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To cite this article: Berit Rönnåsen, Kerstin Möller, Claes Möller, Björn Lyxell & Agneta Anderzen-Carlsson (2016) Aspects of learning from the perspective of people with Alström syndrome, *Disability and Rehabilitation*, 38:7, 644-652, DOI: [10.3109/09638288.2015.1055381](https://doi.org/10.3109/09638288.2015.1055381)

To link to this article: <https://doi.org/10.3109/09638288.2015.1055381>



Published online: 18 Jun 2015.



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RESEARCH PAPER

Aspects of learning from the perspective of people with Alström syndrome

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Abstract

Purpose: The aim of the study was to explore aspects of learning, from a lifelong perspective, in individuals with Alström syndrome (AS). AS is an autosomal recessive disorder causing early blindness, progressive sensorineural hearing loss, cardiomyopathy, endocrine disorders, metabolic dysfunction, and abbreviated lifespan. **Method:** Eleven individuals with AS participated. The study had a qualitative explorative design, giving voice to the participants' perspectives on their situation. Data were collected using semi-structured interviews, which were subjected to conventional (inductive) qualitative content analysis. **Results:** The analysis revealed in the participants a quest for independence and an image of themselves as capable people willing to learn, but in constant need of support to continue learning throughout their lives to be as independent as possible. **Conclusion:** Based on the levels of functioning, i.e. personal resources, revealed in the interviews, supervisors, caregivers, and teachers are encouraged to allow people with AS to be their own advocates, as they know best how, what, and with whom they learn, and what type of sensory material – tactile, auditory, visual, or a combination – is most helpful.

Keywords

Alström syndrome (AS), deafblindness, dual sensory loss, learning, sensorineural progressive hearing loss, vision loss

History

Received 30 October 2014
Revised 7 May 2015
Accepted 22 May 2015
Published online 18 June 2015

► Implications for Rehabilitation

- Individuals with AS strive for independence, and to be independent they need to continue to learn throughout their lives.
- Individuals with AS know best how they learn, and should be asked what modalities are the most effective for them.
- The tactile modality for learning will continue throughout life and should be emphasized early in the individual's education and rehabilitation.

Introduction

This study focuses on lifelong aspects of learning in individuals with Alström's syndrome (AS). AS is a rare autosomal recessive inherited disorder leading to extreme medical complexity with progressive loss of vision and hearing, and finally to deafblindness. The conditions for life with AS are severe and the need for rehabilitation and support continues throughout life.

Different strategies for learning have been proposed by theorists such as Piaget, who held that people discover and learn new things by trying and testing the world around them, and Vygotsky, who suggested that people learn through social interaction [1]. Both of those strategies are challenged by having AS, since as deafblindness interferes to various degrees with physical, cognitive, behavioural, and social functions, all which are important to learning [2].

Generally, vision and hearing are considered central to gaining information. The ability to see and to hear makes the newborn child aware of the outer world, and engagement with others starts in the social interaction and joint attention in naming objects, which can be regarded as the first steps to a common language and understanding [3]. Without compensation, early vision loss will influence and delay a child's psychological and physical development [4,5] and the impact of a progressive hearing loss on a child's development can also be noticed in various areas [6]. For example, delays in all aspects of language development can arise when necessary input from the environment is hampered by hearing impairment [7]. Speech development requires the ability to discriminate and label speech sounds. Learning to read and write can also be affected by hearing impairment [6,8–11]. A facilitative learning environment is essential for learners to have the opportunity to participate and to take responsibility for their own learning [12].

Alström syndrome is a severe metabolic disorder that includes not only early, progressive, and severe losses of vision and hearing, but also cardiomyopathy (chronic heart failure), obesity,

type 2 diabetes, and other organ dysfunctions. Life span is shortened. The syndrome is rare, affecting approximately 950 known individuals from 56 different countries, including those who are no longer alive [13–15]. Most individuals with AS lose their vision and become completely blind early in life, usually in their teens [16,17]. They also gradually lose their hearing, but not as early. Adults over the age of 30 will be profoundly deaf [14]. The early dual sensory loss and general morbidity in individuals with AS are likely to affect and interfere with their development of individual learning styles and social interactions [15,17,18] and delay their development of theory of mind (the ability to attribute mental states to oneself and others) [19], speech, language, and motor skills [13–15].

There is limited knowledge about the life-long learning process in people with AS, and this study in which the participants share their thoughts and memories of what, how, when, where, and with whom they learned various things throughout their lives is unique. Because such knowledge is necessary both for tailoring educational interventions and for gaining a general understanding of learning in people who are or are becoming deafblind, the aim of the study was to explore aspects of learning, from a lifelong perspective, in people with AS.

Material and methods

The study had a qualitative explorative design, giving voice to the participants' perspective of their situation. Data were collected through semi-structured interviews [20]. Because of the study's explorative design, a conventional (inductive) qualitative content analysis [21] was deemed appropriate.

