

# Iranian Future Healthcare Professionals' knowledge and opinions about rare diseases: cross-sectional study

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# Abstract

**Background:** Rare diseases are a new global health priority, requiring evidence-based estimates of the global prevalence of diseases to inform public policymakers and provide a serious challenge to the healthcare system that must not be ignored. The purpose of this study is to investigate Iranian Future Healthcare Professionals' knowledge and opinions about rare diseases.

**Results:** A total of 6,838 students responded to the questionnaire. Nursing and medical students had the highest participation. Almost 85% of participants rated their knowledge about rare diseases as poor or insufficient. While nearly 70 percent of participants took courses about rare diseases at university. Finally, 72.7% of future healthcare professionals did not feel ready to take care of a patient with a rare disease.

**Conclusion:** The present study has indicated a gap in Iranian medical students' knowledge on rare diseases. The researchers believe that health science policy makers should make a joint effort to improve knowledge about rare diseases. Including courses with regard to rare diseases would be of benefit to future healthcare professionals.

## Introduction

Rare diseases (RDs) are a new global health priority, requiring evidence-based estimates of the global prevalence of diseases to inform public policymakers. When a disease affects less than one in 2,000 persons, it is considered as rare (1). According to available information, 309 RDs have been diagnosed in Iran so far. Currently, around 3 million people with RDs have been identified in Iran and are receiving a variety of support services. Patients cared for by the Rare Diseases Foundation can benefit from a 50% discount on medicines by going to the Red Crescent. One of the most important missions of the Rare Diseases Foundation is to improve the lives of people with these diseases by raising awareness in the family and the community (2).

Rare diseases provide a serious challenge to the healthcare system that must not be ignored. Patients with RDs typically experience a lack of treatment alternatives, psychological stress and, financial burden in addition to the burden of disease (3). Furthermore, these patients face difficulties in diagnosing their illness, with it frequently taking 5–6 years to achieve an accurate diagnosis, and more than three different physicians are involved in the diagnosis process (4, 5). Besides, due to a lack of knowledge about the rare diseases and the challenges in properly diagnosing them, many patients with RDs would have visited multiple clinics or hospitals but only received symptomatic therapy (6).

One of the most common variables leading to the diagnostic odyssey is a lack of RD awareness and expertise among healthcare professionals (7). Previous research has found that RD knowledge is typically considered as inadequate by physicians and other health care professionals (8–10). The majority of them believed their academic expertise was insufficient and unsuitable for diagnosing RDs in everyday practice (7, 11). While physicians are responsible for managing the care processes of patients with RDs, the role of other health care professionals, including nurses and physiotherapists, is increasing.

As a result, the need to increase knowledge and awareness of RD is evident among health care professionals (9). Unfortunately, no study with a similar purpose was found in Iran and the knowledge of health care students is unknown. Due to the lack of basic information about the medical education curriculum in the field of rare diseases, we cannot predict the strength of the executive curriculum in the country's medical sciences universities.

Due to the importance of the subject, the purpose of this study is to investigate Iranian future healthcare professionals' knowledge and opinions about rare diseases.

## Results

A total of 6,838 people responded to the questionnaire. The majority of the participants were women (60.1%). Nursing and medical students also had the highest participation, 34.2% and 40% respectively. The demographics of the participants can be seen in Table 1. In the process of evaluating the content validity, no items were removed from the research, but changes were made in the form and richness of the words for better understanding. The CVR value was 0.5–0.9 for the items and 0.73 for the whole scale.

Table 1  
Socio-demographic characteristics of students

Characteristics	N (%)
Gender	
Male	2731 (39.9)
Female	4107 (60.1)
Field of study	
Medical students	2732 (40)
Nursing students	2338 (34.2)
Midwifery students	637 (9.3)
Dentistry students	425 (6.2)
Physiotherapy students	111 (1.6)
Pharmacy students	490 (7.2)
Occupational & Speech therapist students	105 (1.5)
Grade	
Bachelor degree	2807 (53.7)
Master degree	357 (5.2)
MD & DDS	3674 (53.7)
Marital status	
Single	6003 (87.8)
Married	835 (12.2)
Age (year)	22.7 ± 3.6
Duration of education (year)	5.6 ± 2.3
Domicile	
Under 10,000 inhabitants	423 (6.2)
10–50,000 inhabitants	1219 (17.8)
51–100,000 inhabitants	921 (13.5)
101–500,000 inhabitants	1271 (18.6)
Over 500,000 inhabitants	3.4 (43.9)
Have you ever met a person suffering from RD?	

