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Original article

Exploring Healthcare Practitioners' Knowledge of Rare Diseases in Cameroon

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Abstract

Rare diseases (RD) are complex medical incidents that constitute a burden for health systems globally. In Cameroon, the absence of a national framework for RDs means that very little is known about these diseases. Many health professionals due to the rarity of cases and complexity of symptoms, face challenges in detecting and treating them. The aim of this paper is to examine the knowledge of Cameroonian physicians on RDs in order to highlight the medical constructions of RDs in Cameroon. The study is cross-sectional and associates quantitative and qualitative research approaches with 23 health practitioners who responded to an open-ended questionnaire to (1) evaluate their knowledge of RDs, (2) describe the medical discourse on RDs in Cameroon and (3) identify prevailing RDs in Cameroon. The study showed on the one hand that, health professionals have strong theoretical knowledge of RDs. About 40.9% know that an RD is one that has a very low prevalence. 73.1% have come across a case of RD in their practice. RDs practitioners indicated they have encountered in their practice were mainly Down syndrome, hemophilia, and G6PD. 81% say they have difficulties detecting an RD. On the other hand, there are certain health practitioners who view that RDs are not (yet) a public health concern in Cameroon when there is a rise of non-communicable diseases like high blood pressure, diabetes, etc. Treatment strategies highlighted were mainly multidisciplinary consults, surgery, and specialized care. The study suggests that decision makers and practitioners should design strategies to strengthen expertise for the diagnostic and management of RDs in Cameroon. Also, health practitioners should be trained regularly on how to manage RDs.

Keywords: ***Healthcare practitioner, knowledge, rare diseases, Cameroon***

Introduction

RDs are complex medical incidents that constitute a burden for health systems globally. These conditions are serious, chronic, and disabling, often with a potentially fatal diagnosis. The profound prejudice they cause to patients and their families cause them to go through discrimination and rejection with many living in isolation. According to some authors: « Rare diseases (RDs) are numerous, heterogeneous in nature, and geographically disparate. Few are preventable or curable, most are chronic, and many result in early death » (Nguengang Wakap et al., 2020). Additionally: « They are first manifest in the earliest years of life in more than 50% of cases and are responsible for more than 30% of infant mortalities ». (Boy-Lefevre, 2014, p.2). Despite these facts, there is no visibility of these diseases in Cameroon and there exists no official definition of what an RD is, making the issue ambiguous and difficult to master. Also, in Cameroon, there exists no national registry of RDs that could go a long way to facilitate public decisions. Persons living with a rare disease (PLWRD) face a number of challenges that make them have a diminished quality of life (QoL). QoL is a very subjective term that is only definable in this case by those whose living conditions are infringed upon in some way by an RD. In effect, they encounter late diagnosis, misdiagnosis, the inexistence of treatments, and the incurability of their disease. Many will go to the hospital several times, run a myriad of tests before their condition is finally deciphered. Even after going through huge medical expenses, in a majority of cases, their condition may not even be diagnosed due to gaps in knowledge of their condition.

RDs are extremely rare conditions and therefore pose novel challenges to the medical art and create new barriers for patients. In fact: « the lack of knowledge, misdiagnoses, missed diagnoses and incurability have posed formidable obstacles to physicians and patients in effective diagnosis and treatment » (Ni and Shi, 2017). Therefore, it is justified to say that: « Patients with rare diseases are the orphans of health systems, often without diagnosis, without treatment, without research, and therefore without reason to hope » (Behcet's Syndrome Center, 2013). However, how healthcare professionals describe these diseases can influence the social status of RDs and also influence public decision about these diseases. This calls for an increased literacy of practitioners on RDs especially the general practitioner who has a very determining role in the health system because he is the first point of contact, the primary care doctor who is the first contact with patients and disease. Therefore, in the fight to improve the status of RDs in health systems and policies, careful attention should be laid on how RDs are described or viewed by health professionals as a disease entity is the product of medical discourse. This means that the medical discourse on a disease is very determinant of the status this will be given in society (Turner, 1995). This study therefore offers an ideal vantage point to highlight the medical discourses on RDs in Cameroon while

describing how this discourse can constitute a barrier to the proper management of RDs.

