Abstract

The aim of this research paper is to investigate Theory of Mind (ToM) capacities with Rett Syndrome (RS), which is similar to autism yet with more significant impairments. RS patients can best be described as hardly ever advancing past the sensorimotor stage, or the period described by Piaget between birth and eighteen months during which an infant's knowledge of the world is limited to sensory perceptions and motor activities with behaviours that consist of simple motor responses caused by sensory stimuli. Nevertheless, single-case studies have shown that RS patients can develop a mentalistic understanding of others if properly trained. To further advance this finding, non-trained RS female children completed a false belief task appositely devised with a non-verbal answering mode familiar to RS girls. Results show that RS girls are better on this type of false belief task than a control group of autistic children, whose deficit in ToM understanding is well-documented in literature. Our findings, though preliminary, may be a first step towards a better understanding of the differences between RS and autism in mental state reasoning, opening new topics of research and intervention in ToM with severe clinical conditions.

Keywords: Rett syndrome; Autism; Theory of Mind.
1. Introduction

Understanding the reasons behind human behaviours is crucial for successful human social interactions. Behaviours can be understandable and predictable if the intentions, emotions, desires, and beliefs that motivate behaviour are comprehended. The ability to understand what motivates behaviour is known as Theory of Mind (ToM) (Doherty, 2009), which is fully mastered around four years of age in typical development. Various acquired skills pave the way for a fully functioning ToM, starting with those that develop within the first two years of life. The most relevant of these skills in the first two years of life include joint attention (Baron-Cohen & Ring, 1994), declarative pointing (Camaioni, 1993), and the understanding of pretense (Leslie, 1987). Following two years of age, children develop a more complex understanding of mental states, including, between age two and age three, an understanding of desires and by three years of age, an understanding of true beliefs (Bartsch & Wellman, 1995). By four years of age, they show an understanding of first order false beliefs (Wimmer & Perner, 1983), by seven/eight years of age they show an understanding of second order false beliefs (Perner & Wimmer, 1985), then changes in ToM continue throughout the life-span (Baglio, Castelli, Alberoni, Blasi, Griffanti, Falini et al., 2012; Castelli, Baglio, Blasi, Alberoni, Falini, Liverta-Sempio et al., 2010; Castelli, Pini, Alberoni, Liverta-Sempio, Baglio, Massaro et al., 2011).

As far as atypical development is concerned, ToM has been studied in a wide range of neurodevelopmental disorders, finding that it may be only partially developed, impaired or totally inadequate (Korkmaz, 2011). Among these disorders, autism spectrum disorder represents the most emblematic evidence of severe ToM impairment (Baron-Cohen, 2000; Pelphrey, Shultz, Hudac, & Vander Wyk, 2011), so that the lack of ToM for the autism syndrome has become one of the most studied factors that helps explain the great social difficulties that these patients have. ToM involves meta-representational abilities, namely both lower-order representations (i.e., representations of reality) and higher-order representations (i.e. the ability to form thoughts about attributed thoughts). The delayed acquisition of the ability to manipulate mental meta-representations in autistic patients may lie at the heart of this disorder.

Since Rett Syndrome (RS) patients are often misdiagnosed with autism because of similar symptoms at the early stages of both disorders, including markedly impaired use of nonverbal behaviours to regulate social interaction, loss of speech, inconsolable crying and subtle onset (Olsson & Rett, 1990), an open question is whether RS patients have ToM impairments similar to those of autistic patients.
RS is a childhood neurodevelopmental genetic syndrome caused by a mutation of gene MECP2 on locus Xq28 of the X chromosome (Amir, Van Den Veyer, Wan, Tran, Francke, & Zoghbi, 1999), whose incidence has been estimated from 1/10,000 to 1/15,000 of newborn females (Kozinetz, Skender, MacNaughton, Almes, Schults, Percy, & Glaze, 1993). Typically, the onset of symptoms begins with a severe regression around 18 months of age, and progresses through the three remaining stages of decline that include symptoms like the loss of purposeful hand skills and of spoken language (Stage II - 1-4 yrs. of age), seizures and apraxia (Stage III - 4-10 yrs. of age), and scoliosis and reduced mobility (Stage IV above 10 yrs. of age). All of this leads to a significantly impaired clinical picture, with deep linguistic, social, behavioural and fine and great motor deficits (including repetitive automatic hand movements, i.e., hand stereotypes).

