

Education and Information Need for Emergency Physicians About Rare Diseases in China

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Research

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Abstract

Background: Rare diseases are serious and chronic disease that affect no more than 1 person in 2000. The patients suffering from RD may come to emergency department for life-threatening symptoms, such as acute aortic dissection, intracranial hemorrhage, and severe respiratory distress. Diagnostic delay of rare disease patients is common and often caused by low rare disease awareness among physicians. The main aim of this study was to investigate the Chinese emergency physicians' basic knowledge, information access and educational needs regarding rare diseases. An online questionnaire was completed by Chinese emergency physicians during January and March 2021.

Methods and Results: A total of 539 emergency physicians responded to the questionnaire-based study, including 200 females and 339 males. More than half of respondents were from Tertiary A hospital and had engaged in medical clinical work more than 10 years. Only 4.27% of respondents correctly estimated the prevalence of rare diseases. A few respondents knew the exact number of RD in the first official list of rare diseases in 2018. 98.5% of respondents rated their knowledge about rare diseases as rare or insufficient. Most of emergency physicians preferred to getting information by search engine instead of specialized websites of rare diseases. Lack of practice guidelines or consensus and were considered as the most important reason for diagnostic delay of RD. Practice guidelines or consensus and professional websites on rare diseases were urgently needed for emergency physicians.

Conclusion: The investigation shows poor knowledge of emergency physicians regarding rare diseases. Practice guidelines and professional websites on rare diseases were the prominently urgent needs for emergency physicians. Specialized RD courses should also be added in medical education.

Introduction

Rare diseases (RDs) are diseases affect no more than one person in 2000, According to the European Union (EU), always are serious, chronic, and often life-threatening conditions [1]. It has been estimated that from 6 to 8% of the population will be affected by a rare disease [2], this means that there are at least 16.8 million rare disease patients in China. Due to the high clinical complexity, patients with rare diseases often face lengthy diagnostic delays, some families seeing between 3 and 10 different doctors and waiting >5 years for a definitive diagnosis [3, 4].

Previous studies have shown that health care professionals and medical students lack training and experience on RD [2, 5]. Our study aims to describe Chinese emergency physicians' clinical practice regarding rare diseases, their information-seeking behaviors and their educational needs and preferences.

Materials And Methods

The study was conducted between January and March 2021 among emergency physicians of China. A separate online data-collection questionnaire was used for survey, using a web platform(www.wjx.cn), which permits centralized data collection and limit repetition by mobile number. The survey was

conducted with a standard questionnaire that was constructed from themes based on a review of the literature and the study aim. It included 30 questions: 6 questions that addressed their demographic data and 24 items referring to respondents' knowledge of and attitudes towards RD. The questionnaire consisted of four groups of questions. The first group were six questions referred to demographic information, including gender, career length, hospital level, technical titles, licensed province and whether the respondents had any advanced training experience in other hospital.

The second group of thirteen questions concentrated on the emergency physicians' knowledge of RD. The respondents were asked about the incidence and number of RD. Three questions referred to the current situation of RD in China, including the estimated number of RD patients, if there was any national register system and the exact number of RD in the first list of rare diseases in 2018. Four items related to orphan drug, hereditary and the most common age of onset of RD. In addition, all respondents were asked to indicate which RD they had met from a list of twenty-seven RD. The respondents were asked if they had first diagnosed any RD, in 140 respondent who had first diagnosed RD, they were asked the number of RD they had seen in their career and to indicate which RD they first diagnosed from a list of twenty RD.

The third group were six questions related to the respondents' self-assessment and information access of RD. The respondents were asked how they perceive their knowledge about RD, whether they had any classes on RD, how they learn about it and which access was the most impressed. Two questions referred to which website they prefer to learning about RD and which professional website of RD they had used.

The last group of five questions referred to emergency physician's needs of RD information. The respondents were asked if they would like to learn more about RD and which reason delayed the diagnosis of RD. The authors also wanted to know which aspect of RD the emergency physicians prefer to learn more. Finally, the respondents were asked which specialty need more in-depth training on RD and whether it is necessary to add RD course to their medical school education. Ethics approval and research governance approval were obtained from Peking Union Medical College Hospital.

Statistical testing was performed using SPSS Statistics 25.0. The respondents were divided into different groups according to career length, hospital, and title. The rate of correct was the percent of respondents choosing the right answer in each group. The rate of correct were analyzed with the chi-square test to reveal statistical differences ($p < 0.05$).

