How do patients with rare diseases experience the medical encounter? Exploring role behavior and its impact on patient–physician interaction

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ABSTRACT

Objectives: Empirical research shows that patients with severe illnesses prefer the physician to dominate decision processes and provide the information needed. However, in rare diseases, due to the low prevalence and the lack of expertise, the patient is forced to become knowledgeable about his own disease state. Objectives of this study were to describe the experiences of patient–physician interaction in rare diseases, to develop an empirically derived typology of interaction patterns and to explore the antecedents of these interaction patterns, with a special focus on role behavior. Building on these results, implications for health care policy are made.

Methods: We designed an exploratory study as a series of semi-standardized interviews with patients suffering from rare diseases. We extracted the following six rare diseases: amyotrophic lateral sclerosis, Duchenne muscular dystrophy, epidermolysis bullosa, Marfan syndrome, neurodegeneration with brain iron accumulation and Wilson’s disease. A total of 107 interviews were recorded, transcribed and analyzed thematically in accordance with the grounded theory tradition.

Results: As suggested, insufficient expertise of the healthcare providers proved to be a major problem in the highly specialized treatment process of rare diseases. Here, the patient often becomes an expert in his disease. Therefore, we identified the patient-directed interaction as a widely experienced communication pattern among patients with rare diseases. Our study also showed that role discrepancies have a major impact on communication processes in this context.

Conclusions: People with rare diseases often face challenges, due to the low prevalence and the resulting lack of knowledge of their healthcare providers. Communication processes in this context are mainly affected by the role behavior of both the patient and provider. The present study showed the relevance of the provider’s ability to acknowledge the active role of the patient as an informed, involved and interactive partner in the treatment process. However, allowing the patient to control therapy may require a change of mind-set with some long-standing traditional roles in healthcare.

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1. Introduction

Patient–physician interaction has been a topic of great interest to research in the medical field (e.g., [1–5]). Evidence indicates that effective patient–physician interaction is beneficial to several outcomes [3,6–8]. Moreover, failures in effective communication between physicians and patients can lead to increased medical costs arising from overuse or misuse of medical services [9–11].

Traditionally, the patient–physician relationship focuses on the information asymmetry between the patient and physician [12], assuming that the physician dominates the medical encounter, whereas the patient is given a more passive role in the treatment decision process. Yet, in cases of rare diseases, the patient–physician relationship may not fit with the traditional assumptions of medical care. Although the communication process between patients and their physicians shares most of the general features of the standard doctor–patient interaction, rare diseases may be burdened with additional problems, e.g., the lack of disease-related expertise among the health care professionals, the psychological challenges associated with the diagnosis of an incurable disease and greater geographical distances between patients and physicians [13]. Empirical research shows that patients with severe, incurable illnesses seem to prefer the physician to dominate decision processes and provide the information needed [14]. However, due to the low prevalence and the lack of expertise, patients are forced to become knowledgeable about their own disease and related therapies. There is a significant shift in the traditional roles in the medical encounter and the patient–physician relationship. However, despite these challenges, to the best of our knowledge, little research has empirically explored the nature of communication processes in the context of rare diseases, and it remains unclear what constitutes an adequate partnership between patient and physician in the specific context of rare diseases. The existing literature on rare diseases primarily originates from the research of medical specialists and is therefore highly disease oriented [15].

We aim to contribute to the understanding of the patient–physician interaction in the context of rare diseases. Particularly, we seek to differentiate and to integrate the various taxonomies that describe role behavior in this context and its link to patient–physician interaction. This information is important because knowledge of mutual role expectations and the resulting interaction patterns can shed light on the challenges in patient–physician interaction in rare diseases and thus help to identify appropriate strategies and tools to overcome problems and obstacles in this area and to increase satisfaction of both patient and physician. This, in turn, may help to improve patient outcomes and to reduce costs by increasing the efficiency and quality of health care services.

Accordingly, the following three central research questions will guide the current exploratory study: (1) how does role behavior of both the patient and physician influence the patient–physician interaction in rare diseases? (2) What interaction patterns can be observed between patients suffering from rare diseases and their physicians? (3) What are the consequences of the interaction patterns identified on the treatment process?