Study sample

Participants were selected in collaboration with research centers in Sweden and the United States (US), where the patients with AS were well known. Inclusion criteria were a diagnosis of AS in individuals over the age of 18. The study strived for heterogeneity of age and sex.

In Sweden, a letter explaining the aim of the study was sent to eligible individuals. After a week, the first researcher contacted potential participants by telephone and asked about their interest in participating. One person declined participation. The participants recruited in the US were identified and invited to participate in the study by Alström Syndrome International.

Participants

Eleven participants, six women and five men, aged from 18 to 45 years (median = 29), took part in the study. Their origins were Canada, South Africa, Sweden, the United Kingdom, and the US (Table 1). Self-reported data on vision and hearing during different stages of life are presented in the Results section, in close connection with the findings of learning during the same period, whereas self-reported onset are presented in Table 2.

Data collection

The Swedish interviews were conducted in Swedish by the first author. Based on the preferences of the participants, the interviews were conducted in the participants' homes or at a research centre. In the US the first author and a bilingual (Swedish and English) colleague conducted the interviews together. These interviews were conducted in a hotel room in conjunction with an international Alström conference. The interviews were all oral and lasted between 30 min and 2 h. They were digitally filmed and audio recorded. During the interviews, all participants used their hearing aids.

Table 1. Demographics of the participants.

All subjects n = 11	
Age (years)	
Range	18–45
Mean	28
Gender	
Male	5
Female	6
Education level	
High school	8
University/college	3
Current occupation	
High school	3
Part-time work	1
Disability pension	7
Accommodation	
The parental home	5
Boarding school	2
Own home	4

Table 2. Self-reported age at onset of vision and hearing losses and provision of first hearing aid.

Age range (years)	0–9	10–19	20–29
Discovery of vision loss	11		
Discovery of hearing loss	8	2	1
First hearing aid	3	6	2

The semi-structured interview guide covered topics related to both formal (educational environment, including lessons and playtime with schoolmates) and informal situations (life outside kindergarten or school). The participants were given some general life areas and suggested scenarios to help them focus on their learning in different contexts in which the ability to hear and listen was considered important – in school, at home, at leisure, and in society. The interviewer offered to go deeper into each area if the participant wanted to say more about something specific [21]. In consideration of the participants' general health conditions, the interview was paused whenever the participant requested.

Prior to the data collection, the questions were tested in two pilot interviews with one person with deafblindness and one person with hearing loss, neither of whom had the diagnosis of AS. Prior to the study interviews, the questions were adjusted to focus more precisely on learning.

Ethical considerations

The group with this rare diagnosis is small. Special consideration is, therefore, needed to ensure confidentiality, which is why participants recruited both from Sweden and from an international conference were included. Prior to data collection all participants signed an informed consent. The participants received verbal and written assurances that their participation was voluntary and that they had the right at any time, without giving reasons, to suspend their participation. They were informed that they could have a significant person with them during the interview, which one participant did.

The Regional Ethical Review Board in Uppsala, Sweden, approved the study (2012/515, amendment 2012/515/1). The participants in the US were included in an approved protocol granted by the Jackson Laboratory Federal Wide Assurance and The Human Subjects Institutional Review Board (reference numbers 1112000005 and 1112000006).

Data analysis

The voice recordings were transcribed verbatim by professional transcribers. To ensure trustworthiness each interview was listened to, and compared to the transcript.

The qualitative content analysis began with the researchers reading through the transcripts to get a sense of the whole. Meaning units were then identified and coded according to their content regarding aspects of learning such when and what they learned, how they learned (visually, aurally, tactilely, or some combination), what was learned independently and what in collaboration with others, and what things facilitated or obstructed their learning. In the next step, the codes were categorized according to learning in the educational environment and learning in everyday life. The educational environment included formal learning during lessons as well as informal learning during play with schoolmates. All types of courses and special study groups were considered in the analysis to be part of the educational environment. Everyday life was defined as life outside educational environment. Finally the two categories were divided into decades illustrating the lifelong learning process. The first author conducted the analysis in collaboration with the co-authors. This was a back and forward process, moving between the codes, categories and the entire interviews, in order to increase the credibility of the findings. The results are illustrated by quotations, and short passages, drawn from the interviews, describing the participants' self-reported vision and hearing during each decade.

Results

The results are organized chronologically by life decades to show the participants' decline in visual and auditory abilities and their strategies for learning in everyday life and in the educational environment.

Decade 1: 0–9 years ($n = 11$)

Vision: Visual impairment was detected in some participants as early as the neonatal period, and two participants described themselves as having been blind by the age of 3. Most ($n = 9$), however, stated that during their first decade of life they had had good vision, but needed to adapt to light conditions, distance, size, and contrast. Everyone had experienced glare and light sensitivity.

Hearing: The youngest was 5 years old when hearing loss was detected, although everyone reported progressive worsening of hearing during the toddler years. They all said that problems with hearing increased at 8–9 years of age. One of the participants used hearing aids at age 5, and two others at age 9.