Results

1.Demography

A total of 539 emergency physicians answered the questionnaire. All respondents were from China, including 27 of 34 Chinese provincial administrative regions, with the top five in Hubei (83, 15.4%), Liaoning (70, 12.99%), Guangxi (65, 12.06%), Henan (41, 7.61%), and Jiangxi (38, 7.05%). The demographic information of these respondents was listed in Table 1. Of 539 emergency physicians, 200

(37.11%) were female and 339 (62.89%) are male, while 244 (45.27%) had engaged in medical clinical work more than 15 years. In China, a 3-tier system was established to recognize a hospital's comprehensive abilities in medical care, education, and research [6]. Of 539 emergency physicians, 337 (62.52%) were from Tertiary A (the highest level), while 65 (12.06%) were Tertiary B and 137 (25.42%) were Secondary. Responders in this study included 99 (18.37%) residents, 209 (38.78%) attendings, 170 (31.54%) associate chief physicians and 61(11.32%) chief physicians. 331 (61.41%) respondents had advanced training experience in other hospitals.

Table 1
Demographic information of emergency physicians

characteristic	respondents (n=539)	
	n	%
gender		
female	200	37.11%
male	339	62.89%
career length (years)		
<5	64	11.87%
5-10	86	15.96%
10-15	145	26.90%
>15	244	45.27%
hospital		
Tertiary A	337	62.52%
Tertiary B	65	12.06%
Secondary	137	25.42%
Title		
resident	99	18.37%
attending	209	38.78%
associate chief physician	170	31.54%
chief physician	61	11.32%
Advanced training experience		
no	208	38.59%
yes	331	61.41%

2.Knowledge

Respondents' knowledge about rare diseases was shown in Table 2. Only 23 (4.27%) respondents correctly estimated the prevalence of RD and 115 (21.34%) knew the number of RD. 44 (8.16%) respondents were aware of the number of patients suffering from RD in China, while 199 (36.92%) knew that there was national register system for RD in China. 93 (17.25%) respondents knew the exact number of RD in the first list of rare diseases in 2018, and 105 (19.48%) were aware of the percentage of RD with

orphan drug. 509 (94.43%) respondents knew that not all RD were hereditary, but only 97 (18%) knew the percentage of hereditary disease in RD. 147 (27.27%) respondents knew the most common age of onset of RD. Only 140 (25.97%) respondents had first diagnosed RD. Out of the 140 respondents, 98 (70%) had seen less than ten types of RD, 32 (22.86%) had ten to twenty types and only 10 (7.14%) had more than twenty types. For six core questions on RD, the rate of correct of respondents from different groups were showed in Figure 1. The result of chi-square test shows that from different career length, hospital and title groups, the respondents have similar knowledge rate of RD ($p > 0.05$). Only in the correct of questions "Was there any national register system for RD in China", the respondents from secondary hospital have a lower rate than those from tertiary A or B hospital ($p < 0.05$), and the rate of correct has no statistical difference between group tertiary A and tertiary B ($p > 0.05$).

Table 2 Emergency physicians' knowledge of rare disease

Items	n	%
Incidence of RD		
≤1‰	66	12.24%
1‰-2‰	23	4.27%
≤1‰-10‰	259	48.05%
I do not know	191	35.44%
Number of RD in the world		
600-800	93	17.25%
6000-8000	115	21.34%
60000-80000	18	3.34%
I do not know	313	58.07%
Number of patients suffering from RD in China		
600,000	101	18.74%
6000,000	112	20.78%
60,000,000	44	8.16%
600,000,000	0	0%
I do not know	282	52.32%
Was there any national register system for RD in China		
yes	199	36.92%
no	28	5.19%
I do not know	312	57.88%
The exact number of RD in the first list of rare diseases in 2018		
121	93	17.25%
147	95	17.63%
I do not know	351	65.12%
The percent of RD with orphan drug		
0%	11	2.04%
5%	105	19.48%
10%	41	7.61%

20%	23	4.27%
50%	6	1.11%
I do not know	353	65.49%
Were all RD hereditary?		
yes	30	5.57%
<i>no</i>	509	94.43%
The percentage of hereditary disease in RD		
20%	100	18.55%
50%	72	13.36%
80%	97	18%
100%	2	0.37%
I do not know	268	49.72%
The most common age of onset of RD		
Neonatal period	45	8.35%
<i>Childhood</i>	147	27.27%
Adulthood	37	6.86%
Equal in all age groups	143	26.53%
I do not know	167	30.98%
Have you first diagnosed RD?		
yes	140	25.97%
no	399	74.03%
How many RD have you seen in your career? *		
1-5	54	38.57%
5-10	44	31.43%
10-15	22	15.71%
15-20	10	7.14%
≥20	10	7.14%

The respondents were presented a list of twenty-seven rare diseases and asked to select those they had seen or first diagnosed (Table 3). For the 539 responded emergency physicians, the most common RD

they had seen were Marfan syndrome (49.17%), Hemophilia (46.75%), Idiopathic pulmonary fibrosis (46.01%), Multiple sclerosis (41.37%) and Idiopathic pulmonary arterial hypertension (40.63%). For the 140 emergency physicians who had first diagnosed RD, the most frequent RD they first diagnosed were Marfan syndrome (38.57%), Generalized myasthenia gravis (34.29%), Idiopathic pulmonary arterial hypertension (30%), Idiopathic pulmonary fibrosis (29.29%) and Multiple sclerosis (24.29%).

