Communication and eating proficiency in 125 females with Rett syndrome -

The Swedish Rett Center Survey

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Examensarbete i logopedi, 20 p
Höstterminen 2004

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Abstract

Purpose: To describe communication and eating proficiency and related factors in Swedish females with Rett syndrome, from a parental/caregiver view.
Method: Data from a questionnaire were used and was approached descriptively, qualitatively and comparatively. Parents/caregivers of 125 females with Rett syndrome participated. The mean age of the females was 19.6 years.
Results: Expressive language was limited and the most common way of communicating was by producing motor acts of which 37% was eye gazing. Results showed that these females show communicative behaviour during social activities. Out of eight eating difficulties studied, the majority suffered from one, two or no eating difficulty at all. There were some significant relationships between eating difficulties and respectively breathing dysfunctions and food consistency.
Conclusions: Results from this study concerning eating are generally good but the females’ verbal communication was as expected limited. However for optimal intervention, evaluating the females eating situation and communication ability together with family and professionals would probably be valuable for a majority of the females. The study also shows that these females are in need of and enjoy social interplay.
Ethical approval for this project was granted by the Ethical Committee at the University of Umeå, § 15/97, dnr 97-1.
Acknowledgements

We are grateful to supervisor at Umeå University, Prof Jan van Doorn and our supervisor at Swedish Rett Center in Östersund, speech and language therapist Magdalena Jochym Nygren for excellent supervision and support. We would also like to thank research engineer at the dept of logopedics at the University of Umeå Anders Asplund for technical support and MD, Ph.D. and director of Swedish Rett Center in Östersund Ingegerd Witt Engerström for letting us take part in their project. A special thank you to all the females and their families who enabled us to provide a description of their experiences of eating and communication in Rett syndrome.
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Introduction

Rett syndrome (RS) is a neurodevelopmental disorder that seems to affect females almost exclusively although occurrence of Rett syndrome in males have been reported. It is the leading cause of mental retardation in females, second only to Down’s syndrome, with an estimated prevalence of 1 in 10 000-22 000. In 1999 mutations in the MECP2 gene were described, which has been linked to the presence of Rett syndrome. Huppke and colleagues have found mutations in the MECP2 gene in 77% (n=31) of the females with Rett syndrome. However there is still a need of more research to be sure how common the mutation is among these girls and today Rett syndrome is still diagnosed clinically.

The syndrome is divided into four stages. The stagnation stage (stage 1) generally begins between five and 18 months and is characterized by general developmental stagnation and delay in postural control and balance. The regression stage (stage 2), usually begins between the age of one and four years and there is generally a loss of acquired skills such as hand function, fine motor skills, babbling, words and activity and some girls display autistic like symptoms. Stereotypical hand movements including wringing, washing, clapping and repeatedly moving hands to the mouth begin to emerge at this stage. Breathing irregularities may occur. The onset for the plateau or stationary stage (stage 3) is usually at an age of two to 10 and is characterized by apraxia, motor problems and seizures but may also include ‘reawakening’, return of motivation and improvement in communication and behaviour. Some girls remain in this stage for most of their lives. The last stage, motor deterioration stage (stage 4), includes reduced mobility and severe physical disability. Girls who have earlier been able to walk may lose this ability. Repetitive hand movements may decrease and eye gaze usually improves.

A characteristic feature of Rett syndrome is loss of, or severely impaired impressive and expressive language. There are females with Rett syndrome with preserved speech and these patients are considered to have a variant sub-group of Rett syndrome. Budden et al. found deterioration in verbal and expressive language in all 20 girls in their study regardless of what stage the females were in. Out of 99 females with Rett syndrome, aged 3:6-29:9, Uchino and colleagues reported that more than half spoke some words and their language development level was around one year of age. Almost 90% of those who spoke words in their early speech development ceased to utter words by 40 months. The most common way for the female to express her needs was reported as eye gazing by Leonard et al. Other methods reported were pictures or other forms of assisted communication, gestures, facial expressions and words or sounds with specific meaning.

Several studies report feeding and nutritional problems as common clinical findings in Rett syndrome. One study reported feeding problems in 82% of the 49 cases seen in a clinic. Morton et al. investigated feeding ability in 20 females with Rett syndrome, aged 1:6-33:0 using video-fluoroscopy. All individuals showed reduced movements of the mid and posterior tongue with premature spillover of food and liquid from mouth into the pharynx. Delayed pharyngeal swallow was also observed. Furthermore breathing disturbances during feeding has been reported by Morton et al. Most of the girls are dependent or need to be assisted during feeding as reported in several studies and adjustment of food consistency is also described. Additionally tooth grinding is reported as common in a study by Magalhães and colleagues.
Purpose

The aims of this study were to describe eating proficiency and related factors such as breathing dysfunction, seizures and oral and tooth condition in 125 Swedish females with Rett syndrome. In addition we aimed to investigate any relationships between eating functions and associated functional problems. We also wanted to survey communicative behaviour in these females.

Methods

This study is based on a questionnaire, looking at those sections that relate to communication and eating. It complements another study on the same questionnaire, where other sections relevant to physiotherapy were selected for investigation. An important aspect of the study is that the data represents exclusively the parental/caregiver’s view of the problems that females with Rett syndrome might live through.

Recruitment and participants

In 1996 the Swedish Rett Center in Östersund, designed a survey that was sent to all 178 families in Sweden with a girl or woman who at that time had been diagnosed with Rett syndrome (according to research register, University of Gothenburg). Ethical approval for the project was granted by the Ethical Committee at the University of Umeå, § 15/97, dnr 97-1.