Learning in everyday life

Vision, hearing, and touch were all described as important for learning among those who had residual vision. Those who reported that they were completely blind at an early age described themselves as having no clear memories of vision and stated that in addition to hearing, they also used touch, taste, and smell to learn. Someone explained and demonstrated how to do something, after which they tried themselves.

The family was very important for everyday learning, and could always be relied upon to help the participants learn and prepare for adulthood. Participants received no special treatment because of their visual impairment; rather, they had to learn the same family chores as their sighted siblings. Practical learning through active participation in chores at home could involve family projects, cooking, cleaning, and sorting laundry.

My mom began teaching me to cook at home and, once I got into school you know like she would need to teach us cooking skills, daily living skills, we were expected to make our own beds and clean our rooms. (Male no. 5)

As children, participants typically were curious and dared to try things with the support of their family and friends.

My grandmother was probably a bit more protective, and my aunts... They thought that I shouldn't be near a stove or... but my mother thought that if you get burned then you learn... So I got to try cooking and... it happened, I had to pour really cold water on my hands... (Female no. 2)

In conversations, participants sometimes had to ask people to repeat what they said. Those three who had received their first hearing aids during this period said they made it easier to learn to speak and to understand their surroundings. Asking their parents was one strategy described. The parents gave adapted information, for example about AS, as well as advice about issues such as how to handle unforeseen situations or relationships with others.

Activities such as playing and games were learned by imitating friends or through repetition. They either read the rules of the game themselves, or someone read them to them. Several of the participants made their own individual adjustments to their deteriorating eyesight, for example, by adding Braille to playing cards. Grandparents were also described as being supportive in practical learning in the home environment.

Learning in the educational environment

Five of the participants had attended preschool and all 11 began their schooling in the local school. Furthermore, five of them began in the regular 9-year compulsory school and six in a special school or in special groups at school. The curriculum at school was often individually tailored, but they still had to struggle to achieve the learning objectives. By the end of the first decade, adapted teaching was common in the group. One participant had extra support with home instruction when the family was unable to assist with homework.

Visual impairment was described as the largest obstacle to learning in school, but residual vision also helped in the school setting. Glasses, closed-captioned television, magnifying glasses, enlarged text, binoculars for distance, access to educational materials in Braille, tactile materials, and proper placement in the room all facilitated learning. Painted markings made it easier for these students to learn how to find their way in the school environment. Most began to suffer deterioration in their hearing during these years.

Teachers who were described as beneficial for learning were those who taught in small groups, gave private instruction, explained well, provided audio description, and made tactile materials. They also used concrete materials for purposes such as teaching spatial relationships. They made sure that the schoolwork was completed and facilitated the participants' play with the other children. Peer supporters who were the same age were described as significant both inside and outside the classroom. In school, the peer supporter cooperated in the tasks assigned by the teachers, and during breaks they included the participants in play with the other children.

Special education teachers and itinerant teachers provided targeted support with Braille and with speech therapy, offered because some phonetic sounds were difficult to hear and, therefore, to reproduce. Two of the participants stated that they started learning sign language at this age, although only to a limited degree. Half of the participants said that they learned the basics of Braille in preschool.

...they had to make it really big and really bright, bright letters for me to be able to see it and recognize it, and then they tried glasses with me, and they just weren't working. I couldn't

see the pages and the words, it was just too difficult. So they started teaching me Braille. And I was in kindergarten and they started showing me the alphabet, you know, and different letters and then by the time I was in third grade that's when I started learning contractions, Braille to Braille, and learning how to read it and write it... (Male no. 4)

Optimal learning was a challenge even with, and sometimes because of, the special resource available to them for participants who were the only students with impaired hearing in schools full of children who could both hear and see. They said that they had wanted to be like everyone else and that they did not like to use hearing aids, because it made them look different. Several said that they were bullied by both teachers and other children:

I didn't really have much support. I used to get –from elementary school, all the way to high school – I used to get picked on all the time by other kids, teased all the time, and kids would come in junior high, high school kids, would come up to me and tap me on the shoulder, tap me on the head, and run away and play little tricks on me, games and stuff. And I would get mad. I'd lose my temper. (Male no. 4)

Out of all my elementary school years, third grade was probably the worst... I didn't get along with my third grade teacher. She would not wear my transmitter and she wouldn't let my vision teacher and I... work on anything, Braille, none of that... (Female no. 11)

Decade 2: 10–19 years ($n = 11$)

Vision: Those who had previously been able to see said that their sight deteriorated rapidly in their mid-teens. They could no longer see colors or clear images. Objects were perceived as blurry shadows. Nine of the 11 described themselves as completely blind by the end of this decade, and the other two had severely impaired vision.

Hearing: From the age of 10, hearing impairment was a problem, and the eight participants who used hearing aids felt they were very helpful during this period.