The paper is structured as follows. First, we briefly review theory about patient–physician interaction. Second, we explain the major challenges associated with rare diseases, focusing on role behavior in the medical encounter. Having explained the research context and design, we finally present and discuss the empirical results.

1.1. Theoretical background

1.1.1. Communication in the medical encounter

Several studies have shown that the physician–patient relationship can play a crucial role in directly influencing patient’s health and well-being through the patient’s sense of feeling understood as well as indirectly through continuity of care and adherence to treatment regimens [3,6,8,16]. Physicians’ professional credibility and competence, as well as the appropriateness of communication, significantly determine patients’ psychological status, physiological symptoms and functional outcomes [6,17–19]. The accumulated evidence indicates that patient-centered communication, including information-giving and information-seeking behavior and patient involvement in the treatment process, can alleviate patients’ feelings of uncertainty and can even affect the healing process [20–22]. Although the patient–physician relationship may be influenced by various individual attributes [23–27], it is not only a function of personal influences; according to Street and colleagues, it is also “a process of mutual influence” [28].

Many descriptive models have been applied to relationships between patients and physicians [4,29,30]. They are determined by the degree of control exercised between the patient and the provider. Two prominent approaches are the paternalistic model, with the physician having the dominant role, and the informative model in which the key responsibility of the physician is to provide the medical expertise [4,12,17]. Another widely advocated interpretation of the doctor–patient relationship is the model of shared-decision-making. In this model, the patient and the physician equally exchange information across all stages of the decision-making process [4,12,31]. Moreover, recent conversation analysis studies suggest that communication may differ according to the various phases of the medical interview [32].

The assumption underlying each of these models is that the physician has sufficient expertise regarding the diagnosis and treatment. However, in the context of rare diseases, physicians often lack knowledge and experience, and, therefore, patients are faced with a high degree of uncertainty. Particularly, most treatments are considered experimental, which leads to questions about their safety and effectiveness [33]. Information may be inconsistent or contradictory, and the patient is faced with the numerous treatment option opinions of the involved health care providers. Ambiguity, variety and unpredictability of symptoms represent further problems [34–39]. Therefore, the traditional roles within the patient–physician interaction may change. This, in turn, can lead to differing mutual role expectations resulting in role discrepancies.
1.2. A role theory perspective on the medical encounter

According to Ebaugh, roles are “sets of behavioral expectations associated with given positions in the social structure” that may be achieved or ascribed [40]. Thus, role theory constitutes a key element in understanding relationships between individuals [41–43]. There have been two distinct approaches of role theory in the sociological literature [40]. In the structural tradition, roles are typically defined as socially shared expectations and behaviors associated with given positions in the social structure [44–47]. Through the process of role-taking, the individual recognizes and internalizes those expectations. Interactional theories, in contrast, assume that roles are created and redefined, as individuals actively construct, interpret and uniquely express them. Role-making processes are central to understanding this approach. In the current analysis of roles in the medical encounter, we draw on De Rue and Ashford, who combined these two distinct yet complementary approaches and proposed that identity work is undertaken both by an individual claiming an identity and by others affirming and reinforcing (or not) that identity [36].

Sluss and Ashforth presented a conceptual framework in which role identity constitutes a central attribute of the relationship between two individuals [49]. To the extent that roles become socially constructed in interaction with others, role theory occupies a central place in research on interactions in the service encounter. Each individual in the service encounter has a role to perform [48]. The service encounter in health care may thus be approached as a special case of role performances in which both the patient and physician act as social actors appropriate to their position in this relationship. Once the patient enters the medical encounter, he or she adopts the role of patient, and a role-defined dyadic interaction may begin. The course of the medical encounter is governed by traditional behavior patterns. As such, the role of the physician is to provide information concerning the patient’s illness, whereas the patient is assumed to bring in his or her health history, expectations and needs. If role perceptions vary between doctor and patient, conflicts and “sociomedical dilemmas” [32] may arise and lead to frustration [50].

