant gene) in 5/22 (22.73%) samples and K10 (lysine) insertion in the codon K10 of pvcrt-o (*Plasmodium vivax* CQ-resistant transporter gene) in 5/22 (22.73%) samples.

The occurrence of CQ drug resistance in patients in and around Navi Mumbai could call for reinforced surveillance of drug efficacy. *Plasmodium vivax* CQ resistance may lead to the contribution in the spread of CQ-resistant vivax malaria and the clinical severity of this disease may cause mortality of patients. Our study recommends the use of the molecular technique for early detection of drug resistance and highlights the importance of the CQ-resistant vivax malaria and antimalarial drug-resistant surveillance tests must be conducted on a regular basis to assess the efficacy of the drug.

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ORIGINAL ARTICLE

Knowledge, Attitude, and Practice of Medical Students toward Emergency Contraception

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Abstract

Although India became the first country in the world to introduce National Family Planning policy, still the receptivity of contraception is very poor. Innumerable women irrespective of their literacy status are not aware of emergency contraceptive (EC) methods, which invariably leads to unwanted pregnancies resulting in increasing incidences of abortions. Emergency contraceptive pills (ECPs) can avoid many such pregnancies.

Objective: To assess the knowledge and attitude about ECPs among medical interns and postgraduates (PGs).

Materials and methods: A cross-sectional study was conducted among 174 medical students (interns and PG resident doctors) studying at the tertiary care center in Maharashtra. The period of this study was from January 2018 to June 2018. A pre-structured questionnaire was used to collect the data. The data were analyzed by applying statistical tests.

Results: In this study, a total number of 174 interns and PG resident doctors participated, of which 40% were male and 60% were female. About 80% participants had knowledge of ECPs and 15% had used ECPs. Overall, positive attitude toward ECPs was observed. About 29% participants wanted ECPs to be prescribed by doctors apart from being available over the counter (OTC).

Conclusion: Knowledge of medical students as a healthcare provider was inadequate, so there is need for more emphasis on the emergency contraception (EC) among the young medical graduates and PGs.

Keywords: Abortion, Attitude, Emergency contraception, Knowledge.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1226

Introduction

Emergency contraception (EC) is an effective contraceptive method, which could prevent unwanted pregnancies in many women. In 2002, the Ministry of Health and Family Welfare (MoHFW) in India approved the use of emergency contraceptive pills (ECPs) and they were made available over the counter (OTC) in the year 2005.[1] As no contraceptive method is 100% effective in preventing unwanted pregnancies, this gives women a second chance to prevent pregnancy in the case of failure of conventional methods of contraception, unprotected sex, or in the case of forced sex. ECPs are available in over 140 countries and OTC in 60 countries.[7,8]

The ECP is also known as the morning after pill. The term morning after pill is ambiguous as ECPs can be commenced soon after unprotected sex than morning after, and later can be taken up to 120 hours of unprotected intercourse.[1]

Two main types of EC are available:

- Intrauterine device (IUD/IUCD).
- Pills—estrogen–progesterone pills (Yuzpe regimen), progestin-only pills (levonorgestrel), ulipristal acetate (UPA), and mifepristone.

Copper-T IUD

In 1976, Lippes et al. were the first to use copper-T IUCD as an EC.[9]

This is the only nonhormonal form of EC. According to the guidelines, the IUCD can be inserted within 5 days of unprotected intercourse or it can be inserted up to 5 days after ovulation. The WHO guidelines suggest that copper IUCD can be inserted up to day 12 of the cycle without any restrictions or at any time if the urine pregnancy test is negative. The copper IUD also provides additional contraception for at least 12 years.[1–5,10] From a review of 42 studies, it was revealed that the pregnancy rate was less than 0.1% after insertion of copper IUD for EC.[11]

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How to cite this article: Kadam M, Kadam S, *et al.* Knowledge, Attitude, and Practice of Medical Students toward Emergency Contraception. MGM J Med Sci 2019;6(1):25–28.

Source of support: MGMIHS

Conflict of interest: None

Copper IUD is preferred in women with history of deep venous thrombosis, pulmonary embolism, and coronary events, women on enzyme inducing drugs, or in women with BMI ≥30 kg/m^2. IUD is not suitable for women with pelvic infections and unexplained vaginal bleeding.[11]

Mechanism of Action

Copper-T IUD releases copper ions that have a toxic effect on sperms, which inhibits fertilization. Also, it affects the endometrial receptivity, preventing implantation.

Yuzpe Regimen

Estrogen–progesterone combination pills were first prescribed as ECPs by Dr Albert Yuzpe in the year 1974. This method was effective when used within 72 hours of unprotected sex.[1,6] In this regimen, the birth pills are taken in two doses, 12 hours apart and depending upon the quantity of estrogen in each pill, failure rate ranges from 2.0 to 3.5%.

LNG Pill

This is progesterone-only pill, commonly sold as i-Pill. The initial dose is 0.75 mg, two doses 12 hours apart or 1.5 mg single dose within 72 hours of unprotected sex. WHO trial of the LNG regimen showed no decline in efficacy till day 5.

Mechanism of Action

LNG pill, if taken before luteinizing hormone (LH) surge, prevents and/or delays ovulation. However, it is not effective after LH surge. The LNG pill has no effect on implantation or the endometrium.

UPA

This is the newer and most effective ECP. UPA is widely available in more than 75 countries. It is used in a single dose of 30 mg, within 120 hours of unprotected intercourse.

Mechanism of Action

UPA acts by preventing or delaying ovulation, both before and after LH surge and before LH peak. The failure rate of UPA ranges from 0.9% to 2.1%.[1,5,10]

Side Effects

Decrease or absence of menstrual bleeding, dizziness, hot flushes, etc.

Mifepristone

It is an antiprogesterone drug, commonly used in first trimester abortions. It can be taken in low dose (<25 mg) or mid dose (25–50 mg). It can be consumed within 5 days of unprotected intercourse.

Mechanism of Action

It prevents or delays ovulation and also has an effect on endometrium, if consumed post ovulation.[1,10]

Aims and Objectives

To study the knowledge regarding EC in interns and PGs in tertiary care center in Maharashtra.

Materials and Methods

Type of study: Cross-sectional study
Study period: January 2018 to June 2018
Sample size: 174
Data collection: Using pre-structured questionnaire

Results

A total of 174 participants were included in the study. Majority of them were unmarried (92%). Most of the interns and PG residents were in the age group of 20–25 years, of which 40% were females and 60% were males. From the group of students, 80% students had knowledge of ECPs and they also knew that ECP is also called morning after pill. Students who had used ECPs at some point accounted to about 15%, and 36% students knew the cost of ECPs. When asked about the time during which ECPs are to be used after unprotected sex, 80% suggested up to 72 hours, 9% suggested up to 48 hours, 5% suggested up to 24 hours, 4% suggested up to 12 hours, and 2% did not answer. According to 68% of the students, ECPs should be available OTC, 29% said doctor should prescribe ECPs, and 3% of the students did not know the source of ECPs. About 47% students knew the side effects of ECPs and 53% did not know the side effects. The effectiveness of ECPs was known to 22% only. According to 23% students, ECPs can be a substitute for regular contraception, whereas, for 61%, it cannot be a substitute, and 16% had no idea (Tables 1 to 8).

Various reasons for using ECPs was given as split condom by 64%, after missing oral contraceptive pills by 21%, after sexual assault by 34%, and think if conventional contraception has not been used (more than one options were marked) by 13%. When asked about the attitude toward premarital sex, 43% of the students do not agree with premarital sex, 36% accept premarital sex if one has a boyfriend/girlfriend, and 21% accept if they have plans for marriage in near future. Advertisement on social media was preferred by 84%, 7% differed, and 9% had no opinion.

When physical and mental health after abortion is considered, according to 14% there could be no impact at all, 37% said might

Table 1: Demographic characteristics of study population

Demographic characteristics	*Number (n = 174)*	*Percentage*
Gender		
Male	104	60
Female	70	40
Age		
<25 years	128	74
>25 years	46	26
Marital status		
Married	14	8
Unmarried	160	92

In our study, 92% were unmarried, out of which 15% were in relation and using ECPs and 74% were <25 years

Table 2: Timing of ECPs

Variables	*Percentage*
Within 72 hours	80
Within 48 hours	9
Within 24 hours	5
Within 12 hours	4
Could not answer	2

80% students knew that ECPs had to be taken within 72 hours

Table 3: Used ECPs

Variables	*Percentage*
Used ECPs	
Used	15
Not used	85

85% students had not used emergency contraception

Table 4: Source of ECPs

Variables	*Percentage*
Over the counter	68
Prescribed by doctor	29
Could not answer	3

68% students suggested ECPs should be available OTC

Table 5: Side effects of ECPs

Variables	*Percentage*
Known	47
Not known	53

Side effects were approximately equally known and unknown by students

Table 6: Effectiveness of ECPs

Variables	*Percentage*
Known	22
Not known	78

Most of the students did not know the effectiveness of ECPs

Table 7: Substitute to regular contraception

Variables	*Percentage*
Yes	23
No	61
Do not know	16

Most of the students do not think ECP is a substitute for regular contraception

Table 8: Attitude toward premarital sex

Against premarital sex	*Percentage*
Male	15
Female	28

28% females were against premarital sex

have slight impact, 26% said there could be serious impact, and 23% were uncertain about it. In our study, 15% were not sure whether ECPs protect from sexually transmitted diseases (STD)/reproductive tract infections (RTIs).

