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# Effects of continuing medical education on emergency trainees' rare disease knowledge and attitude: a single-center study

Anlei Liu[1†], Wenlin Hao[1†], Jun Xu[1], Huadong Zhu[1], Hongbo Yang[2*] and Jing Yang[1*]

## Abstract

**Background** Rare diseases (RDs) affect 10% of the global population but have inadequate medical resources. Early detection and treatment are crucial, yet many emergency physicians lack awareness of RDs. This study aims to evaluate the effects of continuing medical education (CME) on the knowledge and attitude of emergency physicians.

**Methods** This retrospective study was conducted from April to June 2023, involving 218 Chinese emergency physicians. The online questionnaire consisted of four groups and 30 questions, covering demographic data, knowledge, and attitudes regarding RDs. Respondents were divided into two groups based on their recent CME training experience with RDs.

**Results** Two hundred and eighteen emergency physicians completed the questionnaire, of which 108 received RD CME training and 110 did not receive RD CME training. Most respondents (98.2%) felt their knowledge about RDs was insufficient. The CME training group showed increased awareness of RD incidence ($p=0.047$) and improved case analysis after training, but only slight improvement in knowledge of RD professional websites. Among the CME training group, CME was identified as the most prominent avenue for acquiring knowledge about RDs, with 72 respondents (66.7%, $p<0.001$). In contrast, in the non-training group, clinical work was identified as the primary source of learning, with 47 respondents (42.7%, $p<0.001$).

**Conclusion** Emergency physicians generally lacked knowledge about rare diseases. CME training can improve their awareness and knowledge of RDs.

**Keywords** Rare diseases, Emergency physicians, Continuing medical education

†Anlei Liu and Wenlin Hao contributed equally to this work.

*Correspondence:
Hongbo Yang
yanghb@pumch.cn
Jing Yang
yangbujing@126.com
[1]Emergency Department, State Key Laboratory of Complex Severe and Rare Diseases, Peking Union Medical College Hospital, Chinese Academy of Medical Science and Peking Union Medical College, No. 1 Shuaifuyuan, Dongcheng District, Beijing 100730, China
[2]Key Laboratory of Endocrinology of National Health Commission, Department of Endocrinology, State Key Laboratory of Complex Severe and Rare Diseases, Peking Union Medical College Hospital, Chinese Academy of Medical Sciences and Peking Union Medical College, No. 1 Shuaifuyuan, Dongcheng District, Beijing 100730, China

## Introduction

Rare diseases (RDs) affect up to 10% of the global population, representing a significant public health challenge. Different countries have defined RDs on factors such as the prevalence and severity, and may have implemented policies to address these conditions [1]. Since the first orphan drug legislation in the United States in 1983, many countries have adoped rare disease plans or strategies [2]. The United Nations has stressed the importance of incorporating RDs into policies, prompting governments to initiate relevant projects in their respective regions [3–5]. China has also made substantial efforts in recent years, including publishing lists of RDs, establishing the National Network of Rare Diseases (NNRD, https://www.nrdrs.org.cn) and publishing the Diagnos is and Treatment Guidelines [6–8]. However, individuals with RDs in China still face significant challenges in accessing medical resources, with long diagnostic delays and extensive travel for diagenosis [9].

Early detection and treatment are essential, as many rare diseases can be debilitating and life-threatening if left untreated [10, 11]. Previous studies have shown that emergency and general physicians often lack awareness of RDs [12, 13], leading to delayed diagnosis, misdiagnosis, or insufficient treatment [14–16]. This is particularly concerning given the large population base in China, where many rare diesease patients may first present to emergency departments [12]. Ensuring that the emergency physicians are adequately trained and knowledgeable is crucial for early detection adn treament [17]. While some countries have incorporated RD education into their national strategies [2, 18, 19], China's efforts in this area are still in the early stages [20], with limited research on the effectiveness of existing training programs [14].

This study aimed to fill this knowledge gap by evaluating the effects of continuing medical education (CME) on the knowledge and attitude of emergency trainee physicians regarding RDs. By assesing the impact of CME programs, this study seeks to provide insights into improving the training of emergency phisicians and ultimately enhancing the diagosis and treament of RDs in China.

## Study design and participants

### Study design

Our study design was informed by the comprehensive framework proposed by Tumiene et al. [21], which emphasizes the need for a holistic approach to rare disease education. This framework underscored the importance of interprofessional collaboration, aligning with our research objectives to enhance healthcare professionals' competencies in managing rare diseases. Our study was a retrospective single-center study in Peking Union Medical College Hospital in Beijing, China (Fig. 1). At the first stage, a CME training program for RD was offered for fellowship physicians in the department of Emergency. At the second stage, an online questionnaire was conducted among these emergency physicians and their non-trained colleagues. All study procedures followed were in accordance with the ethical standards of the responsible institutional committee on human experimentation and with the Declaration of Helsinki of 1975 (revised in 2000). The study protocol was approved by the ethics committee of the institutional review board at Peking Union Medical

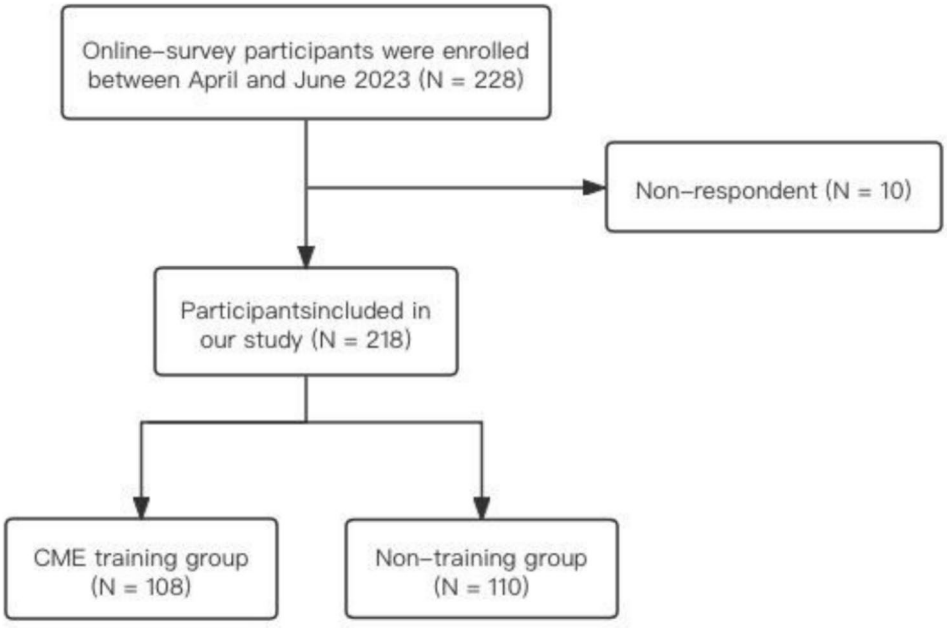


**Fig. 1** The flow chart of online-survey participants

College Hospital (PUMCH). Only those who signed the informed consent form online could participate in this study.