The survey was sent to all 178 families in Sweden with a girl or woman who at that time had been diagnosed with Rett syndrome (according to research register, University of Gothenburg). Participants, including both parents and staff depending on where the female was living at the time, were informed about the aim of the study and that participation was optional. After two postal reminders 125 (70%) questionnaires were returned. A majority of the respondents were the families of the females (94 family, 24 both family and staff, six staff, one was unclear). The age range of the females was 2.5-55.0 years with a mean age of 19.6 years and median of 24.5 years. Regular contact with a speech therapist was reported in 17% of the females.

Questionnaire

The questionnaire included open-ended and closed questions where participants sometimes were also invited to comment. The questions concerned background data, habilitation/treatment, early development, current function and abilities, mobility, behaviour and communication, activities and technical aids. The questions that were chosen for this study were those relevant to a speech and language therapist’s perspective: eating ability including breathing dysfunctions, seizures, tooth and oral conditions and behaviour and communication. The specific sections from the questionnaire can be found in Appendix 1 (translated from Swedish to English).
Data analysis

The responses from all the questionnaires were reviewed before the data were collated. Occasionally, in questions that had yes/no alternatives, the respondents had qualified their answer (e.g. ‘yes, sometimes’, ‘no, not so often’). We have included those qualified responses by reporting them in the appropriate yes/no category. All answers that were ambiguous or where the respondent had selected two answers were consistently excluded, as were all ‘do not know’ answers. All data from the selected sections of the questionnaire were entered into a spreadsheet in Microsoft Office Excel 2003 for further analysis. Descriptive analysis was performed on the closed questions and the open-ended questions were qualitatively sorted into themes and then analyzed descriptively. Chi-squared testing was used to investigate the relationships among variables. In some cases the tables did not meet the criterion for expected frequencies and Chi squared test with Yates’ continuity correction for small numbers was applied.

Results

We report results that involve communication and eating and related factors. As the numbers of internal dropouts vary in the different questions, the total number of respondents are presented in brackets. A small number of respondents tended to report only yes-answers but left out no-answers, so that the number of answers differs from question to question. In three questionnaires there was one page missing.

COMMUNICATION

Questions concerning spoken language were answered by 122 caregivers. The use of one or several words in early childhood was reported in 69% of the females and 19% were still using words at the time the questionnaire was filled in. The use of sentences in early childhood was reported in 12%. One female reportedly used sentences when singing. Most of the females had vocabularies consisting of words consisting of reduplicated syllables such as mamma (mother), pappa (father) and other frequently used words in infants, e.g. titta (look) and lampa (light). For 47 of the females who used words in early childhood there was an age reported when the females stopped speaking (Table 1).

<table>
<thead>
<tr>
<th>Age group</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>0:0-0:11</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>1:0-1:11</td>
<td>16</td>
<td>34</td>
</tr>
<tr>
<td>2:0-2:11</td>
<td>14</td>
<td>30</td>
</tr>
<tr>
<td>3:0-3:11</td>
<td>6</td>
<td>13</td>
</tr>
<tr>
<td>4:0-4:11</td>
<td>4</td>
<td>8</td>
</tr>
<tr>
<td>5:0-5:11</td>
<td>1</td>
<td>2</td>
</tr>
<tr>
<td>6:0-</td>
<td>5</td>
<td>11</td>
</tr>
</tbody>
</table>

At the time when the questionnaire was filled in some females used more than one way to express their will and the 100 respondents reported a total of 186 answers. The ways of expressing will have been classified according to Granlund and Olsson’s system that is
based on which of three mediums is used for communication. Vocal sounds involve producing sounds with voice (e.g. sounds, speech, laughter, screaming, crying), objects (e.g. pounding with hand or object, rattling) or technical aids. Motor acts involve communicating with body movements (e.g. reflexes, mimics, pointing, gestures, signs and sign language). The graphical way of communicating is to mark letters, a picture or graphic symbol by pointing or pressing a button or by writing or drawing.

The most common way to express will, which was reported in 90% of the females, was by producing motor acts. Some of the motor acts could be specified as acts that indicate a developmentally higher level of language\(^1\) (eye pointing, finger pointing and gestures). Additionally a vast majority of these higher level acts consisted of eye pointing. Expressing will by producing vocal sounds (e.g. sounds, laughter, screaming, crying) was reported in 55% of the females. Four per cent of the total number of females used words which also indicates a developmentally higher level of language. Four per cent of the females used a graphical way of expressing their will (all four used Yes/No board).

![Ways of expressing will in 100 females with Rett syndrome.](image)

**Figure 1** Ways of expressing will in 100 females with Rett syndrome. The grey marked area in Motor acts is eye pointing, finger pointing and gestures. The grey marked area in Vocal sounds is use of words. Note that use of Graphical way automatically is classified as a higher level of language. Higher level of language may or may not include lower level of language.

\(^1\) According to Granlund and Olsson\(^2\) higher level of language can be defined as intentional communication with a symbolic system or signals, i.e. spoken language, sign language, pointing at objects/pictures or using technical device. Lower level of language consists of e.g. body language and natural reactions.
When answering a question concerning use of communication aids 40 respondents reported that the female used communication aids and the distribution is shown in table 2. Some of them used more than one communication aid and the total number of aids was 58.

