Original Article

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Knowledge level of medical students and physicians about rare diseases in Lima, Peru

Andrea Flores[1], Sarah Burgos[1], Hugo Abarca-Barriga[1,2,*]

[1] Faculty of Human Medicine, Universidad Científica del Sur, Lima, Perú;
[2] Department of Genetic & Inborn Error of Metabolism, Instituto Nacional de Salud del Niño – Breña, Lima, Perú.

SUMMARY Rare diseases (RDs) affect up to 8% of the world's population, and unfortunately, health professionals have a low level of knowledge regarding the impacts of RDs on the social, psychological, and economic spheres of the patients and their families; hence, RD management is inadequate, consistently empirical, and precarious. The objective of this study was to determine the knowledge level of the medical students from a non-state university and physicians from Lima, Peru of RDs through a virtual survey for an analytical cross-sectional study. A total of 338 medical students and 382 physicians were surveyed. Results showed that several of the respondents (68.1% of students and 48.7% of physicians) had heard of the term "rare disease", but only a few stated that they had received any kind of training specific to it. Of the physicians, 46.6% considered that there should be a course about RDs in medical curricula, and more than 60% considered RDs a public health problem. Most respondents prioritized the planning of a higher budget for common diseases and believe it is convenient to allocate a specific fund for RDs. More than half of the participants had a very poor knowledge level. Due to students and physicians' low level of general knowledge of RDs, it is important to raise awareness and improve their education about these pathologies because this will have beneficial effects for RD patient care.

Keywords rare diseases, orphan drugs, medical students, physicians, knowledge

1. Introduction

A rare disease (RD), also known as an infrequent disease or minority disease, is one in which there is a risk of death or chronic disability and low incidence (*1*), with heterogeneous clinical manifestations such as congenital hypotonia, intellectual disability, autism spectrum disorder, congenital anomalies, altered anthropometry, signs of neuroregression, or the appearance of common diseases that do not correspond to the patient's age group (*2*).

The threshold for defining an RD varies among regions. For example, the European Union defines a disease as rare when it affects no more than 1 person per 2,000 people (≈250,000 people) (*3*), whereas, in the United States, a disease is considered rare when it affects less than 200,000 people (≈1 per 1,600) (*1*). Regarding Asia, in Japan, an RD is a disease that affects less than 50,000 people (≈1 per 2,500) (*4*). Between 3.5-8% of the world population has an RD and approximately 7,000 RDs exist, 80% of which are estimated to be of a genetic origin (*5,6*).

In Spain, future healthcare and non-healthcare professionals have a low level of general knowledge of RDs, and none prioritize the allocation of funds to these diseases (*7*); in contrast, another study conducted on Spanish primary care physicians found a high level of interest in prevention, improvement of the family environment, genetic counseling, and medical education (*8*).

In Norway, 24% of the general population showed little interest in prioritizing RDs (*9*). Conversely, Norwegian doctors showed a preference for prioritizing the treatment of common diseases (77.4%) instead of RDs (*10*).

Counselors of the National Institute for Clinical Excellence (NICE) rarely have a favorable predisposition toward the reimbursement of drugs for RDs, although more than half would agree to reimbursement if the use of the drug is well documented or based on disease severity that is severe (*10*).

The lack of interest in recognizing RDs is a result of inadequate instruction and administrative management of people who are unaware of the prevalence, etiology, and manifestations of RDs as well as the forms of adequate and timely management, which directly harm the patients and their families, affecting them socially, psychologically, and economically and portraying them as rarely able to recover or are beyond recovery. Therefore, it is important to demystify what is related to RDs in order to generate universality and equity in this group of pathologies.

The aim of this study was to determine the knowledge level of medical students and physicians of RDs and its relationship with the need to include training, prioritization of resource allocation, and the degree of interest in these pathologies.