Method

The study was a cross-sectional study that associated both qualitative and quantitative research designs. Data was collected from January to December 2022 in Cameroon. Medical doctors practicing in Cameroon and of all ages regardless of gender were considered for this study. A questionnaire was digitalized and disseminated on various social network platforms like Facebook, WhatsApp, Twitter, etc. Cameroonian medical doctors working out of Cameroon were excluded from the study. A total of 23 participants whose ages ranged between 26 and 49 took part in this study. The mean age of participants was 31. Quantitative data was reported using descriptive statistics, including percentage and mean while qualitative data was transcribed and analysed verbatim.

Results

I. Sociodemographic profile of informants

17 (73.9%) respondents were general practitioners while 5 (21.7%) were specialists. 1 was under specialization. The majority work in private hospitals, that is 9 health workers (39.1%), 8 (3.4%) work in public health facilities, 5 (21.7%) at the central level of the Ministry of public health, and 1 (4.3%) in a confessional hospital. Concerning the experience of these health professionals, 17 have work experience that range from 1-5 years, 4 have work experience that ranges from 6-10 years, and 2 a work experience of 10 years and more. Their ages ranged from 24 to 49 years; 14 (60.9%) had an age that ranged between 24 -30 years while 9 (39.1%) were aged 31 years and above. 12 (52%) participants were males while 11(48%) were females. Possession of pertinent knowledge or not did not correlate with these variables and this will be illustrated in the detailed presentation of results. The table below illustrates the sociodemographic profile of informants.

Table 1: Demographic characteristics of study participants

Variable	**Frequency**	**Percentage**
Age		
24 – 30	14	60.9%
31 and more	9	39.1%
Gender		
Male	12	52%
Female	11	48%
Grade		
General practitioner	17	73.9%
Specialist	5	21.7%
Under Specialization	1	4.3%
Years of experience		
1-5 years	17	74%
6-10 years	4	17.3%
10 years and more	2	9%
Place of work		
Central level services (Ministry of Public Health)	5	21.7%
Public health facility	8	34.8%
Private health facility	9	39.1%
Confessional health facility	1	4.3%

II. Health literacy of health professionals on RDs

We refer to pertinent knowledge on RDs as health literacy of health professionals on RDs. We therefore analyze in this section the knowledge health professionals have on RDs in order to establish if this enables them to handle RDs adequately. In our study we observed that a few physicians consider a disease to be rare when its occurrence is rare not because of its aetiology but because it has been kept under control so much so that it is difficult to come across it in the general population. It is the case of an informant who considers tetanus and rabies to be an RD (4.5%). This poses the problem of having an official definition of RDs per country. Indeed, RDs are defined differently from one geographical area to another but in the absence of an official definition of a health event, the definition used is that of the World Health Organization (WHO). The health literacy of health professionals on RDs is relatively homogenous among Cameroonian health professionals. A majority of health professionals are knowledgeable about RDs and define an RD to be one with a very low prevalence (40.9%) while 18.2% consider rare a disease with a prevalence of less than 0.05% and 9.1% consider a disease to be rare when it affects 1/2000 individuals and one whose occurrence in a population is low with its origins being most often of a genetic nature. This shows strong theoretical knowledge of RDs by practitioners as illustrated in the figure below.