Discussion

To study the knowledge, attitude, and practice of EC, many studies have been conducted. Although EC is not recommended as a regular family planning method, it is a useful method after unprotected sexual intercourse to decrease the chances of unwanted pregnancy.[12] In India, despite a National Family Welfare Program and extensive efforts by the government, the rate of unplanned pregnancies and illegal abortions are high. It is estimated that 78% pregnancies are unplanned and 25% are definitely unwanted. EC is also useful in failure of contraception.

In our study, adequate knowledge of contraception was among 80.4% of medical students, which is much more than the study conducted by Gupta et al.[13] among medical students in North West India which was only 45.1%. A similar study was also conducted in Delhi, which showed 50% of students were having adequate knowledge of EC. A similar study conducted in North Gondar by Fantahun et al. showed the level of knowledge of EC to be 75%.[16]

All the indications of ECP usage were accurately known to 86.6% of students, which is similar to study conducted by Gupta et al. in North West India.[13] In our study, 15% students have used ECPs. This is almost the same (17%) as that of the study conducted by Fantahun et al.[16] in North Gondar and Admina et al. in Nigeria. In another study conducted by Renjhen et al. among medical students in Sikkim, 17% students used ECPs.[15,16]

In our study, 80% students knew the correct time of administration of ECPs which was quite high as compared to the study done by Tessema et al.[17] at Jima University, Ethiopia 2013, where only 61.5% students knew the correct time of ECP administration. In our study, 4% students advised to take ECPs as early as possible.

Regarding the availability of ECPs, 29% participants gave the opinion that it should be prescribed by doctors only, which is very less than the findings of the study by Singh et al. who reported that 65% of medical students wants that ECPs should be sold only on prescription of doctors.[14] In our study, 15% participants were not sure whether ECPs protect from STDs/RTIs, which is similar to the study of Dogra and Wankhede who showed 13.9% were not sure whether ECPs protect from STDs/RTIs.[12,17]

In our study, 23% participants agreed that ECPs are not a substitute to regular contraception, which is similar to the study done in Ghana by Baiden et al. in which 26% participants think so.[18] The attitude of medical students toward premarital sex is studied for the first time in our study, which showed that 43% medical students were against premarital sex.

Conclusion

Participants have positive attitude toward EC. The lack of appropriate knowledge of EC among medical students is a cause of concern. It should alarm the medical teaching system as EC is the only method that can be used to prevent pregnancy after unprotected sex or contraceptive failure. There is more need for awareness of EC.

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ORIGINAL ARTICLE

Enhancement of Scientific Writing Skills of Medical Teachers through Scientific Writing Workshop

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Abstract

Introduction: The twelfth 5-year plan put special emphasis on enhancing the quality research in the health sector. Postgraduate doctors and faculties face difficulties in scientific writing and research methodology. There are many books and journals which help in learning the scientific writing process; however, workshops for writing and peer support are reported as the most successful strategies. Therefore, the present study was conducted with the objectives (1) to assess the learning expectations of medical teachers regarding scientific writing and (2) to assess the increase in awareness related to different issues of scientific writing among participants through workshop.

Materials and methods: A 3-day workshop on scientific writing was conducted from 1st to 3rd December 2014 in SMS Medical College of Jaipur. Out of 30 faculty members, 27 participated in the workshop. All participants' knowledge was evaluated using a question-based pretest performa that was repeated at the end of the training. A reaction form was also filled by participants at the end of the workshop.

Results and conclusion: There is a statistically significant improvement of 2.76 in the score of posttest evaluation (CI is 2.29–3.21). Approximately, 93% of participants revealed that they learned about how to write a simple and effective article, make small but effective sentences, and avoid verbatim and difficult words. Almost two-third of the participants gave feedback on their learning about measurable objectives and 45% of respondents gave feedback about learning of the Vancouver and Harvard method for bibliography. Almost 70% of respondents felt confident enough to conduct such workshops on their own level. All respondents (100%) gave the feedback that they learned about avoidance of repetition and irrelevant things and the importance of "revise, revise, and revise." Do's and Don'ts in writing were the most interesting part for all the participants. All the respondents were concerned to learn more about sample size calculations and 93% of respondents were interested in detailed statistical analysis. The study concluded that workshops on scientific writing skills are enormously helpful and should be organized more frequently.

Keywords: Medical teachers, Scientific writing, Workshop.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1227

Introduction

The twelfth 5-year plan put special emphasis on enhancing the quality research in the health sector. Ideally, 2% of the total budget of health should be spent on research. Tertiary care centers particularly teaching institutes have more responsibility for research activities. It was observed during annual plan presentations of proposed research work under the Research Review Board that our postgraduate doctors and faculties face difficulties in scientific writing and research methodology. Medical Council of India (MCI) has made publications mandatory for the promotion of faculties. This has further accentuated the need for conducting research with appropriate methods and writing work in a scientific manner. This would enhance the probability of publication of our research work.

Although educational innovations in medical education are increasing in number, many educators do not submit their ideas for the publication.[1] Also, there exists a substantial amount of unpublished work due to lack of proper writing skills. Perceived obstacles to writing include time constraints, anxiety, lack of confidence, and procrastination.[2–5]

There are many books and journals which help in learning of scientific writing process. Writing workshops[6–8] and peer support[9] are reported as the most successful strategies. Hence, in light of this background, the present study was conducted to assess learning expectations of medical teachers regarding scientific writing and to assess the increase in awareness related to different issues of scientific writing among participants through workshop.

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How to cite this article: Rathore M, Kashyap A, *et al.* Enhancement of Scientific Writing Skills of Medical Teachers through Scientific Writing Workshop. MGM J Med Sci 2019;6(1):29–31.

Source of support: Nil

Conflict of interest: None

Materials and Methods

Framework of the Workshop

A 3-day workshop on scientific writing was conducted from 1st to 3rd December 2014 in SMS Medical College of Jaipur after getting the approval from the Principal and Controller. It was an initiative of the Department of Community Medicine and Research Review Board in collaboration with the Department of Community Health Administration, National Institute of Health and Family Welfare. Six departments out of 18 were randomly selected. Five faculties from each selected department were enrolled to participate in the workshop. Twenty-seven faculty members participated in the workshop. The facilitators for the workshop were well versed with the scientific writing as well as had vast experience of training.

Experts from community health administration, epidemiology, and executive editor of a publication house were involved in facilitating the workshop.

Interactive sessions, powerpoint presentations, group activities, case studies, and handouts and exercises were used. Topics covered in the workshop were concept introduction, method, and discussion (IMRaD) of IMRaD (lecture–demonstrations, L/D, and exercise), writing introduction, style of writing, how to write discussion and conclusion, how to write results, validity of data and interpretation of statistical tests, group work on methodology and results, communication with journal editor for publication, methodology, writing an abstract, ethical issues in research/scientific writing, writing titles, and references. Lecture–demonstrations and exercises were extensively used to introduce the above-mentioned topics.[10–12]

Expectations of trainees were asked at the beginning of the workshop by a course team. Participants' knowledge was assessed using a questionnaire at the beginning as well as at the end of the workshop. This questionnaire has 10 questions on various aspects of scientific writing and research methodology. It was prepared by a team of three faculties at the Department of Community Medicine, SMS Medical College. A reaction form was also filled by participants at the end of the workshop.

Results

The mean score of pretest evaluation was 4.85 (±1.19) and the mean posttest score was 7.61 (±1.25). There was a statistically significant improvement posttest score by 2.76 (confidence interval 2.29–3.21, paired *t* test = 12.325 at df 26 $p < 0.001$) (Table 1). Postworkshop feedback showed impressive results (Table 2).

Twenty-five (93%) participants said that they improved skills to write a simple and effective article, make small but effective sentences, and avoid verbatim and difficult words. Seventeen (63%) participants stated that they gained clarity of ideas about research, and 12 (45%) respondents reported that they learned the Vancouver and Harvard method for bibliography. Further, 19 (70%) respondents felt confident enough to conduct such workshops on their own level, whereas 8 of them (3%) told that they require assistance. All respondents stated that they learned about avoidance of repetition, importance of "revise, revise, and revise," and "Do's and Don'ts" in writing papers (Table 2).