<table>
<thead>
<tr>
<th>Table 2</th>
<th>Communication aids in 40 females with RS</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N</td>
</tr>
<tr>
<td>Yes/No board (Marie Jensen)</td>
<td>24</td>
</tr>
<tr>
<td>Pictures</td>
<td>14</td>
</tr>
<tr>
<td>Pointing aids</td>
<td>5</td>
</tr>
<tr>
<td>Big Mack</td>
<td>3</td>
</tr>
<tr>
<td>Photographs</td>
<td>3</td>
</tr>
<tr>
<td>Computer</td>
<td>3</td>
</tr>
<tr>
<td>Other</td>
<td>6</td>
</tr>
</tbody>
</table>

One open question in the questionnaire asked the respondents to state the situations in which the female expressed her will. The question was answered by 97 respondents and some of them reported more than one situation and the total number of answers was 101 answers. 60% of the females expressed their will in situations involving eating, 6% when brushing teeth and 5% reported that the female expressed her will in all kinds of situations. Thirty-three per cent of the respondents reported other situations.

**EATING**

**Eating difficulties and eating aids**
The eating proficiency was found to be often impaired. The frequency of different types of eating difficulties can be seen in figure 2 while figure 3 depicts the distribution of the number of eating difficulties. It should be noted that the number of eating difficulties should not be interpreted as a degree of eating disability, e.g. four eating difficulties compared to two does not mean that eating is twice as difficult.

![Frequency of specific eating difficulties](image)

**Figure 2** Distribution of different types of eating difficulties in females with Rett syndrome.
The eating situation may be facilitated by using proper eating aids and 63% (n=122) reported use of one or more eating aids and of these 59 respondents specified what type of aid, which can be seen in table 3.

**Table 3** Occurrence of eating aids in 59 females with RS

<table>
<thead>
<tr>
<th>Aid</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Plate with edge</td>
<td>28</td>
<td>48</td>
</tr>
<tr>
<td>Specially adjusted utensils</td>
<td>25</td>
<td>42</td>
</tr>
<tr>
<td>Trainer cup</td>
<td>17</td>
<td>29</td>
</tr>
<tr>
<td>Feeding bottle</td>
<td>7</td>
<td>12</td>
</tr>
<tr>
<td>Other</td>
<td>10</td>
<td>17</td>
</tr>
</tbody>
</table>

The females reported one or more types of positioning during meals and the distribution can be seen in table 4. “Other” was in all 16 cases specified as wheel chair.

**Table 4** Positioning during meals in 125 females with RS

<table>
<thead>
<tr>
<th>Position</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Specially adjusted chair</td>
<td>77</td>
<td>62</td>
</tr>
<tr>
<td>Ordinary chair</td>
<td>45</td>
<td>36</td>
</tr>
<tr>
<td>Lap</td>
<td>4</td>
<td>3</td>
</tr>
<tr>
<td>Other</td>
<td>16</td>
<td>13</td>
</tr>
</tbody>
</table>

Use of some orthotic device that could have an effect on posture and the ability to move hands and arms when eating was reported by 42% (n=86) and some of the females used several orthotic devices. Corset was used by 19% of the females, elbow restraint by 14% and hand splint by 9%. Two females had earlier used respectively corset and elbow restraint, but did reportedly not use them anymore.
Participation in eating

Out of 121 respondents 69% stated that the female could eat without help in early childhood and of those 83 females 65% had since lost the ability to eat without help. Thirty-six respondents reported the age at which the ability to eat without help was lost (Table 5).

Table 5  Age when the ability to eat without help was lost in 36 females with RS

<table>
<thead>
<tr>
<th>Age group</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>1:0-1:11</td>
<td>11</td>
<td>31</td>
</tr>
<tr>
<td>2:0-2:11</td>
<td>13</td>
<td>36</td>
</tr>
<tr>
<td>3:0-3:11</td>
<td>4</td>
<td>11</td>
</tr>
<tr>
<td>4:0-4:11</td>
<td>3</td>
<td>8</td>
</tr>
<tr>
<td>5:0-5:11</td>
<td>3</td>
<td>8</td>
</tr>
<tr>
<td>6:0-</td>
<td>2</td>
<td>6</td>
</tr>
</tbody>
</table>

The question concerning the female’s present ability to participate in eating was answered by 117 respondents. Eighteen per cent ate without help or sometimes had to be assisted (11 of these females reported independent eating at all time), 21% needed to be assisted when eating or at times fed and 61% were being fed at all times. Three females with PEG (percutan endoscopic gastrostomy) were all being fed when taking food orally. Ages for PEG operation were respectively five and ten years for two of the females and for one female there was no answer.

Food consistency and diet

Almost all girls, 98% (n=124), including the three females with PEG when they were being fed orally, ate ordinary food. Two of the remaining three females needed a special diet free from gluten and lactose respectively and one had problems with the gall bladder and required special diet. Twenty-one per cent (n=122) were given extra supplementation (energy and calories) besides regular food. Two out of the three females with PEG reported that they received supplementary feeds via the gastrostomy.

As many of the females were reported to have difficulty chewing effectively there might have been a need to adjust food consistency. Adjusted food consistency was used in 61% (n=121) of the females. Food consistency was specified by 76 respondents: 71% reported that the females usually were given chopped food only or in combination with mashed or strained food, 18% were given mashed food only or in combination with strained and 11% strained food only.

