



A qualitative evaluation of the National Expert Team regarding the assessment and diagnosis of deafblindness in Sweden

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ABSTRACT

Deafblindness is a rare condition with multiple causes. Given its rarity, professionals generally have limited knowledge of this condition and insufficient experience managing it. Accordingly, in Sweden, a National Expert Team was established to assess and diagnose deafblindness. The aim of this study was to identify the conceptions of persons with deafblindness, parents of children with deafblindness and professionals involved in their care, rehabilitation and education regarding their participation in the assessment and diagnostic procedures performed by this national team. A phenomenographic design was employed. The main findings were illustrated by five descriptive categories: *An opportunity for improvement in daily living, ability to interact with the cream of the crop, personal effort, effects of genetics beyond drawing blood and limited professional interaction.*

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Introduction

The aim of this study was to identify the conceptions of persons with deafblindness, parents of children with deafblindness and professionals involved in their care, rehabilitation and education regarding the assessment and diagnostic procedures provided by the Swedish national team of experts for assessment and diagnosis of deafblindness. The participating professionals will be referred to as ‘professionals’ for the remainder of the text. In the phenomenographic method, the term ‘conception’ means different ways of experiencing phenomena in the surrounding world (Sjöström and Dahlgren 2002). An in-depth understanding of deafblind persons’ individual experiences has been argued to be essential for planning services targeted to this diverse group (Fletcher and Guthrie 2013). By examining the perspectives of deafblind persons with those of the professional networks involved (a triangulation approach), a more comprehensive understanding can be obtained (c.f. Heale and Forbes 2013; Polit and Beck 2012). Knowledge of these perspectives could be used to further refine the organization of the Expert Team and could also be of use for other organizations when planning services for rare populations.

As deafblindness is a rare condition, professionals generally have limited clinical experience with its treatment and management (Van Groenendael et al. 2015). The dual sensory loss involved can be present from birth or can be acquired later in life, and the degree of vision and hearing loss varies between individuals. Furthermore, deafblindness can have multiple causes (Dalby et al. 2009; Dam-meyer 2014; Möller 2003; Wittich, Watanabe, and Gagné 2012). Deafblindness presents a number of evident problems in daily life: difficulties with communication (Fletcher and Guthrie 2013; Hersh 2013; Möller 2003, 2005; Rönnåsen et al. 2016), compromised activities of daily life, lack of

independence (Fletcher and Guthrie 2013; Rönnåsen et al. 2016), risk of isolation (Fletcher and Guthrie 2013; Hersh 2013; Möller 2003, 2005), challenges with mobility (Dalby et al. 2009; Fletcher and Guthrie 2013; Hersh 2013; Möller 2005; Rönnåsen et al. 2016) and difficulty performing household chores (Fletcher and Guthrie 2013; Hersh 2013; Möller 2005; Rönnåsen et al. 2016). Some syndromes resulting in deafblindness also affect other organs (Marshall et al. 2007; Möller 2003; Pagon et al. 1981), which requires multiple contacts with the healthcare and rehabilitation system (Anderzén-Carlsson 2015; Van Groenendael et al. 2015). The 2014 Nordic definition of deafblindness claims that deafblindness can limit activities and restrict full participation in society in such a way that society is required to facilitate specific services, environmental alterations and/or technologies for this population (Nordic Centre for Welfare and Social Issues 2014).

Depending on the complexity of the condition, persons with a diagnosis of deafblindness can need rehabilitation, healthcare (Van Groenendael et al. 2015) and social services support, while those not yet diagnosed may be referred to public healthcare specialists for diagnostic examinations. In Sweden, three multidisciplinary teams operated by the public healthcare system in the three largest cities are available for persons with deafblindness. In addition, there is a national state-funded resource for persons with deafblindness called the Swedish National Resource Centre for Deafblindness (NKCdb, n.d.).

The NKCdb organization includes a well-established team of experts on deafblindness who can support public healthcare providers with assessing and diagnosing deafblindness. This Expert Team has been active since the mid-1990s and uses a multidisciplinary structure (including an audiologist, ophthalmologist, geneticist, paediatric neurologist, psychologist, special education teacher, social worker and administrative coordinator). The team has assessed approximately 150 persons to date (NKCdb, n.d.). In some cases, their extensive familiarity with persons with deafblindness has led to the identification and clarification of genetic causes of deafblindness (Rendtorff et al. 2011).