Table 3
Which of the following rare diseases had been seen or first diagnosed?

Rare diseases	Which of the following rare diseases had you seen? (n=539)	Which of the following rare diseases had you first diagnosed? (n=140)
Idiopathic pulmonary arterial hypertension	219 40.63%	42 30%
Idiopathic pulmonary fibrosis	248 46.01%	41 29.29%
IgG4 related disease	79 14.66%	16 11.43%
Lymphangioleiomyomatosis	45 8.35%	4 2.86%
Marfan syndrome	265 49.17%	54 38.57%
Multiple sclerosis	223 41.37%	34 24.29%
POMES syndrome	76 14.10%	12 8.57%
Porphyria	143 26.53%	24 17.14%
Albinism	204 37.85%	27 19.29%
Atypical hemolytic uremic syndrome	49 9.09%	9 6.43%
Autoimmune encephalitis	157 29.13%	30 21.43%
Castleman disease	42 7.79%	3 2.14%
Congenital scoliosis	125 23.19%	19 13.57%
Diamond-Blackfan anemia	38 7.05%	3 2.14%
Fanconi anemia	35 6.49%	3 2.14%
Gaucher disease	42 7.79%	3 2.14%
Generalized myasthenia gravis	218 40.45%	48 34.29%
Glycogen storage disease	51 9.46%	2 1.43%
Hemophilia	252 46.75%	30 21.43%
Hepatolenticular degeneration	157 29.13%	32 22.86%
Pulmonary alveolar proteinosis	71 13.17%	16 11.43%

Top five are written in bold characters.

Rare diseases	Which of the following rare diseases had you seen? (n=539)	Which of the following rare diseases had you first diagnosed? (n=140)
Amyotrophic lateral sclerosis	115	21.34%
Congenital myasthenic syndrome	63	11.69%
Huntington disease	59	10.95%
Idiopathic cardiomyopathy	61	11.32%
Langerhans cell histiocytosis	74	13.73%
Systemic sclerosis	173	32.10%
Others	10	1.86%
Top five are written in bold characters.		13
		9.29%

3. Self-assessment and information access

Self-assessment and information access of RD were listed in Table 4. Only 1 (0.19%) respondent thought he/she knew well about RD and 7(1.30%) almost, while 158 (29.31%) knew insufficiently and 373 (69.20%) rarely. Most respondents (74.95%) did not have rare disease course in medical education. 376 (69.76%) respondents learn about RD in clinic work, and 232 (43.04%) thought it is the most impressed way. 412 (76.44%) respondents prefer Baidu for more information about RD, only 20 (3.71%) learned from rare disease specialist website. 255 (47.31%) respondents had used national rare disease registry system of China. The answers of "How did you learn about rare diseases?" and "Which websites do you prefer to learn more about RD?" were showed in Figure 2, which were distinguished by different career length and hospital groups. As career length was longer, the proportion of learning RD by studying in medical school was lower. The respondents with longer career length had lesser preference to learn RD through pubmed. The respondents from secondary hospital presented less proportion of studying RD in medical school or through academic website, such as pubmed, CNKI, VIP and WanFang, but high proportion of learning RD by advanced training in other hospital and attending academic conferences.

Table 4
Self-assessment and information access of rare diseases

Items	N	%
How well do you know about rare diseases?		
perfectly	1	0.19%
almost	7	1.30%
insufficiently	158	29.31%
rarely	373	69.20%
Is there any rare disease course in your medical education?		
yes	84	15.58%
no	404	74.95%
I do not know	51	9.46%
How did you learn about rare diseases?		
studying in medical school	248	46.01%
surfing RD website	118	21.89%
working in clinic	376	69.76%
Advanced training in other hospital	216	40.07%
Attending academic conferences	342	63.45%
others	40	7.42%
Never heard	20	3.71%
Which access impressed you most?		
studying in medical school	29	5.38%
surfing RD website	38	7.05%
working in clinic	232	43.04%
Advanced training in other hospital	103	19.11%
Attending academic conferences	124	23.01%
others	13	2.41%
Which websites do you prefer to learn more about RD?		
pubmed	128	23.75%
Baidu	412	76.44%

