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**AUGMENTATIVE AND ALTERNATIVE
COMMUNICATION IN INDIVIDUALS WITH
RETT SYNDROME**

A survey for family members

Faculty of Social Sciences
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ABSTRACT

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Rett syndrome is a rare genetic mutation that is diagnosed to 50–100 people in Finland. Rett syndrome causes varying degrees of intellectual disability and usually severe motor disability that both complicate communication. After a phase of typical development for approximately 6–18 months the learned motor skills, speech, and communication abilities begin to regress, and purposeful hand use is replaced with stereotypies, repetitive movements. Individuals diagnosed with Rett syndrome can benefit from augmentative and alternative communication (AAC). AAC can improve a person's communicative competence that includes linguistic, operational, social, and strategic competences.

The aim of this Master's thesis is to examine how AAC is used in families including an individual with Rett syndrome and what factors influence the success of the communication situations according to the family members' perspectives. This study was a survey study. The respondents were volunteers that had a family member diagnosed with Rett syndrome. The online survey was sent through Rett ry to the members, and 10 people responded anonymously. The questionnaire included both multiple-choice questions and open questions. The quantitative information from the multiple-choice questions was used as it was to describe the study participants, and the qualitative information produced by the open questions was analyzed to answer the research questions. The analysis method for the qualitative material was reflexive thematic analysis. The analysis was inductive, and the material was examined using an experiential and essentialist approach, so the emphasis was on depicting the respondents' individual experiences and their reality.

The qualitative analysis of the material produces four themes: 1. Situations, 2. Eye gaze, 3., Communication partner, and 4. Environmental support. Themes 1 and 2 described the AAC methods used by the families, and themes 3 and 4 the factors influencing the success of the communication situations. AAC enabled communication between the respondents and their family members in various contexts and different functions. The most common AAC methods the respondents used with their family member were eye gaze, body movement and different aids. The most common communicative functions AAC enabled were making choices, answering yes and no questions, requesting, and expressing pain. The respondents' family members' communicative competence was extremely variable. A distinctly frequently mentioned single communication method was eye gaze both as unaided and aided AAC forms. The success of AAC was influenced by the motivation and familiarity of the communication partner and environmental support. Environmental support meant the places and people outside home, such as kindergarten, school, and the professionals working the respondents' family members. Environmental support was seen as a promoting factor for AAC. However, the respondents additionally recognized that only an engaged environment supports the success of AAC. The respondents were mostly content with the support they had received from professionals, but they wished for more support with the technological operation of aids and their personalization. Peer support from other families that include an individual with Rett syndrome was mentioned as a factor that motivated the respondents.

This study offers perspectives to how individuals with Rett syndrome can be offered more efficient communication methods. The study adds knowledge about the syndrome and AAC, and the results can be used as guidelines for professionals, family members, and other people to help communication when the conversation partner has Rett syndrome.

Keywords: augmentative and alternative communication, AAC, Rett syndrome, survey study

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TIIVISTELMÄ

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Rettin oireyhtymä on harvinainen geenimutaatio, joka on diagnosoitu Suomessa 50–100 henkilölle. Rettin oireyhtymä aiheuttaa eriasteista älyllistä kehitysvammaa sekä yleensä vakavan liikuntavamman, jotka vaikeuttavat kommunikointia. Lapsen tyypillisen kehityksen vaiheen jälkeen 6–18 ensimmäisen kuukauden aikana opitut motoriset taidot, puhe ja kommunikointikyky alkavat taantua, ja käsien tarkoituksenmukainen käyttö korvautuu stereotyyppisillä eli toistavilla liikkeillä. Henkilöt, joilla on diagnosoitu Rettin oireyhtymä, voivat hyötyä puhetta tukevista ja korvaavista kommunikointikeinoista (*augmentative and alternative communication*, AAC). AAC-keinoilla voidaan parantaa henkilön kommunikaatiokompetenssia, johon kuuluvat kielellinen, operationaalinen, sosiaalinen ja strateginen kompetenssi.

Tämän pro gradu -tutkielman tavoitteena on selvittää, millaisia AAC-keinoja käytetään perheissä, joissa omaisella on Rettin oireyhtymä, ja mitkä tekijät vaikuttavat kommunikointitilanteiden onnistumiseen perheenjäsenten näkökulmasta. Tutkimus toteutettiin kyselytutkimuksena, ja vastaajiksi haettiin vapaaehtoisia henkilöitä, joiden perheenjäsenellä on diagnosoitu Rettin oireyhtymä. Sähköinen kyselylomake lähetettiin Rett ry:n kautta jäsenille, ja siihen vastasi 10 henkilöä anonymisti. Kyselylomakkeeseen kuului sekä monivalintakysymyksiä että avoimia kysymyksiä. Monivalintakysymysten määrällistä tietoa käytettiin sellaisenaan tutkimushenkilöiden kuvailemiseksi, ja avointen kysymysten tuottamaa laadullista tietoa analysoitiin tutkimuskysymyksiin vastaamiseksi. Laadullisen aineiston analyysimenetelmänä käytettiin refleksiivistä teema-analyysia. Analyysi tehtiin aineistolähtöisesti, ja materiaalia tarkasteltiin kokemuksellisesta ja essentialistisesta näkökulmasta, eli painopisteenä olivat vastaajien yksilöllisten kokemusten ja heidän todellisuutensa kuvaaminen.

Aineiston laadullinen analyysi tuotti neljä teemaa: 1. Tilanteet, 2. Katse, 3. Kommunikointikumppani ja 4. Ympäristön tuki. Perheiden käyttämiä AAC-keinoja kuvailivat teemat 1 ja 2, ja kommunikointitilanteiden onnistumiseen vaikuttavia tekijöitä teemat 3 ja 4. AAC-keinot mahdollistivat kommunikointia vastaajien ja heidän perheenjäsentensä välillä monissa erilaisissa konteksteissa ja erilaisissa tarkoituksissa. Yleisimpiä vastaajien perheenjäsenensä kanssa käyttämiä AAC-keinoja olivat katse, kehon liikkeet ja erilaiset apuvälineet. Yleisimpiä AAC:n mahdollistamia kommunikoinnin toimintoja olivat valintojen tekeminen, kyllä-ei-kysymyksiin vastaaminen, pyytäminen ja kivun ilmaiseminen. Vastaajien perheenjäsenten kommunikaatiokompetenssi oli hyvin vaihteleva. Aineistossa selkeästi toistuva yksittäinen kommunikointikeino oli katse niin avusteettomana kuin apuvälineavusteisena AAC-keinona. AAC:n onnistumiseen vaikuttivat kommunikointikumppanin motivaatio ja tuttuus sekä ympäristön tuki. Ympäristön tuella tarkoitettiin paikkoja ja ihmisiä kodin ulkopuolella, kuten päiväkotia, koulu ja vastaajien perheenjäsenten kanssa työskentelevät ammattilaiset. Tuki ympäristöstä nähtiin AAC:ta edistävänä tekijänä. Vastaajat tunnistivat kuitenkin myös, että vain sitoutunut ympäristö tukee AAC:n onnistumista. Ammattilaisten tukeen oltiin pääosin tyytyväisiä, mutta apua toivottiin lisää apuvälineiden tekniseen käyttöön ja niiden personointiin. Vertaistuki toisilta perheiltä, joissa omaisella on Rettin oireyhtymä, mainittiin vastaajia motivoivana tekijänä.

Tämä tutkimus tarjoaa näkökulmia siihen, miten henkilöille, joilla on Rettin oireyhtymä, voidaan tarjota tehokkaampia keinoja kommunikointiin. Tutkimus lisää tietoisuutta oireyhtymästä ja AAC-keinoista, ja tuloksia voidaan käyttää ohjeina ammattilaisille, omaisille ja muille henkilöille helpottamaan kommunikointia, kun keskustelukumppanilla on Rettin oireyhtymä.

Avainsanat: puhetta tukevat ja korvaavat kommunikointikeinot, AAC, Rettin oireyhtymä, kyselytutkimus

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1 INTRODUCTION

Rett syndrome (RTT) is a severe neurodevelopmental disorder that is typically caused by a genetic mutation in the X-linked *MECP2* gene (Amir et al., 1999; Chahrour & Zoghbi, 2007). The disorder progresses in stages, and core symptoms include loss of acquired speech and purposeful hand use (Chahrour & Zoghbi, 2007; Neul et al., 2010). RTT causes both intellectual and motor disability (Berger-Sweeney, 2011), and apraxia is a prominent symptom (Demeter, 2000; Lotan & Ben-Zeev, 2006). Additionally, RTT is characterized by gait and breathing disturbances, seizures, scoliosis, and features of autism spectrum disorder (Berger-Sweeney, 2011; Chahrour & Zoghbi, 2007; Lotan & Ben-Zeev, 2006). The symptom combination causes difficulties in communication that affect the lives of the individual with RTT and their family members deeply (Townend et al., 2020).