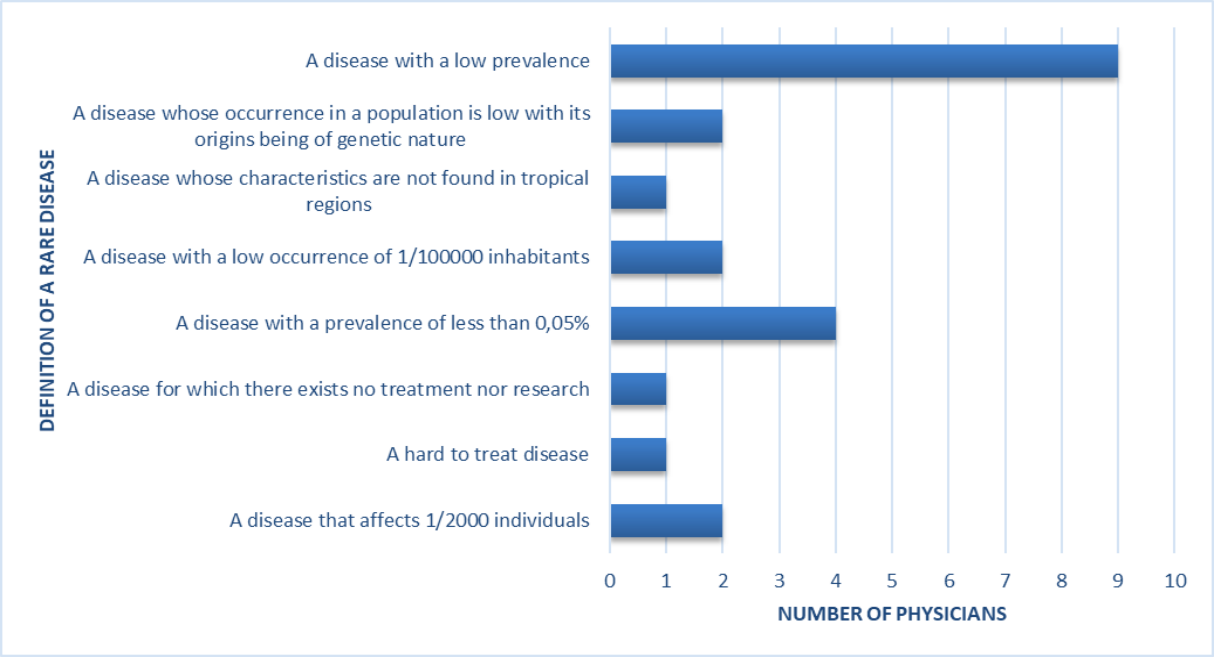

Figure 1: Definition of rare diseases by study participants

III. The medical discourse on RDs in Cameroon

RDs are not common and in most cases, there is no clear knowledge of their occurrence in society. In fact, the true burden of RDs in Cameroon is difficult to estimate, since epidemiological data for most of these diseases are not available. This explains why most RDs attract stigma and rejection. Families go into hiding to protect themselves from going through numerous accusations and ill-treatment from society. The immediate effect of this is that, these conditions become extremely rare in the hospital milieu making it difficult for health professionals to accumulate substantial knowledge and expertise on these diseases. This becomes very determinant of the medical discourse that eventually surrounds RDs in Cameroon. The fact that health professionals do not come across most of these diseases in their daily practice make them to view them as "rare" and uninteresting. Health professionals showed more enthusiasm for diseases that have very high incidences like hypertension and diabetes considering them as pertinent public health priorities. According to them, these are health conditions that need to be invested. However, we noticed that a few health professionals are in favor that RDs should also be viewed as an issue of public health concern because of the burden these diseases cause to families. Another aspect that motivates some health professionals to consider RDs as a health priority in Cameroon is the will to master these diseases that challenge scientific knowledge. That is why, one practitioner in support that RDs should be included in the agenda of important health events sees that: « It will enable monitor their incidence in the general population over a long period and take timely measures to prevent them » PHY (1) while another informant adds that mastery of RDs « will help improve knowledge on RDs and anticipate on the outcome of children and prepare parents to live with a child with a rare disease or a spouse with a rare disease » PHY (3).

On the other hand, some health professionals view that these diseases cannot (yet) be considered as public health concerns in Cameroon because there is very little information on their incidence and prevalence in the general population. For many physicians, RDs are not a major priority that could lead to the elaboration of important health interventions. In the words of some informants: « They are not a priority because we have to handle other health issues, specifically non-communicable diseases like high blood pressure, diabetes, drug addiction, etc that are on a constant rise in our country» PHY (9). Therefore, the point of interest here is the prevalence factor which is still unknown in Cameroon. In fact, for this practitioner RDs have a: « Low prevalence and the presence of other diseases of high prevalence that need to be integrated in public health priority programs do not give a platform for RDs » PHY (4). The medical discourse on diseases contributes to defining RDs as uninteresting and also implicitly suggests that RDs are 'rare' and inexistent in Cameroon.