2. Materials and Methods

The study design was cross-sectional and analytical. The research was carried out between the months of December 2020 and March 2022, among medical students above the age of 18 from the Universidad Científica del Sur (UCSUR) and physicians from Lima, Peru through a virtual survey using Google Forms®.

A survey was designed based on previous reports (*11-15*) and was validated by 11 experts in the field of RDs with a Cronbach's alpha of 0.92. The survey was anonymous and contained 38 questions comprising of open, dichotomous, and multiple-choice questions. The survey was organized into four groups:

i) The first group comprised eight questions about personal information, including sex, occupation, career year of study, medical specialties, career length (years as a physician and years as specialist), level and sector of healthcare in which they practice (the last four were addressed only to the physicians).

ii) The second group comprised 17 questions that investigated the participants' general knowledge of RDs. Among them, polychotomous questions were included on the correct definition of RDs and orphan drugs (ODs), the most frequent etiology, and the estimated number of RDs. Other questions were about the typical manifestations, the percentage of the population affected by these, and treatments available in Peru and worldwide. In addition, participants were asked to mention three rare diseases. A score (between 4-9 points) was assigned to the 17 questions, which gave a total sum of 100 points.

iii) The third group included seven questions related to academic education and participants' self-perceptions of their competence in the RD field. The participants were asked whether they had ever heard the term "rare disease" or "orphan drug", whether they consider it necessary to include a course on RDs in the MD curriculum, and whether they had ever attended a conference on RDs. They were also asked whether they had ever encountered a patient with an RD and whether they felt prepared to care for/treat a patient with any of these pathologies. These last two questions had the same purpose but were formulated differently for the students and the physicians.

iv) The fourth group of questions referred to organizational issues and the participants' attitude towards RDs. It included 2 dichotomous questions asking whether participants consider RDs to be a public health problem and whether they believe that resources should be allocated for RDs and ODs. In addition, three Likert scale-type questions were added about the importance of RD coverage by insurance systems and the importance of the etiological diagnosis (1 = *very unimportant*, 5 = *very important*); and the support of pharmaceutical laboratories in the diagnosis of RDs (1 = *strongly disagree*, 5 = *strongly agree*). Finally, a question was included only for the physicians, asking whether they consider it important to allocate the same budget for both RDs and common diseases.

Ethics approval and research approval were obtained from the Ethics Committees of the Universidad Científica del Sur and the Instituto Nacional de Salud del Niño de Breña. Informed consent was obtained from all the participants included in this study.

The dependent variable was the knowledge level, with a scale of 0-100 points and was categorized as: A - *excellent* (90-100 points); B - *good* (80-89 points); C - *sufficien*t (70-79 points); D - *poor* (60-69 points); and E - *very poor* (0-59 points) (*16*).

The independent variables were sex, occupation, medical specialty, years of experience as a physician, years of experience as a specialist, career year of study, level and sector of healthcare. The specialties were categorized into six groups: *i*) resident doctors and MD without specialty; *ii*) general surgery; *iii*) pediatrics and neonatology; *iv*) internal medicine and related (cardiology, dermatology, endocrinology, gastroenterology, geriatrics, hematology, infectious diseases, family medicine, intensive and emergency medicine, legal medicine, pneumology, nephrology, oncology, psychiatry, rheumatology, genetics); *v*) other surgical specialties (anesthesiology, gynecology and obstetrics, ophthalmology, orthopedics and traumatology, otorhinolaryngology, urology); and *vi*) others (health administration and management, allergy and immunopathology, pathology and laboratory anatomy, epidemiology, radiology, public health).

The sample size was calculated using the OpenEpi version 3 software (*www.openepi.com*). The medical student population at UCSUR in 2020 was 2,759 (*17*); and the medical professional population in Lima in 2019 was 47,465 physicians (*18*). Since there were no references that analyzed global knowledge, the hypothetical frequency was 50%, and a sample of 338 students and 382 physicians was obtained, with a confidence interval of 95%.

