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Social-emotional instability in individuals with Rett syndrome: parents' experiences with second stage behaviour

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Abstract

Background While the medical profession often terms behaviours in individuals with Rett syndrome (RTT) in the second stage as 'autistic-like', parents disagree with this description. The present study focuses on a comparison of parents' experiences with the social-emotional behaviour of the child with RTT in the second and subsequent stages.

Method In collaboration with the Dutch Rett Syndrome Organization, 51 parents of children with RTT in the Netherlands took part in the present study. Parents completed an online questionnaire to clarify their experiences of the social-emotional behaviour of their children during and after the second stage of RTT. Both quantitative and qualitative analysis techniques have been used.

Results The results of the paired-samples *t*-test show that parents see significantly less social-emotional behaviour in the children during the second stage of RTT than in the subsequent stages. Parents reported that their children did not seek as much interaction. From the parents' descriptions, it would seem that the children are willing but unable to interact with their environment.

Conclusions Like previous research, our study leads to doubts about the appropriateness of the label 'autistic-like' for the behaviour of individuals in the second stage of RTT. While behaviours of individuals with autism and individuals with RTT may resemble each other, quality and intentions may differ. Still, future studies are needed for further clarification.

Keywords autistic-like behaviour, emotionally sensitive, parents' experiences, Rett syndrome, social-emotional behaviour

Introduction

Rett syndrome (RTT) is a neurodevelopmental disorder that predominantly occurs in women (Schanen *et al.* 1998; Howlin *et al.* 2011; Banerjee *et al.* 2012). It is characterised by profound intellectual and motor disabilities and is (with a prevalence of 1:10,000 women) one of the most common causes of profound intellectual and multiple disabilities (Ellaway & Christodoulou 2001). Four stages of development are particular to individuals with RTT (Kerr & Witt Engerström 2001). In the first stage, the child's early development appears to be normal to nearly normal, but then it stagnates. The child enters the second stage between the age of 6 and 18 months. This is referred to as the regression stage, because although the majority of the children begin to acquire early speech and walking skills in the first

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stage, these skills are lost now. The second stage is also associated with socially withdrawn behaviour and stereotypic hand movements (Mount *et al.* 2002; Kaufman *et al.* 2012). Epilepsy and respiratory, sleeping and eating problems often occur in this stage (Naidu *et al.* 2003; Halbach *et al.* 2008). The third stage is referred to as pseudo stationary stage, and an improvement can be observed not only in motor and communication skills but also in withdrawn behaviour. While some individuals with RTT remain in the third stage until adulthood, others enter the fourth stage around the time of puberty (Bartolotta *et al.* 2011). The fourth stage is associated with motor deterioration, and most individuals with RTT become unable to walk and need to use a wheelchair. The functional use of their hands also declines, and increasing spasticity and scoliosis may be observed (Kerr & Witt Engerström 2001).

Because of the complex health problems in all four stages, a large number of medical studies of individuals with RTT have been conducted. Several recent studies focused on the detection of the genetic cause for RTT (Neul *et al.* 2010; Percy 2011; Psoni *et al.* 2012) and found mutations on the methyl-CpG-binding protein 2 gene to be responsible for the occurrence of the syndrome in 95% of all individuals with RTT. Other research found that, although many individuals with RTT experience epilepsy (60–80%), there are significant individual differences in age of onset, types of seizures, frequency/severity and drug resistance (Pintaudi *et al.* 2010; Cardoza *et al.* 2011; Bao *et al.* 2013). Several studies have also reported respiratory problems such as hyperventilation and breath-holding, especially at night (Halbach *et al.* 2008; Hagebeuk *et al.* 2012), and other studies have been made of the progress and treatment of scoliosis (Ager *et al.* 2006; Ager *et al.* 2009; Riise *et al.* 2011).

However, the parents of the children with RTT have many questions on how to deal with their child's behaviour on a day-to-day basis, but such questions have tended to take a back seat in the research. Parents formulated these questions and emphasised the importance for them in personal communication with the authors.