Learning in everyday life

A combination of the auditory, tactile, and olfactory senses was the primary means of learning during this period. Most said that they needed to be shown with their hands and to receive verbal instructions in order to understand.

... Well, she usually tries to explain how she is doing it, as well as letting me get the feel of it, because my big thing is touch. I like to do a lot of hands-on stuff, that's how I learn the best, it's by touching things... (Female no. 11)

Mothers or grandmothers taught practical household chores such as washing dishes, cooking, cleaning, and laundry during this decade as well. Participants said that learning such skills instilled in them a sense of capability and inclusion as someone who could contribute to the household by helping with the chores.

Persistence and a desire to influence their situation were important personal characteristics for learning and achieving independence. Participants said that at this time they understood what the best approach to learning was for them. They all said that they had learnt to adapt to their own skill level when learning something new, and to recognize what they could and could not manage. They underscored the importance of not being afraid to ask when they could not see; through this, they had learned that some people were better at explaining than others.

The physical reactions caused by the syndrome, such as headache, dizziness, and fatigue, were described as significant to their own learning about AS. The experience had taught the participants to recognize how they felt when they needed medicine, that strong light hurt and damaged their eyes, and how they could stay healthier by exercising.

The younger participants, presently being in their teens ($n = 3$), expressed a desire to manage on their own and achieve independence. They also described understanding that more everyday knowledge could lead to increased independence, but also that their visual impairment could pose an obstacle to learning. Another obstacle was that because participants learned slowly, people often stopped trying to teach them how to perform various household chores, especially cooking, and just took over and performed the chore themselves. Many said that they could not, or had not learned how to, independently browse and use the internet to search for information or to have their own email account.

Participants said that they had needed more help in this decade than previously, and pointed out that people around them had to be determined and persistent if the participants were to learn something new. For example, taking personal responsibility for the care of their assistive devices and walking to the bus stop to ride on their own were skills that required both repetition and time to master.

Interaction with others became more difficult with increasingly impaired hearing. Participants noted the importance of comparing their life experiences with others' to develop their social skills. Asking questions was one strategy for learning, and the older the participants became, the more questions they asked of others, including their parents' and other adults' advice about their education and future.

Activities such as computer use, ceramics, sports, dance, handicrafts, and games were learned both at home and through courses with friends. The knowledge from those interests was integrated and applied in daily life.

Participants attributed their development on several planes to having dared to experiment, learn, and try to find solutions using trial and error in order to pursue their interests, such as using a computer.

I didn't have my own computer at that point, so I never got to mess with that as much as other people. Then I got mine and I was like, okay! And after that it probably took me maybe like... probably two or three hours to finally figure it out. Oh! This was what I had to do, it was easy! Ok. (Male no. 6)

Learning in the educational environment

All participants stated that at some point during their school years their curriculum was adapted and they were taught in classes with other students with disabilities, mainly in special schools for children who had impaired vision or were blind. One participant attended private school and another had attended class with students who had impaired hearing. Most said that they learned best in the school environment individually or in small groups. The majority ($n = 9$) of participants attended boarding school at this time. In most cases, such schools were far from home and some participants said this made them sad or homesick. Most of these boarding schools were also designed for students with impaired vision or blindness.

Participants stated that during this period, hearing was also important for learning in school.

But when we have PowerPoint presentations... I sit at the front, we sit like this in a circle... Well, they're just

showing the slides and speaking at the same time, so I don't have to strain myself, I can rest and just listen. (Female no. 3)

Losing my sight was probably my greatest barrier, not so much the hearing, not so much anything else, but because with sight I was always used to doing what I wanted... I would say probably in high school that was my greatest barrier. (Male no. 8)

... for the most part I mean, the hearing didn't really affect me till, you know, later in life. (Female no. 10)

Compensatory aids and adaptations used to facilitate learning included computers and auditory technology, microphones and loops, computers with screen readers, and players for reading textbooks. Magnification devices and closed-circuit television (CCTV) were important for learning at the beginning of the second decade, but were described as irrelevant later when eyesight further deteriorated.

Teachers described as facilitating learning were those who challenged the students. They were persistent, spoke loudly enough, used existing hearing technology, and used audio description to explain what was shown on the blackboard. Those teachers who supported the experience of belonging to the class facilitated participation in activities and social interaction. They encouraged friends to help with school assignments and assigned tasks that could be solved by working in groups. In such collaborative efforts, the strengths of all group participants were maximized, and participants felt that their strengths helped to advance the joint efforts.

... If we have a group project, [I say] "Let me write, because you aren't as good at writing, and you can look up the facts so I can write them, and then you can paste pictures," so it could be adapted. Then I could write, because I'm good at writing, and they could look up the facts because they weren't as good at writing. (Female no. 3)

Individual strategies described as important for learning included requesting repetition or explanations, memorizing, listening to instructions, looking things up on their own in a textbook, listening and taking notes, choosing an appropriate seat in the classroom, and trying to read the blackboard.

