

## SHORT COMMUNICATION

## STATE OF KNOWLEDGE ABOUT INFORMATION SOURCES AND HEALTH CARE CENTRES FOR RARE DISEASES AMONG AFFECTED PEOPLE IN GERMANY

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## SUMMARY

**Objectives:** About four million people are affected by rare diseases in Germany and 30 million in the EU. In 2013, a national action plan for people with rare diseases was adopted in Germany which is also aimed at improving the information situation and better gathering of information for affected patients and their families. Since then, various sources of information and medical care structures have been made available. The aim of this study was to evaluate the state of knowledge about information sources and health care centres for rare diseases among those affected.

**Methods:** The study was carried out as anonymous survey among the member associations of the German Alliance for Chronic Rare Diseases (German acronym ACHSE e. V.). For this, a questionnaire was developed which in addition to questions on gender, age and disease comprised free text input referring to knowledge of health care centres or expert centres and source of information on rare diseases in Germany.

**Results:** A total of 484 individuals suffering from 96 different rare diseases participated in the survey. Of these, 74.47% are aware of medical or dental care centres for treatment of their types of rare disease; 69.31% use self-help groups as a source of information, only a few respondents know government-sponsored “se-atlas” and “Orphanet”.

**Conclusion:** The majority of the respondents know medical care centres, most participants use self-help groups as information source, however, government-supported portals are largely unknown so that there is a need for further information in this regard.

**Key words:** rare diseases, se-atlas, national action plan, Orphanet, NAMSE

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## INTRODUCTION

In the European Union (EU), a disease is defined as rare if it affects fewer than one in 2,000 people (1). This definition applies to about four million people in the Federal Republic of Germany and about 30 million people in the EU (2, 3).

The public focus on rare diseases has increased in the EU since 2009 when the Council of the EU recommended that the member states develop and implement plans and strategies for rare diseases at suitable levels or validate suitable measures for rare diseases in the scope of other health protection strategies in order to ensure that patients suffering from rare diseases receive good medical care (3). In particular, member states were called upon to develop and accept a corresponding plan by the end of 2013.

In 2010, cooperation between the German Federal Ministry of Health and its partners: the German Federal Ministry for Education and Research and the Alliance for Chronic Rare Diseases led to the creation of a National Action League for People with Rare Diseases in Germany. The aim of the Action League is to improve the circumstances of people with rare diseases (4). A National Action Plan for People with Rare Diseases was finally

adopted in 2013 (4). The National Action Plan comprises 7 fields of action and 52 proposed measures, including the field of action “Information Management”. The 2017 interim report on the implementation of the National Action Plan for People with Rare Diseases (5) will also include in the Information Management field of action the goal of improving the quality of information and access to information for patients and their families and also for physicians, therapists and nursing staff. For example, since 2015, the online platform “se-atlas” (6) which was financially supported by the German Federal Ministry of Health, has been available in order to make it easier to locate specialist treatment centres for those affected in Germany (7). Information about diseases\* was also transferred to Orphanet (5, 8). A source of information about dental, oral and maxillofacial surgery – the ROMSE specialist database has been available online since 2013 (9, 10). Another recommended action was to raise awareness about sources of information (5). The aim of this study was therefore to survey awareness of sources of information relating to rare diseases among the German Alliance for Chronic Rare Diseases (German acronym ACHSE e. V.) member associations.

\*www.achse.info

## MATERIALS AND METHODS

### Study Design

The study was conducted as an anonymous survey of ACHSE e.V. member associations. It was approved by the Ethics Committee of the Medical Association of Westphalia-Lippe and the Westphalian Wilhelms University of Münster (Ref. No. 2016-006-f-S).

A questionnaire was developed for this purpose. In addition to epidemiological questions about gender, age and disease, it included open questions about awareness of care facilities or centres of expertise and sources of information on rare diseases.

The questionnaire was sent out in electronic form in February 2016 to all 125 member associations of ACHSE e.V. registered at the time.

### Participants

Everyone affected by a rare disease in the Federal Republic of Germany from the age of 16 was eligible to participate. There was no patient selection. All self-help groups registered with ACHSE e.V. were contacted, and everyone with a rare disease who could be contacted via ACHSE e.V. was able to participate in the study.

### Data Source

In addition to age, gender and disease, participants were asked whether they knew of a medical or dental centre of expertise for their particular rare disease. Participants were also asked whether they knew about information sources on rare diseases such as Orphanet, se-atlas or ROMSE.

### Data Evaluation

The data was recorded and evaluated in Excel® (Excel 2013, Microsoft Corporation). The data was graphically represented as bar charts.