The social-emotional behaviour in particular leads to questions in parents of children with RTT. The children's behaviour may not only differ from the behaviour of typically developing children but also

according to the stage of RTT (Kaufmann *et al.* 2012; Wanzek *et al.* 2012). Children in the early stages of RTT especially may display emotional and behavioural disturbances (such as sleeping problems or screaming). However, these may not be specifically related to RTT, but more generally to the occurrence of profound intellectual disabilities (Woodyatt & Ozanne 1992; Mount *et al.* 2003). Many parents report irritability, but are not always able to pinpoint a clear case of this behaviour (Mount *et al.* 2002). They also report withdrawn behaviour, but are unsure how to interpret this. Although these characteristics may diminish over time (Sansom *et al.* 1993), it is especially important to support these parents in the early years of raising a child with special needs.

The medical profession describes the behaviours of children in the second stage of RTT as 'autistic-like' (see, for example, Wolf-Schein 1996; Matson & LoVullo 2009). The frequent socially withdrawn behaviour and stereotypic hand movements even led to the inclusion of RTT as a subtype of pervasive developmental disorders in the Diagnostic Statistical Manual of Mental Disorders, Fourth Edition (DSM-IV; American Psychiatric Association 1994), although it was not included in the DSM-V (American Psychiatric Association 2013). Some doubt has arisen in the recent years about the use of the 'autistic-like' label for behaviours in individuals with RTT. Some authors mention that these behaviours have not been included in the diagnosis for RTT, which suggests some doubt as to whether they are an essential aspect of the syndrome (Mount *et al.* 2003). Furthermore, the authors of several studies concluded that the quality of behaviours of individuals with RTT differed from those associated with autism (see, for example, Olsson & Rett 1990; Wulffaert *et al.* 2009; Guénolé & Baleyte 2011; Percy 2011). For example, the insufficient relation with persons and objects as well as the defence reaction when exposed to these are described as typical characteristics of individuals with autism. In contrast, individuals with RTT show clear social orientation and interest in social interaction by intensive eye contact and smiling (Olsson & Rett 1990; Wulffaert *et al.* 2009).

Parents of children with RTT disagree with the use of the term 'autistic-like' to describe their child's social interaction. Again, no scientific studies are available focusing on this statement of parents. Only personal communication revealed this lack of knowledge. In addition, this difference in how the

behaviour is labelled is important because different labels have considerably different implications for the daily support. An example may be the parents' recognition of withdrawn behaviour in their child. When parents label this behaviour as 'autistic-like', they may diminish the environmental stimulation, because they expect this behaviour to be indicative of personal overload. In contrast, parents who interpret this behaviour as a communicational expression with different possible intention may search for different modifications in the environment.

The present study focuses on the behaviour of individuals in the second stage of RTT compared with subsequent stages. The aim is to clarify parents' experiences of the social-emotional behaviour of their children during the different stages. We hypothesise that parents do observe different behaviours in the different stages of RTT. An additional hypothesis is that differences in parents' experiences of behaviour can, in a next step, be related to differences in labelling. The results of the study should increase the understanding of how children interact with their direct environment in the second and later stages of RTT. The knowledge from the study should also be especially valuable for the direct support persons (DSPs) who, alongside parents, are the most involved with children with RTT and may therefore encounter difficulties in interpreting the children's behaviour. The parents' descriptions should help DSPs learn how to respond to the behaviours of children with RTT. The results should also help the parents of young children with RTT in their interaction with their children, because these parents will be able to learn from the experiences of parents whose children have already passed through the early stages.

Methods

Participants

All 150 members of the Dutch Rett Syndrome Organization (*Nederlandse Rett Syndroom Vereniging*, NRSV), an organization of parents with a child with RTT, were informed of the present study, and those parents whose children had already passed the second stage were asked to participate. Fifty-three of the 150 parents filled in the questionnaire (response rate: 35%), but the parents of two children were excluded from further analyses, because their children were still in the

second stage. The 51 remaining parents were asked which stage of the syndrome their children were in. The parents of 22 children answered that their child was in the third stage (pseudo stationary stage), the parents of 17 children that their child was in the fourth stage (motor deterioration) and the parents of 12 children that they did not know whether their children were in the third or fourth stage of the syndrome. The children with RTT of these 51 parents were between 3 and 42 years of age, with a mean age of 16.3 years.