Special education teachers taught Braille and mobility individually or in small groups. Mobility training included using a cane and learning orientation. Braille was perceived as complicated to learn because there is so much to remember, and the same applied to sign language. One participant did not learn Braille until early adolescence and another participant who still had good vision was not motivated to learn Braille. Some of the interviewees had not learned sign language, even though they wanted to do so.

Those who attended schools with the primary goal of promoting the independence of the students described the benefits of learning with others. In this way, they learned how to perform various tasks in school and in student housing.

Being the only student who needed both visual and auditory adaptations was described as a difficulty, even when attending a special school. Schools designed to adapt to one disability often lacked adaptation for or knowledge about the other. The schools were either adapted for children and young people who had multiple disabilities, or for visual impairment/blindness. Lack of knowledge about combined visual and hearing impairments among friends and professionals posed an obstacle to learning. As a result participants felt discriminated

against, for example when others spoke with their assistant rather than with them.

I was the only one that had a hearing problem in that group. There was nobody else. All the others were just blind... when I asked: I want to do this... want to do that... they said sorry you can't because you have your hearing problem and there's nothing for the hearing... so there were some courses I couldn't do at all. (Female no. 9)

Further obstacles for learning in the school environment were unavailable or inoperative assistive devices and technology, rooms that were not adapted to visual and hearing impairment, inappropriate class sizes, and periods of illness with lengthy absences. Participants also said it took a long time to adjust and feel comfortable in new environments such as boarding school.

Decade 3: 20–29 years ($n = 8$)

Vision: All participants said they were completely blind by this time, but could still perceive light, so they could make out whether it was day or night, and if a lamp was turned on in a room they could see if there were objects, but they could not identify them.

Hearing: They all used hearing aids in this decade, but some felt that the hearing aids no longer provided full compensation and discussed Cochlea Implant (CI; an aid to hearing through electrical stimulation of the auditory nerve) with their doctors.

Learning in everyday life

Tactile, olfactory, and residual hearing continued to be important for everyday learning in this decade. Smell and touch, in that order, were described as being most important for their own hygiene, cleaning, laundry, and cooking. Strategies such as having someone explain and describe things orally, were perceived as helpful. Participants said that meeting others with deafblindness enabled them to relate to the experiences of others and to learn that they could handle more difficulties than they expected. They were aware that their attitude towards life affected their everyday learning, and having a positive outlook on life and being able to talk about things that are difficult made it easier to feel good, which in turn made it easier to learn.

The family was described as a resource that provided support when the participants felt they needed it. A few had moved back in with their parents after the conclusion of their studies. Others lived alone or with a partner in their own apartment.

Personal characteristics described as important for continued learning were persistence, never giving up without a fight, even when times are difficult, and trusting one's own intuition.

One thing I have is that I find it very, very, extremely, incredibly hard to back down when you know I'm still going and I haven't quite completed my goal... I find it very difficult to back down – very difficult to back down – I have to keep going until I have succeeded. (Male no. 7)

Several participants spoke about taking responsibility for breaking their own isolation and participating in various activities and social groups that are part of everyday learning.

...but actually you're opening your heart up to them and actually, well, you know they've been in that situation as well. And you can relate – the cool thing about [discussing these] issues is that I, too, talk them out if they've told me about stuff... related to what I've been through, and that's great. (Male no. 7)

During this decade, participants actively sought knowledge about AS from specialists or the internet and researched various types of assistive technology and support available from social services, charities, and interest groups. Effective hearing aids were very important. Participants described themselves as taking the initiative necessary to achieve their goals, and said that learning is easy when the subject is truly interesting.

A failing memory was seen as a limitation to daily learning during this decade. Participants said that difficulty in remembering things meant that they were no longer always able to manage what they could previously, or that they need to actively practice such things to retain skills such as remembering Braille abbreviations. They also said that it felt like it took longer to learn new things and that a single explanation was no longer sufficient – they needed to hear the same thing several times to retain it.

Learning in the educational environment

Some participants ($n=4$) had studied at the university/college level. Others took short courses in various fields such as sign language, mobility, and using a guide dog.

Individual strategies for succeeding at higher education included taking notes and rereading them several times, using both speech synthesis and Braille. Participants noted that when they were the only ones who needed material and technological support to compensate for their visual and hearing impairment at the university/college level, it was not available. They also felt that teachers lacked knowledge about the impact of deafblindness and about how to compensate for it in the classroom setting.

... they said “we will show you how to use it and once you got used to it we’ll come to school and show your teachers how to use it” ... For a good long while it worked, but when I left school and went to college and it ... just didn’t. (Male no. 7)

Participants felt that the lack of auditory technology (e.g. loop, microphone, classrooms adapted for auditory impairments) hampered studies during this time period.

Decades 4 and 5: 30–49 years ($n = 5$)

Vision: All participants were blind in these decades.