Clinical experience of children with eating difficulties has indicated that there may be relationship between food consistency and eating disability. We wanted to find out if there was any statistical significance regarding this issue and the results are shown in table 6. The percentages shown in the table represent the proportion of females with a specific eating difficulty as a percentage of the number who eat normal or adjusted food respectively, i.e. 5% of females who take normal consistency food are reported to be unable to chew, compared with 40% of those who eat adjusted consistency food.
**Table 6** Relationships among eating difficulties and food consistency

<table>
<thead>
<tr>
<th>Eating characteristics</th>
<th>Normal consistency</th>
<th>Adjusted consistency</th>
<th>Statistics</th>
</tr>
</thead>
<tbody>
<tr>
<td>Can't chew</td>
<td>5%</td>
<td>40%</td>
<td>$X^2 (1, n=107) = 16.4, P&lt;.001^{**}$</td>
</tr>
<tr>
<td>No lip closure when bolus in mouth</td>
<td>49%</td>
<td>35%</td>
<td>N.S.</td>
</tr>
<tr>
<td>Difficulty drinking from a cup</td>
<td>19%</td>
<td>37%</td>
<td>$X^2 (1, n=117) = 4.34, P=.037^*$</td>
</tr>
<tr>
<td>Difficulty swallowing</td>
<td>11%</td>
<td>27%</td>
<td>$X^2 (1, n=113) = 3.94, P=.047^*$</td>
</tr>
<tr>
<td>Holding bolus in mouth without swallowing</td>
<td>33%</td>
<td>49%</td>
<td>N.S.</td>
</tr>
<tr>
<td>Frequent occurrence of aspiration</td>
<td>6%</td>
<td>24%</td>
<td>$X^2 (1, n=119) = 6.04, P=.014^*$</td>
</tr>
<tr>
<td>Vomits during meals</td>
<td>4%</td>
<td>18%</td>
<td>$X^2 (1, n=119) = 4.64, P=.031^*$</td>
</tr>
<tr>
<td>Vomits between meals</td>
<td>4%</td>
<td>16%</td>
<td>$X^2 (1, n=119) = 3.97, P .046^*$</td>
</tr>
</tbody>
</table>

* $p<.05$, **$p<.01$. N.S. stands for non-significant.

**Influence of other factors**

**Seizures**

Seizures of some kind were reported by 74% ($n=123$) of the respondents and some respondents described several types of seizures. In connection to this question there was space to describe what type of seizures and for some of the females both epilepsy and other types of seizures were reported. The answers were divided into two categories: epilepsy specifically mentioned that was reported in 58% of the females and other types of seizures that were reported in 49% of the females. We looked for relationships between the different types of seizures and eating difficulties using Chi-square test but we found no relationships that reached significance.

**Breathing**

Breathing dysfunction is a common feature in Rett syndrome. The occurrence of four types of breathing problems in these girls can be seen in table 7. Forty-six per cent ($n=68$) of the respondents said that they could affect the breathing disturbances in some way, e.g. by distracting with calm talk, singing and/or touching.

**Table 7** Occurrence of breathing dysfunction in females with RS

<table>
<thead>
<tr>
<th>Breathing Dysfunction</th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Abnormally irregular breathing ($n=107$)</td>
<td>39</td>
<td>36</td>
</tr>
<tr>
<td>Apnoea ($n=110$)</td>
<td>57</td>
<td>52</td>
</tr>
<tr>
<td>Hyperventilation ($n=117$)</td>
<td>65</td>
<td>56</td>
</tr>
<tr>
<td>Swallowing of air ($n=117$)</td>
<td>50</td>
<td>43</td>
</tr>
</tbody>
</table>

We looked for relationships between the different types of breathing dysfunctions and the eating difficulties in females with Rett syndrome using Chi-square test. The results can be seen in table 8. The table shows the proportion of females with a specific eating problem as

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ii Some of the descriptions indicate epileptic seizures but as the respondents have not specified with the word “epilepsy” the answer has been categorized as “other types of seizures”. Other types are also laughter and crying attacks, convulsions, hyperventilation and panic attacks.
a percentage of the number who have a specific breathing problem, i.e. 32% of those females who have irregular breathing are unable to chew, while 53% of them do not close their lips when there is bolus in their mouths, etc.

Table 8  Relationships among eating difficulties and breathing dysfunction

<table>
<thead>
<tr>
<th></th>
<th>Irregular breathing</th>
<th>Apnoea</th>
<th>Hyperventilation</th>
<th>Bloating</th>
</tr>
</thead>
<tbody>
<tr>
<td>Can't chew</td>
<td>32%</td>
<td>28%</td>
<td>25%</td>
<td>21%</td>
</tr>
<tr>
<td>No lip closure when bolus in mouth</td>
<td>53%</td>
<td>49%</td>
<td>50%*</td>
<td>56%**</td>
</tr>
<tr>
<td>Difficulty drinking from a cup</td>
<td>31%</td>
<td>27%</td>
<td>18%*</td>
<td>15%*</td>
</tr>
<tr>
<td>Difficulty swallowing</td>
<td>26%</td>
<td>23%</td>
<td>18%</td>
<td>17%</td>
</tr>
<tr>
<td>Holding bolus in mouth without swallowing</td>
<td>36%</td>
<td>48%</td>
<td>35%</td>
<td>27%*</td>
</tr>
<tr>
<td>Frequent occurrence of aspiration</td>
<td>32%**</td>
<td>29%**</td>
<td>23%*</td>
<td>12%</td>
</tr>
<tr>
<td>Vomits during meals</td>
<td>18%</td>
<td>16%</td>
<td>13%</td>
<td>8%</td>
</tr>
<tr>
<td>Vomits between meals</td>
<td>16%</td>
<td>18%</td>
<td>13%</td>
<td>9%</td>
</tr>
</tbody>
</table>

* p<.05, **p<.01

Teeth grinding

Teeth grinding in females with Rett syndrome is common and can cause oral discomfort. Occurrence of diurnal and nocturnal teeth grinding and teeth clenching (point of time not specified) is shown in table 9.