Items	N	%
wikipedia	79	14.66%
uptodate	103	19.11%
Rare disease specialist website	20	3.71%
DXY	329	61.04%
CNKI or VIP or WangFang	114	21.15%
I do not want to know	7	1.30%
others	22	4.08%
Which professional websites of RD have you used?		
OMIM	71	13.17%
national rare disease registry system of China	255	47.31%
Orphanet Portal for rare disease	35	6.49%
ERAM or PEDAM	62	11.50%
others	222	41.19%

4.Information needs

Emergency physician' needs of RD information were taken in Table 5. Almost all the respondents (92.21%) want to learn more about RD. 450 (83.49%) respondents thought lack of practice guidelines or consensus delayed the diagnosis of rare disease. And 466 (86.46%) preferred to learn more information about RD by diagnostic guidelines or consensus. The distribution of information needs with the answer of "Which aspect of rare diseases do you prefer to learn more?" were displayed by different career length and hospital groups (Figure 3). Each group of career length or hospital had similar proportion of information need. The respondents with more than 15 years of career or from secondary hospital had high proportion in needing relevant professional websites and the hospital or specialist that can refer to. Half of the respondents (51.21%) thought every physician regardless of specialization need in-depth training on RD. Moreover, 254 (47.12%) respondents thought emergency physicians need more training about RD, and 475 (88.13%) declared it was necessary to add rare disease courses in medical school education.

Table 5
Emergency physician' needs of RD information

Items	N	%
Would you like to know more about rare diseases?		
Yes	497	92.21%
No	4	0.74%
I'm not sure	38	7.05%
The reasons that delayed the diagnosis of rare disease		
Lack of practice guidelines or consensus	450	83.49%
Do not know how to find relevant professional websites	271	50.28%
Do not know the hospital or specialist that can refer to	252	46.75%
Lack of material or official account to preach to patients or their family	225	41.74%
Do not know the hospital or specialist for genetic counseling	277	51.39%
Which aspect of rare diseases do you prefer to learn more?		
Practice guidelines or consensus	466	86.46%
Relevant professional websites	370	68.65%
The hospital or specialist that can refer to	308	57.14%
Material or official account to preach to patients or their family	267	49.54%
The hospital or specialist for genetic counseling	271	50.28%
Which specialty need more in-depth training on rare disease?		
Pediatrician	156	28.94%
General practitioner	212	39.33%
Neurologist	110	20.41%
Geneticist	186	34.51%
Physician	139	25.79%
Emergency physician	254	47.12%
Every physician regardless of specialization	276	51.21%
Other	9	1.67%
None	4	0.74%
Is it necessary to add rare disease course to your medical school education?		

Items	N	%
Yes	475	88.13%
No	32	5.94%
Depending on the course content	32	5.94%

Discussions

1. Basic knowledge

The questionnaire results indicated that emergency physicians have poor knowledge of RD and that the urgent need of RD information does exist. Although most respondents had medical practice more than five years and half of them had advanced training experience, they lacked basic knowledge about RD. Only 4.27% respondents knew that a disease with an incidence lower than 1 in 2000 was defined as rare diseases [7]. More than half of respondents have no idea about the epidemiology, register system and the first official list of RD in China. 18% emergency physicians knew the that 80% of RD were hereditary, while 27.27% were clear about that the onset of RD most frequently appeared in childhood.

To the best of our knowledge, this is the first research concentrated on the emergency physicians' education and information about rare diseases in China. The similarly poor cognitive level about RD was also reported in physicians [6, 8], surgeon [9], medical students [5], nurses and nursing students [10]. In China, emergency physicians deal with all patients in emergency cases and are responsible for further treatment and referrals, if emergency physicians lack of knowledge of RD, RD patients with acute symptoms may lose the chance of diagnosis. In our research, only a quarter of respondents had experience on first diagnosing RD (140 ,25.97%).