Hearing: Hearing function was dramatically decreased. Everyone used hearing aids, but remained severely impaired even with them. One had had a CI.

Learning in everyday life

Further deterioration of their hearing hampered their daily lives. Even greater demands were placed on their remaining senses of touch, smell, and taste. The need for help with hearing during social gatherings had grown. Technology (hearing aids, microphones, computer-based aids such as software and speech synthesis, etc.) that malfunctioned or constantly changed was a further obstacle to continued learning, as was not having learned how to take care of the home or to cook.

... You know I don’t feel that in school they spend enough time on those social skills. I mean they do a good job with reading, writing, and arithmetic, but you really have to prepare people with disabilities to go out into mainstream society. At some point, I mean you get some training, but at some point we learn to survive or we don’t. (Male no. 5)

Participants said that hearing was most important sense for performing various activities. Hearing aids helped and made it easier to stay focused. Individual strategies for coping with the increasing deafblindness used in interaction with others included talking to one person at a time, steering the conversation when

there were several speakers, or asking participants in the conversation to say their name before they spoke. This made it easier to tell the various speakers apart.

Sometimes I say “What did you say? Speak louder” or ... ‘I’m hard of hearing, I can’t hear. Many people speak very softly and it can be quite hard to hear even on the phone. (Female no 1)

Auditory description made it possible for them to participate in different activities and was helpful for learning about their surroundings. Everyone said that they wanted to manage on their own as far as possible and they did so by using their experience of doing things regularly in a certain order. They only asked for help, usually from their families and mainly from their mothers, when it was clearly necessary. Assistants could also be asked to find or provide information online or by telephone.

The combination of the computer, Braille, and speech synthesis to support reading text made it possible for some of the participants to stay in touch with others, find out more about their illness, and pursue their interests. Others, however, described inadequate computer skills and resource, such as not being able to receive or send email on their own as a barrier to further learning.

Technology that participants felt made their lives easier included the incorporation of FM radio waves in a system to enhance hearing aids, laptop computers, Braille display, and iPods with touchscreen. Vibration alarms and “doorbells” allowed them to experience important sounds in their immediate surroundings.

Learning in the educational environment

Touch and hearing with hearing aids provided the main channels of continued learning at this time. Several of the participants expressed a wish to engage in further studies, partly because of inadequate previous schooling and partly to be able to achieve their dreams. Others wanted to complement previously learned skills. Topics that interested them or that they wanted to learn more about included sign language, Braille, computer skills, and arts and crafts.

I would like to finish you know my schooling, maybe get a degree. And, you know, and go from there, and you know I always – I love art, pottery, and I’d like to go to – I was thinking about going to art school. I’d like to go to art school and learn more, more art, and the different concepts, and study art. (Male no. 4)

Discussion

The results of analyzing the participants’ self-reported lifelong experiences of learning with AS revealed two divergent tendencies that we call *disability* and *functioning*. Under the umbrella term of disability, are both the worsening impairment of both vision and hearing and the negative attitudes sometimes perceived in the educational environment and/or in the family. Functioning was primarily due to the participants’ own energy, commitment, and personal strategies for managing different situations while living with AS. Their reflections revealed a metacognitive competence that enhanced the necessary physical and social facilitating factors in the environment such as hearing aids and supportive parents, teachers, and peers.

On a more detailed level, the results show that sight, hearing, and touch are all important senses for learning among the participants. Visual impairment is consistently described as a greater obstacle to learning than impaired hearing. It affected

early learning even among participants who described their vision as relatively good during the first decade [14]. Tactile support was consistently helpful in learning, both at school and in everyday life. Even in their first decade, when most described their vision as relatively good, participants said that they needed to be shown with their hands or be permitted to feel tactile materials. Another example of early tactile support is the introduction of Braille lessons in preschool, as offered to half of the participants. The ability to read and write Braille was described as the skill taught in school that was most useful in both informal and formal situations later in life. Proficiency in Braille was described by researchers Erin and Wright [22] as a way to ensure learning over time. In the present study several ($n=9$) of the participants first learned to read printed text, and then when their vision deteriorated they moved to Braille. Those two who described themselves as blind before the age of three and began with Braille earliest said that they now mastered it fluently. During the third and fourth decades, however, Braille was perceived by some as complicated and difficult to remember, but also as so important that constant practice was necessary to retain it. Although modern technology can facilitate computer-aided reading and information retrieval for blind people, Braille continues to seem to be an important skill, best learned early, for blind people who will also lose their hearing. Early interventions to teach Braille in kindergarten and school can facilitate lifelong learning and promote feelings of self-esteem, competence, independence, and equality [23,24]. Text-to-speech screen technology may be helpful and complementary, but it cannot replace Braille in allowing everyday literacy for people with deafblindness [25].