#### Continuing medical education of RD

In March 2020, Peking Union Medical College Hospital (PUMCH) established the State Key Laboratory of Complex Severe and Rare Diseases, and all emergency trainee physicians and postgraduates could choose to attend the emergency rare disease CME program including the following:

a. Lectures: Expert speakers and experienced physicians delivered comprehensive lectures on various aspects of rare diseases, including their definition, epidemiology, clinical manifestations, and management.
b. Case reports: Real-life cases of rare diseases in clinical work were presented to trainee physicians. These case reports helped illustrate the challenges of diagnosing and managing rare diseases and provided practical insights into their clinical presentation. Lectures and case reports alternated between regular times each week and lasted two to three hours.
c. Bedside teaching: In daily clinical work, the content of bedside teaching was integrated into the ward rounds of senior physicians. The trainee physicians had the opportunity to learn from senior physicians while attending to patients with rare diseases at the bedside. These hands-on experiences enhanced their clinical skills and decision-making abilities.
d. Multidisciplinary consultation case discussions: Regular case discussions involving a multidisciplinary team of specialists, including geneticists, radiologists, and pathologists, were organized two to three times a month. This fostered a collaborative approach to diagnosing and treating rare diseases.

By participating in the above training, we guaranteed that in addition to approximately 50 h of clinical work and bedside learning opportunities, the trainee physicians had nearly 10 h of lectures and case reports and approximately 6 h of multidisciplinary seminars per month.

#### Participants

The selection of participants in our study mirrored the approach taken by Morgenthau et al. [22], who included medical students from diverse backgrounds to provide a comprehensive evaluation of rare disease education programs. Similarly, our study included emergency physicians from different parts of the country and at different levels of hospitals. This strategy ensured a broad representation and enhances the generalizability of our findings. Upon completion of the CME training, we retained the contact details of these fellowship physicians for further academic discussion. An online questionnaire was distributed to these emergency physicians and their non-trained colleagues (the corresponding ratio of the two groups was 1:1 to 1:2) to compare the responses of the two groups and evaluate the effectiveness of training.

The inclusion criteria of participants were as follows: (1) all the subjects were emergency physicians; (2) advanced study at PUMCH of all trainee physicians had to have been undertaken in the past 3 years; and (3) the subjects of the non-training group were colleagues of someone in the CME training group. The exclusion criteria were as follows: (1) individuals with less than 6 months of training; (2) individuals who declined to participate in the survey; and (3) incomplete or duplicate questionnaires, which were automatically excluded by the online questionnaire platform.

We invited 228 emergency physicians to participate in the survey, and a total of 218 questionnaires were collected, with a response rate of 95.6%. The respondents were divided into two groups based on their recent advanced study experience in the emergency department of PUMCH. A series of rare disease CME training sessions were carried out twice a month for physicians in the CME training group during their advanced learning period, while the non-training group did not receive the above training.

#### Research tools, data collection and questionnaire

This questionnaire survey was conducted between April and June 2023 among emergency physicians in China. The separate online data collection questionnaire was conducted through an internet platform (www.wjx.cn), a widely used web-based survey platform in China, which permits centralized data collection and limits repetition by mobile numbers. The questionnaire was developed for this study and has not been previously published elsewhere.

The development of this questionnaire was primarily based on our group's previous national survey on the awareness of rare diseases among emergency department physicians [12]. The conceptual framework was constructed and the initial question pool was selected based on the textbook Rare Disease Medicine [20]. The questionnaire was independently reviewed and voted on by six experts from our research team. Demographic data is objective data, while knowledge and attitude questionnaires are subjective data. Therefore, this project does not involve validation of validity or reliability.

The questionnaire consisted of four groups and 30 questions, including six questions that addressed the respondents' demographic data and 24 items referring to their knowledge and attitudes regarding RDs (Supplementary Qestionnaire). The first group included

six questions referring to demographic information, including sex, career length, hospital level, technical titles, licensing province, and whether respondents had advanced training experience at PUMCH and its duration.

The second group of four questions focused on the emergency physicians' basic knowledge about rare diseases. They were asked about the incidence of rare diseases according to the 2021 China Rare Disease Definition Research Report, whether all RDs are hereditary, and whether there is any national register system for RDs in China. The respondents were then provided with a list of 19 diseases, including 16 rare diseases relatively common in the clinic and three diseases that could easily be mistaken for rare diseases, and they were asked to identify the rare diseases.

The third group included eight questions about the respondents' self-assessment, information access, and needs for rare diseases. They were asked if they had first diagnosed any RDs, the number of RDs they had seen in their careers, and their self-assessment of their knowledge of RDs. Those who had first been diagnosed with an RD were asked to indicate the exact disease from a list of 21 rare diseases. Then, four more questions asked how they perceived their knowledge about RDs, which access route was the most effective, which website they preferred for learning, and if they would like to learn more about RDs.

The last set of questions consisted of four real case analyses (which had a higher incidence and a higher likelihood of emergency in China), each followed by three questions asking respondents to present possible diagnoses, further supporting examinations, and significant differential diagnoses (Table 1).

### Statistical analysis

For data analysis, SPSS 24.0 software (IBM Corp., Armonk, NY, USA) was used. The chi-square test was employed for comparisons involving proportions to determine whether there were significant differences between the training and non-training groups. The independent-sample t- test was utilized for comparisons involving averages to assess any improvement in the physicians' knowledge after the rare disease CME training. For cases where there were significant differences between groups ($p < 0.05$), subgroup analysis was further conducted based on the duration of training to identify if the training effect varied with the duration of the program. This helped explain whether longer or more intensive training had a more significant impact on improving knowledge about rare diseases among emergency physicians.

