Perceptions of people with osteogenesis imperfecta about the interventions of the occupational therapy and its possibilities of care

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Abstract: Osteogenesis Imperfecta (OI) is a genetic disorder that compromises the collagen formation, which alters in a significant way the body structures causing osseous deformities. Faced with the different possibilities of care for people with OI, stand out the occupational therapy interventions, that even being poorly explored in the context of the national scientific production, show significant practices to compose a team of care for people with OI. Thus, this study aims to identifying the perceptions of people with OI about work of the occupational therapist. It was done a qualitative study based on the application of semi-structured interviews with five adults with OI. Interview data were analyzed by the technique of analysis of thematic content, allowing to cluster the results into three categories: “Knowing the participants: characterization of the sample”; “Living with osteogenesis imperfecta: possibilities and constructions”; and “Occupational therapy and osteogenesis imperfecta: perceptions, fragments, and experiences”. The results lead to the conclusion that these professionals have a wide range of practices that still have to be better explored and points out an encouragement for more scientific production in the area.

Keywords: Osteogenesis Imperfecta, Occupational Therapy, Professional Practice Location.

Percepções de pessoas com osteogênese imperfeita acerca das intervenções terapêuticas ocupacionais e possibilidades de cuidado

Resumo: A Osteogênese Imperfeita (OI) é um distúrbio genético que compromete a formação de colágeno, alterando de forma significativa estruturas do corpo e causando deformidades ósseas. Diante das diferentes possibilidades de cuidado para as pessoas com OI, enfatizam-se as intervenções terapêuticas ocupacionais, que embora sejam pouco exploradas no âmbito da produção científica nacional, apresentam práticas relevantes para compor a equipe de atenção à pessoa com OI. Assim, o presente estudo teve como objetivo identificar as percepções de pessoas com OI acerca da atuação do terapeuta ocupacional. Para tanto se realizou um estudo qualitativo a partir da aplicação de entrevistas semiestruturadas a cinco adultos com OI. Os dados provenientes das entrevistas foram analisados por meio da técnica de análise de conteúdo do tipo temática, possibilizando agrupar os resultados em três categorias: “Conhecendo os participantes: caracterização da amostra”; “Vivendo com osteogênese imperfeita: possibilidades e construções” e “A terapia ocupacional e a osteogênese imperfeita: percepções, fragmentos e vivências”. Os resultados levam a concluir que os profissionais da área possuem um vasto campo de prática para ser mais bem explorado, e pontuam um incentivo para produções científicas na área.

Palavras-chave: Osteogênese Imperfeita, Terapia Ocupacional, Área de Atuação Profissional.
1 Introduction

The imperfect osteogenesis (IO) is known as a genetic and hereditary disorder that compromises the formation of type I collagen, directly related to the bone constitution. Type I collagen due to its structural modifications is not able to interact with hydroxyapatite, being one of the components responsible for the formation of the bone structure, and the interaction of these components is of extreme importance for bone resistance. Genetic alterations affect structures and tissues that contain collagen, causing a variability of clinical manifestations (GLORIEUX, 2008; ESCOBAR et al., 2013). The main characteristics of IO are bone and physical fragility and deformity, osteopenia and osteoporosis, short stature, repetitive fractures, gray or bluish sclerosis, imperfect dentinogenesis (ID), hypoacoustic or deafness, skin hyperelasticity, joint hypermobility, chronic pain and in some cases, Wormian bones present in the skull sutures. Cognition is preserved, and some individuals with IO are able to adapt to the limitations caused by this condition (RAUCH; GLORIEUX, 2004).

Silence and colleagues created the first classification of clinical and radiographic manifestations in 1979, allowing the distinction of four types of imperfect osteogenesis (SILLENCE et al., 1979). In 2004 and 2007, this research expanded, increasing the classification to eight types (VAN DIJK et al., 2010).

It is estimated that there are about 19,075 individuals with IO in Brazil, and only 788 people in treatment, most of them in the Southeast region. This estimate was made by the Brazilian Association of Imperfect Osteogenesis (ABOI) (LIMA; HOROVITZ, 2014). According to this ordinance, the professionals of the multidisciplinary team of the Reference Centers in IO Treatment are nutritionist, nurse, physiotherapist, physiatrist, pharmacist, psychologist and social worker (BRASIL, 2001).