The significance of hearing for learning during the preschool age was shown in the participants' descriptions of asking for repetition and asking their own questions repeatedly. Participants said that their problems with impaired hearing became more pronounced at school in the beginning of their second decade, which may be attributable to age-related increased demands on understanding [26], as well as the hearing deterioration itself. Yet another explanation may be that deafblindness becomes more pronounced during this period when eyesight dramatically deteriorates and 90% become blind [14].

In the third and fourth decades, participants report having had difficulties remembering, this necessitated continued training in previously acquired skills and knowledge. Memory function in deafblindness has received relatively little attention in perceived research. Whether their perceived difficulty with remembering is specifically related to AS, or if it is generally more common among people with deafblindness than those with normal vision and hearing is a topic that requires further research.

Only a few participants highlighted the medical complexity of AS as a factor hampering their learning at school through associated periods of absence. This is somewhat surprising as people with AS are very often physically ill [13–15].

From adolescence, and onwards, the lack of auditory support in various learning environments poses a major obstacle. Participants were heavily dependent in the school environment on properly functioning auditory aids such as hearing aids, assistive technology, and adapted classrooms. Perceived stigmatization was cited as a reason for not using or asking for aids or devices. The lack of motivation among those responsible for providing support was also suggested as an explanation for why necessary assistive devices were not always available or used. Similar findings have been reported in studies of sighted students with hearing impairment who were mainstreamed in school [27]. Rekkedal also found a correlation between teachers' attitudes, how often they used auditory technology, and how students with impaired hearing experienced and used their hearing aids [28].

Without doubt, it is consistently difficult to be the only person with combined vision and hearing impairment in the school environment. In addition to a shortage of technological aids, participants described others' ignorance about both disabilities, their lack of friends, and being bullied. Previous research on students with deafblindness describes similar experiences, suggesting that teachers may have education and knowledge about one of the functional impairments, but not about the second [12,29], and it may be likely that they do not have any knowledge at all about deafblindness as an individual and well-defined phenomenon. Bruce, DiNatale and Ford (2008) [30] report that in experiments with educational programs specifically focused on visual and hearing impairment for teachers who teach these students, such programs were positively received. Earlier research has also identified that information to classmates of children who have deafblindness is often inadequate, which may be a contributing factor to bullying [29,30]. Environmental factors such as lighting and color schemes may also be obstacles for students with deafblindness [12,29], but the present study did not focus on such factors.

Despite the negative experiences described above, participants also reported that they had people in their family or community support networks who had knowledge of their disability and a compensatory approach. These individuals made a difference and played an important role in the participants' own belief in their capacity to learn. They benefited from being able to participate in and carry out practical chores at home early in life. Favorable factors for learning practical things involved a combination of abundant time, clear oral directions, tactile support, and the opportunity for supervised practice. Their mothers were often nearby and actively taught the participants, both in childhood and in adulthood. This social and practical learning brings the idea to what Vygotsky called *scaffolding* [31], which implicates that, together with parents, relatives and/or peers the participants became able to carry out a task that the person could not manage independently. After a while the support is gradually reduced, resulting in an increased independence [31]. The results in the study showed that when the surroundings used tactile instructions, combined with verbal, it facilitated the learning process. The interactive form of learning could possibly contribute to a positive development of theory of mind among persons with AS, as cooperation requires an ability to read the other's intentions.

Learning processes such as the above-mentioned help individuals learn new skills and find their own strategies to carry out tasks independently [31], but they require a partner with insight and understanding to provide non-obstructive support. Some participants said that they did not have the opportunity to learn practical tasks well enough to live an independent life. Independent living proved impossible for four participants, because they had too little knowledge of how to run a home. Previous research has shown that parents' expectations and attitudes towards their visually impaired children and their participation in practical work at home correlates with how independent those children later perceive themselves to be [32]. From a theoretical standpoint, challenges adapted to the level and previous knowledge of the individual are necessary for the individual to develop a sense of competence [1].

Participants were found to need to ask others for advice much more often and until a later age than is usual for those entering adulthood. This may be because in situations when other young people venture into the world to develop their own interests and friends, the participants were faced with major limitations. They did not have the same opportunities as their sighted and hearing peers had to learn and to discover through dialogue with each other physically or online. Their knowledge about computers and the adaptations made for them did not always enable them to

participate independently in social media such as Facebook or in online surfing. Their need for support in learning and using a computer independently, including accessing e-mail, networks, and the Internet, remains high into adulthood. Such skills can be seen as necessary for maintaining contacts and reducing the risk of isolation [33], and were also reported as an important motivator for some participants to continue their quest to learn new information technology.

Curiosity, the courage to experiment, and persistence have been described of all participants as driving forces in learning, especially beginning in the second decade and are reflected in activities conducted both with others and alone. All participants in this study were able to describe their personal learning styles, past and present for example, by listening, touching, or asking questions and then asking again. According to Kolb and Kolb [34], the ability to view oneself as a learner is key to the metacognitive thought processes necessary for learning to take place. In line with this, participants described their plans for continued learning in the future, regardless of age. Thus, learning occurred at all ages, though the content of the learning varied.