## Results

### Demographic characteristics

A total of 218 emergency physicians participated in and completed the questionnaire-based survey. They were devided into two groups: the CME training group (108 physicians with training experience at PUMCH in the past three years) and the non-training group (110 colleagues from the same department without training experience at PUMCH in the past three years).

The 218 emergency physicians included were all from China, including 25 of the 34 Chinese provincial administrative regions, with the top five being Hebei (28, 12.8%), Shanghai (27, 12.4%), Henan (25, 11.5%), Guangdong (20, 9.2%), and Heilongjiang (12, 5.5%). Of the respondents, 113 (51.8%) were male, and 105 (48.2%) were female. Most of the physicians (83.9%) were from tertiary A-level hospitals, and there was no significant difference in sex or hospital level between the CME training group and the non-training group ($p = 0.103$ and $p = 0.595$, respectively).

**Table 1** Case analysis section of the questionnaire

| Case | Case information | Possible diagnosis | Further supporting examinations | Major differential diagnosis |
|---|---|---|---|---|
| Case 1 | A young woman with recurrent hyponatremia, epigastric pain, urination, and constipation whose abdominal CT scan showed no clear organic lesions, and the symptoms were related to the menstrual cycle. | Acute porphyria with syndrome of inappropriate antidiuretic hormone | Uroporphyrinogen and uroporphyrin | Lead poisoning |
| Case 2 | A middle-aged woman with hardening and darkening of the skin, limited mouth opening, malignant hypertension, and acute kidney injury. | Systemic sclerosis with renal crisis | SCL-70 | Vasculitis and systemic lupus erythematosus |
| Case 3 | A woman after an abortion with abdominal pain, vaginal bleeding, decreased platelet and haemoglobin levels, progressive elevation of blood creatine, and fragmented red blood cells on the peripheral blood smear. | Atypical haemolytic uraemic syndrome | ADAMTS13 activity and antibodies | Thrombotic thrombocytopenic purpura |
| Case 4 | An elderly man with a history of sarcoma-like carcinoma who had sudden limb convulsions and loss of consciousness, with a head MRI showing multiple white matter hyperintensities in the bilateral basal ganglia and hemioval centre. The results of the lumbar puncture showed that the number of white blood cells and protein content of the cerebrospinal fluid were elevated. | Autoimmune encephalitis | Blood and cerebrospinal fluid anti-neuroantigen antibody testing | Paraneoplastic syndrome |

**Table 2** Demographic characteristics of the emergency physicians

| Characteristics | Total (*n* = 218) | CME Training Group (*n* = 108) | Non-training Group (*n* = 110) | *p*-value |
|---|---|---|---|---|
| **Gender**, ***n*** **(%)** | | | | 0.103 |
| Male | 113 (51.8) | 62 (57.4) | 51 (46.4) | |
| Female | 105 (48.2) | 46 (42.6) | 59 (53.6) | |
| **Hospital level**, ***n*** **(%)** | | | | 0.595 |
| Tertiary A | 183 (83.9) | 92 (85.2) | 91 (82.7) | |
| Tertiary B | 19 (8.7) | 10 (9.3) | 9 (8.2) | |
| Secondary | 16 (7.3) | 6 (5.6) | 10 (9.1) | |
| **Career length** | | | | |
| Average years(x ± s) | 9.06 ± 4.10 | 8.98 ± 3.02 | 9.15 ± 4.95 | = 0.967 |
| **Title**, ***n*** **(%)** | | | | = 0.056 |
| Resident | 22 (10.1) | 8(7.41) | 14 (12.7) | |
| Attending | 155 (71.1) | 79 (73.1) | 76(69.1) | |
| Associate chief physician | 34 (15.6) | 20(18.5) | 14 (12.7) | |
| Chief physician | 7 (3.2) | 1 (0.9) | 6 (7.2) | |

**Table 3** Basic knowledge of rare diseases of emergency physicians

| Items | Total (*n* = 218) | CME Training Group (*n* = 108) | Non-training Group (*n* = 110) | *p*-value |
|---|---|---|---|---|
| **Incidence of RD (according to the 2021 China Rare Disease Definition Research Report)**, ***n*** **(%)** | | | | 0.007* |
| < 1: 1,000 | 16 (7.3) | 6 (5.6) | 10 (9.1) | |
| < 1: 2,000 | 6 (2.8) | 4 (3.7) | 2 (1.8) | |
| ***< 1: 10,000*** | 48 (22.0) | 32 (29.6) | 16 (14.5) | |
| < 1: 100,000 | 66 (30.3) | 35 (32.4) | 31 (28.2) | |
| I do not know | 82 (37.6) | 31 (28.7) | 51 (46.4) | |
| **Are all RDs hereditary**, ***n*** **(%)** | | | | 0.630 |
| Yes | 16 (7.3) | 7 (6.5) | 9 (8.2) | |
| ***No*** | 202 (92.7) | 101 (93.5) | 101 (91.8) | |
| **Is there any national registry system for RD in China**, ***n*** **(%)** | | | | 0.874 |
| ***Yes*** | 92 (42.2) | 45 (41.7) | 47 (42.7) | |
| No | 11 (5.0) | 4 (3.7) | 7 (6.4) | |
| I do not know | 115 (52.8) | 59 (54.6) | 56 (50.9) | |

Abbreviations: *$p < 0.05$. RD: Rare Disease. Correct answers are indicated in bold and italics

Most respondents (79.8%) were experienced emergency physicians with more than five years of work experience and a title of attending physician or above. The career duration and job positions had no significant difference between the two groups (Table 2). In the CME training group, the largest number of physicians had six months of training on rare diseases (76, 70.4%), followed by those with six months to one year (22, 20.4%).

### Basic knowledge of rare diseases

By answering questions about the definition of rare diseases, their genetic characteristics, and the status of rare disease register systems, this study assessed the basic knowledge of rare diseases among emergency physicians. Thirty-two (29.6%) respondents in the CME training group and sixteen (14.5%) respondents in the non-training group were aware of the incidence of rare diseases according to the 2021 China Rare Disease Definition Research Report, and the correct answer rate in the CME training group was significantly higher than that in the non-training group ($p = 0.007$) (Table 3).