People with complex communication needs, including individuals with RTT, can benefit from using augmentative and alternative communication (AAC; Creer et al., 2016). AAC can supplement existing speech or replace it (Lund et al., 2017). Finding suitable AAC methods for people with RTT can be challenging, but as access to meaningful communication is a basic right of every human (United Nations, 2006), communication and necessary accommodations must be guaranteed. Previous studies have shown that the most used AAC method in people with RTT are eye gaze, pictures, and body movement (Bartolotta et al., 2011; Urbanowicz et al., 2016b).

The aim of this Master's thesis is to discover how AAC is used in families including an individual with RTT. An additional aim is to explore what factors influence the success of the communication situations according to the family members' perspectives. The study participants are family members of people with RTT, and their experiences are examined with the help of an online survey. This study adds to the body of literature about RTT and AAC. The results can be used to help with the development of individualized AAC interventions for people with RTT, so that everyone has access to effective strategies for self-expression.

2 REVIEW OF THE LITERATURE

2.1 Rett syndrome

RTT is a neurodevelopmental disorder with an incidence of approximately 1 in 10 000–15 000 people globally (Amir et al., 1999; Chahrour & Zoghbi, 2007; Laurvick et al, 2006; Panayotis et al., 2023; Wong & Li, 2007), impacting 50–100 people in Finland (Rintahaka, 2023). RTT mainly affects females and is associated with a genetic mutation in the *MECP2* gene on the X chromosome (Amir et al., 1999; Chahrour & Zoghbi, 2007) that mostly occurs sporadically (Neul et al., 2010). A mutation in *MECP2* is present in 86 % of RTT cases (Percy et al., 2007) and the mutation type affects the severity of symptoms (Neul et al., 2008). The gene produces MeCP2 protein that has many functions affecting the development and regulation of the nervous system (Cohen et al., 2011). An *MECP2* mutation in males has previously been thought to result in death (Chahrour & Zoghbi, 2007) or symptoms considerably different from RTT (Schüle et al., 2008). However, male cases of a mutation in *MECP2* co-occurring with classical RTT symptoms have been reported, although rarely (Reichow et al., 2015).

Classical RTT (85.5 % of cases; Percy et al., 2007) is usually diagnosed at the age of 30–36 months (Sandweiss et al., 2020). It can be described to progress in stages (Bartolotta et al., 2011; Berger-Sweeney, 2011; Chahrour & Zoghbi, 2007; Gold et al., 2018; Lotan & Ben-Zeev, 2006). The first 6–18 months the child develops seemingly typically and begins to acquire speech, language, and motor functions age-appropriately. After the stage of typical development begins a period of rapid regression, during which speech and motor skills acquired are partially or completely lost and purposeful hand use is replaced by distinctive stereotypical movements such as hand washing or wringing. Gait disturbances become apparent (Berger-Sweeney, 2011; Chahrour & Zoghbi, 2007; Neul et al., 2010; Lotan & Ben-Zeev, 2006) and other comorbidities might occur. Comorbidities include breathing abnormalities (hypoventilation alternating with irregular breathing and hyperventilation; Ramirez et al., 2013) seizures, scoliosis, and features of autism spectrum disorder (Berger-Sweeney, 2011; Chahrour & Zoghbi, 2007; Lotan & Ben-Zeev, 2006). This stage usually lasts for weeks or months (Lotan & Ben-Zeev, 2006). The regression period is followed by a stage of stabilization, during which regaining lost skills is possible (Neul et al., 2010). The individual may remain in the stationary stage until puberty or adulthood (Bartolotta et al., 2011) before the last stage of late motor deterioration. The final stage includes a decrease or loss of mobility and arise of muscle

rigidity (Hagberg, 2005) and other Parkinson-like features (Berger-Sweeney, 2011; Chahrour & Zoghbi, 2007). Cognition and communicative skills do not seem affected in this phase despite the increased motor dysfunction (Gold et al., 2018; Lotan & Ben-Zeev, 2006).

Atypical RTT (13.5 % of cases; Percy et al., 2007) refers to individuals that fulfil some diagnostic criteria, such as a period of regression, but not all (Neul et al., 2010). Atypical RTT is usually diagnosed at the age of 3–6 years (Sandweiss et al., 2020). The genetic origin in atypical cases might be a mutation in *MECP2* or other locations (e.g., *CDKL5*; Artuso et al., 2010; *FOXP1*; Ariani et al., 2008). The range of observable characteristics classified under the diagnosis of atypical RTT is broad, but however, several distinct forms have been recognized. The preserved speech variant is characterized by similar but milder symptoms compared to classical RTT (Zappella et al., 1998). Many of the people affected can walk relatively steadily. After a regression period, these individuals develop expressive speech, although it might be repetitive or prosodically unusual. The early-onset seizure variant's symptoms include epileptic seizures beginning between the 1st and the 10th weeks of life (Artuso et al., 2010). The stereotypical hand movements are prominent. In the congenital variant the classical RTT symptoms, such as loss of hand use and communication skills, occur noticeably early, usually during the first 6 months of life (Rajaei et al., 2011).

RTT has been described to cause severe cognitive impairment (Berger-Sweeney, 2011). The extent in which RTT affects cognitive abilities is, however, extremely difficult to assess (Ahonniska-Assa et al., 2016; Berger-Sweeney, 2011; Demeter, 2000; Djukic & Valicenti-McDermott, 2012; Ward et al., 2021) as most of the formal tests rely on speech or motor responses when completing the assessment tasks (Berger-Sweeney, 2011; Lotan & Ben-Zeev, 2006; Rose et al., 2013; Ward et al., 2021). Studies have explored the use of eye gaze tracking computers in assessment of cognitive potential when traditional means of testing are not adequate (Wilkinson & Mitchell, 2014) and the research extends to specifically people with RTT (Ahonniska-Assa et al., 2016; Djukic & Valicenti-McDermott, 2012; Rose et al., 2013). There seem to be remarkable individual differences in the cognitive abilities of people with RTT ranging from severe impairment to average skills (Ahonniska-Assa et al., 2016; Ward et al., 2021). In addition to differences between individuals, cognitive performance fluctuates within one person between days or moments (Demeter, 2000; Ward et al., 2021). Regardless of the assessed intelligence level, people with RTT are able to learn, enhance, and maintain new skills (Lotan & Ben-Zeev, 2006). Thus, the cognitive potential of people with RTT has likely been underestimated (Ward et al., 2021).

One of the complicating factors in the assessment of RTT is apraxia (Demeter, 2000) that occurs in the rapid regression stage, staying prominent also through the stationary stage (Lotan & Ben-Zeev, 2006). Apraxia is a disorder in the cognitive side of motor control (Goldenberg, 2013, p. 228) that manifests a difficulty to perform specific actions and intentional, learned movements on demand (Lotan & Ben-Zeev, 2006; Zadikoff & Lang, 2005). It causes a delay in movement initiation, problems with imitating movements, handling objects purposefully, and timing and sequencing of actions (Zadikoff & Lang, 2005). In RTT, all functional areas, such as feeding, speech, and ambulating, are affected by apraxia (Lotan & Ben-Zeev, 2006).

The stages and the clinical presentations of RTT are highly individual, and comorbidities might develop at different times for different people (Gold et al., 2018). Individuals with RTT are likely to survive well into adulthood (Laurvick et al., 2006; Tarquinio et al., 2015; Wong & Li, 2007), the leading cause of death being cardiorespiratory issues (Tarquinio et al., 2015). RTT is so far incurable, but gene therapies have appeared potential in preclinical studies (Panayotis et al., 2023; Qian et al., 2023). Currently, RTT treatment is highly focused on managing symptoms and comorbidities as no therapies specifically targeting the syndrome are available (Gold et al., 2018).

2.2 Communication and AAC

Communication is both an essential life skill (Chung & Stoner, 2016; Creer et al., 2016; McEwin & Santow, 2018) and a human right, regardless of the form and necessary accommodations (United Nations, 2006). Communicative rights, such as having the opportunity to build relationships through social interaction, request and refuse, and express personal preferences and opinions, extend to people with different disabilities (Brady et al., 2016). People with complex communication needs can use diverse means of augmentative and alternative communication (AAC; Battye, 2022, p. 1; Beukelman & Mirenda, 2013, p. 4; Higginbotham et al., 2007; Ogletree, 2021, p. 5). Augmentative forms of communication support and supplement existing speech and/or speech comprehension, whereas alternative communication replaces spoken language entirely (Lund et al., 2017). Groups of people benefitting from AAC are extremely heterogeneous (Creer et al., 2016; Fjeldvang et al., 2023) as the need for AAC can be temporary or permanent, developmental or acquired, and it can affect people coming from different age groups and backgrounds (Beukelman & Mirenda, 2013, pp. 4–5).