The answers obtained on Google Forms® were automatically transferred to Google Sheets, and the database was then relocated to Microsoft Excel for encoding. The statistical analysis was performed with Stata, with a statistical significance of $p < 0.05$ and a 95% confidence interval. First, the frequencies of each of the questions in both populations were calculated, and the means and standard deviations of the years of experience and scores were obtained. The bivariate analysis was performed using the student's *t*-test and the variance analysis test and the corresponding post-hoc analysis. In addition, robust Poisson regression was used to calculate the crude and adjusted prevalence ratio (PR). To carry out this analysis, the dependent variable (knowledge level) was divided into two groups: deficient (0-69 points) and sufficient (70 points or more), and the PR was calculated for this new variable with the following questions: "Do you consider that RDs are a public health problem?"; "Have you ever attended a course, workshop, or educational congress on RDs?"; and "Do you think the government should designate a fund specifically for RDs and ODs?".

3. Results

3.1. Sociodemographic characteristics of the participants

The survey was answered by a total of 338 students and 382 physicians, the majority of whom were women (in both groups; 61.5% and 53.4%, respectively). As shown in Table 1, 59.4% of the physicians worked in the public sector and 78.3% belonged to the third level of healthcare.

Regarding knowledge level, 75.2% of the medical students and 61.8% of the physicians obtained a score reflecting a very poor level (Table 1). The mean score achieved was 51.3 (SD: 12.6) for the students and 55.6 (SD: 12.6) for the physicians.

3.2. General knowledge of RDs

About 87% of both groups provided the correct definition of RD. However, less than half of the participants knew that the most common cause of RDs is genetic (48.5% of the students, 41.9% of the physicians). Only 9.5% and 6.3% of the students and physicians, respectively, knew the frequency of the prevalence of RDs, while 25.2% of the students and 20.7% of the physicians correctly estimated the number of RDs that exist. Similarly, a low number of participants in both groups knew that RDs mostly affect children (11.8% of the students and 19.4% of the physicians). More than 50% of the students and physicians believed that RDs did not discriminate against countries with low or high resources (Table 2, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

More than 90% of the participants in both groups knew that the treatment of RD is expensive. The majority was not aware of the existence of a law on RDs in Peru (75.1% and 50.5% of the students and physicians, respectively).

Among the typical manifestations of RDs, of the 11 options given, the most recognized by the students were manifestations from birth (congenital anomalies), and in the case of the physicians, it was altered anthropometry (Table 2, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

Table 1. General characteristics of the medical students and physicians

Items	*n* (%)
Students (*n* = 338)	
Gender	
Female	208 (61.5)
Male	130 (38.5)
Year of studies	
First year	35 (10.4)
Second year	35 (10.4)
Third year	35 (10.4)
Fourth year	40 (11.8)
Fifth year	60 (17.8)
Sixth year	88 (26.0)
Seventh year	45 (13.3)
Knowledge level	
Very deficient (0-59 points)	254 (75.2)
Deficient (60-69 points)	57 (16.9)
Sufficient (70-79 points)	23 (6.8)
Good (80-89 points)	3 (0.9)
Excellent (90-100 points)	1 (0.3)
Physicians (*n* = 382)	
Gender	
Female	204 (53.4)
Male	178 (46.6)
Medical specialty	
Residents doctors and MD without specialty	185 (48.4)
Internal medicine and others medical specialties	89 (23.3)
Other surgical specialties	42 (11.0)
General surgeons	25 (6.5)
Pediatric and neonatology	25 (6.5)
Other	16 (4.2)
Healthcare sector	
Public sector	227 (59.4)
Both	90 (23.6)
Private sector	65 (17.0)
Healthcare level	
First level	49 (12.8)
Second level	34 (8.9)
Third level	299 (78.3)
Career length (years of medical doctor)	
0-5	159 (41.6)
6-10	80 (20.9)
11-15	38 (9.9)
16-20	23 (6.0)
More than 20	82 (21.5)
Career length (years of specialist)	
0-5	65 (17.0)
6-10	34 (8.9)
11-15	25 (6.5)
16-20	26 (6.8)
More than 20	46 (12.0)
No specialty	186 (48.7)
Knowledge level	
Very deficient (0-59 points)	236 (61.8)
Deficient (60-69 points)	89 (23.3)
Sufficient (70-79 points)	42 (11.0)
Good (80-89 points)	15 (3.4)
Excellent (90-100 points)	0 (0.0)