Table 9  Occurrence of teeth grinding and teeth clenching in females with RS

<table>
<thead>
<tr>
<th></th>
<th>Diurnal teeth grinding (n=115)</th>
<th>Nocturnal teeth grinding (n=103)</th>
<th>Teeth clenching day/night (n=98)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>N</td>
<td>%</td>
<td>N</td>
</tr>
<tr>
<td>Severe</td>
<td>19</td>
<td>17</td>
<td>5</td>
</tr>
<tr>
<td>Moderate</td>
<td>23</td>
<td>20</td>
<td>12</td>
</tr>
<tr>
<td>Light</td>
<td>27</td>
<td>23</td>
<td>11</td>
</tr>
<tr>
<td>None</td>
<td>46</td>
<td>40</td>
<td>75</td>
</tr>
</tbody>
</table>

We studied possible relationships between teeth grinding and the specific eating difficulties using Chi-square test but we found no relationships that reached significance.

Dental/oral problems

Table 11 shows the occurrence of seven common oral problems in 121 females with Rett syndrome. Some females suffered from several of these problems.

Table 11  Occurrence of oral problems in 121 girls with RS

<table>
<thead>
<tr>
<th></th>
<th>N</th>
<th>%</th>
</tr>
</thead>
<tbody>
<tr>
<td>Injuries on teeth from falling</td>
<td>41</td>
<td>33</td>
</tr>
<tr>
<td>Incorrect bite</td>
<td>39</td>
<td>32</td>
</tr>
<tr>
<td>Bleeding gingiva</td>
<td>36</td>
<td>30</td>
</tr>
<tr>
<td>Bad breath</td>
<td>25</td>
<td>21</td>
</tr>
<tr>
<td>Caries</td>
<td>9</td>
<td>7</td>
</tr>
<tr>
<td>Periodontitis</td>
<td>5</td>
<td>4</td>
</tr>
<tr>
<td>Ulcer in mouth mucosa</td>
<td>2</td>
<td>2</td>
</tr>
<tr>
<td>Other</td>
<td>3</td>
<td>2</td>
</tr>
<tr>
<td>No problems</td>
<td>26</td>
<td>21</td>
</tr>
</tbody>
</table>
These oral problems were tested for relation with each eating difficulty, using Chi square-test. We found no relationships between these oral problems and eating difficulties that reached statistical significance except between difficulty swallowing and oral problems.

A majority of the females, 88% (n=119), had not received any treatment to avoid injuries on teeth caused by teeth grinding. Nine of the females had been treated with bite plates, two with plate and one with teat. Five per cent (n=114) had received some kind of oral motor treatment by a speech and language therapist or a dentist.

The question concerning dental treatment was answered by 117 caregivers. Dental treatment carried out well in 70% of the females, less well in 15% and badly in 15%. Out of 122 females tooth brushing was easy in 33%, caused some problems in 57% and caused major problems in 10%. In conjunction with the latter question 66% (n=122) described how tooth brushing was carried out. Some of the respondents described several types of issues. 61% reported that the female did not want to open her mouth or bit the toothbrush, 16% showed struggling and/or defending behaviour, for 10% there were problems in reaching different areas in the mouth e.g. due to the tongue being in the way, 8% showed signs of oral hypersensitivity and 8% used some kind of aid to facilitate tooth brushing. Five answers could not be put in any of these categories, e.g. there was one girl who had small seizures during tooth brushing. Six respondents described positive experiences of tooth brushing.

BEHAVIOUR/ACTIVITIES

Some questions in the questionnaire concerned behaviours and activities of the females. The questions were open-ended and we selected the answers that were relevant for the purpose of this study which were those concerning communicative behaviour and eating except the question about how the females express their will which has already been reported under results for communicative behaviour. The answers were categorized into four groups (eating, social interplay, being at the centre of attention, participation). As some respondents reported several answers that belonged to different groups the total number of answers exceeds the number of respondents.

The question in the questionnaire that concerned situations in which the girl is most active was answered by 112 caregivers. Twenty-nine per cent reported that the females were most active during meals, 17% during social interplay, 3% when the female was at the centre of attention and 1% when she was able to participate in an activity. Fifty-four per cent of the respondents answered activities unrelated to communicative behaviour or eating and thus were not of interest for this study. The question concerning what situations the females liked most was answered by 114 caregivers. Thirty-nine per cent of the respondents described social interplay situations as the females favourite, 18% answered eating, 10% being at the centre of attention and 3% where the female could participate. Other situations that did not involve eating or communicative behaviour were described by 42% of the respondents. Regarding situations that the females liked least four per cent reported situations involving eating. Seventy-four per cent answered situations that did not involve

iii Our view of communicative behaviour is broad and includes both verbal and non-verbal communication and we have referred to it as social interplay. With our definitions verbal communication is talking, reading or singing to the female and non-verbal communication includes close contact, cuddling and joking around with the female.
eating or communication. One-hundred-and-six caregivers answered the question concerning best contact situations. Fifty-three per cent reported social interplay as situations when having best contact with the female, 17% situations answered situations involving eating, 7% reported “all situations”, 3% when the female is at the centre of attention and 1% when the female gets to participate. Twenty-four per cent reported situations that did not involve eating or communicative behaviour.