Table 1: Evaluation of pre- and posttest scores of participants

S. no.	*Pretest score*	*Posttest score*	*Level of significance*
1	5	6.5	Average improvement of score = 2.76 (confidence interval = 2.29–3.21)
2	4.5	6	Paired *t* test statistic = 12.325
3	4	6.5	df = 26
4	5	8	$p < 0.0001$
5	5	8	
6	6.5	10	
7	5	8	
8	6	8	
9	5.5	9	
10	4	6.5	
11	3.5	6	
12	5	6	
13	4	5.5	
14	4.5	7.5	
15	4	6	
16	3	6.5	
17	4	7.5	
18	2	7	
19	7	8	
20	5	9	
21	6	8	
22	4	10	
23	6	8.5	
24	4	8	
25	5.5	7.5	
26	6	9	
27	7	9	
Mean score (SD)	4.85 (1.19)	7.61 (1.25)	

Discussion

The Indiana University School of Medicine conducted an in-depth 2-day workshop plus individual tutorials designed to enhance faculty academic writing skills, the majority (87%) of faculties perceived that there was a significant improvement in the confidence level after completing the workshop. In our study also, approximately, 70% of the participants agreed that they are confident enough to conduct such workshops on their own level after completing this workshop.[10–12]

In a study by Bydder et al.,[6] an improvement in the required writing skills and understanding the structure of scientific articles were observed. Participants found the workshop useful and recommended more of these workshops in the future. Half of the participants felt that the workshop has motivated them to publish. Similarly, Guilford[13] revealed a dramatic increase in understanding of publishing process and peer review. Students strongly agreed that the quality of their papers would improve as they have come to know how to write a manuscript for publication in a professional "style." However, very few students agreed that they have acquired the desired skills for writing a scientific article, the majority said, they need more practice to change the way they write in the future. All students felt that the experience was helpful and practical. Ninety-one percent of students responded favorably to peer review. Seventeen percent of students indicated that writing the paper and engaging in peer review were their favorite aspects of the course. In contrast, when the term paper approach was used, none of the students listed the writing of the paper as their favorite aspect of the course.

Conclusion and Recommendations

The study concluded that workshops on scientific writing skills are enormously helpful to increase writing skills of the medical professionals. So these types of workshops should be organized more frequently. This may lead to an increase in the number of future publications. Along with this, unpublished work may get an opportunity to get recognition by overcoming the obstacles of lack of enough skills.

Table 2: Participants' feedback

S. no.	*Feedback*	*No. of participants (n = 27)*	*%*
1	*Writing skills they learnt*		
a.	How to write simple and effective article. Make small sentences, avoid verbatim, difficult words	25	92.59
b.	Clarity of idea of research, measurable objectives	17	62.96
c.	Repetition has to be avoided/how to avoid irrelevant things	27	100
d.	Writing sequence/flow of writing with taking one idea of research to all the section of article	11	40.74
e.	Do's and Don't of all section of article writing	24	88.88
f.	Vancouver and Harvard method for bibliography	12	44.44
g.	Importance of "revise, revise, and revise" in writing	27	100
2	*Teaching skills learnt*		
a.	Group discussion or interactive sessions	24	88.88
b.	Exercise based hands on practice	19	70.37
c.	Two way of communication	27	100
d.	How to teach difficult topic in a simple manner	9	3.33
3	*What did they like most about the workshop*		
a.	Do's and Don't of each section of writing	27	100
b.	Flow of delivery of concepts and idea	12	44.44
4	*Whether they find themselves confident to conduct such workshop at their level*		
a.	Yes	19	70.37
b.	Need little assistance	8	2.96
5	*Skills they would like to learn more about?*		
a.	Detailed sessions on statistical analysis	25	92.59
b.	Sample size calculation	27	100

Acknowledgment

Financial assistance: Academic Section of SMS Medical College, Jaipur, and by NIHFW, Delhi.

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REVIEW ARTICLE

Genetic Race: Prevalence of Diseases and Guidelines for Prevention

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Abstract

Background: Endogamy in races and ethnic communities contribute to the prevalence of genetic diseases, especially recessive monogenic disorders. The demographic and epidemiological transition also increases the incidence of noncommunicable diseases in underdeveloped and developing countries, including India.

Observation: Among all, hemoglobinopathies, cystic fibrosis, Tay-Sachs disease (TSD), etc., prevail in populations of Jewish, Mediterranean, African, and Asian descends. The prevention of births with congenital anomalies of genetic etiology is possible through genetic counseling, carrier screening, and prenatal and/or preimplantation genetic diagnosis. Guidelines have been established by reputed organizations such as American College of Obstetrics and Gynecology (ACOG) and American College of Medical Genetics (ACMG) of different countries with a view to protect the genetic information and misuse of diagnostic samples, selection of the appropriate technologies, clinical application and interpretation of the test results, etc.

Conclusion: The integrated approach of community education and counseling in public health assessment can increase awareness about genetic disorders, and also establish an accurate estimate of its prevalence.

Keywords: Genetic counseling, Genetic disorders, Prenatal and preimplantation genetic screening, Prevention of genetic defects, Racial/ethnic origin.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1228

Introduction

Genetic race or ethnicity refers to different factors including ancestry, social identity, phenotype, genetic makeup, and living practices. Racial groups are susceptible to disease burden mainly due to social inequality and health disparities and, thus, intrinsically affected with high morbidity and mortality.[1,2] Ambiguous distinction between race and ethnicity could be resolved by the fact that the terms race, genetic population, ethnicity, ancestry, geographic population, etc. are their interchangeable use in a situation of application in different disciplines. Racial health disparities define population-specific differences in the prevalence of diseases, access to health care and outcomes, and life expectancy. Racial health inequity is mainly associated with socioeconomic status, education, gender, and access to medical care, diagnosis, and treatment.[3,4] The race of understanding the race has started with scientific basis of inheritance and technological challenges for investigation, which is concentrated on deciphering the genetic patterns in some races or ethnic groups. The modern-day admixture of racial groups is creating complexities in self-identification to one ancestry, biological research, and public health policies.[5] Biological ramifications and genetic intersection of race have established a database on the genetic predisposition of human diseases in different racial populations of different geographical provinces.[6]

Diseases of racial groups can perpetuate different biological effects that are not predetermined by biology such as lack of basic understanding of life and living, and, thus, adoption of preventive care. Sharing a genetic makeup among individuals from a common ancestry also result in share of certain propensity or resistance to specific diseases. In general, human individuals have 99.9% common genepool, and the difference of 0.1% causes a wide spectrum of variations in both the phenotype and the genotype.[7] In epidemiological risk-calculation of genetic disorders, the race is considered as an adjunct useful tool for the assessment of the variation in disease risks, which considers the genetic ancestry of a population such as Askenazi Jewish (AJ) population. Genetic information is largely determined by inheritance from parents in a 1:1 ratio, which is often confronted by imprinting and incomplete penetration, and thereby causing variation in health-risk. Genetic diseases can be ascribed to a single error in a single gene or more errors in multiple genes wherein environmental and epigenetic factors contribute to significant extents. Globally, over 10,000 human diseases (10/1,000 birth) are monogenic, which could be dominant, recessive, or X-linked (dominant/recessive) for inheritance. Some of the prevalent monogene disorders include thalassemia (most prevalent in populations having Mediterranean ancestry to the point that the disease's name is derived from Greek *thalasson*, "sea"), sickle cell anemia (most prevalent in populations with sub-Saharan African ancestry but also common among Latin-American, Middle Eastern populations as well as those people of South European regions such as Turkey, Greece, and Italy), hemophilia, cystic fibrosis (the most common life-limiting autosomal recessive disease among people of Northern European heritage), Tay-

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How to cite this article: Ganguly BB, Kadam NN. Genetic Race: Prevalence of Diseases and Guidelines for Prevention. MGM J Med Sci 2019;6(1):32–41.

Source of support: MGMIHS

Conflict of interest: None

Sachs disease (TSD) (an autosomal recessive disorder more frequent among AJ than among other Jewish and non-Jewish populations), lactose intolerance (affects over lifetime as many as 25% of Europeans but up to 50–80% of Hispanics, along with AJ, but nearly 100% of Native Americans), fragile X syndrome, Huntington's disease, etc., of which thalassemia stands the most prevalent.[8] Multifactorial polygenic diseases differ in frequency between different populations and are highly complex due to the interaction of both genetic and environmental factors. Races are categorized as low or high risk for some polygene disorders depending on their exposure to certain risk factors. Beyond genetic factors, socioeconomic culture as well as the past and present environmental exposures influence a population's risk for specific diseases.

The present review article has a notion to understand the racial prevalence and the frequency of genetic disorders and their prevention. The genetic abnormalities cannot be treated as of now. Therefore, relying on the principle of "prevention is better than cure," recommended guidelines on prenatal (PND) and preimplantation (PGD) genetic diagnosis have been highlighted with a view to reducing the burden of genetic disorders, which further envisage the necessity of regulatory policies and ethical aspects for PND and PGD screening and diagnosis.

Prevalence of Genetic Disorders

Interethnic variation is known for a large number of common conditions, including migration, geographic variation, natural selective pressure, and other environmental variables. Information on the population-specific prevalence of genetic disorders is scanty in most of the developing countries including India. Despite multilingual and multicultural habits, Indian population constitute a distinct cluster having a low level of genetic heterogeneity.[9] A genome-wide search of South Asian populations has demonstrated that 81 different groups of people have descended from a single "founder event," which is more extreme than that of descendants of the AJ and the Finns.[10] Like other developing countries having demographic and epidemiological transitions, India is also experiencing an increase in obesity, diabetes, and heart disease due to the westernization of culture amidst poverty, predominantly in an urban setting. Also, the amalgamation of people from diverse cultural, social, religious, and tribal backgrounds is causing genetic diversity within racial groups.[11] Association of Indian immunological profile, particularly histocompatibility antigens (HLAs), varies significantly for a number of complex medical diseases and confers varying susceptibility to malaria, tuberculosis, HIV, leprosy, and other infectious diseases.[12,13] The incidence of heritable genetic disorders in the Indian subcontinents is largely unknown due to lack of defined and concerted estimation program established by the governmental agencies, and/or underestimated because many of the individuals/families remain undiagnosed and unreported. In India, there is no useful official resource to provide the collective national information on the prevalence and the frequency of genetic disorders for the benefit of medical care and/or offering preventive care of the inherited disorders. The reports available are largely from hospital-based small cohort studies and are insufficient to draw a true figure of the total burden of genetic disorders. Consanguineous and interstate (interreligious) marriages could further influence the incidence of genetic illness.[14]

Congenital anomalies are the leading cause of infant mortality (20%) across the globe.[15] Yearly, an incidence of 7.9 million birth defects is on the rise even in the post-genomic era. Of this, 3.2 million remain disabled for life. Apart from familial inheritance of single-gene and chromosomal disorders and the result of the multifactorial gene-environment interactions, approximately 50% of all anomalies remain undiagnosed.[16] Genetic etiology is characterized as chromosomal, monogenic, and complex polygenic wherein the genetic makeup is determined at conception following the nuclear event of fertilization, and the major developmental defects are manifested in the prenatal environment.