Regarding the studies available in Peru on the diagnosis of RDs, neonatal screening and karyotyping were the most remembered methods by both groups. When questioning the participants about the informatic programs that recognize the clinical diagnosis, the best-known platform among the students and physicians was PubMed. On the other hand, Phenomizer, OMIM, FDNA (Face2Gene), and Possum were the least known (Table 2, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

The answers to the question about the most recognized treatments available, in Peru as well as worldwide, were special formulas and gene therapy. An open response was placed on this question where another treatment mentioned was immunoglobulin therapy (Table 2, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

Finally, regarding the open-ended question that asked the participants to mention three RDs, 10% of the participants did not answer or did not know the answer. Among the most reported RDs were Marfan syndrome, Prader Willi syndrome, fragile X syndrome, amyotrophic lateral sclerosis, and phenylketonuria (Table 2, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

Table 3. Frequency of the general opinion on RDs among the medical students and physicians

Characteristics	*n* (%)	
	Students *n* = 338	Physicians *n* = 382
Have you ever heard the term rare diseases or orphan drugs?		
Rare diseases	230 (68.1)	186 (48.7)
Orphan drugs	15 (4.4)	30 (7.9)
Both	45 (13.3)	131 (34.3)
None of the above	48 (14.2)	35 (9.2)
Do you consider that there should be a subject about rare diseases in medical curricula?		
Yes	247 (73.1)	178 (46.6)
No	14 (4.1)	64 (16.8)
Maybe	77 (22.8)	140 (36.7)
Have you ever been on a conference, course, or congress about rare diseases?		
Yes	31 (9.2)	86 (22.5)
No	307 (90.8)	209 (77.5)
Do you consider rare diseases as a public health problem?		
Yes	220 (65.1)	233 (61.0)
No	118 (34.9)	149 (39.0)
Do you consider that the government allocate a specific fund for rare diseases and orphan drugs?		
Yes	312 (92.3)	308 (80.6)
No	26 (7.7)	74 (19.4)
Likert scale questions		
Do you think is important for coverage of rare diseases by insurance systems?		
Very Unimportant	1 (0.3)	7 (1.8)
Unimportant	3 (0.9)	12 (3.1)
Neutral	30 (8.9)	63 (16.5)
Important	76 (22.5)	110 (28.8)
Very Important	228 (67.5)	190 (49.7)
Do you consider important the etiological diagnosis?		
Very Unimportant	3 (0.9)	14 (3.7)
Unimportant	4 (1.2)	24 (6.3)
Neutral	30 (8.9)	80 (20.9)
Important	85 (25.2)	104 (27.2)
Very Important	216 (63.9)	160 (41.9)
Do you agree that pharmaceutical laboratories support the diagnosis of rare diseases?		
Strongly Disagree	12 (3.6)	14 (3.7)
Disagree	46 (13.9)	37 (9.7)
Neutral	62 (18.3)	102 (26.7)
Agree	62 (18.3)	109 (28.5)
Strongly Agree	155 (45.9)	120 (31.4)

3.3. General opinion on RDs and ODs

Although several of the respondents (68.1% of students and 48.7% of physicians) had heard of the term "rare disease", only a small percentage of the doctors and students claimed to have received any kind of education about it. Most of the participants were of the opinion that a course about RDs should be included in medical curricula and more than 60% of them considered RDs to be a public health problem. On the other hand, a considerable number of participants were in favor of allocating a specific fund for RDs and ODs (Table 3).