Besides these professionals, the person diagnosed with IO can benefit from the care offered by the professional occupational therapist as evidenced in the work of Cheung and Glorieux (2008), which discusses the actions of this professional in positioning, motor development, mobility, function in daily life, social integration and school performance. According to Bishop e Walsh (2014), reduced mobility due to IO can be the object of occupational therapy intervention through improvements in the instrumental activities of daily living, often in simple adjustments, such as improving the route of going from home to work.

Considering these observations, it is necessary a rescue carried out through the Federal Council of Physical Therapy and Occupational Therapy (COFFITO), on the concept of OT as an area of knowledge focused on the prevention and treatment of cognitive, affective, perceptive and psychomotor changes, whether or not resulting from genetic, traumatic and/or acquired disorders, through the systematization and use of human activity as a basis for the development of specific therapeutic projects (CONSELHO..., 2014).

Thus, considering the different possibilities of care for people with IO, emphasis is placed on OT interventions, which, although little explored in the context of national scientific production, present relevant practices to compose the care team for people with IO. Thus, this study aimed to identify the perceptions of people with IO about the work of the occupational therapist.

2 Method

This is a descriptive exploratory study with a qualitative approach. The initial procedures were carried out by inviting a group of people with IO, professionals such as physiotherapists, occupational therapists (OT), psychologists, among others; the use of biphosphonate drugs responsible for inhibiting the process of bone resorption (BASEL; STEINER, 2009; MONTI et al., 2010).

Treatment for IO has been available in the Brazilian public health network since 2001, with the institution of Ordinance 2305, developed by mobilizations carried out by the members of the Brazilian Association of Imperfect Osteogenesis (ABOI) (LIMA; HOROVITZ, 2014). According to this ordinance, the professionals of the multidisciplinary team of the Reference Centers in IO Treatment are nutritionist, nurse, physiotherapist, physiatrist, pharmacist, psychologist and social worker (BRASIL, 2001).

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through the director of ABOI and groups of social networks related to the theme.

The following inclusion criteria were defined for the development of the sample selection: individuals with an IO diagnosis, over 18 years old and who agreed to participate in the study. Thus, the sample universe was composed of five (5) individuals with IO who have or have not undergone occupational therapy.

A self-administered questionnaire was developed by the researchers to collect data based on previous studies on the individual. The questionnaire developed contains eight open questions and includes personal data, questions about the perceptions of occupational therapy, about what is OT and about the family relationship.

The questionnaire was sent to the virtual groups indicated by the sources already mentioned. For the development of the instrument, the study of Flick (2009) on online questionnaires was considered, which points out the need for instructions that are detailed, so the participant of the research alone can answer the questions without any doubt. The use of this type of method has the advantage of being able to reach a large number of people, even if they are in other far regions.

Data from the questionnaire were analyzed through content analysis, thematic type, described by Bardin (2004). According to this author, content analysis is a research technique by objective, systematic description of the manifest content of the communication, focusing on historical psychological and sociological variables Bardin (2004).

This paper was approved by the Ethics and Research Committee (CEP) of the Federal University of the Triângulo Mineiro (UFTM), under the protocol number 788.562/2014. An Invitation Letter with clarification data replaced the Free and Informed Consent Term. During the sample survey, a total of fifteen individuals with IO, older than 18 years old were enrolled. However, only five participants answered the e-mails and were included in the study.

3 Results

The study sample consisted of five female participants, aged between 26 and 47 years old. Pseudonyms were associated with flower names (Margarida, Violeta, Lírio, Rosa, and Frésia) to refer to the interviewees throughout the work.

Regarding the educational level of the participants, it was verified that one of them had complete secondary education and the others had complete tertiary education.

In the professional context, three participants worked with administration, one with journalism and another was unemployed.

Discussing the family composition, four participants reported that they were single and lived with relatives (parents and children) and one was married and lived with the partner.

Among the clinical conditions of IO-related participants, there were short stature, scoliosis, lordosis and kyphosis, spine problems, chronic asthma, right hip arthrosis, body aches. As an assistive technology resource, the use of the wheelchair for long distances and crutch for short distances was mentioned. It should be noted that the types of IO were not questioned since the objective of the study was not focused on the specificities of clinical limitations.