Instrument

An adapted version of the Screen for Social Interaction (SSI; Leone 2009) was used to investigate the parents' experiences of their children's social-emotional behaviour. The SSI is a parent/caregiver questionnaire, which focuses on the child's social interaction skills while minimizing the impact of language ability. It can therefore help to describe the behaviour of young children with autism spectrum disorders in particular. Previous studies have shown the SSI to be a reliable and valid instrument for measuring basic social interaction skills (Ghuman *et al.*, 1998a,b; Ghuman *et al.* 2011).

As the aim of the present study was to clarify the experiences of parents with their children's social-emotional behaviour, only the items in the domain 'social approach/interest' were included. Thereby, 'social approach/interest' is operationalized in nine items focusing on different types of social interaction as well as the expression and recognition of emotions in self and others (Fig. 1). These nine items were complemented by nine additional items, which were formulated in collaboration with five parents of children with RTT who acted as an expert group. They screened the questionnaire in the light of their specific experience of children with RTT and formulated additional items. The instrument is shown in Fig. 1.

Eighteen items in the questionnaire were multiple-choice questions with four possible answers: (1) never; (2) some of the time; (3) most of the time; and (4) almost all of the time (Leone 2009). Higher scores therefore reflect a higher occurrence of social-emotional behaviours. Testing the reliability of the two sets of 18 multiple-choice questions of the questionnaire for the present study revealed a Cronbach's α of 0.851 and 0.866, respectively. This reflects a high internal consistency. Item 19 was an additional open question, which allowed parents to

1. Does your child interact with you during meals by making sounds or talking?*
 2. Does your child interact with you during personal care by making sounds or talking?
 3. Does your child look distant or removed?*
 4. Does your child recognize other persons except you as parent(s) or family?
 5. Does your child express preferences for other persons?
 6. Does your child express disapproval of other persons?
 7. Does your child show interest in other children by watching them?*
 8. Does your child show interest in other children by moving towards them?*
 9. Does your child show interest in other children by staying close to them?*
 10. Does your child show interest in other children by joining in the play if they invite him/her to?*
 11. Does your child show interest in other children by showing concern if another child is upset?*
 12. Does your child avoid other children?*
 13. Does your child copy other persons' emotions?
 14. Is your child shy around people s/he does not know well?*
 15. Does your child enjoy being the focus of the others' attention?
 16. Does your child react to other persons' talking about him/her?
 17. Does your child look forward to an event in the future or remember an event in the past?
 18. Does your child respond to an earlier event in which your child did not seem to have interest back then?
- Optional:
Would you like to add any comments about the social-emotional behaviours of your child?

Note. All questions derived from the SSI have been marked with an asterisk.

Figure 1 'Emotional sensitivity' questionnaire.

add any comments relating to social-emotional behaviours of their child.

In order to clarify whether there are behavioural differences between the second and later stages, the parents were asked to answer each question twice: once thinking back to when their child was in the second stage and once for the present situation. The present situation was defined as the situation of the last 3 months.

Procedure

Information about the study appeared in the NRSV newsletter, and the parents were sent an e-mail inviting them to complete the online questionnaire. A reminder was sent out 2 weeks after the initial e-mail.

To protect the privacy of the parents, the invitation and reminder were sent collectively by the NRSV, which meant an individual reminder was not possible. The parents who formed the expert group to develop the questionnaire did not fill in the questionnaire and therefore were explicitly excluded from the study.