Characteristics	N (%)
Yes	3105 (45.4)
No	2154 (31.5)
I do not know	1579 (23.1)
Values are presented as n (%), mean ± standard deviation.	

Table 1. Socio-demographic characteristics of students

Although almost all of the participants had heard of the term “rare disease” (96.4%), they were not sufficiently aware of its prevalence in the community (3.9%). only 8.5% correctly estimated number of RD (Table 2). Students' knowledge about the number of RD patients in the world (6.2%), Asia (3.8%), and Iran (3.8%) was low. A quarter of future healthcare professionals knew the percentage of RDs that were genetic (24.8%). Most participants used the “I do not know” option when responding the questions.

Table 2  
Students' knowledge about rare diseases

Items	N (%)
1. Have you ever heard the term 'rare diseases'?	
Yes	6589 (96.4)
No	249 (3.6)
2. Rare disease is the one that affects less than:	
1 person in 1 000	820 (12)
<b>1 person in 2 000</b>	<b>264 (3.9)</b>
1 person in 3 000	219 (3.2)
1 person in 5 000	327 (4.8)
1 person in 10 000	2200 (32.2)
I do not know	3008 (44)
3. What is the estimated number of rare diseases?	
100–500	512 (7.5)
1000–2000	679 (9.9)
3000–5000	437 (6.4)
<b>6000–8000</b>	<b>584 (8.5)</b>
9000 - 1000	196 (2.9)
Over 10 000	388 (5.7)
I do not know	4042 (59.1)
4. At what age group do rare diseases most frequently appear?	
Newborns	1787 (26.1)
<b>Children</b>	<b>1634 (23.9)</b>
Adolescents	200 (2.9)
Adults	312 (4.6)
They are present in all age groups equally	1001 (14.6)
I do not know	1904 (27.8)
5. How many people suffer from rare diseases worldwide?	

Items	N (%)
10–15 000 000	390 (5.7)
50–75 000 000	477 (7)
100–150 000 000	496 (7.3)
200–250 000 000	344 (5)
<b>300–350 000 000</b>	<b>421 (6.2)</b>
Over 500 000 000	178 (2.6)
I do not know	4532 (66.3)
6. How many people in the Asia suffer from rare diseases?	
10–50 000 000	686 (10)
50–100 000 000	481 (7)
100–200 000 000	328 (4.8)
<b>200–250 000 000</b>	<b>259 (3.8)</b>
250–300 000 000	168 (2.5)
I do not know	4916 (71.9)
7. How many people suffer from rare diseases in Iran?	
10–50 000	675 (9.9)
100–300 000	625 (6.9)
300–500 000	469 (6.9)
1–1 500 000	535 (7.8)
<b>Over 2 000 000</b>	<b>263 (3.8)</b>
I do not know	4271 (62.5)
8. What is the most common cause of rare diseases?	
Infectious and bacterial	189 (2.8)
<b>Genetic</b>	<b>4829 (70.6)</b>
Autoimmune	654 (9.6)
Mitochondrial	89 (1.3)
Environmental	122 (1.8)
I do not know	955 (14)

Items	N (%)
9. What percentage of rare diseases are of genetic origin?	
5–10%	677 (9.9)
20%	697 (10.2)
50%	906 (13.2)
<b>80%</b>	<b>1693 (24.8)</b>
100%	0 (0)
I do not know	2865 (41.9)

Table 2. Students' knowledge about rare diseases

Participants selected rare diseases from a list of 28 diseases (Table 3). The most recognized RD were: Sickle cell anemia (30.2%), Marfan syndrome (28.6%) and Gaucher disease (28%). Least often were indicated Pompe disease (10.5%) and Osteogenesis imperfecta (9.9%). In contrast, the diseases most frequently confused with RD were Crohn's disease (31.6%), Cerebral palsy (25.7%), and Glaucoma (21%).

Table 3  
Which of the following diseases are considered to be rare in Iran?