AAC can be divided to unaided and aided forms, the latter including low- or light-tech and high-tech means (Bailey et al., 2006; Baxter et al., 2012; Light & McNaughton, 2014; Marshall & Goldbart, 2008; Moorcroft et al., 2019). Unaided AAC forms mean different nonverbal natural communication (Moorcroft et al., 2019; Ronski & Sevcik, 2005) utilizing the user's own body to convey messages (e.g., gestures and facial expressions; Baxter et al., 2012). Aided AAC refers to the use of a separate tool as support for communication (Moorcroft et al., 2019; Ronski & Sevcik, 2005). Low-tech systems include non-electronic aids (e.g., communication boards), and high-tech systems, by contrast, encompass technological communication devices (e.g., speech generating devices). Several different strategies and aids can be used depending on various factors such as the context or the communication partner, and the most suitable AAC methods might change over time (Marshall & Goldbart, 2008).

AAC interventions aim to improve communicative competence. Communicative competence of individuals using AAC consists of four domains: linguistic, operational, social, and strategic competences (Battye, 2022, pp. 211–215; Light & McNaughton, 2014). The linguistic domain focuses on the individual's receptive and expressive skills in both the language of the society they are a part of and their own AAC system. Operational skills involve the ability to use different AAC strategies, and social competence refers to the skills being used in appropriate contexts. The strategic domain describes the different compensatory strategies the individual uses to navigate the speaking world. In addition to the four domains described the efficiency of AAC is also determined by psychosocial and environmental factors such as motivation and support received from family.

Communicative competence is not a single constant trait that an individual possesses, but a dynamic production of social interaction (Teachman & Gibson, 2014; Tsai, 2016). Therefore, communicative competence is co-constructed (Tsai, 2016). Co-construction is a process during which meanings are created together by the participants in the communication situation (Solomon-Rice & Soto, 2011). It involves the communication partners working to elicit, prompt, and question the other one. Co-construction requires a developed theory of mind, the ability to take the perspective of the communication partner (Stadskleiv et al., 2022). All conversations are co-constructed, but AAC requires more effort from the more competent communicator (Ferm et al., 2005), which increases the role of co-construction.

To ensure the discovery of the most fitting AAC strategy a multidisciplinary assessment is required (Battye, 2022, pp. 15–16; Fjeldvang et al., 2023; Moorcroft et al., 2019; Ogletree, 2021, pp. 4–5). The team working with a person when introducing and supporting an AAC strategy can include

among others speech-language pathologists, occupational therapists, and physical therapists (Battye, 2022, pp. 15–16; Ogletree, 2021, pp. 4–5). Collaboration between the people present in the AAC user’s life is crucial (Uthoff et al., 2021), including professionals, family, friends, and carers (Battye, 2022, p. 13; p. 61; McNaughton et al., 2019; Ogletree, 2021, p. 4). Each AAC intervention must be individually tailored to support the person’s communicative needs and to fit the available family resources (Bailey et al., 2016; Marshall & Goldbart, 2008; McNaughton et al., 2019).

2.3 Communication and AAC in individuals with RTT

Individuals with RTT can use a wide range of communicative behaviors (Didden et al., 2010; Hetzroni & Rubin, 2006; Sigafos et al., 2011). These include, for example, alternating eye gaze and facial expressions, such as smiling as a response to another person. People with RTT also have preferred looking at people’s faces rather than objects in studies (Djukic & Valicenti-McDermott, 2012; Fabio et al., 2006; Rose et al., 2013), which indicates an orientation for communication.

The foundations for social competence and successful use of AAC are joint attention and communicative intent (Battye, 2022, pp. 28–32; p. 212). People with RTT have shown joint attention behaviors in studies (Djukic & Valicenti-McDermott, 2012; Fabio et al., 2006). Intentionality in communication is difficult to assess among people with severe disabilities (Iacono et al., 1998), but according to caregivers, their family members with RTT express desire to communicate meaningfully in everyday life (Bartolotta et al., 2011; Urbanowicz et al., 2016b). There is a disconnect between the quantitative and qualitative results of the developmental level of individuals with RTT as caregivers describe their family member’s communicative abilities as higher than assessed in clinical tests (Demeter, 2000). In addition to joint attention and intentionality, AAC requires sufficient motor and cognitive skills (Higginbotham et al., 2007), which can make the discovery of a suitable AAC method challenging due to the symptom combination of RTT. A trait typical for conversations with people with RTT is that they need a considerably long time to generate an answer (Townend et al., 2020) – most need a response time of 11 seconds or more (Bartolotta et al., 2011). Thus, it seems like individuals with RTT are willing to communicate, but their options for self-expression are limited due to their motor disability and apraxia (Bartolotta et al., 2011; Djukic & Valicenti-McDermott, 2012; Vessoyan et al., 2018).

People with RTT can, therefore, benefit from AAC (Creer et al., 2016). There is strong evidence for the effectiveness of individually tailored AAC interventions for people with RTT (Amoako & Hare, 2020) and it is recommended to be included as early as possible (Townend et al., 2020). People with RTT can use both unaided and aided, low-tech and high-tech, AAC strategies. The most used AAC methods are eye gaze, pictures, and body movement (Bartolotta et al., 2011; Urbanowicz et al., 2016b). The use of multiple AAC methods and devices can also benefit this population.

Of the variety of AAC methods people with RTT can utilize, eye gaze seems to be the most reliable (Bartolotta et al., 2011; Didden et al., 2010; Hetzroni & Rubin, 2006; Townend et al., 2018; Urbanowicz et al., 2016a; Vessoyan et al., 2018). This is in line with the diagnostic criteria of RTT, where intense eye gaze is mentioned as a supportive criterion (Neul et al., 2010). Eye function is not affected by the motor challenges associated with RTT as people with RTT performed as well as the control group in a study testing the accuracy of eye movements (Townend et al., 2018). Eye gaze is used diversely as an AAC method. People with RTT can use ‘eye pointing’ and looking intensely at something they are interested in (Bartolotta et al., 2011; Urbanowicz et al., 2016a). Eye gaze can also be utilized with eye gaze tracking (EGT) technologies which automatically pick up where a person’s eyes fixate while they are viewing a visual image (Wilkinson & Mitchell, 2014). Caregivers have reported that they are satisfied with EGT as AAC for their children with RTT (Townend et al., 2016; Vessoyan et al., 2018) and AAC interventions using EGT cause perceived improvement in communication skills (Vessoyan et al., 2018).

The perspectives of family members have been an important source of information in previous research regarding communication and AAC in individuals with RTT (e.g., Bartolotta et al., 2011; Townend et al., 2016; Urbanowicz et al., 2016b). Family members’ reports offer data on authentic situations in familiar contexts from people that regularly communicate with the person with RTT, thus allowing indirect observation of communicative behaviors present.

3 AIMS OF THE STUDY

The aim of this study is to describe how AAC is used in families including an individual with RTT and explore what factors influence the success of communication according to the family members' perspectives. Developing new speech, occupational, and physical therapies is vital in the care of individuals with RTT (Sandweiss et al., 2020), and the results of this study can help in the search of suitable AAC interventions conducted by these professionals. This study aims to improve access to AAC and thus ensure the actualization of communicative rights. Family members of individuals with RTT were recruited as participants because they are the most likely communication partners and experienced AAC users in everyday life. This study is the first to examine the views of family members of people with RTT on AAC in Finland.

The research questions were formed as follows:

1. How is AAC used in families including an individual with RTT?
2. According to the family members' perspectives, what factors influence the success of AAC?

4 METHODS

4.1 Study design

The role of qualitative research in the understanding of human communication is crucial (Simmons-Mackie & Damico, 2003) as describing the complexity of communication situations with only the help of statistics is not adequate (Tetnowski & Franklin, 2003). This has led to a shift in interest to a more holistic approach to the research methodology in the field of speech-language pathology experts and researchers. In qualitative research formal hypotheses are avoided (Eskola & Suoranta, 1998) or the initial hypothesis is revised and refined multiple times during the research process (Tetnowski & Franklin, 2003) to serve the purpose of the methodology. According to Tetnowski and Franklin (2003) the ideal situation for a qualitative approach is when the objective is to familiarize experiences of individual people – Simmons-Mackie and Damico (2003), in turn, define the goal of qualitative methodologies to be not only describing but explaining the results detected.

This study was conducted as a cross-sectional online survey. Survey research is flexible and allows the use of mixed methods (Andres, 2012, p. 18), so both quantitative and qualitative data were collected with a questionnaire including multiple-choice and open-ended questions. The data from the multiple-choice questions were used to gather background information of the participants and to set the context for the qualitative data, which was analyzed further. Completely open-ended questions are advantageous as they give the respondents the opportunity to bring up new issues and opinions not foreseen by the researchers (Andres, 2012, p. 70; Sue & Ritter, 2016, p. 56). In addition to its compliance to flexible use of mixed methods, survey research has other benefits especially in an online form. Online surveys are fast and efficient (Andres, 2012, p. 50; Sue & Ritter, 2016, p. 5). They are also low-cost and remove geographical limitations for participation (Sue & Ritter, 2016, p. 5). Both Andres (2012, pp. 43–44) and Sue and Ritter (2016, pp. 3–4) have defined the phases of doing survey research: initially, survey objectives are to be defined and a literature review and preliminary research are to be conducted. Next, a model for the study is designed – this phase consists of choosing the survey method and drafting the questionnaire and invitation. Before the invitation is sent, possibilities for misunderstandings must be minimized by pretesting the questionnaire (Andres, 2012, p. 86; Sue & Ritter, 2016; p. 73). After launching the survey and receiving the data an analysis is conducted and the results are presented in a report (Sue & Ritter, 2016, pp. 3–4).