Only two of the five interviewees received occupational therapy intervention, but they all knew the profession in some way. Thematic categories were elaborated that sought to group the sense cores of the texts coming from the interviews to explore the participants’ perceptions about the performance of this professional. In this way, the categories were entitled: “Living with osteogenesis imperfect: possibilities and constructions” and “Occupational therapy and imperfect osteogenesis: perceptions, fragments and experiences”, the second one being fragmented into two subcategories: “Fragments and experiences about Occupational Therapist Care” and “Perceptions on Occupational Therapy Care”.

3.1 Living with imperfect osteogenesis: possibilities and constructions

This thematic category has the purpose of presenting the data regarding the participants’ perceptions about the daily life with IO, how they deal with the condition and their daily activities.

In general, participants were shown to engage in multiple activities such as self-care, work, leisure, household activities, sexuality (mentioned by a participant), and general care (such as caring for pets, partners, family, and children). They referred to these activities independently and autonomously. Only one interviewee reported having a helper assisting her in household chores.

[...] I have a girl so I give her orders to help me at home with some obligations, she does not always do everything, with something on the street, I come home at 5 p.m. or later it depends on what I
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have to do, the times step in the market or buy pet food. At home when I arrive I go on the net and always make my dinner I usually dine on the net and I usually go to sleep about 11 p.m. or later (Margarida).

It should be emphasized that the interviewees were actively involved in activities with important social components, such as religious groups, study groups, and friends. One participant was the president of the Disability Support Group Council, another participant was responsible for organizing meetings of the Reference Center on Imperfect Osteogenesis (CROI), and another was a volunteer.

Besides these activities, they also performed practices for the improvement of their physical condition, such as hydrogymnastics, physiotherapy, massage therapy, swimming and medical monitoring for comorbidities associated with IO. Regarding the use of assistive technology for locomotion, only one of them reported using wheelchairs and crutches for short distances, as verified in the following quotation.

The days are very varied because the profession has irregular periods. The only routine is swimming 2 times a week. The schedules also vary according to the physical layout of the day or the importance of each activity. There are days when I'm much more physically tired, but I usually wake up at 8 a.m. and go to bed at 11 p.m. [...] (Frésia).

The causes of the non-performance of some activities were lack of time; the participant Lily reported that she did not perform some activities due to the lack of money, and Frésia referred to restrictions due to current emotional issues experienced by unemployment. Violeta reported the lack of accessibility for the tasks she would like to accomplish, as described below:

I feel the need to try out activities such as skateboarding, skating, rappelling, parachuting, cable-car ride between mountains and various other activities. The non-achievement of activities is the lack of ACCESSIBILITY preventing its performance (Violeta).

The participants’ speeches referred to the need for constant care, which produced outcomes such as feelings of overprotection, fear, too much care and constant search for improvements in quality of life. Margarida, for example, reported that she was always very afraid of overprotection, also said she had many restrictions, and the deformities caused by IO bothered her because she was a pretty vain woman. Violeta reported that there was always something she had to adapt to use, but she could not and did not stop living because of IO.

I confess that it is a tragic and delightful challenge, that sometimes I am very angry at living, and at other times, I do not know what it would be like if I had not been born with this physical condition (Frésia).

IO for me today is ok but I have always been very afraid, I think by overprotection. It means a lot of restrictions and living with the deformities that always bothered me so much because I was a woman and vain has always bothered me (Margarida).

The relationship of family and friends with IO was positive in the participants’ reports: Margarida, Violeta, Lírio, and Rosa. However, they mentioned discrimination outside their family and social nucleus. Frésia reported suffering prejudice from 90% of her relatives and some friends, and it is possible to observe the prejudice in the family context.

Today it is a little better, but I still face much prejudice and discrimination of 90% of the family and some friends (Frésia).

I could not have had a better creation than my family gave me, and I could not have found better friends in this life (Rosa).

My relatives at first have always been very afraid and difficult to handle the person with IO, because of the fragility due to the complexity that the disease brings when still very young, but at no time they refused to learn, to seek to give without embarrassing always supporting even repeating careful that due to repetition, brings a little fear of in making decisions, of having initiatives. Always surrounded by friends, many of them lay in relation to illness, but always open to learning with little prejudice and taboos, but with coexistence, it is possible to modify concepts in relation to the disease (Violet).