The respondents were provided with a list of 19 diseases, including 16 rare diseases and three diseases that could easily be mistaken for rare diseases, and they were asked to identify the rare diseases. The average number of correctly identified rare diseases in the CME training and non-training group were 5.53 types and 4.99 types, respectively, with no significant difference ($t = 1.502$, $p = 0.136$). In the CME training group, the most easily identified rare diseases were porphyria (63, 58.3%), Castleman disease (58, 53.7%), POEMS syndrome (56, 51.9%), Langerhans cell histiocytosis (56, 51.9%), and hepatolenticular degeneration (48, 44.4%), while the least easily identified diseases were haemophilia (15, 13.9%), generalized myasthenia gravis (15, 13.9%), and autoimmune encephalitis (18, 16.7%). Similarly, the most easily identified rare diseases in the non-training group were porphyria (56, 50.9%), Castleman disease (54, 49.1%),

**Table 4** Identification of rare diseases by emergency physicians

| Rare Diseases, *n* (%) | Total (*n* = 218) | CME Training Group (*n* = 108) | Non-training Group (*n* = 110) |
|---|---|---|---|
| ***Idiopathic pulmonary arterial hypertension*** | 47 (21.6) | 26 (24.1) | 21 (19.1) |
| ***Idiopathic pulmonary fibrosis*** | 37 (17.0) | 20 (18.5) | 17 (15.5) |
| ***IgG4-related disease*** | 90 (41.3) | 45 (41.7) | 45 (40.9) |
| ***Lymphangioleiomyomatosis*** | 70 (32.1) | 39 (36.1) | 31 (28.2) |
| ***Marfan syndrome*** | 92 (42.2) | 39 (36.1) | 53 (48.2) |
| ***Multiple sclerosis*** | 43 (19.7) | 23 (21.3) | 20 (18.2) |
| Multiple myeloma | 21 (9.6) | 11 (10.2) | 10 (9.1) |
| ***POEMS syndrome*** | 105 (48.2) | 56 (51.9) | 49 (44.5) |
| ***Porphyria with SIADH*** | 119 (54.6) | 63 (58.3) | 56 (50.9) |
| ***Atypical hemolytic uremic syndrome*** | 58 (26.6) | 32 (29.6) | 26 (23.6) |
| ***Autoimmune encephalitis*** | 44 (20.2) | 18 (16.7) | 26 (23.6) |
| Systemic lupus erythematosus | 10 (4.6) | 3 (2.8) | 7 (6.4) |
| ***Castleman disease*** | 102 (46.8) | 58 (53.7) | 54 (49.1) |
| ***Generalized myasthenia gravis*** | 33 (15.1) | 15 (13.9) | 18 (16.4) |
| Acromegaly | 27 (12.4) | 16 (14.8) | 11 (10.0) |
| ***Hemophilia*** | 29 (13.3) | 15 (13.9) | 14 (12.7) |
| ***Hepatolenticular degeneration*** | 88 (40.4) | 48 (44.4) | 40 (36.4) |
| ***Alveolar proteinosis*** | 74 (33.9) | 44 (40.7) | 30 (27.3) |
| ***Langerhans cell histiocytosis*** | 105 (48.2) | 56 (51.9) | 49 (44.5) |

Rare diseases are indicated in bold and italics

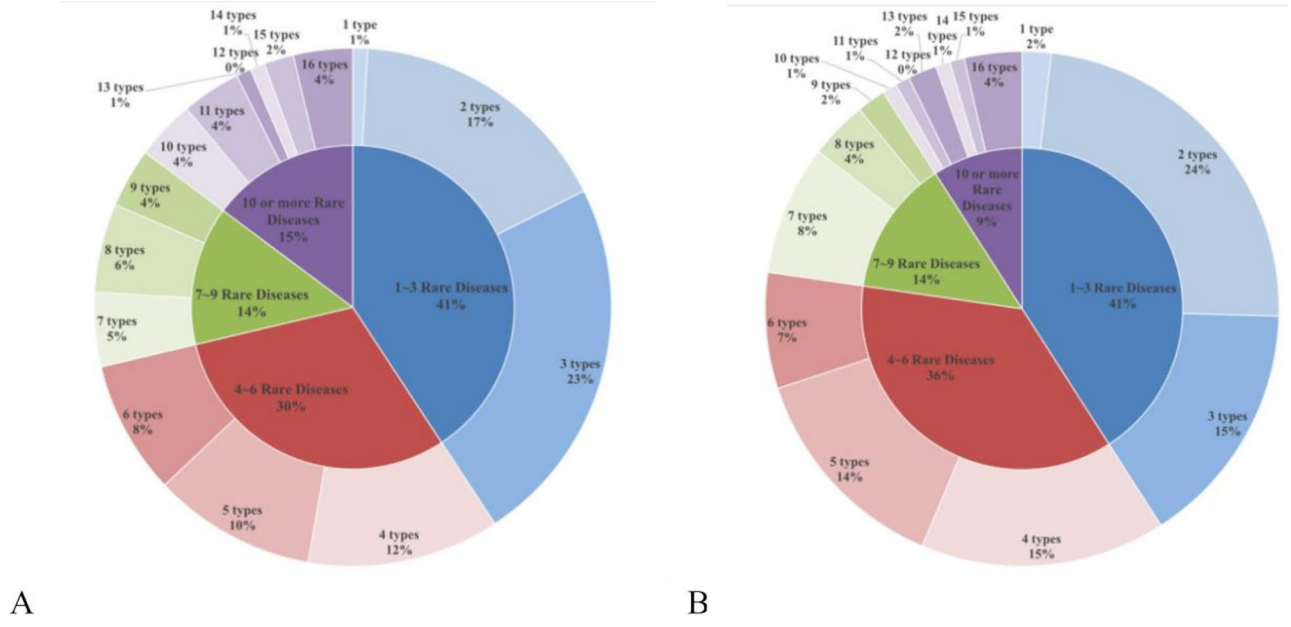


**Fig. 2** Identification of rare diseases by emergency physicians. (**A**) Distribution of the number of rare diseases correctly identified in the CME training group. (**B**) Distribution of the number of rare diseases correctly identified in the non-training group

Marfan syndrome (53, 48.2%), Langerhans cell histiocytosis (49, 44.5%), and POEMS syndrome (49, 44.5%), and the least identified diseases were haemophilia (14, 12.7%), idiopathic pulmonary fibrosis (17, 15.5%), and generalized myasthenia gravis (18, 16.4%). Acromegaly was the most likely misidentified disease in both groups (14.8% and 10.0%, respectively) (Table 4). We further divided the number of correctly identified rare diseases into different intervals and compared the distribution of respondents in each interval between the two groups (Fig. 2), finding no significant difference between the groups ($p = 0.562$).