Analysis

The first step in the analysis was to use descriptive statistics to analyse the data. Means and standard deviations provided an overview of the answers of the multiple-choice questions. Thematic analysis (Joffe & Yardley 2004) of the open question provided a

summary of the parents' comments. After familiarization with the data, codes were formulated and analysed in number and content. Throughout the analysis, interpretations were validated with the two co-authors. The next step was to enter the results of the multiple-choice questions in SPSS 20 and to use paired samples *t*-tests to clarify any differences between the answers relating to the second stage and those relating to the present situation. Paired samples *t*-tests were conducted for the overall mean (based on the mean of the 18 multiple-choice questions per parent) and for each individual question. To account for the multiple analyses, a significance level of $\alpha = 0.05/19 = 0.003$ according to the Bonferroni correction was used for all tests.

Results

With regard to the multiple-choice questions, the parents answered that most of the social-emotional behaviours occurred 'never' or 'some of the time' (35.5% and 37.9%) during the second stage. The overall mean of the answers for the second stage was 1.99, which thus means a low to moderate occurrence of social-emotional behaviours observed by the parents. In contrast, the distribution of the answers concerning the present situation was very different: the parents saw most of the behaviours 'most of the time' (30.6%). However, they did select the other three categories of answer frequently as well (20.9% for 'never', 25.7% for 'some of the time' and 22.8% for 'almost all the time'). The overall mean of the parents' answers for the present situation was 2.55. The parents therefore observe social-emotional behaviours quite frequently in the present situation.

Differences were found between the individual questions: in answer to questions concerning interaction with other people, the parents often selected 'some of the time' for the second stage and 'most of the time' for the present situation. However, the distribution of answers concerning emotional expressions (such as copying other persons' emotions) was similar for both situations (i.e. the second stage and the present situation). Parents indicated that these behaviours mostly occurred 'some of the time' or 'never'. An overview of the percentages and means of all answers is provided in Table 1.

The paired samples *t*-test of the overall mean revealed a significant difference between the answers concerning the second stage and those concerning the present situation ($t(50) = -8.67$; $p < 0.001$). The parents saw significantly more social-emotional behaviours in the present situation than in the second stage. These differences also became apparent when the questions were considered separately (all $P < 0.001$ with exception of item 18 being $P < .01$; Table 2). Only the answers to the following two questions did not differ significantly between the two situations: 'Is your child shy around unknown people s/he does not know well?' and 'Does your child express disapproval of other persons?' ($P = 0.302$ and $P = 0.532$, respectively). The first question was derived from the original questionnaire by Kaur-Ghuman and the second question was added by the parents of a child with RTT.

Thirty-four of the parents answered the open question (67%) inviting them to comment on the social-emotional behaviours of their child. Fourteen of these provided additional information about their children's development, 13 mentioning that their child often screamed and cried during the second stage: 'She was often very sad; we couldn't calm her. We didn't understand why she was so sad'. Several of these 14 parents said that they had always witnessed their children expressing the entire range of emotions (from happy to sad), but that this was less explicit in the second stage. Three parents stated that their child sought interaction more often in the present situation than in the second stage. However, another six parents said that they thought that their children were interested in others, but were physically unable to move towards them. All of them said that their children seemed to enjoy interaction with other people, with children in particular, and some said that they seemed to enjoy interaction with animals more in the present situation than in the second stage. With regard to the present situation, a large number of parents described their children as happy individuals who laughed a lot to express this happiness. Twenty-two parents noted how their children responded to specific situations. A recurring example (five parents) was the following: 'She laughs about situations that are not funny to others, such as when somebody hurts themselves or is punished, or when something breaks'.