Chromosomal Disorders

Chromosome abnormalities in the form of aneuploidy and/or structural alterations in autosomes and sex chromosomes could be acquired *de novo* or inherited in the form of balanced or unbalanced rearrangements.[17–19] Of all, trisomy 21 is the most common one (1 in 700 births) causing the Down syndrome (DS) followed by trisomies 13 and 18 resulting in Patau and Edward syndromes, respectively (1 in 10,000 births each). Each one is having severe disabilities with specific characteristics and commonness of mental retardation and susceptibility to chronic illness.[20] DS patients can survive till 60 years, but other two die soon after the birth or at the neonatal age.[21,22] Sex chromosomal aneuploidy commonly includes monosomy X and XXY complements in Turner and Klinefelter syndromes, respectively.[17,23] Chromosomal aneuploidies occur due to meiotic nondisjunction during gametogenesis, and often influenced by the advanced maternal age. Unbalanced structural alterations could be clinically serious to fatal for a successful live-born delivery.[17] Moreover, ~95% of conceptuses with aneuploidy or structural alteration do not get embryonic development resulting in the first-trimester miscarriage.

Single-gene Disorders

Single-gene defects are inherited in a dominant or recessive manner from one or both parents. The central dogma of one-gene–one-enzyme hypothesis underlies the majority of single-gene defects such as *PAH* gene on chromosome 12, which encodes phenylalanine hydroxylase (PAH) enzyme and its malfunctioning causes phenylketonurea (PKU). There are many such metabolic disorders caused by single-gene defects, which are heritable and prevalent among ethnic groups such as sickle cell anemia reported prevalent among Africans, Indians, and Mediterranean descents (Table 1).

TSD and many other single-gene disorders are prevalent in AJ population, which are grouped as Jewish genetic diseases (JGD) (Table 2).[8] TSD is caused by an autosomal recessive mutation in the *HEXA* gene on chromosome 15 resulting in deficiency in hexosaminidase protein which affects the fatty buildup of the brain and eventually the nervous system. TSD mainly affects young children leading to progressive neural degeneration followed by death during the first few years of life.

Cystic fibrosis is another highly prominent single-gene defect in different racial groups with high frequency and detection rates (Table 3). The culture of endogamy and relatively low admixture increases the inheritance of heterogeneous recessive mutations. Around 1.5 billion population of South Asia has many small endogamous groups, which has presented 81 unique genetic groups with a higher incidence of recessive diseases.[10] The spectrum and the diversity of genetic diseases in this population may actually require a living lab.

Multifactorial Polygenic Disorders

Approximately, 50% of the birth defects remain undiagnosed and are characterized as multifactorial and polygenic. Polygenic defects

Table 1: Genetic disorders by ethnicity

Genetic diseases	*Ethnic population at highest risk*	*Assay system*
Sickle cell anemia	African, Southeast Asian, Mediterranean, Caribbean	Hb-electrophoresis
α-Thalassemia	African, Chinese, Filipino, Southeast Asian	CBC with indices
β-Thalassemia	African, Indian, Southeast and East Asian, Mediterranean	CBC with indices
TSD	Ashkenazi Jewish Eastern European, French Canadian, Cajun	Hexosaminidase A
Cystic fibrosis	Non-Hispanic Caucasian, Descendents of Ashkenazi Jewish communities (North America)	DNA mutation

Table 2: Carrier frequency of Jewish genetic disorders and detection rates

Disorders	*MIM #*	*Carrier frequency*	*Detection rates*	*Disease incidence*	*Residual risk*
Gaucher disease	230,800	1 in 15	0.95	1 in 900	1 in 281
Cystic fibrosis	219,700	1 in 23	0.94	1 in 2,500–3,000	1 in 368
Familial dysautonomia	223,900	1 in 31	>0.99	1 in 3,600	1 in 3,001
Canavan disease	271,900	1 in 55	>0.97	1 in 6,400	1 in 1,801
TSD	272,800	1 in 27	0.98	1 in 3,000	1 in 1,301
Fanconi anemia group C	227,645	1 in 100	0.99	1 in 32,000	1 in 9,901
Bloom syndrome	210,900	1 in 134	0.99	1 in 40,000	1 in 13,301
Mucolipidosis IV	252,650	1 in 89	0.95	1 in 62,500	1 in 1,761
Niemann-Pick disease A	257,200	1 in 115	0.97	1 in 32,000	1 in 3,801
Maple syrup urine disease	248,600	1 in 97	0.95	1 in 50,000	1 in 1,921
Glycogen storage disease Ia	232,200	1 in 64	0.95	1 in 20,000	1 in 1,261
Dihydrolipoamide dehydrogenase deficiency	248,600	1 in 107	>0.95		1 in 2,121
Familial hyperinsulinism	256,450	1 in 68	0.90	1 in 18,000	1 in 671
Nemaline myopathy	256,030	1 in 168	>0.95	1 in 47,000	1 in 3,341
Usher syndrome type IF	602,083	1 in 147	≥0.75	1 in 80,000	1 in 585
Usher syndrome type III	276,902	1 in 120	>0.95	1 in 45,000	1 in 2,381
Joubert syndrome	213,300	1 in 92	>0.95	1 in 34,000	1 in 2,200
Spinal muscular atrophy	253,300	1 in 40-60	~0.90	1 in 10,000	1 in 600
Walker-Warburg syndrome	236,670	1 in 149	>0.95	1 in 60,500	1 in 2,400

are mainly caused by gene-environment interactions, epigenetic factors, teratogens, use of anti-epileptic drugs, lifestyle, etc. The contribution of such multiple factors is known to cause abnormalities of the brain and spinal cord including anencephaly (lack most of the brain development), spina bifida (incomplete closure of the spinal cord), etc., which are collectively known as neural tube defects (NTDs). India has an estimate of over 30 million individuals with some kind of neurological disorders.[25] Centers for disease control and prevention (CDC) has presented the US National estimates for selected major birth defects (Table 4). NTDs are common with varying clinical significance and sometimes associated with trisomy 18. Micronutrient deficiencies such as folate and iodine have been attributed to various malformations including NTDs and mental retardation. Cleft lip and/or palate, autism-like behavioral disorders, etc., are not caused by a single gene but contributed by a set of genes and environmental factors.[26]

Table 3: Prevalence of cystic fibrosis in different ethnic groups[24]

Racial groups	*Detection rate (%)*	*Carrier risk before testing*	*Carrier risk after –ve test result*
Ashkenazi Jewish	94	1 in 24	1 in 380
Non-Hispanic White	88	1 in 25	1 in 200
Hispanic White	72	1 in 58	1 in 200
African American	64	1 in 61	1 in 170
Asian American	49	1 in 94	1 in 180

Table 4: CDC data on incidence of cephalic defects and syndromes (adjusted for maternal race/ethnicity)[27]

Birth defects	*Incidence per birth*	*Annual incidence*
Anencephaly	1 in 4,859	859
Spina bifida without anencephaly	1 in 2,858	1,460
Encephalocele	1 in 12,235	341
Omphalocele	1 in 5,386	775
Cleft palate	1 in 1,574	2,651
Cleft lip with/without cleft palate	1 in 940	4,437
Transposition of great arteries	1 in 3,333	1,252
Atrioventricular septal defect	1 in 2,122	1,966
Tetralogy of Fallot	1 in 2,518	1,657
Rectal and large intestinal atresia/stenosis	1 in 2,138	1,952
Gastroschisis	1 in 2,229	1,871
Diaphragmatic hernia	1 in 3,836	1,088
Down syndrome[a]	1 in 691	6,037
Patau syndrome[a]	1 in 7,906	528
Edward syndrome[a]	1 in 3,762	1,109

[a] Adjusted for maternal age

Prevention of Genetic Disorders

The catalog of the recessive mutations shall provide useful guidance on how to prevent or control the transmission of genetic disorders through generations. Such cataloging exercise of JGD has established the "Dor Yeshorim" program that follows premarital screening of AJ and Sephardi Jews with a view to preventing transmission of disease-causing mutations, and that has reduced TSD.[8] However, the burden of genetic diseases can be controlled through integrated approach including community education, population screening, genetic counseling and carrier screening, screening of the newborn, and PND and/or PGD. Carrier screening of hemoglobinopathies and glucose-6-phosphate dehydrogenase (G6PD) deficiency has lowered the incidences. However, the magnitude of the genetic disease and the feasibility of prevention program in a cost-effective manner stand important for controlling the alarming public health issue of birth defects. Nevertheless, awareness about the impact of parents' age, sequel of micronutrient deficiencies, vaccination for rubella and other infections, effects of smoking and alcohol intake, syphilis, other sexually transmitted diseases, etc., on the acquisition of new mutations would combat the development of complex polygenic disorders.

