ORIGINAL ARTICLE

Prevalence and Clinical Presentation of Primary Immune Deficiency Diseases: Knowledge among Doctors

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ABSTRACT

Background: Primary immunodeficiency diseases (PID) are a group of rare, chronic disorders in which part of the body's complex immune system is either missing or functioning improperly.
Aim: To collectively assess the knowledge of health care providers about Primary immune deficiency diseases.
Methods: A cross-sectional study design was used with a 49-item questionnaire consisting of various close-ended questions on etiology, 26 clinical situations associated with PID and laboratory data presentation regarding PID patients was circulated to be filled by training doctors in various hospitals of Punjab. Responses collected and scores created with the percentage of correct responses employed as a knowledge indicator.
Results: A total of 103 training doctors participated in the study: 66 medical students (64.1%),18 house officers (17.5%), 11 medical officers (10.7%) and 8 post-graduate trainees (7.8%).The mean percentage of the correct responses on etiology expertise of primary immune deficiency was (73.4%) , knowledge regarding clinical symptoms in both adults and children was (66.51%) and (67.9%)respectively while lab analysis cognition was seen in (49.56%) of the respondents.
Conclusion: This study showed that there is insufficient knowledge among training doctors regarding etiology and presentation of primary immune deficiency diseases. Inceptive correction of education skills is the premier strategy by which long term adversity resulting from lack of knowledge can prevent adversities in patients of primary immune deficiency.
Keywords: Primary Immunodeficiency Diseases, Training Doctors and Knowledge.

INTRODUCTION

When part of the immune system is either absent or not functioning properly, it can result in an immune deficiency disease. When the cause of this deficiency is hereditary or genetic, it is called a primary immunodeficiency disease (PID). Researchers have identified more than 150 different kinds of PID with over 120 genes involved.[1]

Furthermore, in some types a single part of the immune system is affected; while in other PIDD one or more components of the immune system are involved. PIDs therefore can be termed as group of heterogeneous disorders with immune system abnormalities characterized by various combinations of recurrent infections, autoimmunity, lymph proliferation, granulomatous process, atopy, and malignancy.[2]

Research shows that the most common manifestation is Immunoglobulin A deficiency which is defined as decreased serum level of IgA in the presence of normal levels of other immunoglobulin isotypes.[3] Most individuals with IgA deficiency are asymptomatic and identified coincidentally showing that even the most common manifestation of PIDD is identified late or circumstantially, signifying that there might be a diagnostic delay in the case of primary immunodeficiency diseases[4]. While many reasons can be sited for this: lack of awareness regarding primary immune deficiency in doctors is one of them[5,6]. This lack of awareness has been studied in various hospital settings proving that in many cases, PIDD are not in the list of differential diagnosis made by doctors for potentially affected patients[7].

In Pakistan a cross sectional study on physicians was done to assess their knowledge base regarding the diagnosis of primary immune deficiency and was concluded that health care knowledge needs to be increased regarding this globally prevalent issue.[8]This diagnostic delay is particularly pertinent in developing countries, as evident from the long interval between the onset of clinical symptoms and the diagnosis.[9] Delay in diagnosis of PID patients is one of the important reasons in occurrence of the permanent sequels and consequent complications, while also leading to increased mortality and morbidity rates**.**[10]

This shows that the general population will suffer if diagnosis of primary immune deficiency is not done earlier. Thus we planned the current project with aim to collectively assess the knowledge of health care providers about Primary immune deficiency diseases. It will be helpful for the health care community to see the gap of clinical knowledge regarding presentation and etiology of primary immune deficiency which is limiting the approach of training doctors to serve these patients.

MATERIAL AND METHODS:

The study was cross sectional and was aimed at reaching hospitals (both private and public) in cities throughout Punjab and was conducted between April and May 2020. Hospitals were taken into consideration based on accessibility. The sample was random sampling and the criteria to include a participant was studying or training to become a medical specialist. The instrument used for data collection was a questionnaire drafted on the basis of a

similar survey done in Iran and Brazil.[11,12] These scores were converted into percentages and three categories were considered i.e. less knowledge (lower than 50%), moderate (50% to 75%) and good knowledge (greater than 75%).