### Self-assessment, information access and needs of rare diseases

We assessed emergency physicians' perceptions of rare diseases by investigating whether they had first diagnosed a rare disease in a patient, how many rare diseases they had seen in their careers and their self-assessment

of the knowledge of rare diseases. Most respondents had never diagnosed RD cases (180, 82.6%) and considered their knowledge to be insufficient or minimal (214, 98.2%). Among physicians who had diagnosed an RD, the most recent diseases were autoimmune encephalitis (13,6.25%), porphyria (12, 5.76%), and multiple sclerosis (11, 5.29%). Compared to the non-training group, physicians in the CME training group experienced a higher number of rare diseases in their careers ($p = 0.047$), but there was no significant correlation with the duration of training attended ($p = 0.096$) (Table 5).

Regarding ways to learn about rare diseases, the non-training group was more inclined to learn about them through medical school education and clinical work

**Table 5** Self-assessment, information access and needs of rare diseases according to emergency physicians

| Items | Total ($n = 218$) | CME Training Group ($n = 108$) | Non-training Group ($n = 110$) | *p*-value |
|---|---|---|---|---|
| **Have you first diagnosed an RD in a patient, *n* (%)** | | | | 0.136 |
| Yes | 38 (17.4) | 23 (21.3) | 15 (13.6) | |
| No | 180 (82.6) | 85 (78.7) | 95 (86.4) | |
| **How many RDs have you seen in your career, *n* (%)** | | | | **0.047*** |
| 1 ~ 5 types | 167 (76.6) | 75 (69.4) | 92 (83.6) | |
| 6 ~ 10 types | 40 (18.3) | 26 (24.1) | 14 (12.7) | |
| > 10 types | 11 (5.0) | 7 (6.5) | 4 (3.6) | |
| **How well do you know about RDs, *n* (%)** | | | | 0.147 |
| Well or pretty much | 4 (1.8) | 1 (0.9) | 3 (2.7) | |
| Insufficiently | 82 (37.6) | 35 (32.4) | 47 (42.7) | |
| Minimally | 132 (60.6) | 72 (66.7) | 60 (54.5) | |
| **How did you learn about RDs, *n* (%)** | | | | |
| Studying in medical school | 121 (55.5) | 44 (40.7) | 77 (70.0) | **< 0.001*** |
| Browsing RD websites | 42 (19.3) | 16 (14.8) | 26 (23.6) | 0.079 |
| Working in clinic | 129 (59.2) | 53 (49.1) | 76 (69.1) | **0.001*** |
| Advancing training in other hospitals | 94 (43.1) | 93 (86.1) | 1 (0.9) | **< 0.001*** |
| Attending academic conferences | 119 (54.6) | 58 (53.7) | 61 (55.5) | 0.621 |
| Others | 17 (7.8) | 6 (5.6) | 11 (10.0) | 0.199 |
| Never heard of them | 7 (3.2) | 2 (1.9) | 5 (4.5) | 0.457 |
| **Which access was most effective for you, *n* (%)** | | | | **< 0.001*** |
| Studying in medical school | 25 (11.5) | 3 (2.8) | 22 (20.0) | |
| Browsing RD websites | 4 (1.8) | 0 (0.0) | 4 (3.6) | |
| Working in clinic | 69 (31.7) | 22 (20.4) | 47 (42.7) | |
| Advancing training in other hospitals | 79 (36.2) | 72 (66.7) | 7 (6.4) | |
| Attending academic conferences | 33 (15.1) | 9 (8.3) | 24 (21.8) | |
| Others | 8 (3.7) | 2 (1.9) | 6 (5.5) | |
| **Which websites do you prefer to use to learn more about RDs, *n* (%)** | | | | |
| PubMed | 83 (38.1) | 40 (37.0) | 43 (39.1) | 0.714 |
| Baidu | 119 (54.6) | 49 (45.4) | 70 (63.6) | **0.005*** |
| Wikipedia | 19 (8.7) | 7 (6.5) | 12 (10.9) | 0.238 |
| Uptodate | 106 (48.6) | 78 (72.2) | 28 (25.5) | **< 0.001*** |
| Rare disease specialist website | 0 (0.0) | 0 (0.0) | 0 (0.0) | - |
| DXY | 115 (52.8) | 58 (53.7) | 57 (51.8) | 0.834 |
| CNKI or Weipu or WanFang | 56 (25.7) | 23 (21.3) | 33 (30.0) | 0.130 |
| I do not want to know about them | 3 (1.4) | 1 (0.9) | 2 (1.8) | 1.000 |
| **Which aspect of RDs do you prefer to learn more about, *n* (%)** | | | | |
| Practice guidelines or consensus | 190 (87.2) | 96 (88.9) | 94 (85.5) | 0.665 |
| Relevant professional websites | 119 (54.6) | 56 (51.9) | 63 (57.3) | 0.335 |
| A hospital or specialist that I can refer to | 140 (64.2) | 78 (72.2) | 62 (56.4) | **0.021*** |
| Material or official account to disseminate to patients or their family | 83 (38.1) | 40 (37.0) | 43 (39.1) | 0.674 |
| A hospital or specialist for genetic counselling | 83 (38.1) | 44 (40.7) | 39 (35.5) | 0.483 |
| Methods of conducting relevant research and publishing articles | 52 (23.9) | 25 (23.1) | 27 (24.5) | 0.750 |
| I do not want to learn about them | 4 (1.8) | 1 (0.9) | 3 (2.7) | 0.627 |

Abbreviations: *$p < 0.05$. RD: Rare Disease

experiences ($p < 0.001$ and $p = 0.001$, respectively). In contrast, the CME training group was more likely to learn through advancing training at other hospitals ($p < 0.001$). In addition, some respondents in each group obtained relevant knowledge through browsing rare disease professional websites, attending academic conferences, and other ways. The diversity comparison of information acquisition between two groups of emergency physicians is shown in Fig. 3A. However, the two groups had no significant difference in the variety or richness of ways to learn about rare diseases. The most impressive access to rare disease learning in the CME training group was advanced training (72, 66.7%), while in the non-training group, it was clinical work (47, 42.7%). There was a significant difference in the most effective type of access between the two groups ($p < 0.001$) (Table 5).

