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Rare Diseases: Awareness Amongst Pharmacy Students in Karachi, Pakistan

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ABSTRACT

Rare diseases remain habitually chronic, degenerative, progressive, and often life-threatening. 80% of rare diseases have identified inherited origins. Further rare diseases are the consequence of infections (bacterial or viral), allergies and environmental causes, or stand worsening and proliferative. Rare diseases exist characterised by a broad diversity of disorders and symptoms that fluctuate not solitary from disease to disease, but as well from patient to patient suffering from the same disease. Relatively communal indicators can hide fundamental rare diseases, leading to misdiagnosis. Rare disease patients face common problems like delay or lack of access to correct diagnosis, Lack of quality information and scientific knowledge on the disease. Objectives: This study was premeditated to identify the perception of pharmacy students towards rare diseases. Study Design: It stayed cross sectional, qualitative study. Setting: Pharmacy final year students of different public and private sector universities of Karachi were included in the study. Period: Data was collected between January, 2016 to March,2016. Method: Appropriate and Significant information was collected using inquiry form with open-ended questions. 200 final year participants were incorporated in this survey. Results: Response rate of final year students was found (100%, n = 200). Students level of awareness about rare diseases was found (90%, n =181).Conclusion: The results of this study demonstrate that pharmacy students of final year in public and private sector universities of Pakistan are cognizant with particular basic knowledge of rare diseases, but it is a need of time to incorporate more contents of such aspects in program of study.

Keywords: *Rare diseases, degenerative, genetic, scientific knowledge, pharmacy students*

INTRODUCTION

A disease is a damage of health or a circumstance of abnormal functioning. It is a pathological condition of a part, organ, or system of an organism consequential from numerous causes, such as infection, genetic defect, or environmental stress, as well as is characterised over and done with a distinguishable group of signs or symptoms. [1] Rare diseases are one of the most scientifically multifarious health tasks of our time. There are presently 7,000 known rare diseases, half of which affect children. [2] "A rare disease, compatibly point out to as an orphan disease, [3] is a disease that occurs infrequently or seldom in the common residents".[4] Rare syndromes are ordinarily genetic. [5] "Awkwardly, the epidemiological data that are available are inadequate on behalf of furthermost rare diseases to give secure facts on the figure of patients through a precise rare disease. In general people through a rare disease are not listed in records. Numerous rare diseases are summed up and around as "other endocrine and metabolic disorders" and as a concern, with few exemptions, it is difficult to record individuals with a rare disease on a

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countrywide or worldwide foundation, and in a trustworthy, synchronized way".[6] Research publications emphasize rare diseases that are prolonged or incurable, while many temporary medical circumstances are likewise rare diseases. [7] Rare diseases are characterised by a extensive variety of symptoms and signs that differ not solitary from disease to disease but similarly from patient to patient suffering from the similar sickness. [3] Symptoms of numerous rare diseases seem at birth or in childhood, comprising Infantile Spinal Muscular Atrophy, Neurofibromatosis, Osteogenesis Rett syndrome, Imperfecta and most metabolic diseases, such as Hurler, Hunter, Sanfilippo, Mucolipidosis Type II, Krabbe diseases, Chondrodysplasia. Other rare diseases, such as Huntington disease, Spinocerebellar Ataxias, Amyotrophic Lateral Sclerosis, Charcot-Marie-Tooth disease, Kaposi's Sarcoma and thyroid cancer, are specific to adulthood. [4]

Rare diseases similarly fluctuate widely in terms of harshness, but in average the life expectancy of rare disease patients is significantly condensed. The influence on life anticipations how a discrepancy greatly from one disease to the other; some cause death at birth, many are deteriorating or life threatening, although others are compatible with a normal life if make a diagnosis on time and appropriately managed and/or treated.