Antenatal screening techniques were described in the 19th century for the management of various genetic disorders and congenital malformations. Evidently, PND has become a norm with exponential evolution from biochemical triple screen to quadruple and then first-trimester double screening in the maternal blood along with ultrasound imaging (Tables 5 and 6).

Discovery of cell-free fetal DNA (cff-DNA) in the maternal blood-plasma in 1997 for the screening of trisomies (13, 18, and 21), which is similar to double screening but with a higher level of precision and sensitivity, has gained importance in prenatal management (Table 7). However, guidelines have been set for considering cff-DNA by ACOG and others (Table 8).[28,29] Altogether, reproductive screening relies on an acceptable test protocol, because the aim is not just early diagnosis for prevention and treatment but also to facilitate reproductive decision making.[28,29]

There is tremendous technological and knowledge-based advancement for screening genes and mutations associated with a growing number of diseases. Analytically, microarray-based comparative genomic hybridization (aCGH) and single-nucleotide polymorphism (SNP) array techniques have validated screening of several disease-causing genes. However, microarray techniques are not capable to detect balanced rearrangements. Sanger and next-generation sequencing (NGS) may generate extensive information on the genes and mutations with variants of unknown significance and potential incidental or unsolicited findings and, thus, the clinical use of NGS requires extensive validation. Nevertheless, time-consuming processing and analysis, and high cost debar its use in a clinical setting with the present status.[30,31]

Traditionally, biochemical assessment of hexosaminidase A in serum or leucocytes was followed for the screening of TSD. As the JGDs are caused by a small number of mutations, DNA-based carrier screening has been facilitated by the technological advancement with higher sensitivity and detection rates. Familial dysautonomia was characterized by two mutations in the *IKBKAP* gene with 99% exclusivity of type 1 mutation in Jewish descendents. Likewise, sequencing of the whole genome or exome may facilitate identification of more disease-causing mutations in the present era. Such development has improved the speed and sensitivity of detection rate at much lower cost.[32–34] Of 1,700 mutations identified in the *CFTR* gene, 23 mutations are recommended for carrier screening.[33] Complete analysis of the *CFTR* gene by DNA sequencing is not appropriate for clinical screening as it may yield unsolicited results that are difficult for interpretation. Therefore, ACMG has strictly recommended appropriate validation of the genetic techniques before their incorporation into routine clinical care for screening or testing. Sequencing of the entire *CFTR* gene would be meaningful for cases with a family history of the disorder, males with congenital absence of vas deferens (CAVD) or newborns with a positive screening, and when mutation testing with 23 mutation-panel results appears negative.[35]

Molecular screening of spinal muscular atrophy (SMA) carrier status involves mainly the deletion of exon 7 of the *SMN1* gene in 95% of cases; however, such diagnosis cannot detect heterozygous deletions. Gene dosage analysis has been found to be beneficial to many families with an affected offspring. Quantitative PCR assays have also been implemented for the detection of SMA carriers. Additionally, ~5% of the normal population is reported to carry three copies of *SMN1*, two copies in one chromosome and an *SMN1* deletion on the other resulting in balanced gene dosage. Therefore, the risk of false-negative condition and interpretations of SMA screen shall be managed by experienced genetic professionals and qualified laboratories having validated technologies.[36]

ACMG has established standards of care for preconception and prenatal screening of CF and SMA, and several other recessive conditions for reproductive decision-making for ethnic families.[34,35,37] Screening of an unprecedented quantity of disease-specific genetic variants is possible through preconception screening and also in a time-frame suited for PND.

Myths of PND and PGD

To know the genetic composition of an unborn child for a decision-making, it raises pertinent questions on the benefits and harms of the PND and/or PGD (Table 8). The religious and racial culture might have myths to pose an additional restriction on the approach. The first anxiety of the risk of a miscarriage of invasive sampling (CVS: 1–2% and amniocentesis: 0.5%) is an additional concern of the expecting parents.

To alleviate the fear of inaccuracy of the diagnosis, and selection of the appropriate diagnostic samples and techniques, standard guidelines have been set by the authorized organizations (Table 9).

Conclusion

The prevalence of diseases has a significant link to diet, environmental, and cultural habits. India having one-sixth of the world population, several thousands of endogamous groups indicate a strong potential for recessive diseases and birth defects. Miscarriages with genetic and congenital anomalies contribute to a considerable amount of perinatal and neonatal mortality, on the one hand, and blood-loss and anemia of the mother, on the other hand. Community genetics services for extending counseling, carrier screening and antenatal and PND/PGD could reduce the prevalence of genetic diseases. Clinical diversities of recessive and dominant mutations in the vast human population pose an urge to changing the system of genetic research. Genetic investigation of natural genetic information in human shall replace the manipulation of knock-out genes in experimental primates. Information on the disease prevalence and frequencies in racial and ethnic cultures has governed major attractions in the postgenomic research. Congenital malformations

Table 5: Prenatal screening techniques

Name of the test	*Sample*	*Technique*	*Time of screening*	*Detection*	*Likelihood of diseases*	*Non-/invasive*	*Risk: false +ve/–ve*	*Recommendation*	*Insurance coverage*
NIPT	Cell free fetal DNA (cff-DNA) in maternal plasma	Molecular techniques (a-CGH/ SNP-array/ sequencing)	Any time after 8 weeks	Validated for trisomy 13/18/21 (detection of gene mutations possible)	Down/Patau/Edward syndromes (does not detect NTD)	Noninvasive	Low 99% accurate for Down syndrome	• Integration with first trimester NT screen, and CVS/amniocentesis for confirmation • Recommended only for ≥35 years women and/or having h/o high risk, and not for low-risk women (ACOG 2015)	Most likely not covered by insurance companies. In absence of national coverage determination, varies on gestational age/ resource/countries
Double screen	Maternal blood serum	Biochemical screening	10–13 weeks	hCG, PAPP-A	Down/Edward syndrome, cardiac defects	Noninvasive	High	Integration with first trimester NT screen	May be covered as a part of routine prenatal care
Nuchal translucency (NT) screening	Expecting mother's womb	Ultrasound imaging	10–13 weeks	Collection of fluid beneath the fetal skin in the region of the fetal neck	Trisomy 21/other chromosomal anomalies	Noninvasive	High	Integration with first trimester double screening	Covered as a part of routine prenatal care
Triple screen: normally considered for ≥35 years women or other history and indicative NT/double screen	Maternal blood serum	Biochemical screening	16–18 weeks	AFP, hCG, EU	↑AFP: spina bifida or anencephaly; ↓AFP and abnormal hCG/ EU: Down/Edward syndrome, other chromosomal aberrations	Noninvasive	High	CVS/amniocentesis for confirmation; ultrasound imaging	Most likely covered by insurance companies
Quad screen	Maternal blood serum	Biochemical screening	14–20 weeks	AFP, hCG, EU, inhibin A	Chromosomal aberrations and NTD	Noninvasive	High	CVS/amniocentesis (less accurate than integrated screen for which time has gone)	Most likely covered by insurance companies
Diagnostic testing: in cases with indicative NT/NIPT/double screen, and women of ≥35 years age	CVS	Genetic (tissue culture)	11–13 weeks	Chromosomal aberrations and gene mutations (does not detect NTD and anatomical defects)	Numerical/structural aberrations, and recessive/dominant disorders	Invasive (transvaginal or transabdominal); carries risk of miscarriage	Nil (~99–100% accurate)	NT screen for NTD, genetic counseling for whether it's right for the parents and/or the best next steps if the results are positive	Generally covers CVS but it may not if women is below 35, "low-risk" for certain problems, and/or normal results from first trimester screens
Diagnostic testing: in cases with indicative NT/NIPT/ double/triple/quad screen, and women of ≥35 years age	Amniotic fluid	Genetic (tissue culture)	16–20 weeks	Chromosomal aberrations, gene mutations, and NTD (does not detect anatomical defects)	Numerical/structural aberrations, and recessive/dominant disorders; NTD	Invasive (transabdominal); carries less risk of miscarriage	Nil (~99–100% accurate)	NT screen for NTD genetic counseling for whether it's right for the parents and/or the best next steps if the results are positive	Generally covers CVS but it may not if women is below 35, "low-risk" for certain problems, and/or normal results from first or second trimester screens

AFP, α-fetoprotein; CVS, chorionic villus sampling; EU, unconjugated estriol; hCG, human chorionic gonadotropin; NIPT, noninvasive prenatal testing; NT, nuchal translucency; NTD, neural tube defects (spina bifida)