In practice, persons with deafblindness can apply to receive an assessment by the Expert Team. Based on an anamnesis, for example, of vision and hearing, the team decides whether it is appropriate to begin a full assessment and diagnostic process. If the full process is warranted, the person signs an informed consent that allows the team to collect all medical records from her/his regular healthcare contacts. These documents form the basis of the team's assessment and search for a genetic or medical diagnosis. Additionally, one of the experts usually meets with the person with deafblindness. The team members then decide on which additional examinations or tests are necessary. For example, genetic tests are routinely recommended to identify the genetic causes of deafblindness. The team meets approximately four to six times per year to discuss new results and decide on next steps. They maintain contact with the person being assessed through personal consultations, by letter or by telephone. The duration of the process varies, but it is not unusual for an assessment to take more than a year, especially if advanced genetic analyses are needed to reach a diagnosis. At the end of the diagnostic process, the team, together with the person with deafblindness, invite next of kin and relevant professionals to a formal meeting, in which the members of the team provide an oral summary of the medical history, share the results, summarize the identified needs and suggest rehabilitation and social services support. After this meeting, the information provided is summarized in a formal document that is distributed to those who participated with the approval of the person with deafblindness.

The present study is part of a formal evaluation of the team's work and includes an examination of the individuals' and professionals' perspectives.

Methods

Design

A descriptive qualitative design was employed. The data were analyzed using phenomenographic analysis (Sjöström and Dahlgren 2002). In phenomenography, the focus is on the 'second-order

Table 1. Interview guide for persons with deafblindness/parents.

Topic guide for persons with deafblindness/parents

1. Please tell me where and when you obtained information about the opportunity to be assessed by the Expert Team
2. Please tell me about how the assessment was initiated
3. Please tell me about your experiences with the assessment, from the very beginning until the end
4. Please describe the concluding meeting and your experiences with it
5. Please tell me about your experiences with the written summary
6. Please describe how you perceived your role in the assessment process
7. Please tell me about your experiences with any uses you/your family had with the assessment performed by the Expert team
8. Please tell me about your/your child's professional contacts prior to the assessment period and how they are today
9. Would you like to offer any suggestions to improve the assessment process conducted by the Expert team in the future?
10. Demographic data

Probing questions were continually asked to elicit further explanations of the participant's answers to help the researcher capture their conceptions of the assessment and diagnostic process

perspective', which concerns how the phenomenon of interest is conceived, in contrast to 'the first-order perspective', which more describes 'how things really are' (Marton 1981). The phenomenon under study was involvement in the assessment and diagnostic processes provided by the Swedish national team of experts for assessment and diagnosis of deafblindness. For the person with deafblindness, this involvement pertained to being assessed with the aim of finding a genetic or medical diagnosis; for parents, it meant assisting their child and having contact with the experts and for the professionals, it meant collaborating with the experts in the assessment and diagnostic processes.

Sample and setting

In total, 13 persons were interviewed in the present study (3 persons with deafblindness, 4 parents of 2 children with deafblindness and 6 professionals). All participants were native Swedish speakers.

Two purposive samples (Patton 2002) were obtained; first, 11 persons who had attended the formal meeting after the Expert Team's assessment and had received a written summary of the results between 1 and 3 years prior to the interview were contacted by letter and informed about the study. Three adults who had received an assessment (all women, aged 30–51 years) and four parents of two children aged 9 and 17 years agreed to participate in the semi-structured interviews. The aetiology of deafblindness varied within the sample; however, the three adults were identified as having acquired deafblindness, and the children had congenital deafblindness.

Second, based on recommendations provided in these interviews, 11 professionals were nominated to be approached by the researcher and invited to participate in an interview. Professionals were included if they had ongoing professional contact with the person with deafblindness at the time of the interview, had also had such contact prior to the assessment period and had been present at the concluding meeting at the end of the assessment. Six professionals agreed to

Table 2. Interview guide for professionals.