Rare diseases are responsible for 35 percent of deaths in the first year of life and 30 percent of children through a rare disease will not alive to perceive their fifth birthday. [13] It is also to be underlined that relatively common illnesses can hide fundamental rare diseases, e.g. autism (in Rett syndrome, SotosCerebral Gigantism, Usher syndrome type II, Angelman, Fragile X, Adult Phenylketonuria, Sanfilippo,…) or Epilepsy (Shokeir syndrome, Feigenbaum Kohlschutter Tonz syndrome, Bergeron Richardson syndrome, Dravet syndrome…). For several conditions designated in the historical as clinical ones such as psychological deficiency, autism cerebral palsy, or psychosis, a genetic origin is now suspected or has even now been described. Actually,a rare disease can be masked by a host of further circumstances, which might lead tom is diagnosis. 4 Despite this great diversity, rare diseases have some major common traits. The main characteristics are as shadows:

- Rare diseases remain severe to very severe, chronic, often degenerative and life-threatening;
- The onset of the disease transpires in infantile for 50% of rare diseases;
- Disabling: the quality of life of rare diseases patients is frequently compromised by the dearth or loss of autonomy;
- Highly painful in expressions of psychosocial liability: the suffering of rare disease patients and their relatives is provoked by psychological despair, the nonexistence of therapeutic hope, and the nonappearance of practical provision for daily life;
- Irrepressible diseases, typically deprived of effective treatment. In certain cases, symptoms can be treated to expand superiority of life as well as life expectancy;
- Rare diseases are actual problematic to succeed: families come across massive complications in finding adequate treatment. [4]

Because rare disease patients are a minority, there is a dearth of public attentiveness;these diseases do not epitomize public health precedence, and little research is performed. [4] The rare disease patient is the orphan of health coordination, frequently without diagnosis, without treatment, without research: therefore, without reason to hope. [8] The main objective of Rare Disease Day is to raise awareness surrounded by the common public and decision-makers round rare diseases

and their influence on patients' lives. [9] The first Rare Disease Day was apprehended in European well as Canada in February 2008 to raise awareness for rare diseases. [10,11] It is intended to be observed on the last day of February every year. [12] In Pakistan, Rare Disease Day has been celebrated since 2012. [9] The FDA has approved 400 new managements for rare diseases, and improvements in science and technology, such as personalized medicine, are producing new occasions to improve and expand research into rare diseases and the development of new treatments. Even though this, only 5% of rare diseases currently have a treatment, and there is much more work to be done. [13]

The extensive occurrence of intermarriages in Pakistan means that our population is at an increased threat of rare genetic diseases for instance inborn errors of metabolism (IEMs). In the West the incidence of rare diseases is taken to be 1 in 5,000 births. However in countries wherever intermarriage is common the incidence rises dramatically. For example in Qatar, where the frequency of intermarriages is 40 percent, cases of rare diseases stand at 1 in 1,300. In Pakistan, the frequency of intermarriages is 60 percent that means we have only seen the tip of the iceberg when it comes to rare diseases. At present, there is very little data in Pakistan on the prevalence of rare diseases, but statistics point to a worrying state of affairs. IEMs result in babies being unable to break down carbohydrates, fats or proteins. The condition indications to toxic substances gathering in their bodies that can cause complete developmental arrest and even early death, unless the disease is diagnosed quickly. [14]

The challenges to the progress of diagnosing IEMs in Pakistan such as enormous discrepancies in ethnic people, the high prevalence of malnutrition and infections, the co-existence of very diverse simulations of public health services, unsteady socio-economic and political conditions, and the difficulties in assimilating the numerous stakeholders. Mothers will present to their obstetricians with a history of recurrent miscarriages or loss of child in initial childhood. Exaggerated children ordinarily suffer from common and non-specific symptoms: repeated infections, letdown to increase weight, seizures or convulsions, and if remain untreated with cerebral palsies. Desired outcomes can only be achieved through a close association among paediatricians, chemical pathologists, dieticians as well as metabolic specialists.[15]

METHODS

Study Design: The design of this qualitative study was observational, and cross-sectional.