Table 6: Prenatal genetic testing

Sample	*Tests*	*Detection system*	*Target detection*	*Sensitivity*	*Genome coverage*
CVS/amniotic fluid	Diagnostic: chromosome analysis	Microscopic and imaging of chromosomes	Aneuploidy, structural aberrations	CVS: ~99% Amniotic fluid: >99%	Screens all chromosomes structurally and numerically
CVS/amniotic fluid	Screening: aneuploidies by FISH	Microscopic and imaging of interphase nuclei	Aneuploidy (13/18/21)[a]	~99%	Screens numerical changes of selected chromosomes
CVS/amniotic fluid	Diagnostic in cases with known hereditary history	PCR/chromosome microarray/sequencing	Single gene disorders (recessive/dominant mutations), e.g., thalassemia, Huntington disease	~99%	Targeted genes/copy number variations (CNVs)/uniparental disomy (UPD)
Cff-DNA	NIPT (molecular)	Digital PCR/massively parallel sequencing (MPS)/targeted sequencing/shotgun sequencing	Fetal chromosome dosage/ Single gene disorders	~96–99%	Targeted genes/allelic ratios

[a] Prenatal test of sex chromosomes is restricted in India; Cff-DNA, cell-free fetal DNA; NIPT, noninvasive prenatal testing

Table 7: Cell-free fetal DNA (cff-DNA)-based noninvasive prenatal screening

Features	*Strength*	*Weakness*	*CLIA-approved NIPT tests*
Fetal cell trafficking in pregnancy: 1/10,000 maternal cells; equivalent to 20 cells in 20 mL maternal blood (1 µg DNA in 20 mL blood)	It relies on noninvasive prenatal sampling and does not contact the growing fetus directly	Isolation and characterization of less-frequent fetal cells in maternal blood is beyond the practice in different geographical regions due to lack of resources	Sequenom MaterniT21™ Plus: trisomy 13/16/18/21/22, del (1p/4p/5p/8q/11q/15q/22q/) for microdeletion-syndromes: MPS reports as positive or negative
~6–10% of cff-DNA of total cff-DNA in maternal plasma	It is possible as early as 8–10 weeks	Tests are unable to perform in cases with ≤4% DNA	Ariosa (acquired by Roche in 2015) Diagnostics Harmony™ test: direct DNA analysis for +13/18/21 as risk-score
It is a screening test and should not be considered as diagnostic test, and should not be considered in isolation from other clinical findings	Detects chromosomal aneuploidies with 98–99% sensitivity in high-risk pregnancies (1:200)	Unable to detect all-chromosomal alterations at genomic level	Natera Panorama™ prenatal tests risk-score for trisomies and microdeletions on SNP-technology
This critical distinction shall be explained to every patient carefully. NIPT for +21 is not recommended for low-risk pregnancies	Detects microdeletions/CNVs/ uniparental disomies	Prenatal detention of microdeletion-syndromes are not recommended for NIPT in many countries, and not validated clinically	Illumina (formerly Verinata Health) verify® reports not detected/ detected/suspected +13/18/21 based on MPS
Confirmation by invasive karyotyping is mandated by ACOG and other regulators (false positives occur because of DNA sequenced can represent both maternal and fetal origin, and the fetal fractions derives from placenta as well as the developing fetus)	Detects recessive and dominant mutations with reliable detection limit	The techniques are not yet validated for calling disease-specific mutations and interpreting unwanted mutations	Integrated genetics (LabCorp Specialty Testing Group) InformaSeqSM prenatal test uses illumina platform following similar reporting
Analytic validity, clinical validity and clinical utility shall be established for cff-DNA prenatal test	High sensitivity and specificity of +21 in high-risk singleton pregnancies	Sequencing-based detection of +13/18 and sex chromosomes requires validation on large data	Quest Diagnostics QNatal™ tests aneuploidies
Possible confounders: early gestational age, maternal obesity, multiple pregnancies, placental mosaicism, maternal chromosomal aberration	May require fewer cases of invasive diagnosis and sampling-related miscarriage	Limited data is available on +21 in twin, discordant and multiple pregnancies	Quality control and analytic performance metrics of clinical sequencing is not standardized/regulated by US-FDA
NIPT clinical trials: PreNATUS using SNPs (NCT01545674); NCT01597063 on low-risk women; PEGASUS (NCT01925742)	ACOG (2012) recommended for +21 screening for high-risk singleton pregnancies	May miss Down syndrome and other aneuploidies, though less, due to risk of false negatives of NIPT	In-house laboratory-developed marketable NIPT tests shall meet the general regulatory standards of Clinical Laboratory Improvement Act (CLIA) and approved by CLIA

Cff-DNA, cell-free fetal DNA; NIPT, noninvasive prenatal testing

Table 8: Benefits and harms of prenatal genetic screening and testing

Screening/testing	*Purpose*	*Benefits*	*Harms*
Screening: collects indication of genetic abnormality, but does not confirm its presence	Maternal blood sample is screened to know whether there are chances of developing genetic disorders, like Down syndrome, NTD, and trisomy 18	Noninvasive sample collection does not increase any risk of fetal demise	The results can only indicate whether the baby is at a high/low risk of developing one of these diseases. In other words, the test cannot show accurately whether the baby will definitely acquire this disease or not
	For an indicative screening result, testing shall be recommended for confirmation of the presence of the genetic impairment in the fetus	Likelihood of having a child with chromosomal disorders (Down/Patau/Edwards/Turner/Klinefelter syndrome), thalassemia and cystic fibrosis are detected. Two carrier parents must test the fetus for the expected genetic condition	If the screen only detects indicative genetic markers, this can create a difficult situation to make a decision of approach towards management
			A false-positive screening and/or straightforward approach towards invasive sampling for a diagnosis might affect the normal fetuses negatively
Testing: confirms the presence or absence of a genetic condition with known clinical significance	Couples who have a family history of a genetic disease; have child with genetic defects; have gone through more than one miscarriage in the past or woman is >35 years old shall consider genetic testing	*In utero* rectification of problems based on prior knowledge If this is not possible, the doctors/parents can prepare themselves to manage the condition as soon as the baby is born or for a special delivery	Tests are very expensive and not all insurance companies cover the expenses of prenatal/preimplantations tests
	Women above the age of 35 are recommended for prenatal test because such women are at a high risk of developing genetic disorders in the child	Parents have a choice to terminate the pregnancy if they are not ready to take the responsibility of a child who has genetic abnormalities	Terminating a pregnancy just because there is risk of developing a clinical complication is not acceptable by many. Religious sects believe that only God has the right to take the life of a person (child)
	Testing is accompanied with counseling sessions that help the parents to deal with the situation and to take the final decision	Many parents do not want their baby to suffer from genetic defects and die from extreme sufferings (e.g., TSD). Hence, these parents opt to abort their pregnancy	A positive screen/test result and associated anxiety are the biggest disadvantages
	Using tests to prepare for	Sometimes, screen/test is meant for preparing themselves mentally to take care of the child if he/she is born with abnormalities, but not to abort. Predelivery preparation of medical management for pregnancies with known genetic aberrations can save time and money	A mutation of unknown clinical significance, if detected in an unborn baby, does not surely correlate with disease-development
		This may include: special needs for a delivery or nursing an infant; immediate medical insurance facility; taking out life insurance policies on their child; having a donation fund set up well in advance	Accuracy of the test always raises a question before terminating a life. Also it depends on information about the abnormality is presented to the parents and interpretation of the screening/testing result
		Prenatal diagnosis is helping to-be-parents to abort a child based on any indication of a birth defect, no matter what the severity is	Tailoring of tests can lead to controversy, particularly on gene-markers that indicate the tendency for homosexuality, autism, and violence. This has led to a public outcry to stop the screening and/or research
Direct-to-consumer (DTC) test	Many people who opt for this test even when their doctors do not recommend it. This type of testing is called DTC, i.e., direct-to-consumer genetic testing	People have a direct access to genetic testing along with the privacy of the results	People may misinterpret the results that causes a lot of anxiety which may also lead to improper decisions

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Screening/testing	*Purpose*	*Benefits*	*Harms*
		Better to get these tests done only under the supervision of a health practitioner	The present-day practice of selection of genetically normal/affected embryos for delivery/termination is diminishing the research for a cure of a genetic illness in the scientific communities or funding agencies Social acceptability of abortion, pressure from health insurance companies, medical professionals and government agencies are all possible negative consequences of PND Psychological costs, including anxiety, loss of confidence about the pregnancy, negative attitude towards the baby may be detrimental People may increasingly become intolerant and hostile towards disabled child and their parents