IV. Cataloging prevailing rare diseases in Cameroon

RDs are numerous and unknown to many health professionals. The number of years of practice do not have any significance with a health professional's capacity to identify a rare condition. In fact, it is the absence of a database or national registry of RDs in the country that poses the greatest challenge. There is very little knowledge of the sociodemographic and epidemiological characteristics of RDs in Cameroon. This makes it difficult to draw an action plan for the management of these diseases. In our study, we however asked health practitioners to tell us what RDs they have come across in their years of practice in order to have an insight of prevailing RDs in Cameroon. A few health professionals have not come across an RD in their practice, about 26.9% while 73.1 % have come across an RD in their practice. From the table below, diseases that were commonly seen by practitioners were Down syndrome, hemophilia, blood cancer, and G6PD deficiency.

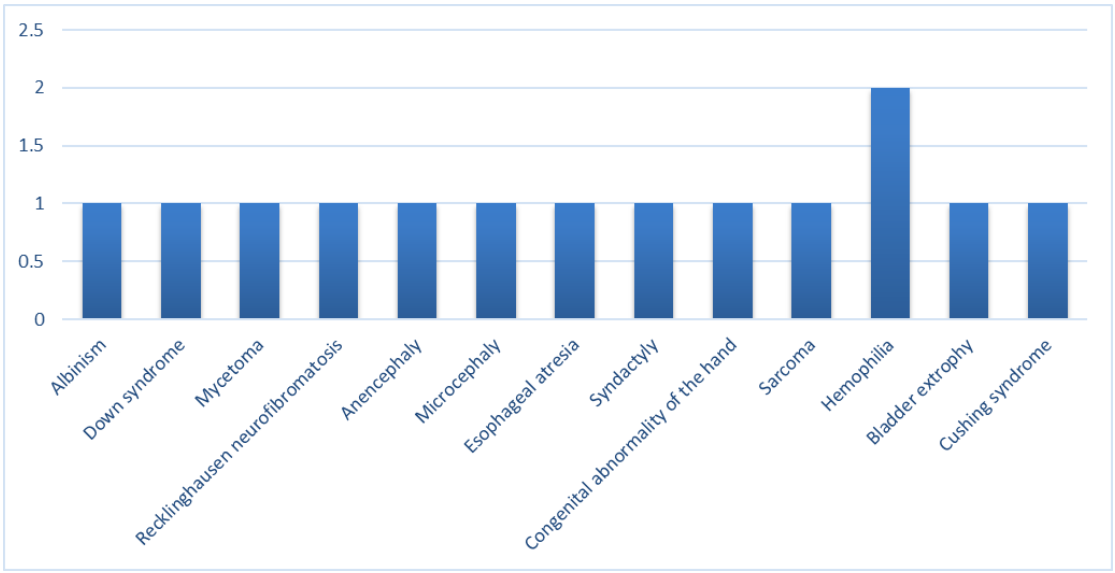

Figure 2: Diseases experienced by study participants

On the other hand, we equally asked our informants to say which diseases they think are the most prevailing in the hospital milieu in Cameroon. Down syndrome (11.1%) has the highest ranking followed by hemophilia (8.3%) and blood cancers then G6PD deficiency (5.5%) while 11.1% do not have an idea of prevailing RDs in Cameroonian hospitals.