Topic guide for professionals

1. Please tell me when you obtained information about the assessment performed by the Expert Team
2. Please tell me about your experiences with the assessment, from the very beginning until the end
3. Please tell me about the concluding meeting and your experiences with it
4. Please tell me about your experiences with the written summary
5. Please describe how you perceived your role in the assessment process
6. Please tell me about your professional contacts with the person with deafblindness/his or her parents prior to the assessment period and how they are today
7. Do you have any prior experience with the assessments conducted by the Expert team?
8. Would you like to offer any suggestions to improve the assessment process conducted by the Expert team in the future?
9. Demographic data

Probing questions were continually asked to elicit further explanations of the participant's answers to help the researcher capture their conceptions of the assessment and diagnostic process

Table 3. Presentation of the analysis process.

Methodological steps (Sjöström and Dahlgren 2002)	Procedure conducted
Familiarization	All interview transcripts were read in their entirety to help the researcher become acquainted with the text
Compilation of responses	All identified conceptions (i.e. responses) related to the aim of the study were marked in each interview transcript, starting with all the persons who were assessed by the team, followed by the parents of the children who underwent an assessment and finally, by the professionals
Condensation of the individual responses	The most significant views in each interview were summarized
Preliminary grouping of similar responses	All identified conceptions at the interview level were combined at the group level (i.e. including material from more than one interview) and abstracted into preliminary categories of descriptions based on similar content
Preliminary comparison of categories	The categorization was refined – all identified conceptions were re-read, and some revisions were made
Naming the categories	The preliminary labels were refined, resulting in the final names of the descriptive categories
Contrastive comparison of categories	The unique characteristics of the categories and the resemblances between them were described

participate in the interviews. They were all women and included one occupational therapist, two low-vision therapists, one physician, one social worker and one special education teacher. One nominated professional declined to participate, and another four were unable to be reached.

Data collection

Semi-structured interviews were performed in line with phenomenographic methodology (Bowden and Green 2005) and covered the participants' experiences and conceptions related to the assessments of the Expert Team. Questions within three main areas guided the interviews: (1) the assessment process, (2) the formal meeting with the Expert Team at the conclusion of the assessment and (3) the impact on daily life and rehabilitation after the assessment had been completed (Table 1). The participants were also asked to suggest how the work of the Expert Team could be improved. Individual interviews were conducted with persons with deafblindness. All interviews were in spoken Swedish, but one interview was also translated into sign language by a professional interpreter. The interviews with parents were performed in pairs. Three interviews were held in the participants' homes, and two were conducted at a research centre. The individual interviews with the professionals covered the same topics, but the responses provided related to their professional perspectives (Table 2). The majority of these interviews occurred at the professionals' workplaces. All interviews lasted approximately one hour and were performed by the author of this paper (a paediatric nurse and researcher experienced with the methods used). The researcher was previously unknown to all but one of the participants. The interviews were audiotaped and transcribed verbatim by an experienced secretary.

Data analysis

The analysis was performed by the author in accordance with the steps described by Sjöström and Dahlgren (2002) (Table 3). The QSR NVivo 10[®] software (QSR International Pty Ltd, Doncaster, Victoria, Australia) was used as a tool in the data analysis.

The results of the study are provided in descriptive categories that illustrate the variations of the world experienced by the participants, as outlined by the method used. This differs from phenomenology, for example, in which an essence of the invariant meaning is sought (Sjöström and Dahlgren 2002).

Although the method implies a stepwise abstraction of data, in practice, there was constant interaction between the steps (Dahlgren and Fallsberg 1991).

Ethical approval was granted by the Regional Ethics committee in Uppsala, Sweden (dnr: 013/056). All participants signed an informed consent.

Findings

The data analysis yielded five descriptive categories: *An opportunity for improvement in daily living, ability to interact with the cream of the crop, a personal effort, effects of genetics beyond drawing blood and limited professional interactions.*

An opportunity for improvement in daily living

This descriptive category included the expectations and hopes, as well as successful outcomes, of the diagnostic process. It also covered the disappointment with and lack of apparent effects of the services provided in daily life after the assessment.

The assessment was considered an opportunity to obtain a diagnosis, which was an opportunity that those affected (persons with deafblindness/parents) could not resist. The reasons for this varied; having an assessment was important for being taken more seriously when applying for different services, as it could facilitate one's position within the group of people with deafblindness, and there was also hope that the diagnosis could lead to a cure. Furthermore, a diagnosis was important for professionals in conducting their ongoing visits with patients, and the fact that the public healthcare system lacked the knowledge and resources to perform complete and thorough assessments of deafblindness was also highlighted.