More than 75% of the students and physicians believed that RD coverage by insurance systems is important. A minority (2.1% of the students and 10% of the physicians) considered that the etiological diagnosis of RD is not important (Table 3). More than 60% of the students agreed that laboratories support the diagnosis of RDs, while a few of them (17.5%) thought the opposite and 18.3% had a neutral stance (Table 3).

The participants were also asked whether they had

Table 3. Frequency of the general opinion on RDs among the medical students and physicians (continued)

Characteristics	*n* (%)	
	Students *n* = 338	Physicians *n* = 382
Only for students		
During your studies have you ever suspected a rare disease in your medical practice?		
Yes	177 (52.4)	
No	79 (23.4)	
Maybe	82 (24.3)	
Do you think that your training gives you the ability to care for a patient with a rare disease in the future?		
Yes	143 (42.3)	
No	195 (57.7)	
Only for physicians		
Have you ever treated a patient with a rare disease?		
Yes		215 (56.3)
No		51 (13.4)
Maybe		116 (30.4)
Do you feel prepared to care for a patient with a rare disease?		
Yes		121 (31.7)
No		88 (23.0)
Maybe		173 (45.3)
Do you think a budget should be assigned for rare diseases?		
It does not matter if you assign a higher budget for one or another		22 (5.8)
Higher Budget for common diseases		236 (61.8)
Higher Budget for rare diseases		30 (7.9)
Same Budget for both		94 (24.6)

ever come across a patient with an RD and whether they felt prepared to care for patients with any of these pathologies. Regarding the former, more than 50% of both the students (52.4%) and physicians (56.3%) reported having known a patient with an RD during their training or professional practice. And regarding the latter, 23% of the physicians considered themselves not trained to care for a patient with an RD, while more than half of the students (57.7%) considered that their training did not give them the skills to care for a patient with an RD in the future (Table 3).

Finally, a question was included on whether the participants considered it important to designate the same budget for both RDs and common diseases, taking into account that the treatment of some RDs is more expensive than that of common diseases, but this was addressed only to the physicians. Most of the physicians (61.8%) believed the largest budget should be allocated to common diseases, and only 7.9% thought that it should be allocated to RDs (Table 3).

3.4. Analysis of the knowledge level compared to previous education, degree of empathy, and general characteristics of the participants

Among the students, it was observed that men had a greater knowledge about RDs in comparison to women (p = 0.02) (Table 4). On the other hand, no significant differences between the sexes regarding the knowledge level were found among the physicians (p = 0.18). The students and physicians who indicated having received some type of training with respect to RDs had better scores on the survey (Table 4).

Regarding the scores obtained by the students in relation to whether they considered RDs to be a public health problem, no significant differences were found (p = 0.83). Conversely, there was a significant difference (p < 0.001) among the physicians, as those who did consider it a public health issue obtained a higher score in the survey (Table 4).

When we asked if they agreed that the government should designate a fund specifically for RDs and ODs, the mean knowledge level was higher in the group that agreed with the question in the students and physicians (Table 4).

3.5. Analysis of the knowledge level regarding the allocation of resources, demographic characteristics, and the participants' self-perceptions about their competencies in RDs

Both the students and physicians who positively answered the question "Have you ever heard the term RD or ODs?" obtained a significantly higher score on the survey (p < 0.001) (Table 5 *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

For the question of whether they thought a course on RDs should be included in the medical curricula, no significant differences (p = 0.56) were observed in the general scores of the physicians. However, among the students, those who disagreed with the need for a course on RDs were the ones who obtained the lowest scores (42.9) (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

Regarding the relationship between the knowledge level and importance of the etiological diagnosis of RDs, no significant differences were found (p = 0.35) between the physicians who did not consider it important and those who believed it to be very important (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

Regarding the relationship between the knowledge level and whether the participants considered that pharmaceutical laboratories should support the diagnosis