Currently, the Internet has become an essential way for clinicians to obtain knowledge, so we also investigated the habits of emergency physicians who used the Internet to learn about rare diseases. As the results showed, the CME training group was more willing to use UpToDate ($p < 0.001$), while the non-training group used Baidu at a higher rate ($p = 0.005$). In addition, the two groups of respondents had a similar proportion of using Chinese medical forums such as DXY (www.dxy.cn) or domestic and foreign databases such as CNKI (https://chkd.cnki.net/), Weipu (https://wwwv3.cqvip.com/), Wanfang (https://www.wanfangdata.com.cn/), and PubMed/Medline. However, none of the respondents used rare disease professional websites (Table 5). The two groups had no significant difference in the number of websites they chose to use (Fig. 3B).

The responses to information needed for rare diseases were similar in both groups (Fig. 3C), except that more physicians in the CME training group wanted to know about hospitals or specialists to which they could refer patients ($p = 0.021$). The vast majority of emergency physicians (214, 98.2%) wanted to learn more about rare diseases, with the most urgent need being practice guidelines or consensus (190, 87.2%), followed by hospitals or specialists that they could refer patients to (140, 64.2%) and relevant professional websites (119, 54.6%) (Table 5).

### Case analysis of rare diseases

The questionnaire set up four case studies to objectively evaluate the knowledge of rare diseases in the two groups by making the correct diagnosis, indicating the main auxiliary examinations, and identifying the principal differential diagnosis. In case one, 90 (83.3%) respondents in the CME training group made the correct diagnosis, and 87 (80.6%) of them opted for the corresponding auxiliary tests, which was a higher proportion than the 74 (67.3%) and 71 (64.5%) respondents in the non-training group, respectively (Fig. 4A). In case two, there was no significant difference between the two groups in the distribution of answers regarding diagnosis, auxiliary

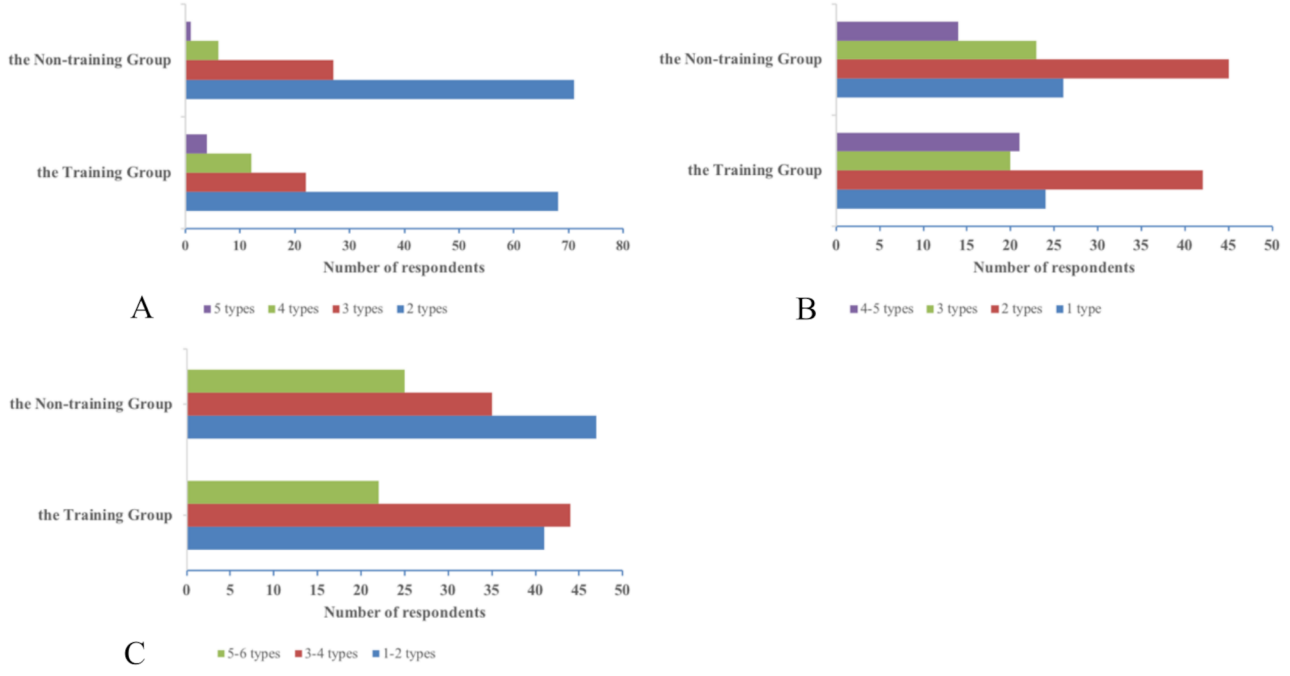


**Fig. 3** Information access and needs of rare diseases according to emergency physicians. (**A**) Distribution of the number of types of information access in both groups. (**B**) Distribution of the number of websites used for rare disease knowledge in both groups. (**C**) Distribution of the number of information needs in both groups