Receiving a diagnosis or simply being enrolled in an assessment was highly important for persons with deafblindness/their parents. It was considered a relief that made their life easier in many ways. New interventions and support services were suddenly available:

So for me, it was like, when I came to see NN [member of the Expert Team], he rubber-stamped me, like the doll that has its OK-stamp in Santa's Workshop, [a Disney film], [he said I had] deafblindness, and then all the doors were opened wide. (Person 5)

For some, the diagnosis also provided a sense of revenge against officials who had previously refused to provide the services for which they had applied. The assessment provided an increased awareness of their own rights. The majority of persons with deafblindness/their parents used the formal written summary to inform their next of kin and professional network and to claim their rights.

By attending the formal meeting with the Expert Team at the end of the assessment, the professionals became aware of the medical history and needs of the person with deafblindness. This understanding was a relief to persons with deafblindness/their parents, as they did not have to retell their story to the professionals and argue for their needs.

The actual everyday benefits mentioned by persons with deafblindness/parents as well as by the professionals included better access to social services and adaptations to the school environment. Professionals' increased awareness of other members of the professional network after having met at the formal meeting seemed to have an impact on their cooperation. For some persons with deafblindness/parents, this reduced their responsibility and the energy needed to coordinate healthcare, rehabilitation and social services. The outcome of the assessment also had an impact on the daily lives of the next of kin; some were relieved by the increase in social services provided, which in turn lessened the burden of the person with deafblindness.

From the professionals' point of view, the increased knowledge of the diagnosis and prognosis obtained from the assessment and the Expert Team was important for providing a more tailored rehabilitation:

Well, this prognosis could include a loss of sight within two years. What will she need then, in terms of her hearing? [After becoming aware of the diagnosis and prognosis, I asked myself] How should we plan ahead? Should we quickly start mobility training to prepare her? Should we introduce a guide dog; is that possible with her current hearing?

What are her computer skills? How will she be able to receive information? Is she confident in sign language; should we introduce tactile signs; well, we have to discuss these kinds of things. So, therefore, I believe that my job, our job within the deafblind-team, is secondary to the work of the Expert Team. (Professional 10)

The formal meeting deepened the professionals' understanding of their fellow professionals' responsibilities, which was considered potentially beneficial to their work and thus also to the person with deafblindness. Furthermore, they found that by meeting other professionals face to face, it was easier to initiate future contact: 'We have been introduced to each other now, and as strange as it might seem, it is easier to make a call when you know who is on the other end of the line' (Professional 7). The professionals found the opportunity to ask the experts questions and to have discussions with other professionals at the formal meeting valuable. They also greatly appreciated the formal written summary that they received. They attached it to their medical records, etc., and at least for some time, it was used as the basis for planning interventions and providing organizational learning. It was re-read as needed to refresh the professionals' memory.

Although meetings with other professionals were believed to be valuable, very few examples of actual new collaborations between professionals with different backgrounds were expressed by the participants. Likewise, only a few obvious effects were experienced in the healthcare and rehabilitation sector, as a consequence of the assessment and diagnostic process.

There were some accounts of disappointment expressed by persons with deafblindness/parents when, for example, the professionals at the concluding meeting were reluctant to assume responsibility for the suggested follow-up activities for the person with deafblindness: 'I had the impression that everybody was looking around and [thinking], well, who will take on the responsibility? That's often how things are. They looked at each other, and finally someone agreed to take me on' (Person 1). Furthermore, some professionals agreed to assume a coordinating role for future rehabilitation services but later failed to act. There was also disappointment that some professionals did not show up to the meeting. The persons with deafblindness perceived this as a lack of interest, while the professionals highlighted the importance of sharing information about the content of the meeting at a later stage with those who did not attend. Finally, the diagnosis itself could be disappointing either because there was no cure or because the initial diagnosis was later found to be incorrect. Most of the negative conceptions were shared by persons with deafblindness and the parents and were less often expressed by the professionals.