**Discussion**

As data in this study were collected from questionnaires that were filled in by parents and/or caregivers of females with Rett syndrome it is important to have this in mind when comparing results from this study to those that are based on clinical/professional examinations. In most cases the parents have filled in the questionnaire even though the female might have lived in a group residence and thus did not have daily contact with the parents. Only a few studies have previously been conducted with this parental/caregiver’s view design.\(^{13,23}\)

**COMMUNICATION**

Usage of one or several words in early childhood was reported in 69% and 12% used sentences in early childhood. At the time when the questionnaires were filled in only 19% still used words. Uchino and colleagues\(^{12}\) have reported a result where more than half of 99 females spoke meaningful words and that 14.5 % of these females spoke two-word sentences in early childhood and most of these females ceased to utter words. A comparison of the results in our study show that even though 19% reported usage of words (which also may be use of words in a non-communicative way), only four reported use of words when expressing their will. In our study 66% stopped speaking before 36 months of age similar to Uchino et al’s.\(^{12}\) result of 64.6% at 40 months.

The expressive language ability in these females is often severely impaired and as the result shows motor acts is the most common way of expressing will which indicates that using body movements probably is the easiest way for these females to express themselves. A vast majority of the 50 females that used a higher level of motor act reported eye pointing. This result can be compared to the result of Leonard et al\(^{13}\) where eye gazing was reported as the most common method for the females to express their will in 41.9% of the cases. The results from our study indicate some use of a developmentally higher level of language, especially in the case of motor acts where these are eye- and finger pointing and gestures whereas use of higher level of vocal sounds is very limited.

Sixty per cent of the 40 females that were reported to use communication aids had been provided with Marie Jensen’s communication aid that consists of a board with a red and green circle for ‘yes’ and ‘no’. This is an eye-pointing device especially designed in the middle of the 1990s by a special educationalist for females with Rett syndrome. There is probably a relation between the time of the questionnaire being sent out and the time when this communication aid was distributed and this might explain why this was reported as the most common communication aid. Other communication aids were also reported but were not very common.
An interesting observation is that when reporting way of expressing will in the females, only four out of 100 stated that the female used a graphical way while as many as 40 females had been provided with a communication aid which most of the time involves communicating in a graphical way. There are several possible explanations to this. When introducing a communication aid it is important that professionals carefully try it out with the female and people in the immediate surroundings and one have to take into account the female’s and surroundings needs and ability. To support optimal language development and communication it is vital that the alternative communication method is introduced as early as possible and in consensus with the surroundings. When everybody is familiar with the communication aid it is important to maintain the ability to use the aid and the responsibility for this lies mainly on the surroundings. If this is not achieved it might be difficult to make alternative communication aids work in daily life.\(^{24}\)

Many of the answers from the questions concerning behaviour and activity indicate that these females are in need of and enjoy social interplay and interaction with other people. This is encouraging to see as there are other results in this study that indicate that most of the females are limited in their communicative behaviour.

**EATING**

As there are several studies that report extensive problems with eating\(^{14, 15}\) our expectations were that these females would have several eating difficulties. Almost 80% reported one or more eating difficulties but as a majority had only one, two or no eating difficulty at all, the results indicate a better eating ability than we had expected. The most common impairments were absence of lip closure when having food in mouth and holding bolus in the mouth without swallowing. The least common difficulties were vomiting during and between meals. The fact that only 26% in our study could not chew is remarkable as all 20 females in a study by Morton et al.\(^{15}\) were reported as having mild to severe problems with chewing. Worth noticing is that this question in our questionnaire is ‘Can your daughter chew the food?’ and thus degree of difficulty can not be specified. When it comes to occurrence of aspiration the 16% (n=123) in our study is comparable with that of Morton et al.\(^{15}\) where aspiration of food was seen in 15% (n=20). It is important to remember that the data in our study is based on the caregivers’ view of how frequently the females were aspirating whereas Morton and colleagues observed frequency of aspiration on videofluoroscopy. Motil and colleagues\(^{18}\) reported that 62% of 13 females had poor lip closure during feeding while only 39% of 113 are reported having this problem in our study. The differences in our results compared to the studies mentioned might be explained by the fact that professionals with more demanding criteria have made the judgements in studies by Morton and Motil while it has been judged by caregivers in our study. Another factor that might affect the results is the difference in number of subjects.

Over half of the females in this study had lost the ability to eat independently. Additional comments made by parents/caregivers showed that the female might need help with initiating the eating or that only eating with hands or spoon is possible or that there were more spills than earlier. This indicates a general deterioration with increasing age which is supported in a study by Morton et al.\(^{15}\) However, in our study one girl had improved in her eating and had acquired the ability to eat with a spoon at an age of ten years. This confirms results about possible learning and relearning in Rett syndrome that have been reported in studies earlier.\(^{18, 25, 26}\)
The need for assistance during eating is in this study categorized into three groups on the basis of best reported ability. Sixty-one per cent of the females in our study who needed to be fed is in accordance with the 59% that has been reported by Reilly and Cass. Based on the same questionnaire as our study but a different section focusing on ability before and after regression Larsson reported inability to self-feed in 57% of the females. In addition, Motil et al. reported inability to self-feed in 92% out of 13 females. When it comes to independent eating the 9% in our study that could eat without help indicate a better preserved ability than the 3.4% that has earlier been reported. As eating might cause problems for many of the girls it is encouraging that as many as 60% of the females could express their will in situations involving eating and 17% of the respondents felt that situations involving eating provided best contact with the female. This indicates that these constantly occurring situations create opportunities for the females to have an influence on their living conditions and to make conscious choices. In addition almost one third reported that the females were most active during meals. Only 4% stated eating situations as occasions the females liked the least.