Table 4. Analysis between the knowledge level and sociodemographic characteristics, previous training, and level of empathy of the medical students and physicians

Variables	Sample survey					
	Students			Physicians		
	Mean	SD	p	Mean	SD	p
Gender						0.18
Female	50.2	12.4	0.02	55.0	13.1	
Male	53.2	12.8		56.2	12.6	
Have you ever been on a conference, course, or congress about rare diseases?						< 0.001
Yes	57.0	12.1	0.009	61.1	12.7	
No	50.8	12.6		53.9	12.5	
Do you consider rare diseases as a public health problem?						< 0.001
Yes	51.4	13.2	0.83	58.2	13.2	
No	51.1	11.5		51.3	11.1	
Do you consider that the government allocates a specific fund for rare diseases and orphan drugs?						< 0.001
Yes	51.5	12.7	0.50	56.1	12.9	
No	49.7	11.5		53.3	12.5	
Do you have a specialty?						0.05
Yes				56.6	13.4	
No				54.4	12.3	

of RDs, physicians with the highest score in the survey were in favor of pharmaceutical laboratories supporting RDs ($p = 0.003$). Despite this, no significant differences were found among the students ($p = 0.92$) since almost all of them obtained the same score in the knowledge survey regardless of how much they agreed or disagreed (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

The physicians who worked in both public and private sectors had greater knowledge about RDs (59.2 points) in contrast to those who worked only in the public or private sector (54 points) ($p = 0.01$), (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

For the relationship between the healthcare level (I, II, or III) and the score achieved in the RDs survey, no significant differences were found ($p = 0.84$), with a similar score (approximately 55 points) (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

No differences were found regarding the knowledge level of general practitioners and specialists ($p = 0.05$). Regarding the years of experience as a physician and the knowledge level, there was a significant difference ($p = 0.003$) between those who had more than 20 years of experience and those who had between zero and five years of experience (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*). The years of experience as a medical specialist did not affect the knowledge level (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

In reference to the question addressed to the physicians about whether they had ever treated a patient with an RD, a statistically significant difference was found ($p < 0.001$), as those who stated that they had treated a patient with one of these pathologies obtained a higher score on the survey than those who had not treated a patient with an RD. The students were asked whether, at any time during their training, they had suspected any RD in their clinical rotations, and significant differences were found ($p < 0.001$) since those who had commented on it in their pre-professional practices were those with the highest scores (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

We also assessed the relationship between the score achieved and the ability physicians feel to care for a patient with an RD; no significant differences were found ($p < 0.001$) since those who felt qualified had greater knowledge (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

The additional question addressed to only the physicians revealed that those who were indifferent to providing a supporting budget to RDs obtained the lowest scores (47.7 points) (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

When comparing the variable *year of studies* with the score achieved in the survey, the knowledge level increased slightly from the first to sixth year; however, in the seventh year the knowledge level dropped to 51.7 ($p < 0.001$) (Table 5, *http://www.irdrjournal.com/action/getSupplementalData.php?ID=121*).

11% of the students and 25% of the physicians who previously attended training on RDs demonstrated a sufficient knowledge level. Only 6% of the students and 17% of the physicians who considered RDs to be a public health problem had a sufficient knowledge level. In the case of whether the government should allocate a fund specifically for RDs and ODs, only 8% of the students and 3% of the physicians who were in favor of it showed scores indicative of sufficient knowledge (Table 6).

4. Discussion

This research confirms the previous findings of other countries that have shown that both medical students and health professionals lack knowledge of RDs. For example, research carried out in La Rioja, Spain concluded that future healthcare and non-healthcare professionals have a low level of general knowledge of this subject and none of them prioritize the allocation of funds to RDs (*7*). Also, another study conducted in Kazakhstan showed deficient knowledge if the epidemiology of RDs, since only 5% of the physicians had mastered the frequency of prevalence and correctly estimated the number of RDs (*19*). In this research, 92% of the students and 85.6% of the physicians obtained scores indicative of an insufficient or low level of knowledge, as demonstrated in a study carried out in Spain, where the primary health care professionals lack knowledge about RDs and this lack is consciously perceived by physicians (*20*).