The majority of the conceptions corresponded between the person with deafblindness and the professionals with whom she/he had a professional relationship. For example, they shared the conception that obtaining social services support had been difficult prior to the assessment and became easier afterwards. On the other hand, some persons with deafblindness and parents expressed a total lack of outcomes after the assessment, while the professionals within their professional network described an increased understanding and the provision of tailored interventions based on the outcome of the assessment.

Ability to interact with the cream of the crop

Meeting the Expert Team was considered encountering the Swedish elite within the area of deafblindness. These types of responses included conceptions of the experts as having knowledge and power, as well as a professional manner. Nevertheless, the Expert Team received some criticism, as certain aspects of the diagnostic process were considered sub-optimal. Suggestions were made to improve the team's efforts.

Involvement with the experts elicited emotions from both the persons with deafblindness/parents and the professionals. Those affected by deafblindness felt as though they were VIPs: 'Well, in fact, I felt privileged to be selected by the Expert Team and offered this opportunity [to be assessed]; I really appreciated that' (Parent 2). They were thankful for being invited to be assessed and felt that the experts were genuinely interested in them. However, the substantial interest expressed in them

also led some to feel as though they were research objects. Additionally, interacting with the experts occasionally left the professionals feeling inadequate.

At the formal meeting at the end of the assessment period, the experts adopted a professional approach, took the lead, spoke up for the person affected by deafblindness and argued for her/his needs: 'NN [member of the Expert Team and chair of the meeting] was quite forceful in ensuring that my needs were met! [He said] "Who will assist her? Who will take on that responsibility?" Well, [questions] like that' (Person 5). Persons with deafblindness/parents and the professionals expressed that the experts provided information about the assessment in a clear and confident manner. The person with deafblindness was considered holistically, although the major focus was on concerns specifically related to deafblindness. When the information was presented in a positive way, it created hope for the person with deafblindness.

Using the written summary to argue for one's rights was explicitly encouraged by the members of the Expert Team: '... and finally, I received a document with the names of the professors. NN [member of the Expert Team and chair of the meeting] said, "The names will make a significant difference; this document will be very important for you"' (Person 3). The professionals also acknowledged the influence of the experts and found their written summaries and other documents useful when assisting the person with deafblindness apply for services.

The participants believed that there were no closed cases for the Expert Team. Although the diagnostic process had ended, the cases were still considered open. For some persons with deafblindness/parents, this meant that it was acceptable for the team to further investigate their genetic samples when team members became familiar with new research of relevance. Furthermore, contact with the Expert team was perceived to be ongoing; this was natural in some cases, as members of the Expert Team could also have separate professional relationships with the affected persons, e.g. being the doctor in charge. Both the affected persons and the professionals believed that the Expert Team should be responsible for conducting updated assessments and follow-up:

Well, maybe they [the Expert Team] should call and ask 'How are things with you? Is there anything more we can do for you?' and questions like that. However, maybe they are not responsible anymore ... I think that as experts, they should perhaps offer [functional assessments], well, offer advice about rehabilitation - not only a diagnosis but also follow-up of the rehabilitation and healthcare provided. (Professional 6)

The unique knowledge of the Expert Team was useful for professionals in their daily work. They understood that it was acceptable for them to contact the experts at any time, and having the national team as a resource was considered important:

... They are the ones who have the knowledge and skills relevant to this condition [deafblindness] in Sweden. It is really hard to get things to work on a daily basis. Even in a special school where one would expect the staff to know and understand, they don't. In such cases, it is important to have access to the collective knowledge of the Expert Team. (Professional 11)

Expertise in the area of deafblindness implied that the experts would have international contacts, as well as knowledge about and participation in the forefront of research. The Expert Team was viewed as an extraordinary resource. Suggestions were made to use the Expert Team for additional purposes, such as holding lectures in connection with formal meetings, helping establish contact with other persons with the same diagnosis or establishing an official website with information about rare deafblind conditions.

The professionals acknowledged that applying for a diagnostic process incurred costs for the local healthcare system (e.g. for genetic tests, MRI scans), which could make them reluctant to refer a person for assessment. There was also awareness that there could be additional costs following the assessment, after the recommendations for various forms of support and services had been made. However, the professionals believed that there was no alternative to accepting these expenses: '... Well, if you go as far as engaging the Expert Team, you must accept the cost. You send someone for an assessment because you can't manage it yourself. Therefore, you have to accept the consequences' (Professional 9). One professional suggested that the Expert Team should be part of a state organization rather than based on locally employed experts organized by the NKCDdb.