4.2 Data collection

The online questionnaire was created using Tampere University's Microsoft Forms application. The questionnaire was a mix of closed- and open-ended questions related to the AAC strategies that are used in families including a person with RTT. To make the questionnaire clearer for the respondents, it was divided into sections (see Andres, 2012, p. 86). The four sections were under the headlines *Background Information* (5 questions), *Unaided Communication Strategies* (3 questions), *Communication Aids* (5 questions) and *Challenges and Support in the Use of AAC* (4 questions). Before sending of the study invitation the questionnaire was piloted and refined with the help of a group of Tampere University logopedics students and teachers.

A Finnish association, Rett ry, was contacted to reach family members of individuals with RTT. The criteria for the respondents were age above 18 and to have a family member with RTT. An invitation to the study was sent via an association member email list and shared in the Rett ry Facebook group. The email including the invitation, a link to the online survey (see Appendix 1), the study handout (see Appendix 2), and the confidentiality report was sent on December 16th, 2022. The survey was open until January 15th, 2023.

An exhibition of the open-ended questions providing the qualitative data relevant to this study's research questions and their English translations is shown in Table 1. The original questionnaire in Finnish is shown in Appendix 1.

Table 1. The original open-ended survey questions and their English translations

Question number	The original question	The English translation
8	Kuvaile arjen tilanteita, joissa kommunikoit omaisesi kanssa käyttäen kysymyksissä 6 ja 7 valitsemiasi menetelmiä [ei-avusteiset AAC-keinot]. Kerro keskustelu ympäristöistä ja keskusteluiden aiheista. Missä ja mistä keskustellaan? Kuvaile 1–4 tilannetta.	Describe everyday situations where you communicate with your family member using the AAC strategies you chose in Questions 6 and 7 [unaided AAC strategies]. Narrate conversation environments and conversation topics. Where do you discuss and about what? Describe 1–4 situations.
13	Kuvaile arjen tilanteita, joissa kommunikoit omaisesi kanssa käyttäen kysymyksissä 9, 10 ja 11 valitsemiasi apuvälineitä [avusteiset AAC-keinot]. Kerro keskustelu ympäristöistä ja keskusteluiden aiheista. Missä ja mistä keskustellaan? Kuvaile 1–4 tilannetta.	Describe everyday situations where you communicate with your family member using the AAC strategies you chose in Questions 9, 10 and 11 [aided AAC strategies]. Narrate conversation environments and conversation topics. Where do you discuss and about what? Describe 1–4 situations.
14	Onko mainitsemiesi ei-avusteisten AAC-keinojen ja apuvälineiden käytössä ollut haasteita? Jos kyllä, millaisia?	Have you faced challenges with the use of aforementioned unaided and aided AAC strategies? If yes, what kind?
17	Mitkä tekijät ovat edistäneet AAC-keinojen käyttöä? Miksi kyselyssä kuvailemasi menetelmät ja apuvälineet ovat olleet toimivia?	What factors have promoted the use of AAC strategies? Why have the particular strategies and aids you have mentioned in the survey been effective?

4.3 Participants

A total of 10 people completed the survey. The response time average was 25 min 55 s, and response times varied between 10 min 45 s and 61 min 20 s. All respondents informed that they were the parent or caregiver of an individual with RTT. The ages of respondents' family members with RTT ranged from 5 to 28 at the time of the survey.

The types of unaided and aided AAC strategies the respondents used with their family members were evaluated with multiple-choice questions. The most used unaided strategies were facial expressions (9/10), and body movement (9/10), and the least used were signs (0/10). The most used communication aids were pictures (9/10), and the least used were other communication devices (1/10). None of the respondents chose the “I do not use any communication aids with my family member” option. The number and variety of unaided strategies were greater compared to the communication aids in use according to the responses to the questions mapping the preferred AAC strategies. Other AAC strategies in use in the respondents' families were the communication apps

GoTalkNow, MOI and Sanoma 48, and a yes-no-something else -fan. Figures 1 and 2 visually depict the unaided and aided AAC strategies the respondents used with their family members.

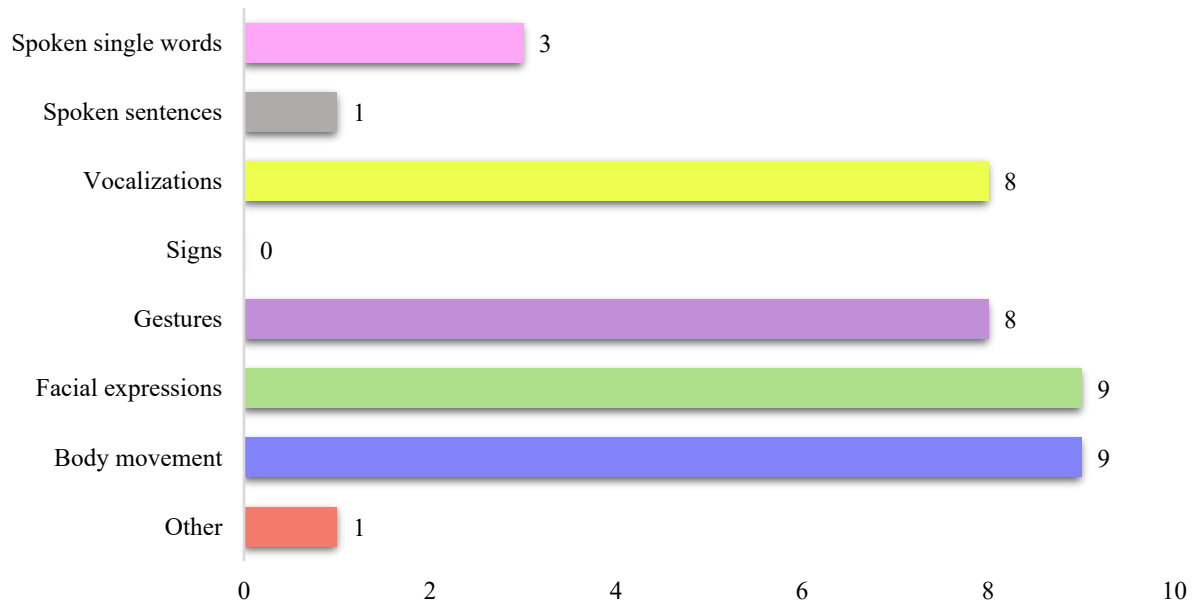


Figure 1. Descriptive statistics of unaided AAC strategies respondents used with their family member with RTT

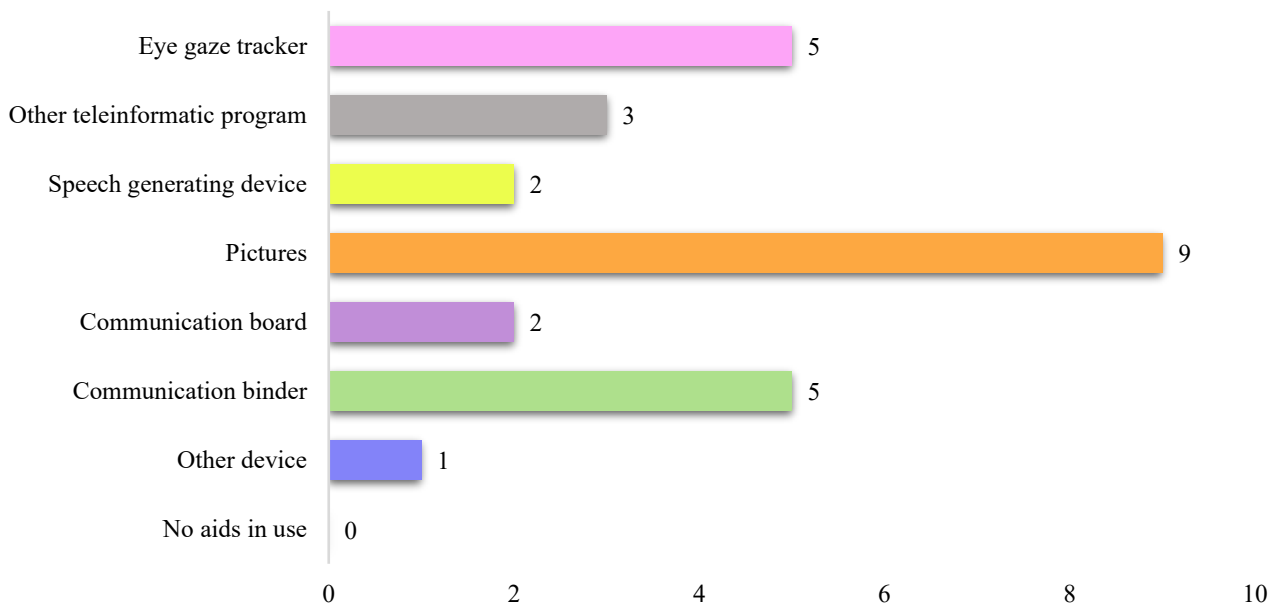


Figure 2. Descriptive statistics of aided AAC strategies respondents used with their family member with RTT

4.4 Data analysis

The data from the open questions of the online survey (see Table 1) were analyzed qualitatively using instructions for reflexive thematic analysis defined by Braun and Clarke (2006; 2022, pp. 34–35). The first phase of the analysis is immersing oneself to the material. After a meticulous familiarization of the data, the researcher creates preliminary codes and searches for themes, meaning the recurring patterns found in the data (Braun & Clarke, 2022, pp. 69–70). The identified themes are then developed and reviewed, and finally defined and named. These phases might happen simultaneously instead of following strictly one after another (Terry & Hayfield, 2021, p. 30).