However, in rare diseases, the medical encounter is influenced by changes in the traditional role structures. Due to poor knowledge of rare diseases and the limited therapeutic options of treatments, physicians may not act as competent technical experts who provide relevant information to the patients. The patients are thus forced to become experts on their own disease state. Therefore, both parties must revise their role identities and mutual expectations. However, professional role identity is highly resilient and resistant to change [51,52]. As a result, in an environment without existing routines and limited experience, the physician’s role concept probably differs from that projected by the patient [53]. In light of research indicating that the associations between mutual relational expectations and the communication process in the medical encounter are complex, we aim to offer new approaches to understand this link [28].

<table>
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<tr>
<th>Variable</th>
<th>Total</th>
<th>Participants suffering from ALS</th>
<th>Participants suffering from DMD</th>
<th>Participants suffering from EB</th>
<th>Participants suffering from muscular dystrophy</th>
<th>Participants suffering from Wilson’s disease</th>
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<td>15</td>
<td>15</td>
<td>17</td>
<td>9</td>
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<td>59.7 (12.5); 31–88</td>
<td>15.1 (9.2); 4–37</td>
<td>14.2 (14.8); 25.1 (26.6); 1–64</td>
<td>15.9 (10.9); 10–45</td>
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<td>Female</td>
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Table 1 Characteristics of participants included in the study.
2. Materials and methods

To answer the abovementioned explorative research questions, an interpretive, qualitative approach was selected. This approach allowed us to describe and understand patients’ behavior and its implications for social interaction [54]. We considered this approach consistent with both our research goals and the methodology and assumptions used in recent research on related topics (e.g., [55–57]).

The study was conducted in a sample of patients in Germany. Through expert interviews with various physicians who specialize in the care of rare diseases and with representatives of self-help organizations, we extracted the following six rare diseases that mainly differ in care intensity, patients’ age of disease outbreak (children vs. adults) and prevalence: amyotrophic lateral sclerosis (ALS), Duchenne muscular dystrophy (DMD), epidermolysis bullosa (EB), Marfan syndrome, neurodegeneration with brain iron accumulation (NBIA) and Wilson’s disease. These diseases are characterized by regionally dispersed expertise, limited experience, a degree of uncertainty due to an absence of knowledge and routines and extraordinary individual health care demands. Patients were sampled via ACHSE (Allianz Chronischer Seltener Erkrankungen), which is a non-profit association for the needs of people with rare diseases, and through centers of expertise and specialized hospitals for rare diseases.

Our analysis includes a total of 107 semi-standardized interviews with patients from Germany. Table 1 displays selected demographic and health characteristics of the subjects.

In some cases, family members were integrated in the interviews, e.g., in cases of pediatric patients or patients with limited speech ability. In sum, we conducted 34 interviews with parents of ill children and 73 interviews with adult patients. The questions that guided the interviews were broad and open-ended and pertained to the experiences patients had in the medical encounter and to the interaction processes with their healthcare providers. New questions emerged with time as a function of the evolving results of the analyses. The interviews lasted between 30 and 45 min and were tape-recorded and transcribed verbatim.

The analysis was designed in accordance with recommendations of qualitative research and followed the grounded theory approach of comparing and contrasting the data and the emerging theory throughout the data collection and analysis process [58–60]. We analyzed the transcribed interviews using Atlas.ti version 6.2, software for content analysis. In a first step, we derived in vivo codes inductively from the interviews and subsequently created first-order categories. As our coding progressed, associations between these initial categories were determined, and further subcategories emerged. Finally, by continually aggregating and consolidating our categories, we obtained abstract theoretical dimensions that revealed interaction processes between patients and their physicians, as well as their antecedents and consequences. In order to ensure that the emerging categories fit the data, a representative sample of 10 transcripts was double-coded by a second researcher. Fig. 1 shows the development of the categories.