3.2 Occupational therapy and imperfect osteogenesis: perceptions, fragments, and experiences

In this category, the objective was to present the participants’ perception about the professional occupational therapist’s performance. Thus, it was observed the need to divide this category into two
subcategories, entitled: “Occupational therapy for those who experienced the care of this professional” and “Occupational therapy for those who did not experience the care of this professional”.

3.3 Fragments and experiences about Occupational Therapist Care

The data presented below are related to Margarida and Violeta, who came into contact with occupational therapy intervention at some point in their lives.

Margarida has made treatments for IO throughout her life, attending OT sessions for three years, and she has stopped doing this treatment for eight years.

It is noteworthy that Margarida is 41 years old and her clinical conditions are stable. She reported great autonomy in her daily activities, relying only on someone to assist her in her residence. Her contact with the profession of OT was through handicraft workshops, where they were made cross-stitch, works with newspapers, painting in wooden box, biscuit, and imitation jewelry. The participant was not able to directly point out the objectives of OT, although she pointed out the benefits that the therapy provided during the interventions.

In addition to taking my time, because getting to bed recovering is not easy after I recovered also helped me for my nervous anxious behavior that therapy calms me (Margarida).

Violeta, 26 years old, received occupational therapy during sporadic hospital admissions. During her sessions, she developed activities such as workshops for crafts and music. When questioned about the knowledge of the profession, Violeta said that since she was 5 years old, she had been doing follow-ups with psychologists, apparently presenting a lack of distinction between the professions.

This participant did not answer the question about what OT’s objectives were in the activities performed, but in an account about the importance of OT in her life, she was able to bring some aspects about the objectives of occupational therapy.

Occupational therapy has a power to detach you from suffering whether it is sudden, temporary, or continuous. It is able to contribute and fortify a basis so you are not afraid to face so many other activities, properly it works the occupation it provides a diverse exploration feeling that often become inertia due to lack of stimulation (Violet).

3.4 Perceptions on occupational therapy care

The data below are related to Lírio, Rosa, and Frésia, who did not have direct contact with OT.

Although not attended by the occupational therapist, the participant Lírio reported believing that this could help in the quality of life, referring to the lack of opportunity to know the profession. Lírio believed that OT could help with activities related to physical condition.

I believe it is activities that improve the physical conditions of those who need it (Lírio).

The participant Rosa in her perception about OT said she believed it was a very important and good work, but she reported never having needed it because of the way her family treated the IO problem.

I believe it is a very important job and it brings good results, but in my case, it was never necessary because of the way my family has always faced the disease and created me. I did a couple of years of group therapy with other children who had no pathology followed by a psychologist because they were afraid they might have an existential crisis as they grew up. But since I never had this crisis, after a few years the psychologist ended up dispensing (Rosa).

Frésia reported knowing OT only in theory, and said that she always thought that the profession was directed to the care of “intellectual problems”.

4 Discussion

When discussing the thematic category “Living with imperfect osteogenesis: possibilities and constructions”, reflections on how is to have IO for the participant, how they deal with diagnosis and what it is like to have physical disability in today’s society have emerged.

According to the National Policy on the Health of People with Physical Disabilities, the term physical disability is defined as the concept that reflects the close relationship between the limitations that people with disabilities experience, linking the structure of the environment and community attitudes (BRASIL, 2010).

According to the results, the participants showed physical alterations typical of the clinical picture of IO but did not refer them as limiting factors for the accomplishment of a large part of the significant daily activities, although relevant difficulties were considered.
Locomotion and accessibility in spaces, for example, were frequently cited by participants. These statements have raised concerns about what kind of care and social support has been offered to people with IO and how society is preparing to deal with the diversity of characteristics of the population.

These reflections are closely related to the observations of the participants about the prejudice experienced in the family and social context. Prejudice arose in all lines, being more frequent in the social context, especially in the family context. Prejudice constitutes an assignment of quality to a particular group or individual, without first knowing it, demonstrating a hostile attitude and aversion (PINHEIRO, 2011). The physically handicapped still faces stigma by society and family, not only focusing on distancing reactions, but also indulgence reactions, attributing “pity”, “pain” and lack of capacity.