The results of our study indicate that, although almost all the participants knew the correct definition of RDs, they tend to underestimate the epidemiological burden of RDs. Additionally, less than 25% of the participants did not know the current number of existing RDs, and very few students knew that RDs develop mainly in the childhood population. Similar findings were reported in China, where doctors did not know how many RDs exist (58.1%), and that the most frequent age of onset is in childhood (30.9%) (*21*).

The results regarding the participants' knowledge level show that the more knowledge the participants have, the more sensitive or empathetic they are to support RDs in terms of diagnosis by pharmaceutical laboratories and coverage of these diseases by insurance systems. This idea was based on a previous study carried out in China among nine RD experts, where all of them expressed concern about the high cost of ODs and the majority of them (seven out of nine participants, 77.8%) supported the creation of a special insurance program for RDs (*22*).

A study carried out in Norway indicated that almost

Table 6. Analysis between insufficient knowledge level and academic education, perception of RDs as a public health problem and the allocation of resources of the medical students and physicians

Items	PR	*p*	95% CI
Have you ever been on a conference, course, or congress about rare diseases?			
Students			
Crude	1.11	0.08	0.95-1.29
Adjusted	1.11	0.21	0.95-1.29
Physicians			
Crude	1.28	< 0.001	1.11-1.48
Adjusted	1.25	0.002	1.08-1.45
Do you consider rare diseases a public health problem?			
Students			
Crude	1.06	0.06	1-1.13
Adjusted	1.06	0.06	1-1.12
Physicians			
Crude	1.19	< 0.001	1.11-1.29
Adjusted	1.17	< 0.001	1.08-1.26
Do you consider that the government allocates a specific fund for rare diseases and orphan drugs?			
Students			
Crude	1.09	0.12	1.06-1.13
Adjusted	1.08	< 0.001	1.04-1.11
Physicians			
Crude	1.08	0.14	0.99-1.18
Adjusted	1.03	0.43	0.95-1.13

The statistical significances are bold. PR: prevalence ratio.

half of the participants (48.3%) believed that funds should be allocated to common diseases, since this group of pathologies is the most prevalent; however, a lower percentage (44.4%) agreed with assigning a small portion of the funds to RDs (*9*). The same occurred in our study: although more than 90% of the participants believed that RDs incur high costs, they prioritized the allocation of a larger budget for common diseases. However, they also believed it is convenient to allocate a specific fund for RDs and ODs.

In Peru, although the subject of genetics is included in the respective curricula of medical careers, it only encompasses general topics and occasionally touches on some genetically-related RDs, but no workshop or course specifically covers the topic. Therefore, the participants in our study believe that the inclusion of RDs in medical study programs is necessary. Similarly, in a previous study conducted among medical students in Poland, almost half of the participants (46.5%) agreed with adding an extra course on RDs in medical curricula (*13*). In this study, more than 50% of the participants considered that their training did not give them the skills to care for a patient with an RD, which coincides with research carried out in Spain, Iran, and Poland that showed that most future healthcare professionals (81%, 73%, and 92%, respectively) did not feel prepared to care for patients with an RD (*20,23,24*).

Another finding was that, although most of the participants perceived RDs as a public health problem, less than half were aware of the existence of a law in Peru related to RDs, which reflects the paltry effect that this rule has on the care for patients with RDs (*25*).

Regarding the studies available in our country, neonatal screening was one of the diagnostic methods for detecting RDs that was most recognized by both groups; however, this technology is limited to a group of only approximately 50 conditions, out of more than 7,000 RDs (*5*), although these pathologies are potentially treatable (*26*).