Despite the general admiration for the Expert Team, shortcomings were also expressed. The professionals most frequently mentioned the lack of written consent from the person with deafblindness for the professionals to cooperate with the team. Other shortcomings included the experts being inaccessible, not considering deafblindness when arranging the formal meeting (not employing optimal light or aural environment, etc.) or focusing on vision and hearing loss as two separate entities, instead of treating it as a unique condition. The timeframe of the meeting was also perceived to be too short, not allowing the local professionals to properly interact and make plans: 'Many [professionals] tried to talk to each other across the table, saying "Hey, I would like to talk to you" and conversations like that. [The response was] "Well, I'll have to call you later" (Parent 4).

While most of the criticism came from the professionals, some of those affected also expressed shortcomings, primarily regarding unfulfilled promises by the team or the extended duration of the diagnostic process.

Personal effort

The diagnostic assessment by the Expert Team had noticeable effects on the person with deafblindness, or in the case of a child, her/his parents. The assessment invaded their normal life, as persons with deafblindness had to undergo numerous examinations and engage with the team. It was emotionally challenging, and receiving a diagnosis often resulted in new questions. These conceptions were mainly expressed by the persons with deafblindness.

None of the persons with deafblindness were prepared for the impact the diagnostic procedure had on their daily life:

... Well, I didn't exactly know what embarking on this would mean. It was possible to believe that it would be like simply standing there and consenting, and then someone else doing the rest. Instead, for me, it meant bone density measurements and computed tomography and MRI [magnetic resonance imaging] scans. I had to include so much stuff in my daily life and find enough time for it. So then I began to wonder what I had done [entering into the diagnostic process]. (Person 1)

Apart from the physical invasion into their life, for some, the assessment and later diagnosis led to existential thoughts and feelings about their own identity being in jeopardy, as the diagnosis was rare and there was little information available. However, persons with deafblindness/parents stated that they did not regret their decision to be involved in the diagnostic process. Despite the efforts required, they said they would gladly do it again. Nevertheless, the importance of being offered professional support was highlighted. Having support provided by the local deafblind team was preferable, but the Expert Team was considered to be more aware of the needs and assumed the responsibility for insisting that emotional support was provided.

At the conclusion meeting, some persons felt that they were expected to play an active role by describing their situation and needs. Providing this description was challenging, as they felt they had to tone down their story to prevent those deciding on official support from considering them too independent and consequently withdrawing services. However, the overall conception was that when the persons with deafblindness assumed an active role, the other participants paid full attention to them, and no services were negatively affected.

The persons with deafblindness or parents of children with deafblindness felt included in the process of choosing which next-of-kin and professionals to invite to the meeting, as well as in determining the content of the written summary. Families also became involved during the process; the parents acted as coordinators and provided information to the team.

Effects of genetics beyond drawing blood

Genetics was considered a delicate issue by both persons with deafblindness and professionals.

Some family members became involved in the assessment and diagnostic process because they were asked to participate in genetic testing, and some of them declined to have their genes tested. This refusal could be disappointing to persons with deafblindness.

Genetic testing also led to persons with deafblindness having questions about heredity, and from the professionals' perspective, one concern was the risk that genetic testing might reveal unwanted information that would have to be shared with the persons being tested. It was therefore considered important to thoroughly inform those participating in the diagnostic process and ensure that they were fully aware of what their consent entailed. As genetic findings in a rare population are closely related to new scientific endeavours, there could be a conflict of interest between team members' ongoing research and the patients' right to receive information about their test results and speak freely about them.

Limited professional interaction

The diagnostic process did not occur in a vacuum. During the assessment, there was some limited cooperation between the Expert Team and local professionals, but the professionals' routines remained mostly business as usual.

There was no common pattern regarding the activities within the local environment during the process. There were reports of healthcare and rehabilitation efforts continuing as usual during the assessment, as well as reports that these activities were put on hold to some extent. The professionals perceived that the latter occurred because of knowledge that the assessment process was ongoing, while the former occurred because the professionals were unaware of the ongoing process or considered it a parallel and unique resource. Some interventions were offered during the diagnostic process due to recommendations provided by the team members, such as fitting new hearing aids and adapting the home environment.