3. Results

To help make sense of the resulting dimensions of interaction processes between patients and physicians in cases of rare diseases and their relationships in the current data, we propose a theoretical model (Fig. 2). The model shows how the role behavior and individual competencies of both the patient and physician combine to form the patient–physician interaction in the medical encounter.

In describing these findings, we begin with a discussion of role behavior and individual competencies of both the patient and physician. Next, we continue by addressing how these individual factors are intertwined with communication processes in the medical encounter. Finally, we outline four distinct interaction patterns that were identified in the current study.

3.1. How are role behavior and patient–physician interaction in rare diseases related?

Role behavior can be described by the role identity and role expectations of patients and physicians. Role identity is defined as a constellation of goals, values, beliefs, norms and experiences that are related to a role [61]. Therefore, certain patients show more passive attitudes in the medical encounter [62], whereas others regard themselves as experts seeking an active role in medical treatment and decisions:

“To a large extent, I am my own expert, because I know best what I need and what helps me.” [MA21]

We found that the majority of physicians acted as the patient’s partner in the medical encounter. However, a minority of physicians rejected the patient’s assertive behavior, demonstrating an almost arrogant attitude toward the patient:

“The school doctor thinks that he knows more than we parents do, and he always tries to make our decisions look bad.” [DU20]

Role expectations reflect expected behaviors of actors in complementary social positions [50]. Missing congruence between the perceived and the expected behavior leads to role discrepancies and conflicts. If both the patient and physician share mutual role expectations, they enact their prescribed roles of patient and physician through the process of role taking. However, the current data indicated that contextual uncertainty may influence these ritualized roles and lead to role transitions. These, in turn, have an impact on the patient–physician interaction. Our study revealed that, in the context of rare diseases, role discrepancies arise: physicians had to cope with lacking, insufficient or ambiguous knowledge. Patients also found themselves in a dilemma. Although they approached the physician to receive help, they were forced to assume the expert role and provide information to the physician:
First-order Categories  (examples)  Second-Order Themes  Aggregated Theoretical Dimensions

- Patient providing the physician with information; well-informed physician
- Shared decision-making; physician taking the decision; no need for decisions
- Physician accepting patient’s expertise; rejecting patient’s objections
- Technical expert; patient’s partner; patient’s guardian
- Physician’s advisor; passive consumer
- Wishing to be taken by the hand by the physician; accepting physician’s authority
- Heterogeneity of symptoms; difficult diagnose; inconsistent information
- Prognosis; progression; curability
- Experience with patient’s disease; disease-related knowledge
- Information-seeking initiatives; creation of new ideas
- Ability to cope with death; sensibility; empathy; communicational abilities
- Information-seeking initiatives; adaptation to uncertainty; proactive behaviour
- Disease-related knowledge; experience
- Communicational abilities; assertiveness

- Information-transfer processes
- Decision-making processes
- Physician’s role expectations
- Physician’s role identity
- Patient’s role identity
- Patient’s role expectations
- Ambiguity
- Severity
- Disease-related expertise
- Uncertainty management
- Social competencies
- Uncertainty management
- Disease-related expertise
- Social competencies
- Descriptions of patient-physician interaction
- Physician’s role behavior
- Patient’s role behavior
- Disease-related characteristics
- Physician’s competence
- Patient’s competence

Fig. 1. Development of coding categories.

Fig. 2. Patient–physician interaction and its antecedents.
Another challenge resulting from role discrepancies arises if the patient, although not a medical expert, is better informed about possible treatment options than his physician. Because the patient is well aware of the physician's higher authority, this situation might constitute a problem for the patient:

“I am not a medical expert, and I cannot simply tell her that I would like to try that out because it might work ... Actually, you need a lot of fingertip feel.” [NB14]

Because of these differing expectations and the resulting discrepancies, role-taking processes must be completed by role-making processes. Consequently, both patients and physicians are involved in an ongoing process of actively creating, adapting and modifying their roles based on the situation and their mutual expectations [63]. Particularly when confronted with patient assertiveness and engagement, physicians often need to adjust their professional role identity and behavior. The present data revealed that some physicians feared that patient empowerment would threaten their social power. Because of their limited experience, they were not able to cope with the newly emerging role of the “expert patient” [64], as indicated by the following statements:

“It has frequently been my experience that the doctor asked me, who was the doctor - him or me.” [EB05]

In addition, the patient also had to adjust to the newly emerging roles in the medical encounter. We found evidence that the majority of patients with rare diseases learned to fulfill their role as experts in the treatment process, actively pursuing multiple types and sources of information for new data related to their disease and sharing these insights with the physician:

“I have explained the disease to my GP.” [EB02]

3.2. What interaction patterns between patients suffering from rare diseases and their physicians can be observed?

The study revealed a variety of patients’ experiences in the medical encounter. We aggregated the descriptions of these experiences and finally identified four interaction patterns. The main factors that differentiated these patterns were their particular conceptions of the underlying processes of information-exchange and decision-making.

Table 2 shows the distribution of the interaction patterns among the patients.

We address these patterns in the following sections.

The paternalistic pattern of patient–physician interaction has been widely described in the literature [4,12,30,32]. This model assumes that the physician discerns what is in the patient’s best interest, therefore minimizing the patient’s participation. In the current study, patients often preferred not to participate in the treatment’s decision-making process, believing that decisions require expertise and medical experience that they lack. Thus, they relied solely on the doctor’s opinion. As such, the paternalistic approach that emerged from the present data was characterized by a mutual approval of the physician’s authority and thus by role congruence. This finding is in line with prior research that suggests that being confronted with a serious and potentially life-threatening illness leads to a delegation of responsibility to the physician [29,67]. The current findings strongly support this implication:

“We did not discuss it; it was the doctor who decided for us. We did not question his opinion; he is the expert.” [DU10]

“Actually, you’re getting told what to do next, and I think that the physicians know more than I do.” [MA27]

The collaborative pattern is distinguished by equal levels of medical information exchange between the parties and a mutual agreement on the treatment. Thus, this interaction type corresponds in the most part to the widely known concept of shared decision making [4,12,32,68–70]. However, this pattern includes another important aspect, namely, the patient’s self-acquired expertise and the physician’s acknowledgment of this expertise. The present research supports previous findings that patients attempt to develop relevant knowledge to manage their illness and conditions through information-seeking activities [71]. The majority of physicians encourage the patient’s engagement because it may contribute to a common definition of the medical problem and build a collaborative relationship between the parties [72]. Thus, there is mutual comprehension of the roles both parties play in the medical encounter. The following examples illustrate this type of interaction:

“We are always working out together what is right in therapy.” [AL12]

“I am getting involved by my doctor; he asks me what I want and how I would like to proceed.” [DU14]

The patient-directed interaction pattern is apparent in the patient’s engagement and the large amount of information
he provides the physician. In contrast to the collaborative pattern, in this case, the physician does not have any expertise about the disease. Therefore, the patient needs to acquire knowledge on his or her own disease state and instruct the physician on the treatment options. In this situation, the physician is a passive participant, and his involvement is often limited to providing consent for the treatment advocated by the patient or to writing prescriptions. Therefore, the patient and physician both actively create and modify their roles based on the situation:

“Our family doctor does not know anything about my disease, and he relies on my opinion.” [DU15]

“As actually, the way it works in EB is that you go to a physician and you explain to him what you want and what you need.” [EB09]

Finally, the confrontational interaction pattern reflects a conflict situation. In this case, the patient seeks an active role in the medical encounter. Because of his expertise, the patient is able to evaluate and even challenge the physician’s competence [73]. The physician, however, attempts to reassert control by authoritatively making decisions and ignoring the patient’s suggestions and preferences. This inter-role conflict is attributable to different role expectations [41, 74]. In extreme cases, the physician questions the patient’s knowledge and skills, as the following quotes show:

“Actually, there are few physicians who admit their lack of knowledge. Many feel offended, and that’s why it’s getting so difficult: because you have to fight against them.” [MA06]

“He barked at me that I should stop acting up. And then they carried out the operation in a way that was against my wish.” [EB15]