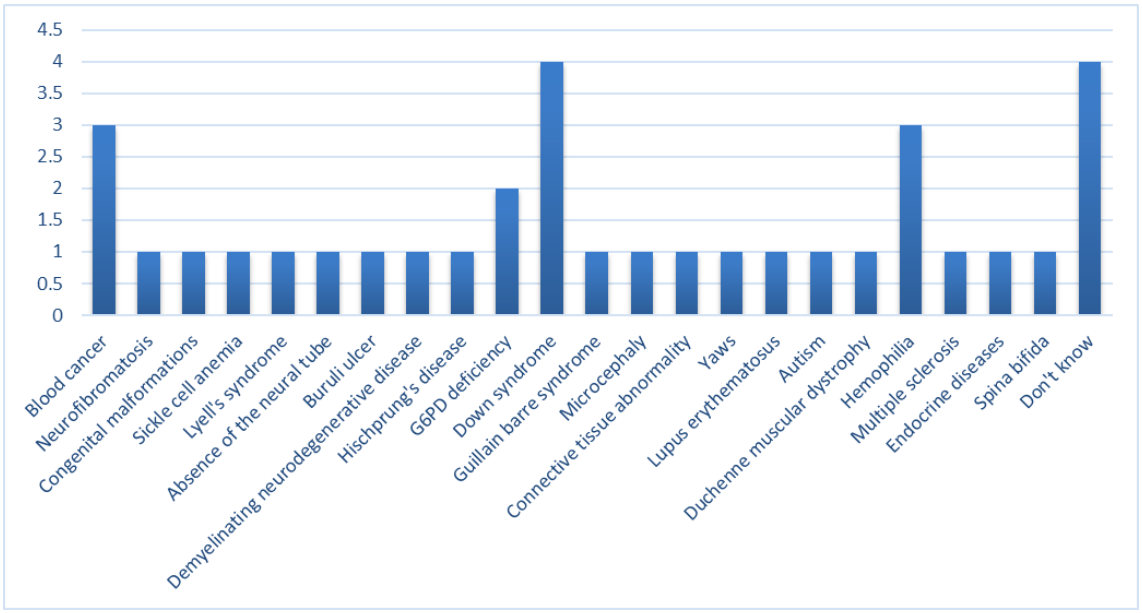

Figure 3: Diseases identified as prevailing in Cameroon hospitals by study participants

The figure above, shows similarities with the diseases practitioners declare they have come across in their practice, especially for hemophilia where 7.7% declare they have dealt with it and 8.3% declare it is one of the prevailing RD in Cameroon hospitals. 11.1% of respondents say Down syndrome is prevailing in Cameroon hospitals while 3.8% have come across DS. A responses catalogue of thirty-two RDs was drawn out by the responses of different health practitioners. These include: Blood cancer, Absence of the neural tube, Neurofibromatosis, Buruli ulcer, Congenital malformations, Demyelinating degenerative disease, Sickle cell anemia, Hirschsprung's disease, Lyell's syndrome, G6PD deficiency, Down syndrome, Guillain Barre syndrome, Microcephaly, Autism, Connective tissue abnormality, Duchenne muscular dystrophy, Yaws, Hemophilia, Lupus erythematosus, Multiple sclerosis, Endocrine disease, Spina bifida, Albinism, Cushing syndrome, Bladder exstrophy, Congenital abnormality of the hand, Syndactyly, Esophageal atresia, Recklinghausen neurofibromatosis, Sarcoma, Anencephaly, Mycetoma.

V. Detection of rare diseases

The detection of RDs is still a challenge for many health practitioners even for those with long years of experience. The majority of health practitioners who responded declared that it is difficult for them to easily identify and detect an RD case. In fact, this highlights the view that RDs are uninteresting to many physicians, firstly because they have a very low prevalence and secondly because in their practice, they hardly come across these health conditions. According to many health professionals, emphasis was never laid on these conditions during their years in medical school because they were viewed as conditions that are hard to occur in an African context. One health professional, talking of the difficulty in identifying an RD justifies this by saying: « We were not really taught how to manage a rare disease because we were always told cases are very rare in our country but are more visible in European countries» PHY (6). The majority of health professionals confirm that they are unable to easily identify an RD; that is 18 health professionals while only 4 said they were able to identify an RD. Out of the 4 health professionals, 3 were general practitioners while 1 was a specialist. Out of the 18 who responded they are not capable of easily identifying an RD 4 were specialist while 14 were general practitioners.