The feeling of being “differentiated” is also identified when discussing the care they demand from family members. At that moment, not with a pejorative connotation, the participants mentioned about “overprotection” and “special care”. Dogba and colleagues (DOGBA et al., 2013) point out that the trauma and bone deformities resulting from IO lead to limitations, physical disabilities and the need for care usually taken by relatives, such as mothers.

Margarida, Violeta, Lírio, and Rosa said that their relatives demonstrated an active role in IO care and had no stigma within the family and social support networks, but they reported prejudice outside of this scope. In contrast, Frésia reported suffering prejudice within and outside the family unit.

It is inferred that the difficulties reported do not cause the members of the study to lose the desire to get involved and participate in meaningful activities. In the routine of the interviewees, there was great involvement in activities related to work and social participation, inside and outside the family. Three participants reported engaging in activities such as Imperfect Osteogenesis Reference Center (CROI), Disability Support Groups and other voluntary activities. The activities reported reflected the involvement in different political and social roles.

The significant participation of the interviewees in organizations guaranteeing the rights and citizenship of people with IO prompts reflection on their access to the virtual groups that composed the sample. The importance of these movements and the legitimacy of the struggle of these participants to rescue from the social invisibility of thousands of people with IO who still have few opportunities of treatment and social participation is emphasized.

The dedication to such activities was possible through resources that favor access. The participant Frésia, for example, reports using the assistive technology to get around due to physical disabilities, using the wheelchair for long distances and crutch for small distances, which allows her independence for locomotion, also having an accessible environment in her house and in the spaces frequented. Accessibility is defined as the possibility of access to places, people and objects that the individual needs and/or wants at work, study, leisure, among others, for an insertion into society. Accessibility is the possibility of using, with security and autonomy, public, private and private buildings, their spaces, furniture and urban equipment, providing the greatest possible independence (GONZALEZ; MATTOS, 2014).

It is observed that devices and environments with accessibility are not available to all, as can be verified in the case of participant Lírio, who due to difficulty in locomotion caused by arthrosis in the right hip, she would remain most of the time at home. Violeta feels lack of accessibility to practice extreme sports.

Sports and physical activities have been the subject of studies for adaptations and accessibility. The involvement of some participants in physical activities, such as swimming practiced by Frésia, water aerobics by Violeta and massage by Rosa was verified. The activities performed by the participants were consistent with the indications for the diagnosis of lower impact due to the possibility of bone and joint injuries.

The activities in the water allow free movement, without overloading the joints, preserving the bones and strengthening muscles. These activities also contribute to the relaxation of the body, improving self-esteem, self-image, and motivation (GOMES et al., 2014; MAZO et al., 2006).

Performing regular physical activities can bring benefits such as control or reduction of body fat; maintenance or increase of muscle strength and bone mineral density; improvement of flexibility; reduction of resting heart rate and blood pressure; decreased risk of developing cardiovascular disease, stroke, hypertension, type II diabetes, osteoporosis and obesity; improvement of self-esteem and decrease of insomnia, muscular tension, stress, anxiety and depression (MAIA, 2006).
It is considered relevant to highlight that one of the participants mentioned sexuality as one of the activities that make up their daily lives. Sexuality is understood as one of the activities of daily life that is part of people’s lives and that can promote health and well-being. In the context of IO, particularly in physical disability, there are some factors that can be listed as limiting the experience of sexuality: biological, psychological and sociocultural factors (MAZO et al., 2006). There are reports of the difficulty of relating to the own body image and also to sexuality due to physical limitations and deformities (STARR et al., 2010).

Although fear and stigma can become barriers in this context, the participant in referring to a healthy sexuality experience demonstrates that this issue can be developed in a satisfactory way individually with the partner or with the help of a professional.

Regarding the subcategory *Fragments and experiences about the care of the occupational therapist* with the results obtained and explained, it is possible to observe that only two participants, Margarida and Violeta, had direct contact with the OT.

Margarida reported having participated in craft workshops, in which she learned cross-stitch, newspaper works, painting in wooden box, biscuit and imitation jewelry. In the OT sessions that Violeta performed sporadically in the hospital context, participation in artisan workshops and music workshops was mentioned.

From this experience, the use of artisan activities as a resource of OT in its practice is verified. The activity constitutes for the occupational therapist the diagnostic and therapeutic resource of his intervention, being the centrality of this professional’s investments (QUARENTEI, 1994).