The general conception held by the professionals was that they played a minor role in the diagnostic process, with which they were fairly content. They were able to inform the person with deafblindness about the Expert Team, assist in writing applications for the assessment, submit medical records to the team and facilitate blood testing or implementation of the team's various tests or functional assessments in the child's natural environment. They were also approached by the Expert Team to help arrange locations for the formal meeting after the diagnostic procedures had been completed and were invited to participate: 'Well, when it was over [the diagnostic process], of course we were invited. Or rather, we were asked to help with the practical arrangements for the meeting ...' (Professional 10). However, had they been asked, some of the professionals would have been happy to assist in the assessment, for example, by providing information based on their knowledge of the person with deafblindness.

Discussion

The findings reflect the importance of having a national team with expert knowledge in the area of deafblindness. In many ways, the assessments conducted by the team reassured concerned individuals with deafblindness and increased professionals' knowledge. The value of accumulated knowledge is not surprising, as persons with deafblindness are a rare and heterogeneous population (Dammeyer 2014; Möller 2003; Wittich, Watanabe, and Gagné 2012) with whom audiology and ophthalmology professionals do not interact on a daily basis, as confirmed by this study. The importance of identifying the needs of the deafblind population at the individual level has been reported (Dalby et al. 2009; Dammeyer 2012). The identification of these needs also seems to be one of the greatest benefits of the assessments conducted by the national deaf blindness Expert Team, both from the perspective of persons directly affected by deafblindness and from local professionals who were responsible for providing tailored interventions. In a previous interview study with participants from six European countries (the majority from Poland), the results indicated that persons with

deafblindness had received their diagnosis but not any further information and were left to identify services and training and cope with their diagnosis on their own (Hersh 2013). This is not the case in the Swedish healthcare system, nor was it expressed in the present findings.

Formally centralizing the primary responsibility for the healthcare and rehabilitation of the deafblind population through a state organization, as suggested by one informant in this study, might enhance its quality and equality. Thus, further investigation of the benefits of centralizing the primary responsibility for the healthcare and rehabilitation of the deafblind population in Sweden is warranted, as examples within deafblind services in the UK (e.g. NHS England Alstrom syndrome national service) and Norway (e.g. Nasjonal kompetansetjeneste for døvblinde [National Competence Centre for persons with Deafblindness]) demonstrate promising outcomes. In the UK, there are two multi-professional national centres for persons with Alström syndrome (a syndrome causing deafblindness, among other dysfunctions) (Van Groenendael et al. 2015). In Norway, the services for persons with deafblindness are organized into four regional and two national teams, all of which are affiliated with the National Competence Centre for persons with Deafblindness (the coordinating unit) and organized within the public healthcare system (“Nasjonal Kompetansetjeneste for døvblinde” 2015). Adopting a regional centralization within the public healthcare system could mean that the costs associated with assessment referrals may no longer be an issue, while knowledge and equality in terms of the quality of care and rehabilitation could be maintained due to collaboration between teams. Furthermore, it could facilitate sharing of experiences and knowledge building (Government of Sweden, Ministry of Health and Social Affairs 2014). Although some alternative organizations have been presented here, reorganization should not be performed ad hoc; it should instead be evidence-based and planned in collaboration with people with deafblindness, representatives from deafblind organizations and professionals working with this specific population.

The identified expert status of the Expert Team members is most certainly related to their extensive clinical experience and professional interest in the deafblind population. Nevertheless, the fact that the team members were almost glorified by the informants is interesting, but not unique. A strong affinity to service providers with whom a relationship has been established has been described elsewhere (Donohue et al. 2009; Sandy et al. 2012). As those affected by deafblindness felt acknowledged and were grateful for being offered the opportunity to be assessed, they often excused the identified shortcomings of the experts and instead blamed the professionals for a lack of results. This dichotomization could possibly be decreased by establishing a closer cooperation between the experts and professionals within the local healthcare organizations.