Items	N (%)
<b>Fragile X syndrome</b>	<b>1365 (20)</b>
Alzheimer's disease	1499 (21.9)
<b>Progeria</b>	<b>1006 (14.7)</b>
<b>Duchenne muscular dystrophy</b>	<b>1466 (21.4)</b>
Crohn's disease	2164 (31.6)
<b>Niemann-Pick disease</b>	<b>1029 (15)</b>
<b>Huntington disease</b>	<b>1342 (19.6)</b>
Cerebral palsy	1758 (25.7)
<b>Marfan syndrome</b>	<b>1957 (28.6)</b>
<b>Phenylketonuria</b>	<b>1165 (17)</b>
Munchausen syndrome	438 (6.4)
<b>Sickle cell anemia</b>	<b>2064 (30.2)</b>
Down syndrome	1070 (15.6)
<b>Acromegaly</b>	<b>1077 (15.8)</b>
<b>Osteogenesis imperfect</b>	<b>680 (9.9)</b>
<b>Pompe disease</b>	<b>715 (10.5)</b>
Schizophrenia	322 (4.7)
<b>Mucopolysaccharidosis</b>	<b>1782 (26.1)</b>
<b>Gaucher disease</b>	<b>1916 (28)</b>
Halitosis	857 (12.5)
<b>Haemophilia</b>	<b>1618 (23.7)</b>
<b>Craniodiaphyseal dysplasia</b>	<b>1312 (19.2)</b>
Glaucoma	1438 (21)
<b>Neurofibromatosis</b>	<b>1037 (15.2)</b>
<b>Achondroplasia</b>	<b>874 (12.8)</b>
<b>Cystic fibrosis</b>	<b>1482 (21.7)</b>

Items	N (%)
Acquired immunodeficiency syndrome	1317 (19.3)
Fibromyalgia	532 (7.8)

Table 3. Which of the following diseases are considered to be rare in Iran?

Future healthcare professionals' practical information about RD is shown in Table 3. Only 9.8% of participants were aware of the Iranian website that provided information about RDs. This is while about 59% of the participants were aware of the central register of RD patients in Iran and about 43% of them were aware of the existence of a national plan for RDs in Iran.

Table 4. Students' knowledge about healthcare system for RD patients

Table 4  
Students' knowledge about healthcare system for RD patients

Items	N (%)
11. What percentage of rare disease can be treated with drugs?	
0%	500 (7.3)
<b>5%</b>	<b>524 (12.1)</b>
10%	630 (9.2)
15%	428 (6.3)
20%	477 (7)
50%	196 (2.9)
I do not know	3783 (55.3)
12. When is Rare Disease Day celebrated?	
January 26	189 (2.8)
<b>February 26</b>	<b>1231 (18)</b>
March 26	185 (2.7)
April 26	242 (3.5)
I do not know	4991 (73)
13. The Iranian non-governmental patient's organization in the field of rare diseases is:	
Ghoghnoos	324 (4.7)
<b>Iranian Rare Diseases Foundation</b>	<b>1470 (21.5)</b>
Zanjireh Omid International Charity Institute	481 (7)
I do not know	4563 (66.8)
14. What is the name of the Iranian website providing information about RD and orphan drugs?	
<b>Radoir</b>	<b>669 (9.8)</b>
NORD	413 (6)
EURORDIS	136 (2)
RARE	433 (6.3)

Items	N (%)
Orphanet	143 (2.1)
Global Genes	83 (1.2)
I do not know	4961 (72.6)
15. Is Iran a member of The world Organization for Rare Diseases?	
<b>Yes</b>	<b>2683 (39.2)</b>
No	510 (7.5)
I do not Know	3645 (53.3)
16. Is there a central register of RD patients in Iran?	
<b>Yes</b>	<b>3751 (54.9)</b>
No	403 (5.9)
I do not know	2684 (39.3)
17. Are orphan drugs reimbursed in Iran?	
Yes	1925 (28.2)
<b>Yes, some</b>	<b>2834 (41.4)</b>
No	0 (0)
I do not know	2079 (30.4)
18. Is there a national plan for rare diseases in Iran?	
<b>Yes</b>	<b>2925 (42.8)</b>
No	1670 (24.4)
I do not know	2243 (32.8)

Students' perceptions of their knowledge of rare diseases are shown in Table 4. Almost 85% of participants rated their knowledge about rare diseases as poor or insufficient. While nearly 70 percent of participants took courses about rare diseases at university, a small number of them declared their

knowledge because of the university courses. 60.3% of participants indicated that rare diseases are a serious public health issues and, in their opinion, family physicians (49.3%) and geneticists (64.6%) should be trained specifically in rare diseases. Finally, 72.7% of future healthcare professionals did not feel ready to take care of a patient with a rare disease.