The intimacy of occupational therapy with “activities” as a working tool has expanded the repertoire of therapeutic activities that today includes the diversity of human actions such as sharing, listening, grouping, knowing, learning... along with cooking, writing, walking, painting... Conceptual magnification that operates in activities while acting and vice versa (QUARENTEI, 1994, p. 26).

It is possible to identify the subjective and phenomenological aspects in the activity analysis, as well as the symbolism, meanings and personal preferences, style, feelings and effects of the environment in each activity performed. The activity allows the individual to undergo transformations and reflect the discovery about the malleability of the human being, experimenting with new experiences that allow a process of structuring and restructuring, permeating the feeling of freedom to explore its full potential (ARIOLI, 2008; BENETTON, 2008). In the case of Margarida, she attributed a feeling of tranquility as a result of the intervention. Violeta believed that occupational therapeutic interventions could result in decreased suffering and empowerment of the subject to engage in activities.

In this perspective on the activity, the craft, mentioned by Margarida and Violeta, emerged as a possibility of a promoter of cultural rescue, social experience, as a means of expression, communication, leisure, income generation, among other elements that may have done part of the clinical reasoning of the professional responsible for the intervention.

In the specific case of Violeta, the hospital was the therapeutic intervention environment. Within the health area, one of the contexts in which the OT performs its practices is the hospital, and may be inserted in Intra-hospital care, Outpatient care, Palliative care, ICUs, Infirmaries, Outpatient clinics, Neonatal care, among others (CARLO; LUZO, 2004). Besides to its activities in the health area, OT has occupied and strengthened its actions in different contexts, such as educational, cultural, social, leisure, etc.

In this way, the construction of reflections about the interventions with the people with IO develops from the understanding that the OT can follow any process of rupture in the different moments and phases of the person’s life. As examples, care can happen from the gestation, from guidelines and reception of the relatives to deal with the IO and its possibilities; early stimulation of the baby, offering all the necessary stimuli for development, since it is in this phase that larger amounts of fractures occur, due to the bone development and its plasticity; performance in school inclusion, preparing professionals in this area to properly receive patients with IO; there are works done in the adaptation of furniture and environments offering greater autonomy and freedom for adequate mobility in different spaces, and prescription and confection of assistive technology resources.

In the second subcategory *Perceptions on care of occupational therapy*, reports of participants Lírio, Rosa and Frésia, who did not have contact with the OT professional, contributed with their
perceptions about the occupational therapy and the importance of this in the care of IO.

In the results obtained in this topic, it was possible to perceive that the interviewees had difficulties in saying what is the role of OT in the context of IO, but they pointed out actions that the professional performs, pointing out that there is understanding of certain aspects of professional action, even without direct contact.

The participant Lírio reported believing that this professional could help improve the lives of many people and also act in activities that improve the physical condition of those who need it. In Rosa’s reports, the OT’s representation emerged as a very important and successful work, but in her interview, she showed no distinction between the OT professional and the psychologist (all other participants also portrayed this perception). Frésia said that she always believed OT was aimed at intellectual problems. One of the hypotheses about this devolution of participants may refer to the term “therapy” used in both areas of knowledge. On the other hand, the statements of the participants that focus on the physical and mental areas refer to concepts related to the genesis of the profession, in which two processes were basically strengthened: from the occupation of chronic mental patients to long-term hospitals and from the restoration of the functional capacity of the disabled people (MOREIRA, 2008).

Conceptual distancing about what the profession is, as well as the resources used, can be associated with a relatively new field of knowledge, constantly growing and transforming, and having a wide area of intervention, which can hamper in the precise delineation of professional action.

5 Conclusion

From the results obtained, it can be observed that the possibilities of intervention of the OT with the people with IO have great fragilities for deepening and discussion, particularly in the national publications.

Therefore, they conclude that the professionals of the area have a wide field of practice to be better explored since the identified needs of the people with IO meet the proposals of occupational therapeutic interventions. Thus, the importance of the appropriation of the occupational therapist next to this population is emphasized, seeking to guarantee new contexts of practice.

It is expected that the results of this work may contribute as incentives for the encouragement of the discussions to the care of people with IO, as well as for the production of new publications.

References


Author’s Contributions

All authors also contributed to the drafting and writing of the text, as well as its final version.