In line with previous research, which found that communication between the patient and the physician is influenced by personal attributes, the current study also considers the impact of the individual competencies of both parties on their interaction. A physician’s communication skills shaping patient-centered care seem to be fundamental competencies leading to a better patient–physician relationship. Patients expect the doctor to be a competent partner in discussions:

“It’s important for me that I can talk to my doctor.” [MA08]

“I would like the doctor to also ask some counterquestions.” [MA20]

The physician’s professional competence, especially his task-related and disease-specific knowledge and experience, is another crucial factor that influences mutual agreement [65]. However, in the case of rare diseases, the lack of knowledge constitutes a major challenge for patients. If the medical providers do not have any knowledge about the disease, the patient often takes the initiative. Thus, patient-directed interaction is mainly predicted by the physician’s lack of expertise in rare diseases, as indicated by the following quotes:

“If you have something to do with people who don’t know the disease, you have to lay the groundwork for them.” [MA03]

“I have set myself the goal of informing the doctors.” [MA07]

Most patients were realistic about what their physician could provide, and they appreciated honesty about a physician’s lack of knowledge [66]. The following quotes illustrate this attitude:

“I would not assume that my GP knows my disease.” [AL16]

“This is simply a lack of knowledge, and you cannot blame anyone for this.” [MA03]

Finally, other personal attributes of the physician that patients attached importance to are empathy and ability to cope with imminent death [67]:

“I wished to have a doctor who accompanies me throughout the illness, without any fear of death.” [AL22]

In addition, the results suggest the patient’s individual competence as another antecedent to satisfactory role enactment and interaction in the medical encounter. For example, the patient’s ability to cope with the disease-related uncertainty in conjunction with well-developed information-seeking skills may result in higher self-esteem in enacting the role as an expert patient:

“I am very well informed, and that’s why I can talk with the doctor at the same level. Actually, I am on a par with him.” [AL28]

Our results also show differences in the patient–provider-interaction processes between adults who themselves were affected by the disease at those who had a child with a rare disease. As such, the patient-directed interaction pattern is more apparent among parents who interact with the physician on behalf of their child than among adult patients. Parents do often engage in seeking information on the disease and providing it to the physician.

Patients’ diagnosis and health status, along with their attitude toward their illness, are also likely to play an important role in the medical encounter. Therefore, the degree to which patients with rare diseases want to be involved may vary according to individual or contextual characteristics, such as the complexity of the illness or its prognosis. Patients suffering from Marfan’s syndrome live in constant fear of an aortic dissection that can happen at any time. This unpredictability about individual disease trajectories contributes to patients’ uncertainty and their fear of observational oversight, increasing their need for intensive consultation. The findings of this study also reveal that patients who face a fatal and rapidly progressive disease such as ALS or NBIA usually interact with their physician in a cooperative way, and they often take the initiative and seek information. In addition, due to the extremely low prevalence of NBIA, both patient and physician are confronted with the lack of knowledge about the
disease. Consequently, they work out the best treatment plan together. Thus, the confrontational interaction pattern was not observed in these participants.

Another interesting finding worth noting is the heterogeneity of the clinical picture, which may lead to uncertainty among physicians and thus to confrontations between patients and physicians. If a patient’s appearance or his symptoms do not correspond phenotypically to the clinical picture as commonly described and known, the patient may face a lack of understanding by his health care providers, which can lead to conflicts. Often, physicians might even question the diagnosis, particularly in the case of patients with EB or Marfan’s syndrome. As the following participant stated:

“I don’t look like a typical Marfan patient, and only few know that there is a wide heterogeneity in appearance.” [MA31]

If there exists a therapeutic option that might slow the progression of the disease, patients with rare diseases are often faced with the problem of numerous opinions among the providers involved in the treatment. In particular, this can be observed in parents of children with Duchenne muscular dystrophy, for which steroids are offered as a treatment option. Although steroids can be beneficial, there are also various negative side effects that should be considered when making the decision to use this treatment. Therefore, participants in the study often reported uncertainty, and they wished to discuss the benefits and risks of such an intervention carefully with the physician to reach a decision, as indicated by the following statements:

“One professor says this, the other says that.” [DU20]

“We went back and forth discussing it with our doctor. Unfortunately, it also has many side effects. However, it was all discussed with us in detail.” [DU22]

4. Discussion

The objective of the current study was to explore the experiences of patients suffering from rare diseases in the interaction with their physicians. The qualitative analysis outlined a variety of problems these patients must face. As suggested, the low prevalence of the diseases and the related insufficient expertise of the healthcare providers proved to be major challenges in the treatment process. The experienced contextual uncertainty influenced the way the parties interacted with each other, as discrepancies in mutual role perceptions arose.

Further, we derived four interaction patterns. By identifying the patient-directed interaction pattern characterized by the role of the patient as an expert on his own disease state, we extend existing ideas and theories on patient–physician interaction. In the specific context of rare diseases, the patient often becomes the driving force in the treatment and information-seeking processes. Therefore, the traditional patient–physician relationship undergoes a significant change. Finally, our research suggests that role transitions encounter greater resistance by physicians than by patients.

As such, we make three contributions. First, by showing the impact of role behavior on communication between the physician and patient, we combine role theory with ideas about interaction in the medical encounter and thus advance the medical socialization literature. Particularly, we found that role discrepancies have a major impact on interaction processes in the medical encounter. This finding leads to an extension of the existing models of patient–physician interaction. Second, we studied patient–physician interaction in rare diseases, a context that has received comparatively little research attention to date. In doing so, we highlight the challenges and problems these patients must face. Finally, we contribute to studies on patient satisfaction by elucidating the relevance of the provider’s ability to acknowledge the active role of the patient as an informed, involved and interactive partner in the treatment process.

The study has also several limitations that should be considered in future research. The findings are based on patient self-reports, thereby raising a question about external validity. Complementary to the perceptional approach, further studies using behavioral assessment of patient experiences (e.g., via audiotape or videotape) are warranted to better understand the interplay between communication and patient satisfaction [68]. Research designs should also measure both the patient’s and the physician’s perspectives and explore the physician’s decision-making style. This will enable to analyze the congruence between decision-making styles of physician and patient and its impact on patient satisfaction. Another appropriate avenue for future research on uncertainty in rare diseases would be to conduct interviews with health care providers to ascertain how they cope with the perceived limits of medical knowledge. Furthermore, the cross-sectional design of our study does not allow us to reliably assign any causation to the observed associations. Therefore, we strongly encourage researchers to obtain multisource data in longitudinal studies on the physician–patient relationship in rare diseases [69].

Another limitation worth noting is that we did not examine the influence of cultural, ethical and sociodemographic issues, although these issues have also been found to contribute to the different perceptions and behaviors of patients [23–27]. We suggest that future research should consider these variables as further potential predictors in patient–physician interactions in the context of rare diseases. Finally, future research is urgently needed to analyze and understand the congruence between the actual level of participation and the preferred pattern of participation of patients with rare diseases, since this constitutes a major prerequisite for patient satisfaction [68,70].

4.1. Policy implications

We believe the results allow us to make several contributions that are particularly relevant to health care practice and to highlight fruitful avenues for both research and practical application. The findings are consistent with the contention of previous research that communication in the medical encounter is a multilayer construct, resulting in various distinct interaction models. The analysis
presented in this study extends the understanding of patient–physician interaction and its elements by revealing role behavior as a crucial predictor. In particular, we propose that the interactional behavior of both the patient and physician may differ according to their role identities and mutual role expectations. As such, our model tentatively suggests that a focus on individual role-related phenomena can illuminate the factors that influence both parties to participate in decision-making and to engage in information-seeking processes.