Table 5. Students' self-assessment of their knowledge about RD

Table 5  
Students' self-assessment of their knowledge about RD

Items	N (%)
Do RD constitute a serious public health issues?	
Absolutely yes	2004 (29.3)
Yes	2120 (31)
No	718 (10.5)
Definitely no	164 (2.4)
I do not know	1832 (26.8)
Which physicians should be uniquely trained in RD?	
Family physician	3370 (49.3)
Pediatrician	2376 (34.7)
Neurologist	1237 (18.1)
Geneticist	4418 (64.6)
Psychiatrist	1165 (27.4)
Immunologist	1876 (27.4)
Have you had any classes about rare disease during your studies?	
Yes	4746 (69.4)
No	1468 (21.5)
I do not know	624 (9.1)
How would you rate your knowledge about rare diseases?	
Very good	125 (1.8)
Fair enough	868 (12.7)
Insufficient	3060 (44.7)

Items	N (%)
Very poor	2785 (40.7)
Would you like to broaden your knowledge about rare diseases?	
Yes	5481 (80.2)
No	823 (12)
I do not know	534 (7.8)
Do you think that there should be a mandatory course on rare diseases in medical curricula?	
Definitely yes	1494 (21.8)
Rather yes	3444 (50.4)
Rather not	1034 (15.1)
Definitely not	489 (7.2)
I do not know	377 (5.5)
Where do you get your knowledge about RD from?	
Mandatory courses at the university	1715 (25.1)
Faculty courses at the university	470 (6.9)
Scientific literature and research	1635 (23.9)
Scientific conferences, symposia	724 (10.6)
Internet	3803 (55.6)
I do not search for such information	1332 (19.5)
Do you feel prepared for caring over a patient with a rare disease?	
Definitely yes	378 (5.5)
Rather yes	1495 (21.9)
Rather not	2112 (30.9)

Items	N (%)
Definitely not	2117 (31)
I do not know	736 (10.8)

## Discussion

According to the literature review, no studies were found in Iran examining the knowledge of future healthcare professionals about RD. Due to the growing understanding of the medical community about genetically based diseases, there is a need for educational programs in the area of RD in particular. As a result, future healthcare professional often do not receive the necessary training in RD (12).

Consistent to previous studies (12–15), the results of the present study showed that future healthcare professionals receive little education about RD. Although almost all of the participating students were familiar with RD and most believed that RD was due to genetic factors, they had little knowledge of the epidemiology of RD. Many students have used the "I do not know" option and the problem of insufficient education is evident during their studies. In Iranian medical universities, basic sciences including biochemistry, physiology as well as genetics are taught in the first year. Regarding to the major, it may continue or not. Numerous RDs such as PKU, CF, Huntington disease, and sickle cell disease are included in genetics courses. In addition to the diseases, diagnostic tests and major clinical manifestations are also taught. We suppose that medical sciences students (MSSs) learn the knowledge on RDs randomly and casually. The premise is according to the following reasons: 1) while most of students were aware of their knowledge deficits, they had unsatisfactory level of information on RDs. 2) RDs are not taught as a separate subject and in a systematic comprehensive form. Rather, it is considered a subset of genetics. So all of MSSs are deprived of training in relation to RDs. In line with our findings, previous studies (9, 16) authenticate the fact that the retention of MSSs' knowledge and skills is compromised.

In the present study, some of MSSs has selected the following diseases as RDs mistakenly: Crohn's disease (31.6%), Cerebral palsy (25.7%), and Glaucoma (21%). The above-mentioned diseases are kind of those which are well discussed in the classes. Confusing them with RDs shows that despite MSSs would be able to present a simplified way of dealing with a given disease, frequently they do not identify them correctly. More interestingly, in high school education some basic information about genetic disorders and RDs is already present, therefore this problem is rooted in educational system which is consistent with Williams' findings (17) These results indicate that medical sciences curriculum is likely to be disrupted and inappropriate.

Moreover, there is not a specific course, guideline or recommendations at Iranian universities pertaining to RDs. Our medical education system usually focuses on more prevalent disorders and prepare students for facing conventional diseases rather than rare ones. More studies in this field is needed to determine whether an elective or mandatory course on RDs should be included in the medical education curriculum.

Additionally, preparing a comprehensive content including multidimensional information about RDs like prevalence, incidence, relevance of RDs to everyday medical care, early detection, potential strategies to dwindle it and the last but not least, the challenges that healthcare workers face with during admission of RD patients is definitely necessary. Healthcare workers have to know sources of information and support for RD patients to help RD patients' family and caregivers.

McKay's (18) suggest that teaching programs should not focus on any particular RD. There are near 6,000–8,000 different types of RDs and focusing on the whole RDs is actually impossible. Trying to raise public awareness about cursory knowledge on the prevalence and incidence of RDs would be of profound importance. Pisklakov acknowledges that health professionals' false beliefs may interfere with their attitude and disrupt the situations. This is of key importance because health professionals' false beliefs in their knowledge and skills makes it difficult to change the situation of patients with rare diseases (19) Alawi et al. (20) suggest to use RDs as a teaching model to transfer the basic sciences and clinical practice to students.