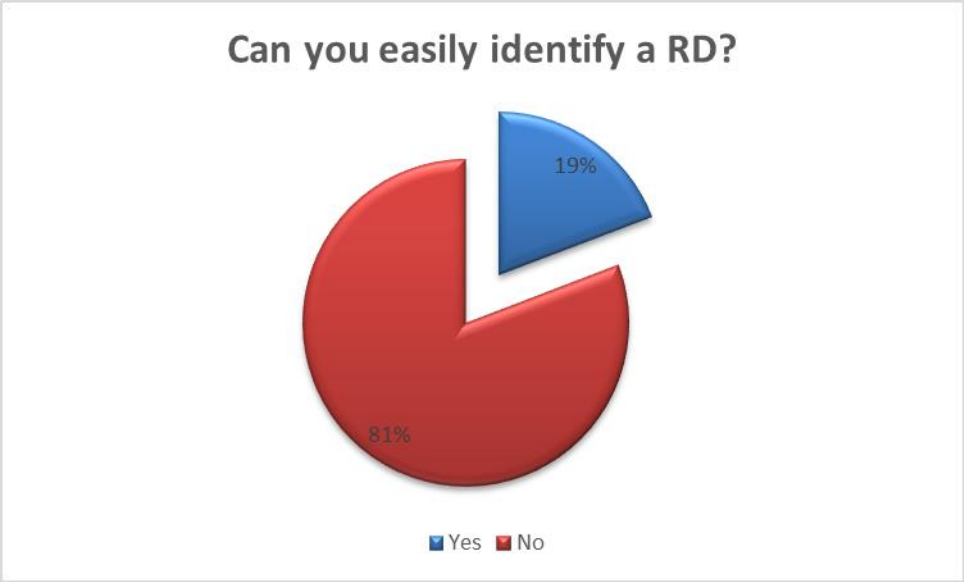

Figure 4: Study participants capable of detecting an RD

The above figure shows that RDs detection is a great challenge for many health care practitioners. This may suggest that there are no budgetary allocations for RDs to foster research on them. Furthermore, this may also imply that there is a lack of expertise with the requisite knowledge to diagnose and treat specific rare conditions. This can lead to poor medication adherence and undermine treatment efficacy.

V. Knowledge on how to manage a case of rare disease

The management of RDs is still very challenging for many health professionals as each has specific clinical signs and symptoms that call for diversified medical approaches to treat them. They are complexe health malfunctions that are difficult to treat because they are usually accompanied by varied symptoms. The diversity of symptoms implies that many health practitioners have to intervene to relieve symptoms. This expertise can be insufficient or lacking due to the rarity of diseases. For many health practitioners, it is rather difficult to have a full mastery of these health conditions for which care and treatment is not yet well understood. In that line, some suggest, that it is necessary to: « Create a care unit for rare diseases and encourage the creation of patients' organizations where they can liaise with their physicians» PHY (6). This collaboration between healthcare providers and patients may be the starting point for developing strategies to effectively design schemes in order to address RDs in Cameroon.

Additionally, a number of practitioners think that a sure way of treating RDs is through the practice of surgery. This can be justified due to the fact that many RDs cause physical abnormalities that often need surgery for repair. Although some practitioners say that: « Each disease has a specific treatment protocol» PHY (7). meaning that it cannot be generalized to all types of RDs. Other health practitioners conclude that: « There is really nothing that can be done. We keep them under observation and accompany them as they die out. What we can do at most is to place an intravenous line consisting of normal saline with ions (Calcium, etc); but this being useless, the most important is a psychosocial follow-up of parent and sensitize families to adopt good behaviors» PHY (10). Other strategies proposed by health practitioners is to focus on sensitization and preventive interventions to reduce the risks of RDs in families and also timely referral of cases to specialists of specific diseases. When practitioners were asked what are strategies can be used to treat RDs, the majority talked of multidisciplinary consults and surgery as illustrated in the figure below.