2. Self-assessment and information access

Of emergency physicians, 98.5% rated their knowledge about RD as rare or insufficient, while Walkowiak et al. [11] reported 94.6% of physicians their perceived knowledge on RDs as insufficient or very poor. Although rare diseases are chronic disease and mostly diagnosed by the specialists, the patients suffering from RD could have come to emergency department for life-threatening symptoms before the first diagnosis of RD. Marfan syndrome could be not diagnosed until an individual present with an acute aortic dissection [12]. For Hemophilia, intracranial hemorrhage is a potentially deadly complication [13, 14]. The patients could present with severe respiratory distress when suffering from idiopathic pulmonary fibrosis, multiple sclerosis, idiopathic pulmonary arterial hypertension, or generalized myasthenia gravis [15, 16]. For our emergency physicians, clinic work and academic conference were the most common and impressed access to learn about rare disease. Baidu (a common search engine in China, <http://www.baidu.com/>) and DXY (a platform for professional medical communication in China, <http://www.dxy.cn/>) were the most popular websites. Only 3.71% of respondents preferred specialist website to learn more about RD and less than half had used the national rare disease registry system of

China (<https://www.nrdrs.org.cn/>). We found that the respondents from secondary hospital presented less proportion of studying RD in medical school or through academic website but high proportion of learning RD by advanced training in other hospital and attending academic conferences, maybe secondary hospitals are closer to community hospitals, and there are fewer opportunities to acquire knowledge of rare diseases from daily work. The convenience and simplicity of information access seems to be the primary consideration for emergency physicians. Moreover, Pauer et al. [17] indicated that the quality of information on the Internet about RD is low.

3.Education and Needs

Diagnostic delay is one of the most common problems encountered while caring for RD patients [18]. A survey of 462 Australian children living with RD showed that 38% consulted ≥ 6 different doctors before receiving the correct diagnosis, 37% believed the diagnosis was delayed and 27% initially received a wrong diagnosis [3]. Yan et.al [19] investigate the diagnosis experience of 1010 adult RD patients in China. The results indicated that 72.97% of patients were misdiagnosed, they waited an average of 4.3 years and visited 2.97 hospitals before the definitive diagnosis of RD [19]. Our emergency physicians regarded the lack of practice guidelines or consensus as the most important reason for diagnostic delay of RD. Practice guidelines or consensus and relevant professional websites about RD were prominently urgent needs for emergency physicians. 47.12% of respondents considered that emergency physicians needed more in-depth training on RD and 92.21% were willing to learn more. Only 15.58% of emergency physicians had RD courses in their medical education and 88.13% thought it is necessary to add RD course. A survey in medical students showed the similarly poor education on RD [5]. In undergraduate or postgraduate medical education, less than a third of the physicians had received training in rare disease in Spain [2]. Vandeborne et al. [8] reported that most physicians had specific information needs regarding rare diseases in Belgium and attending RD continuous training sessions could improve knowledge and awareness on RD. Academic and continuous medical education should focus on “red flags” to increase RD attentiveness and provide easy access to educational opportunities and information resources regarding RD [2, 8].

Limitations

This is a questionnaire-based research of emergency physicians. Some items might be inaccurate as these were answered on account of the respondents' memory and lacking objective medical records. The demographic distribution of respondents was different from all the emergency physicians. The emergency physicians who responded to this questionnaire may be more concerned with RD than those refused to answer. Thus, this study was limited by underlying inaccurate responses to questionnaires and possible selection bias.

Conclusion

In conclusion, the investigation shows poor knowledge and urgent information needs regarding RD in emergency physicians. It is imperative for public health system to improve the training on RD of

emergency physicians, including add specialized RD courses in medical education, providing practice guidelines and popularizing professional websites on RD.

Declarations

Authors' Contributions

Lingli Zhou, Jing Yang: substantial contributions to conception, design and writing.

Jun Xu: drafting the article and revising it.

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Ethics approval and consent to participate

All procedures followed were in accordance with the ethical standards of the responsible institutional committee on human experimentation and with the Helsinki Declaration of 1975 (revised in 2000). The study protocol was approved by the Ethics Committee of the Institutional Review Board at Peking Union Medical College Hospital (PUMCH). A questionnaire and a list of open-ended questions for interviews were designed in Chinese. Potential participants were invited to participate in this study, and only those who signed the informed consent on line could participate in this study.

Consent for publication

Not applicable.

Competing interests

The authors declare that they have no competing interests.

Availability of data and materials

The data used and/or analyzed to support the results of the current study are available from the corresponding author on reasonable request.

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Figures

Figure 1

The knowledge rate of emergency physician on six core questions of RD. A. the rate of correct in each career length groups. B. the rate of correct in each hospital level groups. C. the rate of correct in each title groups.

* p <0.05, the respondents from secondary hospital have a lower knowledge rate than those from tertiary A or B hospital (p <0.05).

Figure 2

The information access of emergency physicians. A. the distribution of information access in each career length group. B. the distribution of preferential website in each career length group. C. the distribution of information access in each hospital group. D. the distribution of preferential website in each hospital group.

Figure 3

The information needs of emergency physicians. A. the distribution of information needs in each career length group. B. the distribution of information needs in each hospital group.