Table 9: Practice guidelines and position statements on cff-DNA-NIPT screening

ACOG and SMFM (2015)	*ASHG/ESHG (2015)*	*NSGC (2013)*	*ACMG (2013)*	*ISPD (2015)*
Risks/benefits/alternatives of various prenatal screening and diagnostic testing methods, including the option of no testing, should be explained to all women	NIPT yield higher sensitivity and specificity for common aneuploidies compared to integrated 1st trimester screening	NSGC supports cff-DNA-screening for woman who wants aneuploidy screening	cff-DNA is a prenatal screening and does not confirm diagnosis	cff-DNA-NIPT leads to high sensitivities and specificities for fetal aneuploidy screening
Conventional screening methods remain the most appropriate choice for first-line screening for most women in the general obstetric population due to limitations of cell-free DNA screening performance, and the limited data on cost-effectiveness in the low-risk obstetric population	A positive NIPT result does not guarantee genetic defects in fetus	The screening shall be considered for high-risk pregnancies	Limitations of cff-DNA-screening for unbalanced chromosomal rearrangements, deletions, duplications exist	Definitive diagnosis of +21 and other aberra shall be achieved through karyotyping in CVS/amniotic fluid
Although any patient may choose cell-free DNA analysis as a screening strategy for common aneuploidies regardless of her risk status, the patient choosing this testing should understand the limitations and benefits of this screening paradigm in the context of alternative screening and diagnostic options	Positive NIPT results shall be confirmed through diagnostic testing before terminating a pregnancy	Cff-DNA-screen shall not be considered as routine first-tier screen for low-risk population	Cff-DNA cannot detect NTD	Maternal age alone is not recommended to assess fetal +21
The cell-free DNA test will screen for only the common trisomies and, if requested, sex chromosome composition	Better NIPT-performance shall be corroborated with better standard of pre- and post-test information and counseling, especially with meaningful options of reproductive choice	The screening shall not be considered as diagnostic	Cff-DNA-NIPT takes longer time than biochemical screening of maternal serum analytes	Integrated NT and serum screening shall be available for considering cff-DNA screening
Given the potential for inaccurate results and to understand the type of trisomy for recurrence-risk counseling, a diagnostic test should be recommended for a patient who has a positive cell-free DNA test result	Information and counseling shall be provided in other linguistics for people with less literate (health) backgrounds	Abnormal screen-results shall be confirmed through conventional diagnostic procedures	Pretest and posttest counseling shall be performed by trained professionals	Quad screen shall be available to women completed 13 weeks 6 days and when cff-DNA-screening can be provided
Parallel or simultaneous testing with multiple screening methodologies for aneuploidy is not cost-effective and should not be performed	Additional information on structural rearrangements (translocations/insertions/deletions) collected during aneuploidy screening shall be made available as a part of information			Markers of 1st and 2nd trimester conventional screening are valid

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ACOG and SMFM (2015)	*ASHG/ESHG (2015)*	*NSGC (2013)*	*ACMG (2013)*	*ISPD (2015)*
Management decisions, including termination of the pregnancy, should not be based on the results of the cell-free DNA screening alone	Clinical relevance and implications of additional findings shall be explained with sufficient details			2nd trimester ultrasound imaging can be a useful adjunct of prenatal screening
Women whose results are not reported, indeterminate, or uninterpretable (a "no call" test result) from cell-free DNA screening should receive further genetic counseling and be offered comprehensive ultrasound evaluation and diagnostic testing because of an increased risk of aneuploidy	Pregnant women's interest on receiving or not receiving specific information on additional findings shall be recorded and act accordingly			Detection of microdeletions and microduplications shall be relevant clinically
Routine cff-DNA screening for microdeletion syndromes should not be performed	Expanded NIPT-based screening and reporting on sex chromosomal anomalies and microdeletions is currently not recommended based on the issues of ethical concerns and counseling-challenges			Rates of false-positive, detection limit, and interpretation of clinical significance of a positive test shall be available
Cff-DNA screening is not recommended for women with multiple gestations				
If a fetal structural anomaly is identified on ultrasound examination, diagnostic testing should be offered rather than cell-free DNA screening				
Patients should be counseled that a negative cell-free DNA test result does not ensure an unaffected pregnancy				
Cell-free DNA screening does not assess risk of fetal anomalies such as NTDs or ventral wall defects; patients who are undergoing cell-free DNA screening should be offered maternal serum alpha-fetoprotein screening or ultrasound evaluation for risk assessment				
Patients may decline all screening or diagnostic testing for aneuploidy				

SMFM, Society for Maternal–Fetal Medicine; ESHG, European Society of Human Genetics; ASHG, American Society of Human Genetics; NSGC, National Society of Genetic Counselors; ISPD, International Society for Prenatal Diagnosis

of genetic and nongenetic etiology can be controlled by the public health and governmental agencies. PND and/or PGD shall be recommended as a routine practice with a view to lower the incidence of heritable genetic disorders. Regulations and policies shall be framed to ensure the judicial utilization of technological advances such as embryonic gene editing by CRISPR/Cas9[38] and to follow the concept of personalized medicine and nutrition.[39]

Acknowledgment

Authors would like to gratefully acknowledge the support provided by The Mahatma Gandhi Mission Trust for generating this review article.

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SHORT COMMUNICATION

A Safe and Easy Method for Hematology Practicals

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Abstract

A new method for hematology practicals, in which a rubber bulb is used to collect blood sample instead of the old method of mouth pipetting, has been described in this paper. The new method is easier and safer. A comparative study of both the methods has been made with a photographic presentation for better understanding and for validating the new method.

Keywords: Glass bottle top dispenser, Hematology practicals, Mouth pipetting, Rubber bulb.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1230

Introduction

In India, hematology practicals are usually undertaken by medical, dental, and paramedical students. Each year, a total of 67,352 medical and 27,000 dental students are taking their admission in medical and dental colleges.[1] At present, mouth pipetting is done in hematology practicals. It involves hemoglobin determination, total red blood cell count (total RBC count), total white blood cell count (total WBC count), and platelet count. It is also a common practice in Primary Health Centers and small private diagnostic laboratories.

Mouth pipetting can cause oral aspiration through pipettes into the mouth and this may lead to infections.[2] Mouth pipetting is hazardous.[3] At present, many practical textbooks still describe mouth pipetting for blood sample collection.[4–10]

Earlier, we have shown the method of dilution for red blood cell count and charging of the Neubauer chamber with a glass capillary.[11] The use of a rubber bulb provides a safe and easy method for the collection of blood, dilution, and charging of the Neubauer chamber in hematology practicals. The collection of blood by mouth pipetting is avoided.

Method

Hemoglobin Pipette (Hb pipette) with Borosil Rubber Bulb

Hb pipette with a rubber tube is used. Only 1.50 cm of the rubber tube is used over the Hb pipette. The remaining part of the rubber tube with a mouthpiece is cut and removed. Over the Hb pipette with a rubber tube, a Borosil rubber bulb (from Borosil bottle dropping with a pipette and RT-30 mL size) is attached. This helps in easy collection of blood from the puncture site over the fingertip. After taking aseptic precautions, finger prick will be done. With minimum pressure on the rubber bulb, blood will be drawn into the pipette by releasing the pressure on the bulb.

The glass bottle top dispenser is used for dilution. The WBC diluting fluid is taken in this bottle (Merck). It is adjusted to dispense 0.4 mL (400 µL) in the 5 mL test tube. Then, 20 µL blood drawn with hemoglobin pipette is added to it. It is mixed by gentle shaking. The dilution is 1:21 after adding 20 µL of blood. The glass capillary tube is used to collect the blood mixed with the diluting fluid to charge the Neubauer chamber. This method is easier in comparison to the conventional method as pipette is not used for charging the chamber. WBC count is done under 10× magnification. The photographs depict about both the conventional method (the usual method) and the new simple method (Figs 1A and B).

The new method can be used for hemoglobin and RBC count determination (4 mL of Hayems diluting fluid; dilution 1:201 after adding 20 µL of blood) and platelet count (dilution 1:201 after adding 20 µL of blood). It is safer and easier for the students to perform hematology practicals.

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How to cite this article: Math MV, Khadkikar RM, *et al*. A Safe and Easy Method for Hematology Practicals. MGM J Med Sci 2019;6(1):48–50.

Source of support: MGMIHS

Conflict of interest: None

Discussion

The new method has been found easier for the students to perform hematology practicals. Since only hemoglobin pipette is used, it is easier to clean as it does not have a bulb. When dispenser bottle

Fig. 1A: Hemoglobin pipette with rubber bulb and glass capillary tube

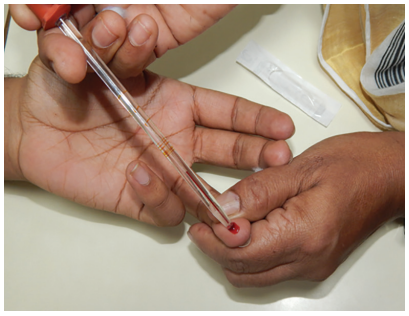
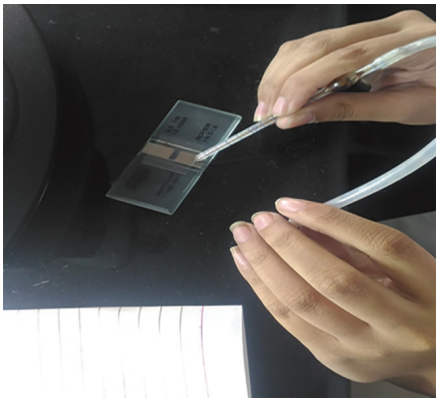
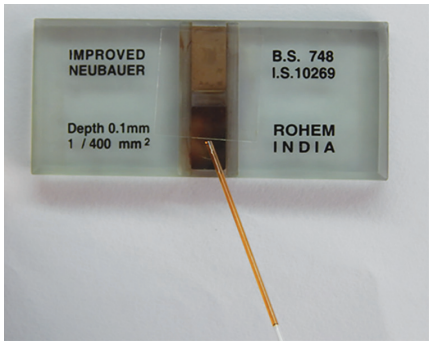
Steps	Conventional method	New modified method
Collection of blood	 Collection of blood sample by mouth pipetting	 Collection of blood sample by using pipette with rubber bulb
Dilution of blood sample	 Mouth pipetting of dilution fluid	 Dilution fluid dispenser
Mixing of blood sample with diluting fluid		
Charging	 Charging with pipette	 Capillary tube

Fig. 1B: Hematology practicals by conventional method and new method

is not available, 1 mL and 5 mL syringes can be used to add the diluting fluid into the test tube to which blood sample can be added.