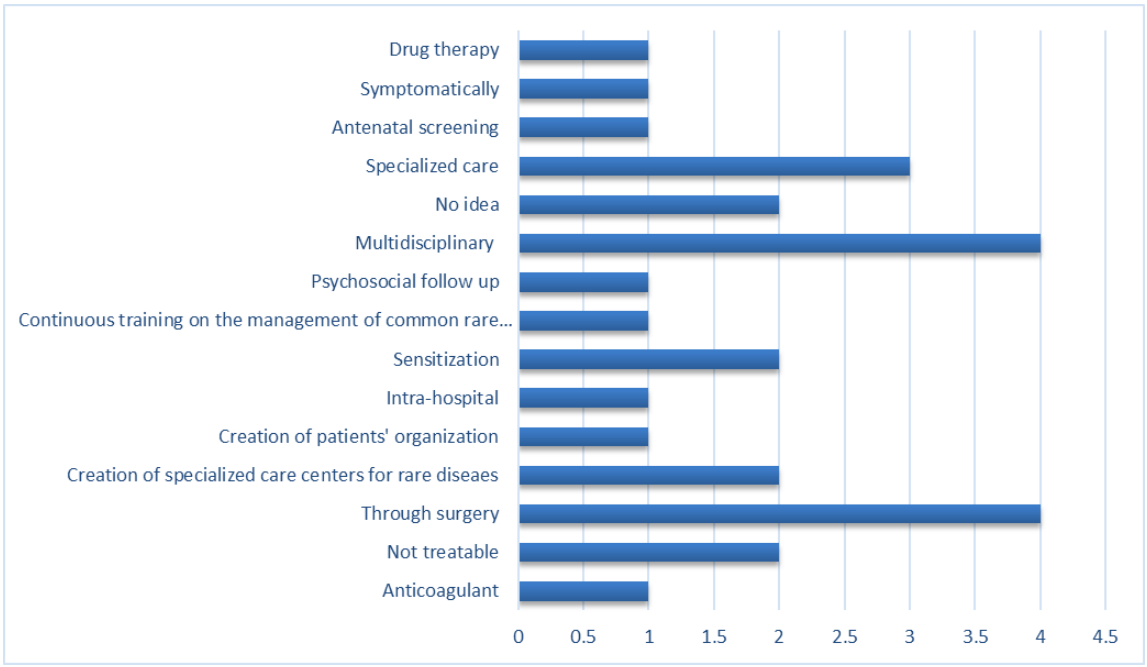

Figure 5: Treatment options for RDs

The above figure highlights a diversity of techniques to manage cases of RDs. However, it is important to note that each disease will respond to a specific approach which cannot be generalized to all diseases. The best approach appears to be a multidisciplinary consult approach which will introduce different expertise to address each case. That is why one practitioner concludes that: « The most important care is a psychosocial follow-up of patients and their families. Concerning the treatment, it will depend on the case you have at hand; it can demand surgery, an excision in the case of Klinefelter syndrome, it can be hormone therapy, or a regular follow-up. As I earlier said, the management of a rare disease will depend greatly on each case. Also, without forgetting multidisciplinarity" PHY (2).