Settings: This study was designed to collect information about rare diseases perception and awareness in Karachi Pakistan. Several public and private sector pharmacy institutes are located in Karachi. This study was planned with the objective to assess the approaches and facts related to rare diseases in selected cohorts of final year (professional) students of different universities.

Study Duration: Study was conducted between January to March,2016.

Study Population: Population of the study comprised of a cohort, enrolled pharmacy final year students.

Study Tool and Data Collection: Data was collected using well-constructed questionnaire containing 10 open ended questions.

Selection of Sample

Inclusion and Exclusion Criteria: Only final year students of different pharmacy school were incorporated. Students of first to fourth year were not recruited in this study.

Sample Size: Sample size of study comprised of 200 students from final year (professionals)

from different academia. Informed consent of each participant was collected with questionnaire.

Quality Pledge of Data

Study tool was elucidated in detail before application. In order to defend the exactness(accuracy) of outcomes, all questionnaires were filled under direction of the evaluators and reviewed and checked carefully before they collected.

Data Analysis

Data is analyzed and represented in the form of pie charts.

RESULTS

Pharmacy schools students of final year from public and private sector universities were incorporated in this study. Data was collected between January to March, 2016 using well-structured questionnaire with 10 open ended questions. Response rate of final year students were calculated and found 100% (n =200). Students altitude and perception about rare diseases awareness was found to be 90% (n =181) in final year students. 76% of final year students have knowledge that which diseases are included under the head of rare diseases, 27% of final year students have knowledge that rare diseases are more common in which age group, 26% of final year students have knowledge about the risk factors, 57% of final year students aware about the initial symptoms, 62% have knowledge about diagnostic and 58% have knowledge about treatment strategies. 81% of final year students have knowledge about the outcomes if the condition is left untreated. Only 3% of final year students have knowledge about rare disease day and 64% of final year students have knowledge about the reason to celebrate rare disease day.

DISCUSSION

Rare disease affect very low portion of population. Symptom varies disease to disease and in fact patient to patient suffering from the same condition. For the following survey we asked first question from 200 final year students about the term “rare diseases”. 181 final year students have knowledge about rare diseases, whereas 19 final year students have no such education. Next question was asked about diseases included in rare diseases. 153 final year students knew about the diseases or conditions included under the head of rare diseases. While 47 final year students have no such learning. The subsequent question was asked about the age group which is at high risk of rare diseases. 55 final year students have knowledge that children and infants are at higher risk of rare diseases. Besides this, 145 final year students have no such edification. Then the question asked was about the cause of rare diseases. 53 final year students have knowledge about the risk factor of rare diseases. Moreover 147 final year students have no such knowledge. The subsequent question was asked about the initial symptoms to 200 final year students. 115 final year students are aware about the initial signs and symptoms of rare diseases. Despite 85 final year students have no such information. Next question in the subsequent survey was asked about the diagnostic criteria of rare diseases. 123 final year students are well acquaintance about the diagnostic measures of rare diseases. On the other hand 77 final year students have no such awareness. Then the question asked was about treatment strategies of rare diseases. 117 final year students have well knowledge about the treatment plans for the patient of rare diseases. While 83 final year students have no such familiarity. The question then asked in subsequent survey was about the outcomes of the rare diseases in case of delayed diagnosis or if left untreated to 200 final year students. 163 final year students have knowledge that theses