Conclusion

It is concluded that medical and allied students should use the new method while conducting hematology practicals as it not only gives accurate and authentic results but it is also easier and safer.

Acknowledgments

We are thankful to Mr Suresh Varghese and Mrs Licy Kunjumon MGM Medical College, Navi Mumbai, India, for extending their help in the Physiology Laboratory for conducting the hematology practicals. A special word of appreciation shall go to Mr Shridhar Tambatkar, photographer, for taking photographs.

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CASE REPORT

Intralobar Sequestration: A Case Report

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Abstract

Introduction: Intralobar sequestration (ILS) or intrapulmonary sequestration lacks visceral pleura and is located within a normal lobe and accounts for 75% of bronchopulmonary sequestration (BPS). The majority of ILS is located in the posterior basal segment of the left lobe. Most commonly patients present with signs of infection later in life.

Objective: Chest X-rays and CT thorax is the commonest imaging done to confirm this diagnosis.

Conclusion: Surgical excision is curative extralobular sequestration, and congenital pulmonary airway malformation (CPAM) and bronchopulmonary foregut malformation (BPFM) are other anatomical classifications.

Keywords: Bronchopulmonary, Sequestered lung, Sequestration, Thoracic, Uniportal VATS.

MGM Journal of Medical Sciences (2019): 10.5005/jp-journals-10036-1231

Introduction

Pulmonary sequestration is a rare congenital abnormality that is also known as bronchopulmonary sequestration (BPS). It is the nonfunctioning lung tissue that does not communicate with the tracheobronchial tree and derives its blood supply from systemic circulation. This condition is broadly classified as extralobar and intralobar BPS. We report a case of intralobar sequestration (ILS) left lower lobe treated surgically.

Case Report

A 38-year-old lady presented with recurrent hemoptysis and fever for 6 months. She had been seen and treated for pneumonia previously from multiple GP clinics with oral antibiotics but her symptoms persisted. In view of her persistent symptoms, she was subsequently referred to our hospital for further management. Her full blood count and baseline blood investigations were unremarkable, tuberculosis screening with the Mantoux test and sputum for acid-fast bacilli were negative and her chest X-ray did not show any abnormalities. Due to the persistent hemoptysis, a bronchoscopy was performed and there was a presence of blood clot over the left lower lobe bronchus. Subsequently, a contrast-enhanced CT thorax with supplementary angiogram was done (Fig. 1) in which a left lobe lesion suspicious of arteriovenous malformation or sequestered lung was seen.

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How to cite this article: Arvind M, Sathiamurthy N, *et al*. Intralobar Sequestration: A Case Report. MGM J Med Sci 2019;6(1):51–52.

Source of support: Nil

Conflict of interest: None

In view of her CT findings, an angiography was done which confirmed the diagnosis of intralobar lung sequestration in left lower lobe with an aberrant artery from the thoracic aorta (Fig. 1). The patient underwent uniportal video assisted thoracoscopic (VATS) and left lower lobectomy, as seen in Figure 2, and was discharged home day 5 postsurgery. Histopathology reported sequestered lung with chronic inflammation. On follow-up, she was well and free from her symptom.

Discussion

ILS is the most common form of BPS.[1] It comprises up to 75% of sequestrations, while the remainder 25% are extralobar

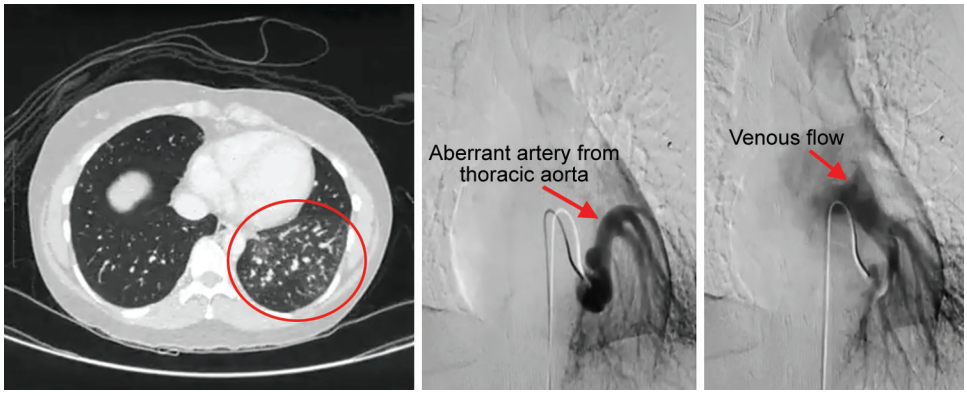

Fig. 1: Axial contrast enchanced computed tomography (CECT) thorax left lobe lesion with angiogram showing aberrant artery

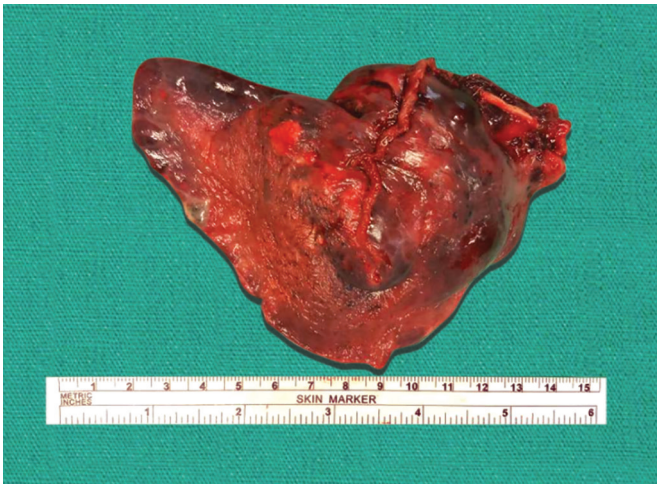

Fig. 2: Medial view of lobectomy specimen with the sequestered lung

Table 1: Differences between intralobar and extralobar pulmonary sequestration[6]

	Intralobar (75%)	*Extralobar (25%)*
Pleural covering	Within normal lung	Own covering
Etiology	Congenital	Congenital
Presentation	Infection	Incidental
Associated anomalies	Rare	Common (65%)
Arterial supply	Systemic	Systemic
Venous drainage	Systemic or pulmonary	Systemic
Macroscopic	Fibrotic with cystic areas	Solid, spongy mass

sequestration (ELS) (Table 1). Genetic predisposition for ILS is equal in both males and females; however, there is a male preponderance for ELS. It is postulated that BPS originates early in the pseudoglandular stage of lung development, during gestation, prior to the separation of the aortic and pulmonary circulations. This would explain its connection to the systemic circulation and the presence of a separate visceral pleura in ELS or the absence in ILS, the occurrence of hybrid lesions with features of BPS and congenital pulmonary airway malformation (CPAM), and the associations with bronchogenic cysts or connections to the foregut, as well as associated anomalies such as congenital diaphragmatic hernia.[2]

The presentation of bronchopulmonary sequestration depends on the type, size, and location of the lesion. Symptomatic lesions can present with respiratory distress in the neonatal period or later in adulthood with lung infection typically fever, cough, hemoptysis, and occasionally pleuritic chest pain. In ELS, diagnosis is often incidental on a chest radiograph. They may present with heart failure due to excessive flow through the aberrant artery or with bleeding.[3] Patients with ELS rarely present with infection.

Both types of BPS can be identified incidentally on a plain chest radiograph. Sequestrations typically appear as a uniformly dense mass within the thoracic cavity and recurrent infections can lead to the development of cystic areas within the mass. The left hemithorax is commonly involved for both ELS and ILS.[4] Computed tomography (CT) is best used to visualize parenchymal abnormalities associated with BPS. They most commonly appear as a solid mass which is homogeneous or heterogeneous with cystic changes. Emphysematous changes at the margin of the lesion are pathognomonic for the diagnosis.[4] However, conventional CT does not routinely demonstrate the aberrant systemic artery.[5] This is better visualized using an MRI which is well suited for the diagnosis of bronchopulmonary sequestration because of its capacity to show the sequestration which may be a well-defined, irregular, or branch-like hyperintense mass with precise anatomic localization as well as to define the size, origin, and course of both the aberrant systemic artery and the venous drainage.[5]

The approach to management depends on whether the patient is symptomatic or asymptomatic. All symptomatic patients should undergo surgical excision which will be curative and has minimal morbidity. In asymptomatic patients, we recommend surgery if the lesion occupies >20% of the lobe or if the mass has multifocal cysts or in the presence of pneumothorax. Low-risk patients are those without the above-mentioned features and can be managed conservatively and planned for elective surgical resection, because they have a moderate risk of developing infection later in life, particularly in ILS. When infection or respiratory symptoms occur, surgery becomes urgent and is associated with a higher risk of complications such as air leak, effusion, and pneumonia, as compared with elective surgery in asymptomatic patients.[1] In our center, we perform uniportal VATS and resection in patients with BPS as demonstrated in this case.

Conclusion

A case of ILS of the lung in left lower lobe in a 38-year-old lady has been presented. Diagnostic methods have been discussed. The patient underwent left lower lobectomy through uniportal VATS. The patient made an uneventful recovery. The prognosis is excellent for patients who undergo elective surgical intervention. Lung parenchyma undergoes compensatory growth and development with normal pulmonary function.

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