An inductive approach was used, so making pre-determined hypotheses was avoided and the material was viewed without a chosen theoretical framework guiding the analysis (Braun & Clarke, 2006; 2022, pp. 50–51; Eskola & Suoranta, 1998; Tetnowski & Franklin, 2003), and the final codes of the themes were not created until the end of the analysis process (Terry & Hayfield, 2021, p. 12). Themes can be semantic, directly observable in the material, or latent, a result of an analysis searching for more underlying meanings (Braun & Clarke, 2022, p. 36). This study took note of both semantic and latent themes. An experiential essentialist approach was chosen, meaning the objective was to explore participants' own perspectives and capture their reality (Braun & Clarke, 2022, p. 36). To account for all the information the respondents provided, the entire data corpus was coded instead of making individual thematic maps for all the questions. Some of the survey questions generated overlapping answers, so proceeding this way made the analysis more comprehensive and truly data-driven. A data extract could be used as an example in multiple different themes that it fit into; the same extract could contribute to the generation of more themes than one. In the reviewing phase of the analysis (see Braun & Clarke, 2006; 2022, pp. 34–35) the responses were reviewed individually to ensure the accuracy of the generated themes. Equal attention was given to all the parts of the material, as suggested by Braun and Clarke (2006).

The purpose of the thematic analysis was to produce connections that would not have been obviously recognizable from the original data (Terry & Hayfield, 2021, p. 50), which is generally a common point of view for qualitative research (Simmons-Mackie & Damico, 2003). Reflexive thematic analysis was the most suitable method for this study because of its flexible nature (Braun & Clarke, 2006; Terry & Hayfield, p. 5) and its ability to highlight both similarities and differences in the data

(Braun & Clarke, 2006). Reflexive thematic analysis is an appropriate method when the areas of interest are the unique experiences and feelings of the participants (Terry & Hayfield, 2021, p. 9).

See Figure 3 for a visual depiction of the analysis process. See Table 2 for an example of the coding process.

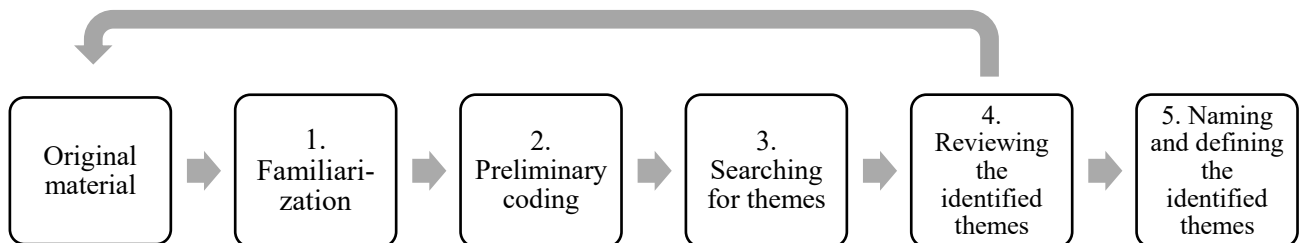


Figure 3. The thematic analysis process

Table 2. An example of a data extract and the preliminary codes assigned to it

The original data extract	The English translation	Codes assigned
Tyttäreni voi sanoa vain vähän sanoja, mutta sanoessaan jotain, pyrimme selvittämään mistä on kyse. Sana on alku asian selvittämiseksi. Tyttärelläni on tapana aloittaa tiivis katsekontakti toiseen silloin, kun hänellä on asiaa, jonka hän haluaa ilmaista.	My daughter has few words, but when she says something, we try to figure out what the matter is. The word is a beginning for the clarification. My daughter will start an intense eye contact to a person when she has something she wants to express.	1. Family members are motivated to clarify what their daughter is communicating 2. The daughter initiates communication using eye contact

4.5 Ethical aspects

When conducting a study, the researcher must be aware of the needed ethical approvals (Andres, 2012, p. 22). The design of this study did not require a preliminary ethical assessment. The permission to recruit Rett ry members as study participants was asked via email from the association chairperson. This study follows the research integrity guidelines by Tutkimuseettinen neuvottelukunta (*Hyvä tieteellinen käytäntö ja sen loukkausepäilyjen käsitteleminen Suomessa*, Tutkimuseettinen neuvottelukunta, 2012).

The privacy of the participants was guaranteed by making the online survey anonymous and not gathering any identity information. The participants were requested not to reveal information from which they or their family member could be recognized. Participant identities must be protected in a way that does not allow even the people closest to the participants to recognize them (Eskola & Suoranta, 1998). This was ensured by not including any more detailed information of the participants in the report than necessary. The dignity of the participants was respected throughout the entire study.

5.1 Theme 1: Situations – “The answering delay typical for Rett makes almost everyone give up too early”

This theme was constructed based on the experiences participants provided about everyday communication situations generally, the more specific examples described, and the possible challenges faced. The questionnaire included a specific question about both everyday communication situations and AAC challenges. However, despite characteristics and challenges were a distinct part of the questionnaire itself, Theme 1 was constructed through a synthesis of all the open-ended survey questions that included descriptions of everyday communication situations and challenges regarding them.

The material indicated that people with RTT were mostly seen as motivated and able to participate in social interaction. Two respondents had contrasting experiences and they described their experiences of their family member’s disinterest in talking by saying ‘She is not particularly interested in talking’ and ‘The girl does not independently initiate telling things’. However, the respondents generally recognized their family member’s behaviors as communicative.

Lapsi on motivoitunut käyttämään kuvia että hänet edes niillä ymmärrettäisiin. / The child is motivated to use pictures to be understood through them at least.

Tyttärelläni on asiaa, mielipiteitä, oma tahto. / My daughter has things to say, opinions, her own will.

Tyttäremme on iloinen saadessaan näyttää mitä on tehnyt koulussa ja samoin toisin päin. Kuvat ja videot ovat todella tärkeitä hänelle. / Our daughter is happy when she gets to show what she has done at school and the other way around. Pictures and videos are very important to her.

The respondents described multiple different means their family members with RTT use to initiate communication and respond to it. The most mentioned were eye gaze (n=7), body movements (n=6), and different aids (e.g., pictures, n=8; EGT, n=5).

Tyttärelläni on tapana aloittaa tiivis katsekontakti toiseen silloin, kun hänellä on asiaa, jonka hän haluaa ilmaista. / My daughter will start an intense eye contact to someone when she has something she wants to express.

Hän saattaa myös kävellä asian luokse tai pyytää huomioimaan tarvettaan tulella luokse. / She might also walk to an object or ask for attention for her need by coming by.

Tyttäreni käyttää painiketta, jossa lause "minulla on asiaa", kun haluaa jotakin selvitettävän/tehtävän. / My daughter uses a button with the sentence "I have something to say" when she wants something to be clarified/done.

It was clear that needs for communicative support varied greatly. Some individuals with RTT had no spoken language, while others could communicate using complete sentences. One respondent highlighted that there were differences between the same person's communicative performance depending on how they were feeling physically and mentally. Thus, in addition to high variability between individuals, there were differences in the same person's communicative performance depending on the time.

Lapsellani on epätyypillinen Rett. Hän puhuu lausein, kävelee, käyttää käsiään jonkin verran toistaiseksi. Puheen tuottaminen on vaikeaa vaikka olo olisi hyvä, välttämättä sana tai oikea sana ei löydy. – – Jos olo on huono, hän puhuu vähemmän eikä löydä haluamaansa sanottavaa. / My child has atypical Rett. She speaks with sentences, walks, uses her hands to some extent for now. Speech production is difficult even if she feels good, she might not find a word or the correct word. – – If she feels bad, she talks less and cannot find what she wants to say.

Two respondents mentioned a delay in answering to be common for communication situations.

Rettissä tyypillinen viive vastaamisessa saa likipitäen kaikki luovuttamaan liian aikaisin. / The answering delay typical for Rett makes almost everyone give up too early.

Kun hän ei katso silmiin kysyttäessä kysymystä niin se on hänen ei vastaus tai en tiedä. Mutta tähän pitää antaa aikaa reilusti ja odottaa vastaus. / When she does not make eye contact when asked a question, that is her answering no or I don't know. But you must give a fair amount of time for this and wait for the answer.