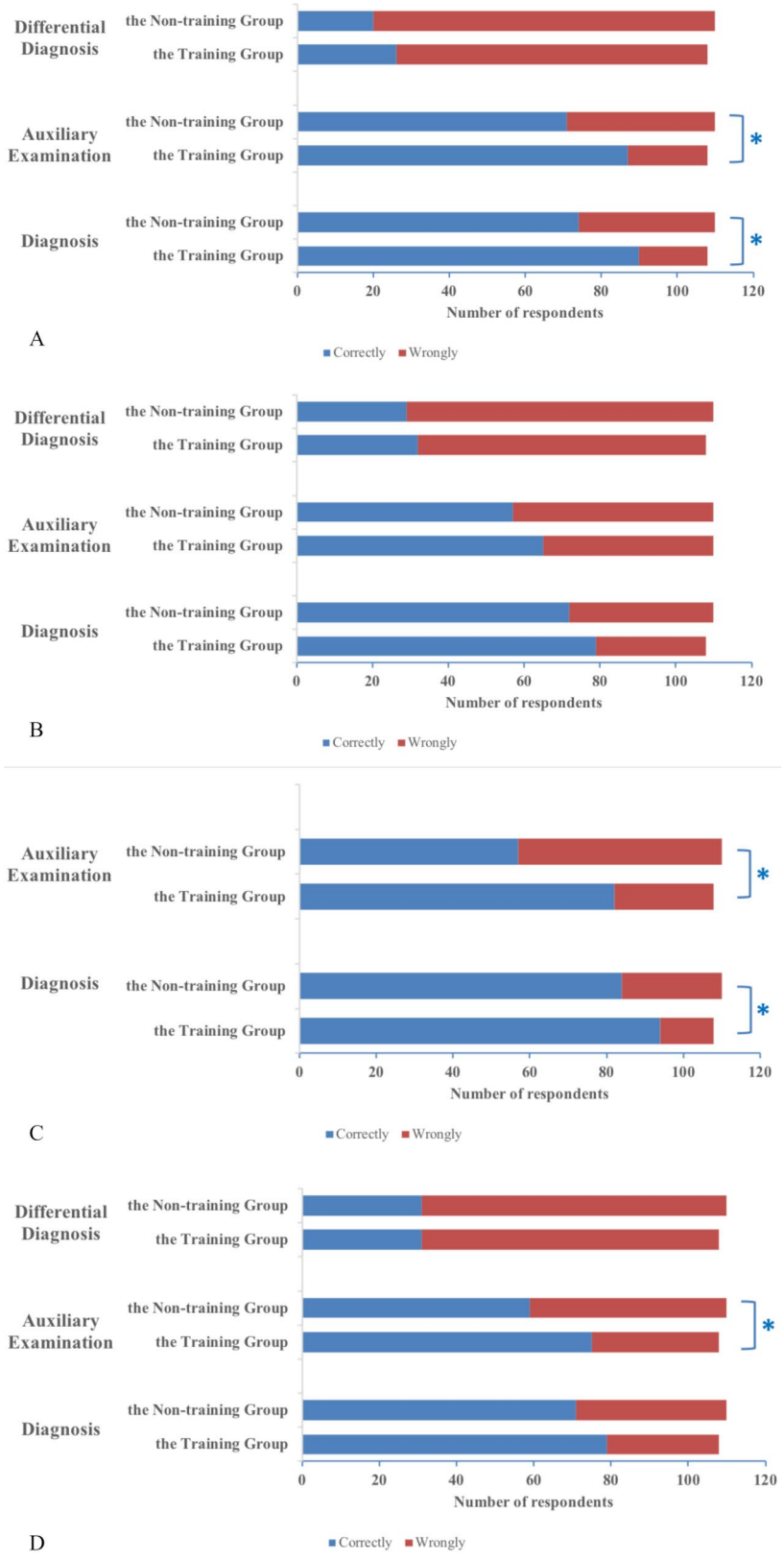


**Fig. 4** Answers to case analysis of rare diseases according to emergency physicians. (**A**) Distribution of answers to Case 1, acute porphyria. (**B**) Distribution of answers to Case 2, systemic sclerosis with renal crisis. (**C**) Distribution of answers to Case 3, atypical hemolytic uremic syndrome. (**D**) Distribution of answers to Case 4, autoimmune encephalitis

examinations, or differential diagnosis (Fig. 4B). In case three, 94 (87.0%) respondents in the CME training group chose the correct diagnosis, and 82 (75.9%) knew the auxiliary examinations to be performed, which was a higher proportion than 84 (76.4%) and 57 (51.8%) in the non-training group, respectively (Fig. 4C). In case four, 75 (69.4%) respondents in the CME training group selected the correct auxiliary tests, which was more than 59 (53.6%) in the non-training group (Fig. 4D). Overall, the CME training group responded better to case analysis than the non-training group.

## Discussions

Previous studies have highlighted the need for physicians across countries and specialties to gain knowledge about rare diseases(RDs). For instance, Walkowiak et al. found that while nearly half of physicians in Kazahstan claimed to have taken RD classes, most still felt their knowledge was insufficient [23]. Similarly, a survey by Kuhne et al. among German dentists revealed that most had little or no knowledge of RDs [24]. Despite this, many healthcare professionals have expressed interest in further training. In Poland, over 80% of physicians surveyed wanted to expand their RD knowledge, and 76.3% believed a compulsory RD course should be part of medical education [25]. In our study, 98.2% of emergency physicians acknowleged their insufficient RD knowldge and expressed a strong desire to learn more, especially regarding practice guidelines.This is crucial because patients with RDs often present to emergency department with life-threatening or undiagnosed symptoms that seriously impact their quality of life. In China, the emergency room is often the first stop of medical contact for RD patients. Therefore, training emergency physicians on RDs is essntial to meet clinical demands and address the needs of most emergency doctors.

Emergency physicians initially had insufficient knowledge of RDs. However, after a series of CME training courses, their understanding of RDs significantly improved. Physicians in the CME training group were more knowledgable about RD incidence, had encountered more RD types, and could more accurately identify RDs based on clinical manifestations and suggest further examinations. Most agreed that advanced learnning through CME training at PUMCH was ideal, highlighting the importance of exposure to real RD cases in clinical practice.

Our questionnaire results indicated that younger physicians in the non-training group believed they learned more about RDs in medical school. This could be due to the rapid development of RD education in recent decades. In China, over 30 RDs were added to undergraduate textbooks after 2006. For example, Hebei Medical University launched an elective RD course for undergraduate students in 2019 [26], and PUMCH started offering graduate elective RD courses in 2020 [20]. In our questionnaire, 18 RDs were mentioned, with 14 appearing in undergraduate textbooks by 2018. The CME training group found that advanced study experiences in other hospitals were effective for learning about RDs. After training, they surpassed the non-training group in RD awareness, further demonstrating the effectiveness of such training.