Sixty-three per cent of the females used one or more eating aids. The eating aids that can be provided are different models of utensils, plates and cups/glasses and feeding bottles. These aids do not have an effect on most of the eating difficulties listed in the questionnaire, although they can facilitate feeding and enable more independent eating for the female and there are certainly still more females that could gain from using eating aids. It is important that eating ability in girls with Rett syndrome is examined in a feeding situation. Teamwork between caregivers and professionals with experience of eating disabilities (e.g. speech therapist or occupational therapist) may be necessary to create a well-functioning eating situation for the female.

Positioning during eating is of great importance, especially for persons who have eating difficulties. Inadequate positioning might cause unnecessary distress such as deficient bolus control and difficulty in reaching the food. In an unpublished master thesis, based on the same questionnaire that we have used, Larsson reported that 74% of the females had developed a scoliosis and kyphosis was reported in 24%. Our results show that about 60% sit in a specially adjusted chair during meals and with Larsson’s results in mind there might be more females that could benefit from using a specially adjusted chair. The wheelchair chairs that are reported might also provide good support when eating, but sitting on an ordinary chair or in someone’s lap might not provide enough support if there are spinal curvatures. It should be noted that not all girls have sufficient difficulties to require a special chair and can use an ordinary chair. Another very important aspect when it comes to feeding is to enable good eye contact, which can be achieved only with optimal seating.

Forty-two per cent of the females used some orthotic device (foot splint not included). A corset can be used to facilitate the female’s balance and provide good support for optimal sitting during meals. Elbow restraints and hand splints may decrease stereotypic hand movements and hand-to-mouth movements that can be disruptive during eating. Although the use of orthotic device is reported in this study it cannot be assumed, due to the nature of the question, that these are used during feeding and thus whether they have an effect on the ability to eat.

Thirty-nine per cent ate food of ordinary consistency and a majority (76%) of the girls that needed adjustment were given chopped food only or in combination with other consistencies. These findings indicate a better preserved eating ability than we had...
expected as Budden et al.\textsuperscript{11} reported that all females in their study were given pureéd or soft food. The prevalence of different types of adjusted food consistencies was in our study categorized into three groups on the basis of the consistency (chopped, mashed or strained). Of those respondents who reported the specific category of food consistency (chopped, mashed or strained), the majority (76\%) reported that the food was chopped, while mashed and strained food were used in 18\% and 11\% respectively. This implies that, of the three types of adjustments, chopped food was generally the most suitable for those females whose eating problems required food consistency to be changed.

When it comes to relationships between food consistency and eating difficulties, and due to the complexity of this issue, our results are somewhat indistinct. Our expectations were that food consistency that is not individually adjusted would have a negative effect on eating proficiency. The relationships that reached significance was food consistency and can not chew/ difficulty drinking from a cup/ difficulty swallowing/ frequent occurrence of aspiration/ vomits during meals/ vomits between meals. From these results it is not possible to come to a conclusion that a certain consistency has an effect on the different eating difficulties but there certainly is a relationship between food consistency and eating proficiency. Adjusting food consistency may not be enough for some of the females that have inadequate oral motor function that might cause considerable eating disability. Although it is known from clinical experience in Sweden that some children with neurological impairments require mashed or strained food to facilitate eating, others might benefit from chopped food that has a more distinct texture. Food consistency is something that should be tried out individually, by a professional in cooperation with the caregivers, for every female. We do not know on what basis the recommendations reported in the survey have been made. When reporting prevalence of adjusted food consistency the caregivers did not have the opportunity to choose thickening of liquids and one need to have this in mind when interpreting the results, but it is still remarkable that as many as six per cent frequently aspirate food and liquids and still do not adjust food consistency.

Thommessen et al.\textsuperscript{19} pointed out that females with Rett syndrome should be provided with substantially more calories than recommended for healthy children to compensate for growth retardation and lack of weight maintenance. Considering this statement it was unexpected that only one fifth of the females in our study were given extra calories besides ordinary food.

In this study we investigated whether breathing abnormality was associated with eating proficiency on the basis that an earlier study reported frequent apnoea during swallowing even in females who do not usually have apnoea.\textsuperscript{16} We examined the relationship between four types of breathing irregularities and eight types of eating difficulties. Hyperventilation was significantly related to no lip closure when bolus is in the mouth and difficulty drinking from a cup respectively, and also between bloating and no lip closure when bolus is in the mouth and difficulty drinking from a cup respectively. Significance was also found between bloating and holding bolus in mouth without swallowing and finally between irregular breathing/ apnoea/ hyperventilation and frequent occurrence of aspiration. Since swallowing is affected by irregular breathing patterns\textsuperscript{16} (among other things) it is not surprising that we found a relationship between three out of four breathing dysfunctions and frequent occurrence of aspiration.

Diurnal teeth grinding was reported to occur to some degree in 60\% of the females and the distribution of severity of teeth grinding (none to severe) was relatively even. Almost a third of the females were reported to grind teeth at night. Magalhães et al.\textsuperscript{20} reported that
Diurnal teeth grinding was seen in nearly all 13 females that were studied and nocturnal teeth grinding was absent in all cases. The differences in results might be due to the different number of subjects. The results of this study shows that more than half of these females clench their teeth to some degree and to our knowledge there are no other reports concerning teeth clenching in Rett syndrome.