The participants reported that AAC is used in various contexts and functions in their families in everyday life. Making choices between options was a common communicative function the individuals with RTT could access using AAC (n=7). They could also express agreement and disagreement, yes and no (n=6). Requesting (n=6) and communicating pain and discomfort (n=6) were also made possible by AAC. AAC allowed the individuals with RTT to communicate in more complex ways, for example talking about their day (n=4), playing (n=1), and commenting during mealtime (n=1). Additionally, AAC was used for social formalities of which greeting people was mentioned as an example (n=1).

Ruokailussa vaihtoehtoisista ruoista valinta yleensä kysytään näyttämällä esim. kahta leipäpakettia, jolloin tyttö osoittaa sormella kumpaa haluaa. / At mealtime, we usually offer food options by showing e.g., two bread packages, and she points the one she wants with her finger.

Tyttäreni ilmaisee kipua ilmeillä ja kehon liikkeillä. Hän ei juuri itke, joten jos häntä sattuu, hänen ilmeestään näkee sen ja hän usein silloin myös vapisee. / My daughter expresses pain with facial expressions and body movement. She does not cry much, so if she is hurting, you can see it on her face, and often she will shake.

There were challenges mentioned regarding communication situations. One of them was the cognitive load (n=4). Respondents mentioned concentration, connecting things, and the general demandingness of using an aid when communicating using AAC as challenging. Misunderstandings or occasional inability to understand the person with RTT was mentioned by three respondents.

Ei pysty keskittymään tehdessä valintoja. / [She] cannot concentrate while making choices.

Another challenging aspect was the clumsiness of the aids in use (n=4). Picture communication was described as slow, finding the right pictures in a folder could be difficult, and the folder itself was experienced to be big and difficult to carry. One respondent characterized teleinformatic communication programs as ‘a little confusing’.

Aluksi käytimme paljon irtokuvia ja katsekehikkoa, tämä toimi hyvin kotona mutta muualla on todella hankalaa ja hidasta etsiä irtokuvia kansioista. / At first, we used loose pictures and an eye gaze frame a lot, this worked well at home but elsewhere it is extremely difficult and slow to look for pictures in a folder.

5.2 Theme 2: Eye gaze – “My daughter will start an intense eye contact to someone when she has something she wants to express”

This theme was constructed with the responses referencing communicative eye gaze. Eye gaze was distinctly the most frequently mentioned single communication method in the material. It was used diversely, both as an unaided AAC strategy (n=5) and as a part of aided communication methods (n=6).

Individuals with RTT could use eye gaze without aids effectively in different contexts. Unaided communicative eye gaze could be used for example to draw attention or make choices.

Tyttärelläni on tapana aloittaa tiivis katsekontakti toiseen silloin, kun hänellä on asiaa, jonka hän haluaa ilmaista. / My daughter will start an intense eye contact to someone when she has something she wants to express.

Syödessä näytän lapselle ruokaa ja maitolasia. Hän valitsee katseella kumpaa haluaa seuraavaksi. / At mealtime I show the child food and a glass of milk. She chooses with eye gaze which one she wants next.

Aided methods utilizing eye gaze referenced in the material were pictures and EGTs. Pictures could be used as either a low-tech AAC form without the assistance of a computer or with help of an EGT. EGTs could broaden the range of communicative functions and conversation topics the individual could access.

Tyttäreni käyttää katseohjattavaa tietokonetta kertoakseen kuulumisiaan tai mitä haluaa tehdä. Hän kertoo katseohjattavan avulla mm. mitä musiikkia haluaa kuunnella. Hän käyttää katseohjattavaa apuna vastatessaan tulkin avulla sähköposteihin. / My daughter uses an EGT to catch up with people or to tell what she wants to do. She uses the EGT to tell what music she wants to listen to, among other things. She uses the EGT as an aid when she answers emails with her interpreter.

5.3 Theme 3: Communication partner – “You must be able to ask the right things”

This theme was constructed based on the significance of the communication partner in dyadic communication highlighted by the respondents either specifically or indirectly. Theme 3 is the most latent of the four, as responses were interpreted to represent this category whenever the respondent described their own actions in enabling communication for their family member with RTT. Therefore, data extracts used while constructing this theme were more indirectly related to one another as their connection could be an underlying message of the importance of individual communication partners in the success of AAC.

It seems like the respondents themselves are very active in keeping the conversation flowing during the dyadic interactions with their family member with RTT. Asking questions (n=9) and generally having a more initiative role in the conversation was apparent. Several responses mentioned the respondent deducing (n=5) or guessing (n=1) things in everyday situations. Respondents reported that they will deduce things based on their family member's appearance and vocalizations. The communicative burden seemed to largely be the responsibility of the communication partner. Respondents would also describe modeling the use of AAC (n=3). Additionally, one respondent mentioned feelings of sadness when they are not able to comprehend what their family member means

or why they are sad, which indicates that they are highly motivated to succeed in communication. One respondent expressed that they hope to get new aids to assist speech.

Kyllä/Ei korteilla pitää osata kysyä oikeita asioita, arvailla yms. Ei voi oikeasti tietää mitä omainen haluaa, ellei sitä osaa kysyä. / With Yes/No cards you must be able to ask the right things, guess, etc. You cannot actually know what the family member wants, if you are not able to ask.

Another factor for the communication situation to succeed was the familiarity of the communication partner. Gestures or facial expressions were not always easily interpretable, and some individuals with RTT had their own means to communicate certain things, such as grabbing the hand of the communication partner to answer yes. Respondents were familiar with their family member and could point out communicative actions they do regularly and consistently that a less familiar person would not understand (n=5).

Eleitä ja ilmeitä on myös muiden vaikea tulkita jos ei tunne ihmistä hyvin. / Gestures and facial expressions are also difficult to interpret if you do not know the person well.

Kysyttäessä iloinen ilme, hymy tarkoittaa kyllä. Myös silmiin katsominen nopeasti tarkoittaa kyllä. / When asked something a happy facial expression, a smile, means yes. Also, a quick look into the eyes means yes.

5.4 Theme 4: Environmental support – “The best motivation and support has been from peer families”

This theme was constructed based on the respondents’ opinions about environmental support present in their own and their family member’s life. Compared to Theme 3, Theme 4’s emphasis is more on the respondents’ experiences about different larger groups interacting with their family member with RTT. Environmental support consists of individual communication partners, hence the responses used to generate themes 3 and 4 overlapped the most. The different nuances in the responses caused the themes to be separated in the final version of the thematic map.

Four respondents mentioned the engagement of their family member’s general nearest environment to be important. People close to the individual with RTT could facilitate AAC by being interested and patient in the communication situation.

Useissa paikoissa ei myöskään ymmärretä, että puhumaton voi ymmärtää ja osata ja haluta viestiä. Asiat puhutaan vanhemmille eikä nuoren vastausta odoteta. / In many places it is not understood either that the nonverbal person can understand and be able and willing to communicate. Things are spoken to parents and the adolescent's response is not expected.

Places and groups that were referenced in the material were school (n=2), kindergarten (n=2), and temporary daycare (n=1). These groups outside home were seen as both promoting factors (n=2) and barriers (n=3) to successful AAC. Based on their answers, the respondents recognize the potential of these groups' role as facilitators of AAC, but they also indicate that it might not actualize until the staff is educated and engaged.

Varhaiskasvatuksen ja perusopetuksen työntekijät [ovat edistäneet AAC:tä], valitettavasti on heissä hyvin persoonakohtainen ominaisuus ketkä edistävät ketkä eivät. / The workers in kindergarten and school [have promoted AAC], unfortunately with them it depends on their personality who promote and who do not.

Professionals mentioned were speech therapists (n=4), occupational therapists (n=1) and other, unspecified professionals (n=2). Professionals were mostly seen as facilitators for AAC (n=3). However, the respondents were not completely content with the professional support they had received for operating the technological aids (n=3) and for personalizing the vocabularies (n=1).

Kuntouttava puheterapeutti on vuosia uskonut tytön taitoihin ja sillä on ollut iso merkitys. / The rehabilitating speech-language therapist has believed in the girl's abilities for years and that has been very meaningful.

Kyllä [on ollut haasteita], katseohjattavan kanssa erityisesti. Kesti kauan ennen kuin saimme ammattilaisen apua kommunikointiohjelman sisällön laatimisessa. / Yes [there have been challenges], especially with the eye gaze computer. It took a long time before we got professional help with designing the content of the communication program.

Two respondents brought up peer support from other families that include a person with RTT. Hearing about other people's success with communication was motivating.

Paras motivaatio ja tuki on tullut muilta vertaisperheiltä. / The best motivation and support has been from peer families.

Se kun on nähnyt että ensin joku muu on saanut oman lapsensa omaa "ääntä" kuuluviin käyttämällä niitä [AAC-keinoja] ja vuosien saatossa on harjoittelu tuottanut tulosta. / Seeing someone else get their child's own "voice" heard first using them [AAC means] and the practice during the years has gotten results.