Some parents noted that they had some difficulty completing the questionnaire. Four parents said they

Table 1 Overview of the percentages and means of all answers

	Second stage					Present situation				
	Never (%)	Some of the time (%)	Most of the time (%)	Almost all of the time (%)	Mean	Never (%)	Some of the time (%)	Most of the time (%)	Almost all of the time (%)	Mean
1	11.8	60.8	19.6	7.8	2.24	2.0	23.5	41.2	33.3	3.06
2	13.7	45.1	31.4	9.8	2.37	3.9	17.6	43.1	35.3	3.10
3	13.7	47.1	37.3	2.0	2.27	0.0	9.8	78.4	11.8	3.02
4	13.7	35.3	33.3	17.6	2.55	3.9	17.6	31.4	47.1	3.22
5	13.7	49.0	27.5	9.8	2.33	5.9	21.3	37.3	35.3	3.02
6	7.8	11.8	47.1	33.3	3.06	5.9	9.8	49.0	35.3	3.14
7	21.6	43.1	25.5	9.8	2.24	3.9	21.6	27.5	47.1	3.18
8	49.0	39.2	5.9	5.9	1.69	37.3	23.5	15.7	23.5	2.25
9	43.1	41.2	9.8	5.9	1.78	27.5	31.4	27.5	13.7	2.27
10	35.3	47.1	11.8	5.9	1.88	19.6	29.4	25.5	25.5	2.57
11	70.6	27.5	2.0	0.0	1.31	51.0	35.3	13.7	0.0	1.63
12	7.8	23.5	43.1	25.5	2.86	0.0	5.9	49.0	45.1	3.39
13	49.0	47.1	2.0	2.0	1.57	21.6	58.8	15.7	3.9	2.02
14	80.4	5.9	7.8	5.9	1.39	78.4	13.7	7.8	0.0	1.29
15	39.2	45.1	11.8	3.9	1.80	19.6	31.4	27.5	21.6	2.51
16	62.7	31.4	3.9	2.0	1.45	29.4	31.4	27.5	11.8	2.22
17	60.8	33.3	3.9	2.0	1.47	35.3	31.4	17.6	15.7	2.14
18	45.1	49.0	3.9	2.0	1.63	31.4	49.0	15.7	3.9	1.92
Total	35.5	37.9	18.2	8.4	1.99	20.9	25.7	30.6	22.8	2.55

Table 2 Overview of the results of the paired samples *t*-tests

Question	T-value	Degrees of freedom
1	-7.194**	50
2	-5.392**	50
3	-7.151**	50
4	-6.216**	50
5	-6.226**	50
6	-0.629	50
7	-6.539**	50
8	-4.040**	50
9	-4.464**	50
10	-6.034**	50
11	-4.093**	50
12	-5.173**	50
13	-4.788**	50
14	-1.043	50
15	-6.913**	50
16	-6.511**	50
17	-5.831**	50
18	-3.125*	50
Total	-8.671**	50

* $P < 0.01$. ** $P < 0.001$.

Note. The results for the second stage and the present situation were entered in the *t*-test for each comparison.

found it difficult to think back to the second stage, and two said they were unsure how to answer some of the questions: 'I don't know if she recognises other people'. Other parents also noted that their interpretation of the child's behaviour might differ from the child's intention: 'She may be able to understand more than she can express to us'.

Of interest were the following comments: 'What could be the effect of the medication on the behaviour of children with RTT?', 'What are the experiences of other parents with the pain their children experience?' and 'I wouldn't call it autism, but I think that children with RTT are angry that they are unable to express themselves'.

Conclusion and discussion

The aim of the present study was to clarify parents' experiences of the social-emotional behaviour of their children during the second stage of RTT. Knowledge about these experiences is necessary in order to understand, respond to and appropriately label the child's behaviour. From the answers in the online

questionnaire, we can conclude that the 51 parents experienced their children with RTT exhibiting significantly less social-emotional behaviours in the second stage than in the present situation. In particular, the parents reported that the children did not seek as much interaction. From the additional descriptions given by a number of parents, it would seem that the children were willing but unable to interact with their environment. Some parents also emphasised that their children had always expressed the entire range of emotions, but that these expressions were less explicit during the second stage.