Previous research has explored the efficacy of training programs for RDs. Groft et al. suggested that various training formats, including didactic lectures, case-based studies, small group discussions, and bedside teaching, could enhance learners understanding of fundamental pathophysiologic principles and imprvoe theire ability to identify RDs in uncommon clinical scenarios [27]. However, their conclusions lacked supporting data. Ramalle-Gómara et al. conducted a survey where 83.6% of respondents who had participated in RD courses reported finding them useful [28]. This finding was based on subjective feedback and did not compare knowledge or skills before and after training. Flores et al. found that students and physicians who had received RDs training scored higher on survey focused on basic RD knowledge [29]. More recently, Regier et al. demonstrated that RD training improved knowledge content by comparing participants' responses before and after the Rare Disease Research Scholar Program [30]. However, this study did not evaluate the program's impact on clinical diagnosis and treatment. Our study innovatively addressed these gaps. We administered a feedback questionnaire to physicians following a series of RD traning courses and assessed the training's effectiveness. By introducing case analysis and combining it with general RD knowledge, we compared the CME training group with the non-training group. This approach provided a comprehensive refelction of the training's impact on RD information mastery. Our findings highlighted the importance of RD training in enhancing clinical knowledge and its potential to improve patient outcomes.

Germany and the United States have distinct approaches to rare disease education. Germany's system is highly structured, with a national plan that includes centers of expertise and multidisciplinary care. These centers provide specialized training for healthcare professionals, focusing on early diagnosis and comprehensive management [19, 31]In contrast, the U.S. model is diverse and innovative, with a robust network of research institutions and clinical centers. The Rare Diseases Clinical Research Network (RDCRN) offers extensive resources like online courses, seminars, and research funding, emphasizing patient-centered care and advanced technologies [3, 32]. The CME program at PUMCH combines elements of both international models while offering

unique strengths. It integrates a comprehensive curriculum with lectures, case discussions, bedside teaching, and multidisciplinary consultations, ensuring thorough training in theory and practice. Similar to Germany, it emphasizes practice-oriented training through direct patient care, enhancing clinical proficiency. Like the U.S., it fosters multidisciplinary collaboration, crucial for managing rare diseases. Additionally, the PUMCH program focuses specifically on clinical manifestations and diagnostic skills, vital for emergency physicians to recognize rare diseases. It also continuously assesses and refines its content through online questionnaires and feedback, setting it apart in the international context.

A previous survey among Malaysian physicians revealed that their low awareness of rare diseases (RDs) was primarily due to the small proportion of RD cases in their clinics or hospitals [33]. A similar situation exists in China, where the uneven distribution of medical resources means that many local and secondary hospitals rarely encounter RD cases. According to the 2014–2015 hospitalization summary reports for 281 rare diseases, the cities with the most RD cases and types were predominantly first-tier or second-tier large cities such as Beijing, Guangzhou, Shanghai, Nanning, and Chengdu [34]. The distribution of tertiary A-level hospitals is a key factor influencing the accessibility of medical resources for rare diseases [10]. Given that the internet has become the leading resource for RD information [25, 35, 36], online courses and e-learning programs should be vigorously promoted. Additionally, cooperation between tertiary A-level hospitals and local hospitals should be encouraged to provide more doctors with training opportunities and resources on rare diseases. This aligns with the needs of most physicians. For example, a survey by Rohani-Montez et al. among 978 clinicians from 16 specialties found that the most preferred RD education formats included comprehensive online learning platforms with current education and resources, as well as case-based, text-based, and short formats [37].

Our study highlights the potential for nationwide training to enhance emergency physicians' awareness of rare diseases (RDs) and address diagnostic and treatment complexities. Integrating RD education into medical school curricula and the national CME framework is essential. We plan to launch a teaching platform using real RD cases and share resources with hospitals across China. Additionally, we will establish teaching alliances with hospitals hosting trainee physicians and conduct regular online RD case discussions. We will also improve our hospital's RD CME training by inviting experts from other specialties to share cases and give lectures. Our research shows that exposure to actual cases leaves a stronger impression on physicians. Many RD patients are highly engaged in learning about their conditions, making patient-led training programs a valuable possibility. For example, the United States' RARE Compassion Program pairs medical students with RD patients to increase exposure and understanding, achieving positive results [22].

The study revealed that training did not significantly increase the variety of learning resources or awareness of professional RD websites. This may be due to the training's focus on case analysis and diagnosis, or because the older CME group was less familiar with the internet. Additionally, the CME course did not cover using online resources. Future training will integrate online research databases and resource navigation courses. In the case analysis section, the CME group did not outperform the non-training group in differential diagnosis, indicating that RD CME training needs further refinement and systematization. Training should not only cover basic RD knowledge but also emphasize collecting diagnostic information and distinguishing RDs from common diseases. Overall, RD CME training has significant room for improvement and requires further research.

## Limitations

This online questionnaire-based study has several limitations. First, responses might have slightly exaggerated emergency physicians' knowledge of RD, as the anonymous and untimed format allowed them to seek external information. Second, some questions were subjective, such as self-assessments of understanding of RD, lacking fully objective data. Third, participants, especially in the non-training group, may have been more interested in RD research, potentially skewing results and limiting representativeness. Fourth, the online survey structure may have deterred some respondents who felt uncomfortable with completing the online questionnaire, introducing selection and information biases. Additionally, this non-prospective study lacked pre-CME data, precluding before-and-after comparisons. The sample size was also relatively small, with only 218 emergency physicians included. We propose to follow up with further research to address these issues. Research in the field of rare disease CME training is gradually attracting attention, and more detailed and comprehensive research is still needed. Moreover, we did not verify the reliability and effectiveness of the questionnaire, which is an important limitation.

## Conclusions

Emergency physicians typically had insufficient knowledge of rare diseases, but after a series of training sessions, their awareness of RDs greatly increased in terms of fundamental knowledge and disease diagnosis. Similar training projects can be extended nationwide, and online courses can be promoted to increase emergency

physicians' awareness of rare diseases and solve the problem of the difficult diagnosis and treatment of RD patients.

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**Author contributions**
AL distributed and filled out the questionnaires. WH conducted statistical analysis of the results and wrote. JY designed the research scheme. HY designed the research scheme. JX drafted the article and revised it. HZ drafted the article and revised it. All authors approved the final Manuscript.

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**Data availability**
The data that support the findings of this study are available on request from the corresponding authors upon reasonable request.

## Declarations

**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 Declaration of Helsinki 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 online could participate in this study.

**Consent for publication**
Not applicable.

**Competing interests**
The authors declare no competing interests.

**Clinical trial number**
Not applicable.

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## Publisher's note