6 DISCUSSION

Using reflexive thematic analysis, shared experiences and opinions of family members of individuals diagnosed with RTT (n=10) have been identified. According to family members, individuals with RTT communicate using various means of AAC. Theme 1 describes communication situations and is thus relevant to the first research question of this study. Theme 2 revealed eye gaze to be the most used and effective AAC method. The second interest of this study was to discover which factors facilitate the use of AAC. Important factors were individual communication partners' contributions examined in Theme 3 and the support received from the environment examined in Theme 4. Generally, the results of this study are in line with previous research. Noticeably, Urbanowicz and colleagues (2016b) used similar research questions in an interview study directed to parents. The researchers identified multiple receptive and expressive communicative behaviors, and additionally facilitators and barriers affecting communication. Despite differences in methods, the results of the current study resemble those acquired by the previous researchers.

6.1 Review of the results

Theme 1 provided descriptions of communication situations in everyday life. Individuals with RTT were seen as motivated to communicate. As expected considering previous study results (Bartolotta et al., 2011; Urbanowicz et al., 2016b), the most used AAC methods were eye gaze, body movement, and pictures. The individuals' communicative competence was highly variable depending on the situation. This variability has been identified by several authors in previous studies (Bartolotta et al., 2011; Urbanowicz et al., 2016b; Townend et al., 2020). The differences in communication skills between individuals could be explained by different types of genetic mutations (Neul et al., 2008; Percy et al., 2007), but this study could not assess this relation as the mutation types of the participants' family members with RTT were unknown.

Individuals with RTT could express a wide range of communicative functions such as making choices, expressing agreement and disagreement, and requesting, which indicates strong skills in the social domain of communicative competence (Light & McNaughton, 2014). A factor negatively impacting their social competence, however, was the delay in answering identified by two respondents. The individuals with RTT additionally seemed to have some skills in the linguistic domain of communicative competence. Some responses indicated clear speech comprehension skills

and a few produced speech. All the respondents could communicate with their family member using some AAC method, which means that their family member with RTT had linguistic judgment, knowledge, and skills in their AAC system's language code. The strategic competence of the individuals with RTT could not be assessed with the material acquired.

Operational competence, in turn, encompasses technical skills in both unaided and aided AAC methods (Light & McNaughton, 2014). Interestingly, only one of the respondents expressed that they had not had any challenges with the use of AAC. Apraxia likely restricted the respondents' family members' operational competence and complicated the use of any AAC form. This could be an answer to why none of the respondents chose the option 'signs' as an AAC strategy they use with their family member in the background information section of the questionnaire. Challenges caused by limited operational competence were highlighted especially with communication devices, as the operation of aids caused difficulties for both the individual with RTT and the respondents themselves. This result implies that AAC devices and apps must be developed further to be easier to use and transport.

Theme 2 indicated that eye gaze was the most clearly noticeable single AAC method used by the individuals with RTT. The respondents expressed that their family member with RTT had excellent operational skills with eye gaze as an unaided AAC method and they were extremely adept in 'eye pointing'. When either low- or high-tech aids were involved, this changed, and the communication was more prone to errors and misunderstandings. EGT was referred to as a demanding form of communication, which is compatible with previous literature – EGT is recognized to require controlled eye movements, which demand effort and high concentration from the user (Battye, 2022, p. 131). The respondents reported frustration with the technology, but overall were still satisfied with their EGT experiences, which is in line with parental opinions from other studies (Townend et al., 2016; Vessoyan et al., 2018). Despite an existing consensus about eye gaze being a superior form of communication for people with RTT in previous research (Bartolotta et al., 2011; Didden et al., 2010; Hetzroni & Rubin, 2006; Townend et al., 2018; Urbanowicz et al., 2016a; Vessoyan et al., 2018), eye gaze might have been overemphasized by this study's results – the respondents' family members with RTT were mostly young (< 19 years of age), and younger individuals are more likely to utilize eye gaze as a means to communicate (Urbanowicz et al., 2016a). However, considering the abundance of existing research, the results of this study can be interpreted to support that eye gaze is the most potential communication method for individuals with RTT.

The communication partner can affect communicative competence (Light & McNaughton, 2014), which was established by Theme 3. The respondents consistently assigned meaning to their family members' behaviors and interpreted them as communicative, which is recommended when communicating with a person with RTT (Townend et al., 2020). Thus, the communication situations were highly co-constructed, which facilitates communication with people with complex communication needs (Solomon-Rice & Soto, 2011). Frequent descriptions of actions that increase co-construction in the material were asking questions and deducing or guessing what the individual with RTT wants to express. The respondents were undoubtedly motivated to succeed in communication. It is recognized that the role of the communication partner is vital in the success of communication with people with RTT (Urbanowicz et al., 2016b), and the respondents' substantial knowledge about their family member's communicative behaviors assisted them in communication. Some respondents' family members had their own methods of communicating certain things consistently, such as subtle changes in eye contact to communicate agreement or disagreement. These behaviors may be difficult to interpret for a less familiar communication partner. The results indicate that a familiar and motivated communication partner is a facilitator to successful AAC.

Effective collaboration of professionals and close people can influence an AAC intervention positively (Bailey et al., 2006; Uthoff et al., 2021) and every individual with RTT should be supported by a multidisciplinary team (Townend et al., 2020). This is demonstrated by Theme 4 of the current study. The respondents mentioned different groups of people working and interacting with their family member to be both facilitators and challenging factors to the success of AAC. Professionals and the staff of school or kindergarten received praise from the respondents and were seen to promote AAC through their motivation and enthusiasm. On the other hand, the lack of knowledgeability and engagement of the workers in these groups could be a factor that complicated AAC and frustrated the respondents, which communicates a need for interventions directed to these people. Peer support from other families including a person with RTT was brought up by two respondents, which could be utilized more as a part of interventions. As implied by Theme 3, the role of the communication partner is crucial in the success of AAC, and thus the resources of the family members that likely are the most common communication partners should be supported and protected by every means available.

6.2 Discussion of the method

The material for this study was gathered using an online survey. The survey was created in in co-operation with a doctoral researcher, and it was piloted with the help of students before sending to ensure its intelligibility and usability. The respondents were informed that participating in the study by filling the survey takes 15–35 minutes, but ultimately the time that was used varied from approximately 11 minutes to over an hour. The respondents that used the least time might not have immersed themselves in the subject as intensely as intended, and on the other hand, the respondents that used the most time may have been engaging in other activities as they were responding. The length of the responses varied as well. A clearer questionnaire layout or more direct instructions before the beginning of the survey could have rendered the resources needed to participate in this study more transparent and resulted in more consistency in response time and response length. The survey reached the respondents via an email list and a Facebook group for family members of individuals with RTT. This may have excluded some families that do not belong in these groups.

The number of respondents ($n=10$) was small, and they did not all answer every question, which made the material scarce. However, as the subject of the study is not well studied, the results provide important information despite the small number of participants. The chosen analysis method was experiential essentialist thematic analysis. The experiential approach makes the results not as easily generalizable to a larger population as it aims to capture people's perspectives (Braun & Clarke, 2022, p. 15), but this study may have served as a platform for the respondents to share their unique experiences. The online survey as a method may have caused interpretation errors in the analysis phase of this study because the researcher could not clarify what the respondents had meant exactly.

The small number of respondents and the small number of people affected by RTT in Finland resulted in a choice to not present the responses as complete sets of answers in this study. The respondents could not be recognized from their individual answers, but the detailed information provided by different questions combined could have allowed that. This procedure protected the anonymity of the respondents but could have led to more ambiguous results as differences and connections between individuals may have been lost. As the survey questions gathered as little personal details as possible, the RTT stage the person was in during the data collection or the specific mutation type and their connections to communication skills could not be assessed. The questionnaire strictly addressed communication situations, and thus conclusions about the connection between motor skills or apraxia and communication could not be made either.

6.3 The clinical impact of the study and ideas for future research

The perspectives of family members of individuals with RTT are an important source of information when assessing communication (Urbanowicz et al., 2016b). This study agrees with previous literature (Bartolotta et al., 2011; Urbanowicz et al., 2016b; Townend et al., 2020) that the communication needs of people with RTT are extremely variable. As the first Finnish study to examine AAC in individuals with RTT, this study suggests that the needs of the families are the same as in other countries, regardless of the culture and language.

According to the results of this study the family members of individuals with RTT would benefit from more support and guidance from clinicians to both the family and their environment. This study encourages family members and professionals working with individuals with RTT to ensure that the environment is fully committed to the AAC intervention and that the communicative rights of the person with RTT are guaranteed in different contexts. Peer support from other families including a person with RTT was mentioned as a motivating factor, and that could be a relatively feasible and low-cost support system to include in AAC interventions.