The results of diagnostic genetic tests elicited strong feelings in some of the participants, as the findings could jeopardize their sense of their own identity. Previous literature has shown that persons with deafblindness can experience a lack of preparedness for the future due to a sense of uncertainty associated with knowing that they have a degenerative disease (Fletcher and Guthrie 2013). Similarly, in more general terms, information about one’s genetic risk for disorders is known to cause stress (Peterson 2005), and this was also found in this study. Thus, it is likely that persons who have undergone genetic testing may be in need of psychological services (Richmond-Rakerd 2013), as expressed by participants in this study. Accordingly, genetic counselling (pre- and post-test) is considered an essential part of the testing process (Nyrhinen et al. 2009; Richmond-Rakerd 2013). It is unclear whether the persons with deafblindness (or parents of children with deafblindness) in the present study were offered any genetic counselling. However, including this type of counselling in the diagnostic procedures is recommended. It has been argued that professionals who provide genetic counselling should receive special education on topics such as psychosocial counselling, how to provide genetic information and ethical issues related to genetic testing (Nyrhinen et al. 2009). A survey of genetic testing in CHARGE syndrome (a cluster of features including deafblindness) indicated that confirming the diagnosis and assisting research were the two most common reasons for genetic testing mentioned by parents (Hartshorne, Stratton, and van Ravenswaaij-Arts 2011). In the present study, the reasons for diagnostic genetic testing were not separately discussed. However, genetic testing was implicitly expressed as the only way to obtain a diagnosis, which was considered

the main purpose of the diagnostic process, but benefiting research was also mentioned. These two reasons highlight the importance of a robust link between clinical expertise and research, as well as the delicate balance between these two entities, as revealed in the current study.

The results regarding multidisciplinary collaboration after the assessment varied, and the conceptions differed, which should be further investigated in future studies. Similarly, the results were somewhat inconsistent regarding the relationship between the outcome of the assessment and ongoing healthcare interventions and rehabilitation. This raises questions about whether the ongoing collaboration between the Expert Team and professional contacts is currently optimal or whether it should be further developed.

Strengths and limitations

The small number of participants in this study can be considered a limitation. Another noticeable limitation could be that the study was conducted by a sole researcher, which could threaten the trustworthiness of the study. However, to increase the credibility and confirmability, quotations were used to illustrate raw data in relation to the results. Furthermore, during the analysis, QSR Nvivo 10[®] software (QSR International Pty Ltd, Doncaster, Victoria, Australia) was used to increase the dependability during the analysis. This software facilitated the compilation of comparable lists of nodes as well as transitions between the original data and the category system. Qualitative software does not analyze the data, but it does facilitate data storage, coding, retrieval, comparison and linking (Patton 2002).

Despite the potential limitations, the knowledge obtained in this study is valuable for the future organization of the Expert Team, as it provides an understanding of how persons involved with the team perceive the assessment and diagnostic process. Together with the results of previous studies, these findings could also stimulate ideas of how to organize the identification, diagnosis and support process for persons with deafblindness in other countries.

Conclusion and clinical implications

Those affected by deafblindness and the professional network involved in their care shared many conceptions related to the diagnostic process. The Expert Team was found to be important, and the opportunity to be assessed by this team made life easier, despite the fact that it generated emotional strain and did not always lead to an obvious increase in rehabilitation or social service support or cooperation within the professional network. Further studies focusing on collaboration within rehabilitation and social service support for persons with deafblindness are warranted.

Based on the findings, it can be suggested that support and genetic counselling should be integrated into the diagnostic process, as undergoing an assessment and receiving a genetic diagnosis could be perceived as an emotional and personal effort. It could also be worth considering the organization of the assessment and diagnostic procedures for rare disease groups, such as persons with deafblindness. The balance between maintaining competence and expertise in rare diagnoses and broadening and perpetuating local professional competence in caring for persons with deafblindness is delicate. The two main reasons for participating in an assessment and diagnostic procedure were obtaining a definite diagnosis and hope for improvement in daily life. As the field of genetics has rapidly advanced in recent decades, genetic testing and diagnosing are likely to become less exclusive and a more routine component of specialized health care. Consequently, knowledge and functional assessments and planning of individual interventions may be conducted at the local level in the future, possible with the support of national experts. Finally, in clinical praxis it is important to have an entity responsible for assisting in coordinating and evaluating the care and rehabilitation provided, as well as to focus on the person with deafblindness as a whole, not only on the various anatomic dysfunctions and impaired functions experienced.

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