The results of the present study confirm those of previous studies. Parents in another study described withdrawn behaviour and little eye contact during the second stage of RTT (Mount *et al.* 2002), and the present study also showed that significantly less social-emotional behaviour is exhibited during this stage. In addition, the present study revealed additional qualitative information: some of the parents described how their children sought less interaction in the second stage than in the present situation, but emphasised that they believed that the lack of social-emotional behaviour was because of an inability in the children to exhibit these behaviours rather than a lack of willingness to seek interaction. These descriptions contrast one of the main characteristics of individuals with autism: the lack of interest in social interaction (Rutgers *et al.* 2004). Furthermore, many individuals with profound intellectual and multiple disabilities are very restricted in their expressions (Petry & Maes 2006; Nakken & Vlaskamp 2007). This may be especially true for individuals in the second stage of RTT. They may consequently withdraw their attention from the environment as a communicational reaction. This behaviour may therefore be their only way of expressing themselves rather than a lack of interest in their environment. Because parents suggested these differences in intentions of the behaviours, we hypothesise that the label 'autistic-like' is not appropriate to describe the behaviour of individuals with RTT in the second stage.

With regard to the expression of emotions, our study also adds important information to the results of previous studies. Previous studies mainly found that individuals with RTT frequently cried and screamed during the second stage (Sansom *et al.* 1993; Mount *et al.* 2002), whereas our study found

that the parents of some children with RTT believed their children had always shown the entire range of emotions. In contrast, individuals with autism have been described as restricted in their emotional expressions during their entire life span (Rutgers *et al.* 2004). The parents in the present study emphasised that the children did not express these emotions as explicitly and distinctly during the second stage as in the present situation and added that the children were more able to express their emotions in the later stages of the syndrome. It could therefore be that the expressive communication skills of individuals with RTT improve during their lives. Again, the differences in quality of behaviour between individuals with autism and individuals with RTT, which have been suggested by the parents, lead to doubts about the appropriateness of the label 'autistic-like' for the behaviour of individuals with RTT in the second stage.

Another finding from our study is that parents see not only regular negative behaviour (such as screaming and crying) during the second stage but also 'non-appropriate' behaviour (such as laughing when something sad happened). This could be related to the brainstem immaturity of individuals with RTT: a large number of previous studies have observed disturbances in brainstem autonomic function (Glaze & Schulz 2001; Trevarthen & Burford 2001; Jackowski *et al.* 2011; Smeets *et al.* 2012; Steinbusch *et al.* 2013). Bergström-Isacson *et al.* (2013) recently described frequent and very frequent abnormal spontaneous brainstem activation (ASBA's) in their participants. They measured brainstem activation while observing emotion and concluded that 'misfiring' of the brainstem can lead to behaviours that resemble, but that are not, the expression of emotions. Differences between true and false emotions could only be observed when taking co-occurring patterns (such as changes in breathing, vocal sounds or body movements) into account. From these findings, we can hypothesise that ASBA occurs more frequently during the second stage of RTT and that these behaviours are not 'autistic-like' characteristics.

A number of limitations need to be taken into account when interpreting the results of our study. Although we approached all parents who belong to the NRSV, only 35% returned the questionnaire. As the inclusion criterion was that the child had to be beyond the second stage, not all parents could be

included. A large number of parents should be included in future studies in order to gain a more representative picture of the parents' experiences. In addition, longitudinal studies including behaviour observations are needed to clarify patterns of social-emotional behaviours in individuals with RTT over time. Thereby, assessment of motor and communication abilities could be a valuable complementation to differentiate between different groups of individuals with RTT. International collaboration may be required to find a large enough number of participants.

The questionnaire that was used in the present study was a combination of the 'social approach/interest' domain of the SSI (Leone 2009) and nine additional questions formulated by a focus group of parents of a child with RTT. Although the SSI has been described as a reliable and valid instrument for measuring social interaction in children, future studies will be needed to analyse the suitability of the adapted questionnaire for the target group of individuals with RTT, and in particular, the two non-significant questions. Even given that preliminary results presented here indicate good reliability, such an analysis seems necessary to evaluate whether these two questions need to be reformulated or whether they are appropriate at all for the target group of individuals with RTT.