AAC interventions require individual assessment and monitoring, and the co-operation of different professionals in multidisciplinary teams is beneficial and should be continued. In addition to healthcare professionals, a technology specialist is an increasingly important part of the team. The respondents of this study voiced a need for more technological support on the operation of communication devices. Especially EGTs are valuable aids in AAC for people with RTT, and issues related to that technology should be prepared for. EGTs in AAC for people with RTT have been studied (e.g., Townend et al., 2016; Vessoyan et al., 2018), but more research about the topic is needed.

In future studies, validity can be improved with a larger sample size compared to the current study. Additionally, a different study method, such as an interview or a focus group study directed to family members of people with RTT, might achieve more in-depth answers from the participants. In addition to methodological adjustments, future studies should consider the variability that is caused by the type of genetic mutation the individual has and explore the connection between motor and communication skills.

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Appendix 1. The online survey in Finnish.

Taustatiedot

1. Olen yli 18-vuotias. Jos vastauksesi on ei, voit lopettaa kyselyn täyttämisen. *

Kyllä

2. Omaisellani on Rettin oireyhtymä. Jos vastauksesi on ei, voit lopettaa kyselyn täyttämisen. *

Kyllä

3. Olen lukenut tietosuojailmoituksen ja ymmärtänyt, että vastauksiani käytetään Tampereen yliopistolla tutkimuskäytössä anonyymina aineistona. Jos vastauksesi on ei, voit lopettaa kyselyn täyttämisen. *

Kyllä

4. Mikä on suhteesi omaiseen, jolla on Rettin oireyhtymä? (esim. Olen huoltaja./Kyselyn täyttivät yhdessä huoltaja ja sisarus./...) *

Kirjoita vastaus

5. Milloin omaisesi on syntynyt? Vastaa **kuukauden tarkkuudella** muodossa **kk/vvvv**. *

Kirjoita vastaus

Seuraava

Sivu 1/4

Ei-avusteiset kommunikointimenetelmät

Ei-avusteisilla kommunikointimenetelmillä tarkoitetaan keinoja, joita käytät omaisesi kanssa kommunikoimiseen ilman apuvälinettä.

6. Millaisia ei-avusteisia kommunikointimenetelmiä omaisesi käyttää kanssasi tällä hetkellä? Valitse kaikki sopivat vaihtoehdot.

Lähde: Bartolotta, T. E., Zipp, G. P., Simpkins, S. D. & Glazewski, B. (2011). Communication skills in girls with Rett syndrome. *Focus on Autism and Other Developmental Disabilities*, 26(1), 15–24. *

- Sanat
- Lauseet
- Äännähdykset
- Viittomat
- Eleet
- Ilmeet
- Kehon liikkeit
- Muut menetelmät

7. Jos valitsit kysymyksessä 6 kohdan "Muut menetelmät", kuvaile omaisesi kanssasi käyttämiä muita ei-avusteisia kommunikointimenetelmiä.

Kirjoita vastaus

8. Kuvaile arjen tilanteita, joissa kommunikoit omaisesi kanssa käyttäen kysymyksissä 6 ja 7 valitsemasi menetelmiä. Kerro keskusteluympäristöistä ja keskusteluiden aiheista. Missä ja mistä keskustellaan? Kuvaile 1-4 tilannetta. (esim. Pesutilanteissa käytämme pääasiallisesti kehon liikettä ja eleitä. Tyttäreni kertoo hanaa kohti kumartumalla ja kädellä osoittamalla, kun vesi on liian kuumaa.) *

Kirjoita vastaus

Edellinen

Seuraava

Sivu 2/4

Kommunikoinnin apuvälineet

Kommunikoinnin apuvälineillä tarkoitetaan erilaisia esineitä tai laitteita, joita omaisesi käyttää kommunikoidessaan kanssasi.

9. Millaisia kommunikoinnin apuvälineitä omaisesi käyttää kanssasi tällä hetkellä? Valitse kaikki sopivat vaihtoehdot.

Lähde: <https://papunet.net/tietoa/apuvälineet>. *

- Katseohjaustietokone
- Muu tietotekninen kommunikointiohjelma
- Puhelaite
- Kuvat
- Kommunikointitaulu
- Kommunikointikansio
- Muut apuvälineet
- En käytä kommunikoinnin apuvälineitä omaiseni kanssa

10. Jos valitsit kysymyksessä 9 kohdan "Muu tietotekninen kommunikointiohjelma", kuvaile omaisesi kanssasi käyttämiä muita tietoteknisiä kommunikointiohjelmia.

Kirjoita vastaus

11. Jos valitsit kysymyksessä 9 kohdan "Muut menetelmät", kuvaile omaisesi kanssasi käyttämiä muita kommunikoinnin apuvälineitä.

Kirjoita vastaus

12. Milloin omaisesi sai apuvälineen käyttöönsä? Vastaa kunkin apuvälineen käyttöönoton ajankohta muodossa **kk/vvvv**.

Kirjoita vastaus

13. Kuvaile arjen tilanteita, joissa kommunikoit omaisesi kanssa käyttäen kysymyksissä 9, 10 ja 11 valitsemiasi apuvälineitä. Kerro keskusteluympäristöistä ja keskusteluiden aiheista. Missä ja mistä keskustellaan? Kuvaile 1-4 tilannetta. (esim. Ennen ruokailua käytämme kommunikointitaulua. Sisareni kertoo kuvaa osoittamalla, mitä haluaa juoda ruuan kanssa. Ruokaillessa apuna on myös äännähtely. Sisareni viestii "mmm"-äännähdyksin, kun ruoka on hyvää.)

Kirjoita vastaus

Edellinen

Seuraava

Sivu 3/4

Haasteet ja tuki AAC-keinojen käytössä

AAC-keinoilla (*augmentative and alternative communication*) tarkoitetaan puhetta tukevia ja korvaavia kommunikointikeinoja.

14. Onko mainitsemiesi ei-avusteisten AAC-keinojen ja apuvälineiden käytössä ollut haasteita? Jos kyllä, millaisia? *

Kirjoita vastaus

15. Miltä tahoilta olet saanut tukea AAC-keinojen käyttöön? Valitse kaikki sopivat vaihtoehdot. *

- Erikoissairaanhoido
- Terveyskeskus
- Kuntouttava puheterapeutti
- Vertaistuki
- Muu taho
- En ole saanut lainkaan tukea

16. Jos valitsit kysymyksessä 15 kohdan "Muu taho", kuvaile muualta saamaasi tukea AAC-keinojen käyttöön.

Kirjoita vastaus

17. Mitkä tekijät ovat edistäneet AAC-keinojen käyttöä? Miksi kyselyssä kuvailemasi menetelmät ja apuvälineet ovat olleet toimivia? *

Kirjoita vastaus

Edellinen

Lähetä

Sivu 4/4

Appendix 2. The study handout in Finnish.

TUTKIMUSTIEDOTE

Tutkimus – AAC-keinot perheissä, joissa omaisella on Rettin oireyhtymä

Pyydämme Teitä osallistumaan tähän tutkimukseen, jossa tutkitaan AAC-keinoja perheissä, joissa omaisella on Rettin oireyhtymä. Voitte osallistua tutkimukseen, mikäli olette yli 18-vuotias, teillä on omainen, jolla on Rettin oireyhtymä, ja käytätte hänen kanssaan AAC-keinoja (puhetta tukeva ja korvaava kommunikointi, *augmentative and alternative communication*). Tutkimuksen tarkoituksena on kerätä tietoja tutkittavien kokemuksista AAC-keinoista ja niiden toimivuudesta perheissä, joihin kuuluu henkilö, jolla on Rettin oireyhtymä. Tämä tiedote kuvaa tutkimusta ja Teidän mahdollista osuuttanne siinä.

Tutkimuksen kulku

Tutkimus on kyselytutkimuksena toteutettava kertatutkimus. Aineisto kerätään O365 Forms -ohjelmalla luodulla kyselylomakkeella. Kyselyn täyttämiseen aikaa kuluu noin 15–35 minuuttia. Kyselyyn vastaaminen on vapaaehtoista. Kyselyn vastaukset vastaanottaa logopedian maisterivaiheen opiskelija Jutta Kivikangas. Vastauksia käytetään Tampereen yliopistolla anonyymina tutkimusaineistona. Tutkimukseen osallistumisesta ei makseta palkkiota.

Luottamuksellisuus, tietojen käsittely ja säilyttäminen

Teistä kerättyä tietoa käsitellään luottamuksellisesti tietosuojalain edellyttämällä tavalla. Tutkimustiedostoa ja tutkimuksen yhteydessä kerättyjä aineistoja säilytetään Tampereen yliopistossa 5 vuotta, jonka jälkeen ne hävitetään. Kyselyssä Teistä kerättäviä henkilötietoja on tieto siitä, että omaisellanne on Rettin oireyhtymä. Omaisestanne kyselyssä pyydetään kertomaan ikä kuukauden tarkkuudella. Yksilöiviä tunnistetietoja ei kerätä. Lisätietoa tietosuojasta tietosuojailmoituksessa.

Lisätiedot

Tutkimukseen liittyviin kysymyksiinne vastaa Jutta Kivikangas.

Tutkijoiden yhteystiedot

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