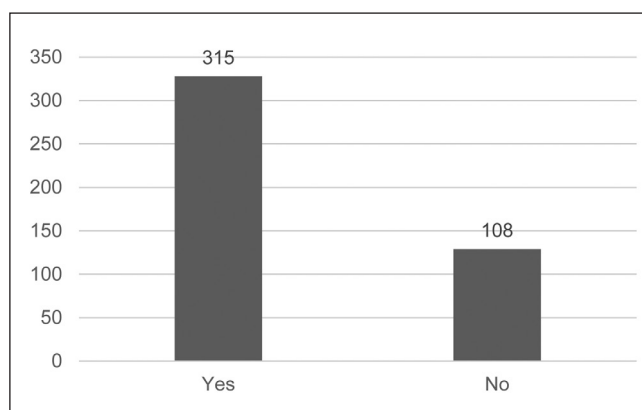
## RESULTS

### Age, Gender and Type of Disease

A total of 484 people with 96 different rare diseases took part in the survey, 313 of whom were women and 171 men. The age range was between 16 and 80 years, and the gender independent average age was 44.57.

### Knowledge of Medical Care Facilities and Centres of Expertise

In response to the question as to whether they were aware of a medical or dental care facility or centre of expertise for treating their respective rare disease, 315 participants (74.47%) answered "Yes" and 108 participants did not know of any specialist facility for their rare disease (25.53%). This was based on information from 423 participants and the graphical representation can be found in Figure 1.



**Fig. 1.** Level of knowledge about medical care facilities or centres of expertise for their own rare disease among people with rare diseases.

### Knowledge about Orphanet Information Resource

Participants were specifically asked about their knowledge of particular sources of information on rare diseases. Of the 391 users who provided information about this, 53 respondents (13.55%) know about Orphanet and 338 respondents did not know about Orphanet (86.45%).

### Knowledge about Se-atlas Information Resource

Based on responses from 391 participants, 17 respondents (4.34%) knew about se-atlas and 374 participants do not know about se-atlas (95.66%).

### Knowledge about ROMSE Information Resource

Five respondents knew about the ROMSE database that specialises in rare diseases with oral components (1.28%) and 386 were unaware of this information resource (98.72%). This was based on responses from 391 participants.

### Self-help Groups as Information Source for One's Own Rare Disease

Self-help groups are mentioned by 271 respondents as a source of information for their own rare disease (69.31%). 120 participants did not mention a self-help group as a source of information for their own rare disease (30.69%). Responses were received from 391 participants.

### Knowledge of Other Information Sources

Other sources of information were known to 148 of respondents (37.85%), 243 of the participants did not mention any additional sources of information (62.15%). This was based on responses from 391 participants.

## DISCUSSION

Specialist medical centres and medical experts are usually required for the medical treatment of rare diseases (11). Infor-

mation about existing medical care facilities for rare diseases in the Federal Republic of Germany can be accessed at se-atlas website (7). Almost 75% of respondents said they were aware of a healthcare organisation and therefore a specialist point of contact for their disease. Se-atlas, a project funded by the German Federal Ministry of Health, has been available online since 2015 (7). Fewer than 5% of respondents knew about the se-atlas website. Overall, it is gratifying that, at the time of the data collection, 74.47% of the respondents knew about an organization specialising in their disease.

The National Action Plan for People with Rare Diseases also promotes the provision of information to patients in the Information Management field of action (4). Around 70% of the respondents use self-help groups as a source of information on rare diseases.

Less well known, on the other hand, are general information sources about rare diseases such as Orphanet (8). Only 13.55% of respondents knew about Orphanet. Only five study participants knew about specialist databases and information sources such as ROMSE (9). However, this information source is specifically designed for rare diseases with oral components (10), and this may be a possible explanation for the low level of awareness.

The interim report of the National Action Plan (5) describes transferring disease information from achse.info to Orphanet as part of the current status of policy proposal 38. The data transfer project was carried out under the supervision of ACHSE e.V. This makes it even more surprising that only 13.55% of the respondents knew about Orphanet, particularly as the project management, ACHSE e.V., is the umbrella organisation for chronic rare diseases in Germany. There is certainly a need for further information about how to raise awareness about Orphanet in the future.

The aim of the government supported “se-atlas” project is to ensure that patients, their families and physicians, but also non-medical staff and the general public can obtain a comprehensive overview of care options and self-help organisations for people with rare diseases in Germany (5). This should not only consider improved information management for patients, but also for medical personnel, who also provide valuable information about the respective disease.

This makes it even more surprising that fewer than 5% of respondents knew about the se-atlas. It seems that there is a significant need to make information about the se-atlas and its contents more widely known. Increased awareness of the se-atlas could also help more people with rare diseases find out about a care facility for their own disease in the future.

We suggest that the European Reference Networks (12) could also contribute to disseminating knowledge about rare diseases on the one hand and to increasing the awareness of information sources in cooperation with the self-help groups on the other.

## CONCLUSION

The majority of the respondents know medical care centres, most participants use self-help groups as information source, however, government-supported portals are largely unknown so that there is a need for further information in this regard.

## Conflict of Interests

None declared

## Ethical Approval

The ethical approval for this study was obtained from the Ethical Review Committee of the Medical Association of Westphalia-Lippe and the Westphalian Wilhelms University of Münster, Germany (Ref. No. 2016-006-f-S). All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and national research committee and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards.

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