Discussion

This study explored the health literacy of Cameroonian health practitioners on RDs using a questionnaire with open-ended questions that aimed at assessing and describing the knowledge health practitioners have on RDs. The study findings reveal that health practitioners have very strong theoretical knowledge of RDs but very little experience in detecting and treating them. Some health practitioners mentioned Blood cancer, buruli ulcer, anencephaly, sickle cell anemia as RDs while according to the catalogue of RDs produced by Orphanet, these diseases do not fall under the category of RDs. However, Orphanet mentions sickle cell anemia as rare but this is only relevant for the European region. Even though there exists abundant literature on RDs, many health practitioners are still lacking in pertinent knowledge concerning their detection, treatment and prevalence. In effect, the distribution of RDs in the general population is unknown in Cameroon. Prevalence of RDs can be obtained differently depending on geographical areas and legislative norms. In that line: « the concept of RDs in the current political and legislative framework is closely linked to a definition according to point prevalence, and existing definitions are explicitly or implicitly based on a prevalence threshold » (Nguengang Wakap et al, 2019:165). This exemplifies the complexity of RDs in many health systems and therefore brings in the phenomenon of competing interest with diseases that have a well-known prevalence. This can be explained by the fact that research on RDs is underfunded due to result constraints (RDs distribution in the general population) and competing priorities. Moreover, there might be insufficient medical infrastructures to efficiently handle these diseases. Previous research showed that physicians and medical students have very little interest for medical genetic due to the absence of dedicated infrastructures for that. The authors say that physicians and medical students they: « do not have the use of genetic techniques for the purpose of diagnosis in mind, since there are no genetic testing laboratories in Cameroon» (Wonkam et al, 2006:336), whereas, the majority of RDs have a genetic origin. This even makes it more challenging for health practitioners who eventually end up viewing them as uninteresting health conditions. In fact: « Most rare diseases are associated with high unmet need due to the lack of available and effective treatments and the relative lack of research to discover and develop such treatments » (Angelis et al, 2014: 4). The challenge most healthcare professionals have in recognizing an RD or treating them is a common difficulty most healthcare professionals face worldwide. In fact: « Rare diseases have major unmet medical needs. Healthcare professionals and rare disease charities often hear tragic stories of patients with rare life-threatening diseases where responsible doctors have been unable to help as they have never heard of the condition nor seen a similar case presentation, nor found relevant knowledge and expertise online ». (Crowe Ashleen et al, 2020: 260). This highlights the need to establish multivariate collaborations that may lead to multidisciplinary consults, data collection strategies on RD patient information to inform health policies, especially at this time where the global commitment is to ensure that « no one is left behind ». Furthermore, the findings from this study suggests that emphasis should be laid on the training of healthcare professionals on the detection and treatment of specific RDs. This will scale up data systems that will lead to a better understanding of these diseases and their distribution in the population. The discourse of health professionals on RDs have serious implications on the status of RDs in society and the organization of health care for persons living with a rare disease. This once more corroborates the idea that: «We can no longer regard diseases as natural events in the world which occur outside the language in which they are described. A disease entity is the product of medical discourses». The view that these diseases do not occur or are not visible in the hospital milieu can contribute in making them to be viewed as events that do not have a significant impact on the health care system and individuals.

Conclusion

The medical constructions of RDs in Cameroon indicates that these health conditions are not well mastered by health professionals and also that the health system is not adapted to manage them. Indeed, the prevalence of RDs is not well known and there are a lot of speculations as to what these conditions are. This situation can have significant impacts on the sustainability of the healthcare system and the QoL of patients and their families. Despite vast improvements in ensuring that no one is left behind in the health care system, PLWRD still face serious hardships as very little is investigated into their condition. Universal health care commitment to which Cameroon has subscribed entails that pertinent health information is made available for health providers and individuals to ensure that patients and families can access quality health care and services. Also, this means that health practitioners are capable of effectively and efficiently addressing the needs of PLWRD and their families. The financial protection of PLWRD remains a challenge even with the recent commitment by the government to achieve that every person can access health care services without facing any financial barrier. This challenge is even more important because of gaps in knowledge on their distribution, detection and treatment.

List of Abbreviations

RD: Rare Disease
PLWRD: Persons Living with a Rare Disease
QoL: Quality of Life

Declarations

Ethical considerations

Twenty-three healthcare practitioners were enrolled and informed consent to take part in the study was obtained from each of them. Participation was based on voluntary and informed consent. Informants were also reserved the right to withdraw from the study at any time they desired. Ethical principles were respected throughout the study. Anonymity of informants was ensured and they were given codes in the place of their real names.

Data availability

The datasets used for this study are available from the corresponding author on reasonable request.

Funding statement

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Conflict of interest

The authors declare no conflict of interest

Authors contribution

R-DN conceived and designed the study, led the writing of the manuscript and data analysis, LMGBE and TYN contributed to the writing of the manuscript and contributed in data analysis. All authors have read and approved the final version of this manuscript.

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