To sum up, the present study shows that insufficiency of medical curriculum about RDs cause knowledge and skills deficits in MSSs, so that they cannot diagnose those diseases or provide appropriate care for patients. It is of key importance to make MSSs aware of their deficits. Then address the desperate desire for improvement of medical education on RDs through obligatory lectures and seminars, journal clubs and group discussions.

## **Conclusion**

The present study has indicated a gap in Iranian medical students' knowledge on RDs. The researchers believe that health science policy makers should make a joint effort to improve knowledge about RDs. Including courses with regard to RDs would be of benefit to future healthcare professionals, the importance of pharmaceutical education on orphan drugs which could be included in the medical curricula; the need to establish closer collaboration with other neighbor countries, the role of machine learning and artificial intelligence that can support the decision process and overcome barriers that affect the diagnostic odyssey and the role of telemedicine and telepharmacy services in providing RD patients the opportunity to continue treatment.

## **Methods**

A cross-sectional survey-based study was conducted in Iran from September 2021 to January 2022 and using convenience sampling. Our target population consisted of students of health care professionals who had completed two years of education and had access to the Internet. Data collection was carried out via online feedback from participants during the survey. An online questionnaire (Google Form) was administered which also contains the motivation letter explaining the purpose of the participants' response, and the hyperlink to the questionnaire was eventually published on social media. The online questionnaire was designed in order that individuals can withdraw at any time. Over 10,000 students

from all medical sciences universities participated in this study, and after removing those who did not meet the inclusion criteria or filled out the questionnaire incompletely, we reached 6838 eligible participants.

## Measures

**Demographic check-list:** Includes: age, gender, marital status, field of study, Grade, Duration of education, and domicile.

## Knowledge And Attitudes Of Rare Diseases' Questionnaire

The questionnaire used in the present study was developed by Domaradzki et al. based on the previous literature review and the aim of the study. The detailed description of the questionnaire and the method of its construction are explained elsewhere (9, 10).

In summary, the questionnaire consisted of 26 questions: the first questions concerned students' basic knowledge of RDs. Students were also asked to name RDs from a list that included twenty-eight diseases: 10 most common condition and 18 RDs. The second section covered questions on organizational issues. The third section consisted of questions on students' awareness of RDs and their self-assessment knowledge and skills in the field of RDs.

The methodological procedures for the translation and cultural adaptation of current questionnaire were employed as suggested by Beaton et al. according to a formal forward/backward translation protocol (21). In the next step, some questions changed based on the culture policies of Iran (questions: 6, 7, 12–18). The prepared version of the questionnaire was sent to 15 experts (Experts included nurse, physician, geneticist, sociologist and physiotherapist) who were invited to give their opinion in order to improve the final version.

Content validity includes Content Validity Ratio (CVR) and Content Validity Index (CVI). Fifteen experts reviewed the questionnaire items and chose one of the essential, useful but not necessary or unnecessary options (22). According to the Lawshe table, the accepted rate is 0.49 (23). None of the phrases have been deleted in this section. After applying expert opinions and enriching the phrases, 15 other experts reviewed the questionnaire to evaluate CVI (24).

## Data Analysis

Data were entered into SPSS software version 18 and reported in frequency, percentage, mean, and standard deviation.

## Ethics

The study procedures were approved by the Student Research Committee of Golestan University of Medical Sciences and Ethical Review Board (IR.GOUMS.REC.1400.194). All participants entered the study with knowledge of the purpose of the study and informed consent and were informed that they could withdraw from the study at any time.

## **Declarations**

### **Ethical consideration**

This study was conducted following receipt of the ethics approval (IR.GOUMS.REC.1400.194) from the Golestan University of Medical Sciences. All students gave informed consent for the use of data for research purposes.

### **Consent for publication**

Not applicable.

### **Availability of data and materials**

All data generated or analyzed during this study are included in this published article

### **Conflict of interest**

The authors declare that there are no conflict of interests.

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### **Authors' contributions**

RJ was responsible for analyzing the data, drafting the manuscript, reviewing the manuscript, and approving the final version. AN was responsible for collecting data, analyzing the data, drafting the manuscript, and approving the final version. MF was responsible for collecting data and approving the final version. SB, MF, KA, AF, SB, MS, AA, KE, NN, FS, HM, EY, SB, NM, EM, SA, and FT were responsible for collecting data. JD and AS were responsible for reviewing the manuscript and approving the final version. LJ was responsible for designing the study, reviewing the manuscript, and approving the final version. All authors read and approved the final article.

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