Experience-based learning was mentioned in relation to learning in adult life. Participants stated that they learned about themselves by comparing their situation with that of others, and by learning about the experiences of others. This finding is interesting because Frölander et al. [19] noted that people with AS may have some difficulties with developing a theory of mind and that such difficulties are greater the earlier blindness occurs. According to Kolb and Kolb [34], understanding and knowledge about personal identity are necessary ingredients for an individual to develop an understanding of others and learn to cooperate. In the results, there were no evidence of learning from peers' experiences in the younger ages, which might be due to the participant usually being the only child with visual and hearing impairment in their childhood and school environments.

To our best knowledge no research had previously been conducted on individual experiences of learning in individuals with deafblindness, nor specifically in individuals with AS. Deafblindness is rare, yet it is associated with about 50 hereditary syndromes, with AS one of the more unusual causes. The most common is Usher's syndrome, which represents almost half of the population with deafblindness [35]. Even within the deafblind community, people with AS are sometimes regarded as unique, in that spoken language is their first way of communication. Visual or tactile sign language is more often the common language among other groups with deafblindness. Because people with AS differ from other people with deafblindness, further research is needed before applying any conclusions from this study to other populations with deafblindness.

Methodological issues

The qualitative explorative design was regarded as appropriate because there was no previous research performed in this area. The small number of people diagnosed with AS worldwide led to the recruitment of participants both in Sweden and at an international conference. Although factors, such as heterogeneity in onset of vision loss and hearing loss, cultural, socioeconomic status of families and different school system in each country, could have influenced the results, the results were surprisingly convergent. According to Pring [36], the qualitative interview is an appropriate method for investigating how people with communications disorders live their lives. The semi-structured interview was chosen to allow the participants to express their experiences in greater detail [20].

Although individuals with AS have early deafblindness, their first learned language is spoken, and thus, verbal interviews were possible to conduct. On one hand, the English-speaking interviewer knew most of the participants from previous conferences, and this may have created a more secure atmosphere for the interviewees. On the other hand, the interviewer may have neglected to ask questions because of previous knowledge, so the use of an interview guide was seen as valuable. Other factors that most certainly had a positive impact on the trustworthiness of the results are the cultural differences in the home countries, communities, and school systems of the participants.

However, it can be criticized that data were collected from memories, which possible not fully correspond with reality [20]. The main function of the memory is to store experiences for later use. The literature describes how written history, based on memories, can influence and change the present perception, in both positive and negative ways. The order of events and other data, like where, with whom and such, may become incorrect or reversed [37]. This can give a contradictory impression, which also was the case in this study. However, the aim with the present study was not to gain objective accounts of learning, but to listen to the participants' experiences and memories of learning, from different periods of life.

Conclusion

Based on the initiative, energy, perception, and persistence shown in the functioning category, i.e. the personal resources the participants revealed in the interviews, supervisors, caregivers, and teachers are encouraged to let people with AS be their own advocates. These personal factors to learn are resources that can actively be utilized for example by inviting the person with AS to participate in planning their course of study and set their individual goals. They know best how, what, and with whom they learn, and what mode (tactile, auditory, visual, or a combination) works best for them.

The present study showed that approaches and attitudes are crucial to facilitate or impede participation in the learning process. Parents and professionals may, therefore, require continuous information about the syndrome and its implications in order to create better opportunities for the learning of the person with AS.

This study also found that touch was important to learning in all four decades reviewed, in both formal and informal contexts. The tactile modality, for reading Braille, for example, seemed to be important throughout the lives of people with AS. Support for tactile learning should be systematically encouraged throughout childhood. Pedagogically, it can be done through tactile games and activities combined with auditory (verbal) support. Adults with AS should always be offered to tactually examine whenever the situation permits it.

In the second decade, emerging problems to remember both in the daily life and in the school environment were described, which reinforces the importance for tactile learning. This can facilitate and relieve the memory. For example, this can be implemented in routines in everyday life, by the use of technical aids, as well as by methods for memory training.

The absence of well-functioning visual and hearing aids hampered learning. The computer is the one tool that makes both educational materials and community information available; therefore, it seems to be crucial having a working computer environment. Aids should function and be continuously maintained to be of use, which requires ambient knowledge of the aids and of the specific needs that exist for a person with AS. People with deafblindness and AS should receive the most technologically advanced tools.

Acknowledgements

We would like to express our sincere gratitude to the participants who generously shared their experiences of learning with us. We would also like to thank Lil Falkensson, The National Resource Centre for Deafblindness, Lund, Sweden and Jan Marshall, Jackson Laboratories, Bar Harbor, Maine and Alstrom International for making the data collection in the US possible.

Declaration of interest

This research was funded by the Swedish Research Council and the Swedish Institute for Disability Research, Linnaeus Centre HEAD. The authors report no conflicts of interest. The authors alone are responsible for the content and writing of this article.

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