Data analysis: The analysis was carried out using Statistical Package for social Sciences (SPSS) version 25.0. For the analysis, difference was considered statistically significant when $p < 0.05$. Final evaluation done by converting these score to percentages.The mean and standard deviation was measured for scale variables. Students 't' test was used to compare two groups with quantitative variables and Levene's test for equality of variables.

RESULTS

A total of 103 participants with general characteristics (Table 1).

Total Score of less than 25 years of age was 182.32±19.62 and more than 25 years of age was 191.43±19.61 but the difference was not statistically significant (p-value 0.060) as shown in table-2

Total Score of males was 185.13±22.28 and female was 183.43±17.95 but the difference was not statistically significant (p-value 0.668) as shown in table-3

Table-1: General characteristics of enrolled patients (n=103)

Gender	**Frequency**	**%age**
Males	45	44
Females	58	56
Correct response on PID	76	74.3
Knowledge of clinical symptoms in adults	69	66.5
Knowledge of clinical symptoms in children	70	68
Lab analysis cognition	51	49.5
Number of participants	Mean ± S.D	
Less than 25 years	82 ± 2.2	
Above than 25 years	21 ± 4.3	

Table-2: Mean number of participants based on Age of enrolled patients (n=103)

	Less Than 25 Age	Above 25 Age
Total Score	182.32±19.62	191.43±19.61
Score 1	30.17±3.93	31.90±5.41
Score 2	45.04±5.92	46.90±4.22
Score 3	53.27±6.61	56.86±6.95
Score 4	10.44±1.59	11.48±1.37
Score 5	29.48±3.73	30.81±4.54

Table-3: Mean number of participants based on Gender of enrolled patients (n=103)

Gender	Male	Female
Total Score	185.13±22.28	183.43±17.95
Score 1	30.93±5.08	30.21±3.59
Score 2	45.53±5.59	45.33±5.74
Score 3	54.42±7.40	53.67±6.34
Score 4	10.62±1.86	10.67±1.37
Score 5	29.91±3.99	29.62±3.90

DISCUSSION

Primary immune deficiency diseases (PIDDs) are rare, genetic disorders that impair the immune system resulting in patients developing debilitating infections and even fatal cancers.[13] Although there are more than 200 different forms of Primary immune deficiency diseases (PIDDs) affecting thousands of people around the globe making them pervasive and intricate.[14] Consequently, diagnosing these disorders remains a salient step. Making a correct and discrete diagnosis, however is crucial for the selection of appropriate treatment since many cases go undetected due to variable clinical presentations.[15]

Incorrect diagnosis results in lack of appropriate treatment and thus rise in disease associated morbidity and patient outcome. Not only diagnosis but lack of complete lab evaluation presents a significant barrier to the treatment of patients.[16] Timely treatment therefore requires high index of suspicion and specialized testing.

Our study highlights these two major concerns regarding PID disorders, that there is a significant need to increase the knowledge regarding diagnosis and lab evaluation among training doctors, as evident by the results being the lowest in the latter i.e.49.56%. The results showed no significance with the gender of participants however individuals older than 25 years were able to accurately answer a few sections. Limited access to specialized diagnostic discussion and equipment's used around the world, lack of communication amongst health care providers to discuss the various presentations of such patients, paucity of public health methods to assess the impact of PID on community health and eventually poor surveillance systems.[17]

To combat this, educational improvement seems to be a priority and a practical solution for systemic correction of this public health problem. The strategies can include organizing educational courses, researching and publishing educational material while organizing workshops regarding PID to give practical training to doctors. Work is being under the domain of research and comprehension, however significant and grueling efforts still need to be made by the health care system to assist local hospitals, countries and the wider world to prevent the impact imposed by these disorders.[18]

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