One point that both the students and physicians agreed on is that there is the same number of RDs in both developed and developing countries. Although the definition of RDs tells us that they are infrequent diseases and that the proportion of people affected could vary according to geographical area, there are no epidemiological studies in that respect. Rather, this assumption is the result of the lack of information, the absence of certain diagnostic methods throughout the world, and an inefficient registration system, as reported in China, where there is still very little documented information on the epidemiology of RDs (*27*).

Since RDs have diverse clinical manifestations and usually take time to be diagnosed, the lack of knowledge resulted in 38% of patients with RDs in Australia having to consult more than six different doctors before receiving the correct diagnosis; 37% believed that their diagnosis was delayed and 27% initially received an incorrect diagnosis (*28*). In Brazil, administrative obstacles for patients and their families caused delays in diagnosis (*29*), which can take between four to six years (*30*), and only 5% of RDs have treatment, as some of these pathologies can incur high costs (*9*).

Therefore, having a greater number of RD specialists would greatly impact the management of RDs. Although in our study, most of the participants considered geneticists to be the specialists to whom a patient should be referred to for a definitive diagnosis of an RD, in our environment the physician-geneticists per million inhabitants is 1,1, which is below the reported figure for Latin American countries (1,9) and worldwide (12,2) (*31-33*).

Some important limitations to note are that this study cannot be generalized to the entire population of medical students or health professionals, since the sample included students from a single private medical university of Lima and only physicians who worked in Lima city, in addition to the fact that the study only represents the opinion of those who agreed to participate, for which further investigations would be necessary. Another limitation of the study is that being a virtual survey, the participants completed the survey without supervision and could have used additional information resources to provide the answers. Nevertheless, this study provides new insights into the knowledge level of RDs in a low- and middle-income country.

Although in 2011, a law was created in Peru that proposed a national plan for comprehensive care and a national registry of patients with RDs as well as budgetary guarantees for treatment, it remains scarcely known. Even the knowledge level of RDs in both future health professionals and physicians is very poor. The United Nations Educational, Scientific and Cultural Organization (UNESCO) tells us that health is one of the fundamental rights of every human being, which includes timely and affordable access to quality health care services (*34*); however, this is not reflected in patients with RDs because they must confront a utilitarian system with many difficulties, which have an impact on various areas of their lives. At a social level, due to the diversity of their condition, an individual patient with an RD experiences different disabilities, which often leads to exclusion with psychological implications. Another important problem is that there is a lack of information both in relation to the disease itself and scientific research, which is reflected in the delays in detection from the moment of the onset of symptoms until receiving the correct diagnosis. There is also a lack of knowledge in the reference centers from which patients can obtain support, which has a serious impact on the economic sphere of the families affected due to the high cost of the few existing medicines (due to the lack of profitability for the pharmaceutical companies) added to the deficit of social benefits and reimbursement due to the deficiency of support from the usual insurance systems. These show the inequity that exists between treating a patient with a common disease and a patient with an RD. Therefore, it is imperative to make medical students aware of RDs and educate them about this regard, since this will have a beneficial effect on the quality of patient care, quality of life, and family environment of those affected by RDs. In addition, there is an urgent need to create a cooperative network with the main hospitals in Lima and other regions, as well as with international institutions to improve the care of these patients.

In conclusion, this study shows the preference of physicians to prioritize the treatment of diseases with the largest/biggest number of patients (*i.e.*, common diseases), even though some of them reserve a small part of resources for RD. On the other hand, the lack of knowledge in both students and physicians causes concern, since most considered RDs as a public health problem, but they did not feel prepared to care for this type of patient, so we consider that the existence of a course that covers the topic of RDs in the curriculum of the medical career is essential. Finally, this study provides new insights into the knowledge level of rare diseases in a low-and middle-income country.

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**Address correspondence to:*
Hugo Hernán Abarca Barriga, Universidad Científica del Sur, Facultad de Ciencias de la Salud, Panamericana Sur Km 19, Villa, Perú.
E-mail: habarca@cientifica.edu.pe