Teeth grinding is a common oral symptom in Rett syndrome and has been shown to cause dental attrition, discomfort and wear of the occlusion.20 Our results did not show any significant relationships between teeth grinding and eating difficulties, but backed up with the well-known consequences of teeth grinding it is still possible that this behaviour in some way could have a negative effect on oral functions.

Dental treatment seemed to go well in a majority of the females in this study while tooth brushing caused more problems. Since dental treatment takes place much less often than tooth brushing the females might not have had so many bad experiences from dental treatment. However, some answered that the female need to be anaesthetized during dental treatment which indicates that these occasions may be hard in some cases.

Limitations

The information from the study has to a small extent been compromised by the variation in the number of answers for different questions. This might be due to several factors, e.g. difficulty remembering after many years or new caregivers. Additionally, some of the questions are very similar to each other so that the respondents might have left out questions they felt they had already answered. Some years have passed since the questionnaire was distributed and filled in by the parents/caregivers, but we believe that most data is still applicable to the females of today who have Rett syndrome.

Conclusions

With this study we have been able to provide a description of how life with a female with Rett syndrome is experienced from a parental/caregiver’s point of view when it comes to communication, eating and related factors. As the number of families that participated in this questionnaire was high we have reason to believe that this study provides a representative picture of the lives of females with Rett syndrome in Sweden in the late 1990s. Hopefully this will lead to improved understanding of the problems females with Rett syndrome and their families might face. It is hoped that the information will lead to further studies and contribute to the development of more efficient therapeutic management for Rett syndrome.
References


21 Larsson G. General development in females with Rett syndrome, focusing on abilities, deformities and management: The Swedish Rett Center Survey. Master thesis in physiotherapy, Dept Community Medicine Rehabilitation, Umeå University, Sweden


Appendix 1

Selected sections from the questionnaire (translated from Swedish to English):

**HABILITATION/TREATMENT**
What other professionals have you been in contact with?
- speech therapist?

**YOUR DAUGHTER’S EARLY DEVELOPMENT**
Could she eat by herself? Yes/No/Don’t know
- with hands
- with spoon
Does she still? Yes/No/Don’t know
If no, at what age did she stop eating by herself?

Did she use any words? Yes/No/Don’t know
Did she use sentences? Yes/No/Don’t know
Does she use words now? Yes/No/Don’t know
Describe!..................................................................................................................................
If no, at what age did she stop using words?

**CURRENT**
*Feeding situation/energy intake*
Can your daughter
- Eat by herself?
- Eat with help?
- Is she being fed?

Does she have gastrostomy/PEG? Yes/No
At what age was it put in?

Does she eat ordinary food? Yes/No/Don’t know
- calorie supplements
- energy supplements

Does the food have ordinary consistency? Yes/No/Don’t know
If not, is it
- chopped
- mashed
- strained

Can your daughter chew the food? Yes/No/Don’t know
Does she close her lips when having food in her mouth? Yes/No/Don’t know
Does she have difficulty drinking from a glass/cup? Yes/No/Don’t know
Does she have difficulty swallowing? Yes/No/Don’t know
Does she keep food in her mouth without swallowing? Yes/No/Don’t know
Does she aspirate frequently? Yes/No/Don’t know
Does she vomit during eating? Yes/No/Don’t know
Does she vomit between meals? Yes/No/Don’t know
Appendix 1

Are eating aids used?
- specially adjusted utensils
- trainer cup
- plate with edge
- feeding bottle
- other

How does she sit when eating?
- in specially adjusted chair
- in lap
- on ordinary chair
- in other way,
  describe!

Teeth/Bite conditions
How does she cope with dental treatment? well less well badly
How does tooth brushing carry out? easy some problems big problems
Describe!

Have your daughter had any of the following problems?
- caries
- bleeding gingiva
- periodontitis
- ulcer in mouth mucosa
- bad breath
- incorrect bite
- injuries on teeth from falling, e.g. during seizures
- other

Does your daughter grind teeth during the day?  None Lightly Moderately Severely
Does your daughter grind teeth during the night? None Lightly Moderately Severely
Does she clench her teeth? None Lightly Moderately Severely

Have she had oral motor treatment by speech therapist/dentist? Yes/No/Don’t know
- bite plane
- other

Breathing
Does your daughter have abnormally irregular breathing? Yes/No/Don’t know
Does she apnoea? Yes/No/Don’t know
Does she hyperventilate? Yes/No/Don’t know
Does she swallow air? Yes/No/Don’t know
Can you affect it? Yes/No/Don’t know
How?
Appendix 1

Seizures
Does your daughter have any type of seizures? Yes/No/Don’t know
Describe!..................................................................................................................................

BEHAVIOUR AND COMMUNICATION
In what situations is your daughter most active? Describe!....................................................
What situations do you experience your daughter liking the most? Describe!......................
In what situations do you experience best contact with her? Describe!................................
How does your daughter express her will? Describe!............................................................
In what situations does she express her will? Describe!..........................................................

MEDICAL TREATMENT
Does your daughter use any of the following orthotic devices? Yes/No/Don’t know
- hand restraint
- elbow restraint
- corset
- other

DAILY LIVING AIDS
What aids does your daughter use?
- communication aids
Which?........................................................................................................................................