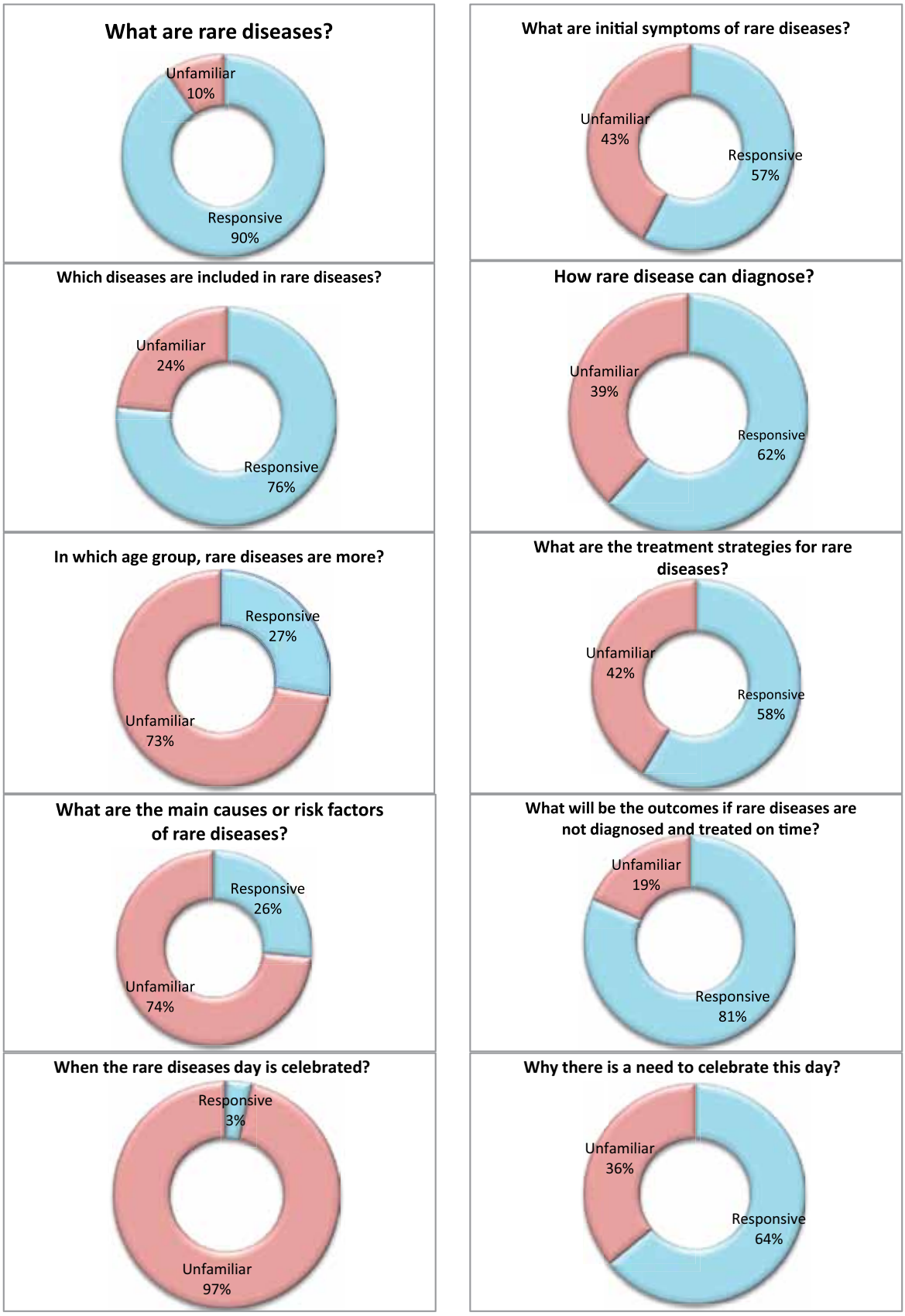

Fig 1: Survey results of rare diseases

rare diseases can lead to death if left untreated. Whereas 37 final year students are unfamiliar to these outcomes. Next question was asked about the rare diseases day to 200 final year students. Only 7 final year students are aware that last day of February is celebrated as rare diseases day in various countries. On the other hand 193 final year students have no such attentiveness. Then in the corresponding survey the last question was asked about the cause to celebrate this day. 128 final year students have awareness that this day is celebrated to increase the level of awareness among general public and health care team to treat the suffering patients and decrease the mortality rate, also to increase the life span and better the quality of life. 72 final year students have no such cognizance about rare disease day.

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