Furthermore, this study tends to substantiate the shift in both theory and practice from the traditional model of patient–physician relationships to one that is characterized by patient participation. Although the notion of ‘expert patient’ is not without controversy, there is broad support for the idea that patients should be actively involved in their health care [75–78]. The present study shows the difficulties and importance of providing information about a rare disease. We found that the vast majority of patients with rare diseases wish to have information and to participate in the treatment process. The patients actively seek information and might even challenge the physician’s authority. Patients should be supported by their providers in developing the confidence and motivation they need to use their own skills and knowledge to take effective control over living with their disease. These patients may benefit from the mutual exchange with their physician by better understanding their own conditions and by participating more effectively in medical interactions. Moreover, the improved patient–physician interaction can also enhance patients’ self-care skills as well as their adherence to treatment [69]. Effective information exchange can also help the physician to understand the patient’s preferences and to find a solution that best matches the patient’s expectations. Mutual collaboration between physicians and patients not only affects the health outcomes of patients, it may also reduce medical costs by increasing the efficiency and cost-effectiveness of medical visits. In particular, it may help to avoid unnecessary tests and re-admissions – factors that contribute significantly to increased medical costs [9–11].

Physicians need to recognize that they have a role in helping patients through the process of accessing and understanding information. Therefore, health care providers can take advantage of this opportunity to promote patients’ awareness of illness. For this purpose, these providers should be trained to deal with patients with rare diseases and they must learn to cope with assertive and well-informed expert patients. In particular, they must become skilled at communicating in ways that build and sustain positive patient relationships by sharing their knowledge openly and by encouraging patients’ self-reliance. Educational associations and institutions in health care should encourage physicians to practice and improve their skills in this area. Our framework suggests that attempts to educate, train and support physicians to promote patient participation need to consider physicians’ underlying professional role identities and communication skill sets. Another crucial factor influencing the satisfaction of patients with rare diseases is related to physicians’ knowledge about the disease. Therefore, health care policy should create the appropriate regulatory environment that helps physicians to stay up to date on the medical literature and new treatments. Examples of providing help in knowledge acquisition and exchange among medical professionals include the implementation of a telematic platform and the pooling of knowledge in centers of expertise.

Although a previous survey conducted on rare disease patients from 16 European countries confirms that the problems that these patients face are similar across the national health care systems, our results must be discussed in view of the health care system level [79]. A major challenge in the German health care system is related to the separation of the ambulant and the inpatient sectors, which makes the coordination and information exchange in the treatment of rare diseases difficult. To improve the treatment of patients with rare diseases, health care policy makers must refocus their efforts on the broader care continuum. The establishment of networks of cooperating experts in various medical specialties may help to relax the traditional lines between outpatient and inpatient care.

Patients in Germany enjoy a high degree of free access to health care and choices among a wide range of specialists. This is especially beneficial for patients with rare diseases because it allows them to change their doctor if his communication style does not correspond with their expectations. However, our findings suggest that approaches that have recently been introduced in Germany to limit the choice of providers for patients may further improve the coordination of treatment for patients with rare diseases. In particular, the gatekeeper role of the GP is crucial in helping patients to find highly specialized providers and to coordinate their treatment. However, a prerequisite of this improved access to treatment is to provide appropriate regulatory structures that enable the GP to perform this task. Because the diagnosis and treatment of rare diseases require more time, health care policy should create the appropriate regulatory environment that allows physicians to spend more time with these patients, for example, by providing more flexibility, improving physician workflow and creating more efficient processes.

Finally, we suggest the transferability of the findings to other diseases. Due to an increasing focus on preventive health care, improved access to medical information and the growth of consumerist attitudes in health care, patients become more actively involved in decisions concerning their treatment. Further studies in this direction could lend insight into how physicians deal with the emerging role of empowered expert patients.

5. Conclusion

People with rare diseases often face challenges, due to the low prevalence of their diseases and the resulting lack of knowledge of their healthcare providers. The present findings reveal that the patient–physician relationship undergoes a significant change that results in role discrepancies. The study shows the difficulties and importance of providing information about a rare disease. The vast majority of patients with rare diseases actively seek information. Healthcare providers should take advantage of this opportunity to promote patients’ awareness of their
illnesses. Finally, policy makers must recognize the unique needs of patients with rare diseases to get support in information seeking and in accessing specialists.

References

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