If we consider the results of the present study, we must emphasise that the interpretation of behaviour is always, to a certain extent, subjective (Hogg *et al.* 2001). Although we were aware of this limitation to our questionnaire from the beginning of our study, the parents confirmed that it is especially true for the behaviour of their children with RTT. In addition, asking parents about retrospective interpretations could lead to memory bias (Hassan 2005). Consequently, we are not able to express a definite opinion about the social-emotional development of children with RTT, but can only outline parents' experiences. Parents, however, are experts on their children's behaviours, so their opinion and experiences should be included in scientific studies as such (Bartolotta *et al.* 2011). Future studies should investigate the relationship between the behaviours of parent and child in order to gain more insight in this area. Such studies would also take into account parents' comments on the impact of pain and medication on their child's behaviours. A more

qualitative approach would therefore be a valuable complement to the present study.

The results of our study have several implications for parents and DSPs. First, our study supports parents in their doubts about the appropriateness of the label 'autistic-like' concerning the behaviour of individuals with RTT in the second stage. This is in accordance with other studies on characteristics of Autism Spectrum Disorder (ASD) in neurodevelopmental disorders of generic origin, like, for example, Angelman syndrome, Prader-Willi syndrome, Cornelia de Lange syndrome or Fragile X syndrome. Moss *et al.* (2013) showed high proportions of meeting cut-off scores for autism in Cornelia de Lange and Fragile X syndrome; however, profiles were atypical and/or revealed a milder presentation of ASD, and adaptive behaviour skills only showed a limited association with severity of autism symptomatology. As in our study, other factors are likely to account for heightened prevalence of ASD in these specific syndromes. Understanding the similarities and differences in the presentation of ASD symptomatology next to looking at the quality instead of the type of behaviour may help parents to facilitate interaction and adapt the environment to the needs of their child. Parents also indicate that a different view on their child's behaviour can improve the quality of life of the child and the family. If they can gain a better understanding of the child's strengths, this may be the first step in facilitating the best outcomes for both child and family (Gillman *et al.* 2000; Whitaker 2002; Poehlmann *et al.* 2005). The same applies for DSPs: if they interpret the few interaction initiatives of individuals in the second stage of RTT as 'unable' rather than 'unwilling', they may try to elicit situations that allow for interaction and communication. Understanding the needs and abilities of individuals with RTT, particularly those in the second stage, may be a precondition for the individual with RTT feeling able to express their interaction and communication abilities. Second, information about false emotions could raise parents' and DSPs awareness on this topic. Interpreting false emotions as an unconscious misfiring of the brain can help parents and DSPs to understand this part of their child's unusual behaviour or atypical development. Consequently, it will help them overcome uncertainty and move beyond focusing on it (Perry *et al.* 2008). Finally, parents should be considered experts on their children's behaviour, and DSPs would therefore be advised to remain in close

consultation with them, especially during the second stage.

In summary, the present study leads to doubts about the appropriateness of the label 'autistic-like' for the behaviour of individuals with RTT in the second stage. This is in line with several previous studies on RTT (Wulffaert *et al.* 2009; Guérolé & Baleyte 2011; Percy 2011). It is also in accordance with other studies on ASD comparing the symptomatology of individuals with Cornelia de Lange Syndrome and individuals with idiopathic ASD (e.g. Moss *et al.* 2012). As the association between RTT and ASD characteristics is predominantly seen in the second stage of this progressive disease, further knowledge is needed on the specificity of the differences and similarities in ASD symptomatology in this stage. Another study that pointed out differences is the one from Schwartzman *et al.* (2013). They used eye-tracking technology to reveal that the social-emotional behaviours exhibited by individuals with RTT differed from those exhibited by individuals with ASDs, and the behaviours of typically developing children and children with RTT largely resembled each other. Consequently, the label 'social-emotional instability' might be a better fit for the behaviour of individuals with RTT, because it implies moments of few pro-social behaviours and quick and irregular shifts in emotions (Carlo *et al.* 2012) and thus accounts for the withdrawn behaviour and the false emotions that individuals express during the second stage of RTT. Still, additional studies are required in the future to confirm our hypotheses.

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