While RS and autism demonstrate a phenotypic overlap and RS girls commonly are misdiagnosed as having autism, especially at the early stages of RS development, in the later stages of RS development autistic traits become less prominent and differences in the quality of autistic behaviour become evident. Over the past decade, RS has become a cross-disciplinary topic of significant interest. Genetic as well as psychopathology research has been done on the twofold goal of defining the RS phenotype and behavioural conditions (Vignoli, Fabio, La Briola, Giannatiempo, Antonietti, Maggolini et al., 2010; Berger-Sweeney, 2011), as well as identifying its commonalities and differences with autism (Olsson & Rett, 1990; Percy, 2011). With reference to behavioural conditions, the neuropsychological profile of RS patients has fallen within the third or the fourth stage of the sensorimotor intelligence, corresponding to a mental age of 4-8 and 8-12 months, respectively (Olsson & Rett, 1990) or at least within the transition to the symbolic stage or the mental age of 18-24 months (Lindberg, 1988).

It is compelling, therefore, to examine ToM abilities in RS individuals in order to understand when ToM reasoning capacities develop in RS individuals. Previous research, for example, has demonstrated that RS girl’s spontaneous non-verbal behaviours, such as gaze control and pointing, which are known to be precursors of ToM abilities (Camaioni, 1993; Baron-Cohen & Ring, 1994) are not different than normal controls (Antonietti, Castelli, Fabio, & Marchetti, 2005). Related research by Antonietti Castelli, Fabio, & Marchetti, (2001; Antonietti, Castelli, Fabio, & Marchetti, 2008; Fabio, Castelli, Marchetti, & Antonietti, 2013) has found that non-verbal training (see also Fabio, Giannatiempo, Antonietti, & Budden, 2009) aimed at teaching basic and complex emotion recognition allowed two RS girls to develop the ability to discriminate, recognize and generalize the expression of emotions, which is considered to be crucially important for understanding the mental states of others. Antonietti et al. (2008) also found that one RS girl was able to communicate non-verbally with her caregivers due in part to a specific
cognitive rehabilitation training program which was designed to successfully overcome a first and a second-order false-belief task, which is the most commonly employed and reliable task to evaluate ToM development (see for example Wellman, Cross, & Watson, 2001). In the first-order-false-belief task, the child is asked to predict the behaviour of another person; for instance, Mary, on the basis of the meta-representation of Mary’s mental states about reality, resulting in a thought like, “I think that Mary thinks that...”. In the second-order-false-belief task, the child is asked to predict the behaviour of another person, on the basis of the meta-representation of Mary’s mental states about reality from another person’s point of view, Susan’s, resulting in a thought like “I think that Susan thinks that Mary thinks that...”.

This previous research formed the basis in reaching the goal for the present research, which is to evaluate ToM abilities in RS girls who have not completed any cognitive rehabilitation training.

2. Methods

2.1 Objectives

The aim of this study is to investigate ToM abilities with a non-verbal false-belief task in not-treated RS girls.

2.2 Participants

Nine RS participants (age range 5 – 19 yrs., all girls were in the IV stage of the disease, except for one girl who was in the III stage) were recruited in northern Italy through the Italian association of RS girls’ parents and families (e.g., AIR, Associazione Italiana Rett). The aim of the research was explained to the president of AIR and RS family contact information was provided to the researchers.

A control sample of autistic children (age range 9.5-10.4 yrs.) was recruited at an educational centre in Milano (Centro per la Cura e lo Studio dell’Autismo e dei Disturbi Generalizzati dello Sviluppo, Azienda Ospedaliera San Paolo, Milano). All had been diagnosed by experienced clinicians with the Autism Diagnostic Observation Schedule (Lord, Rutter, DiLavore, & Risi, 1999) and the Autism Diagnostic Interview Revised (Lord, Rutter, & Le Couteur, 1994). They were given the same tasks and procedure used with RS participants.

Participants were treated according to the principles of the Declaration of Helsinki and to APA ethical standards. Informed written consent was obtained from the “next of kin” on behalf of children participants involved in our study. The research has been approved by the ethical committee of the Department
of Psychology of the Catholic University of the Sacred Hearth, Milano.

2.3 Tasks and Procedure

All the tasks were completed either at the RS girl’s home or school, and at the educational centre for autistic participants. Tasks were the following:

1. Evaluation of the pre-requisites of intentional choice and of the acquisition of the meaning of “happiness” and “sadness”. They were assessed in order to exclude casual gaze-answers (i.e., false positives) during test sessions. This evaluation was done with each participant by giving them three pictures of familiar objects or people, where each picture was presented with a distractor for three times in a randomized right-left order: the participant was asked to choose the target, i.e., the picture of familiar objects or people. Scores were given in the following way: 1 score for two or three correct answers out of three; 0 for one or no correct answer out of three.

2. Coloured Progressive Matrices (CPM) (Raven, 1984) was used to test non-verbal mental age. The set A of CPM was presented: it contains 12 coloured and attractive items representing a series of patterns with a bit missing. Success in Set A is determined by individual’s ability to complete a continuous pattern changing, first, in one direction, then, at the end of the set, in two directions at the same time. Each participant was given the first item and asked to look at it carefully because a piece of the item was missing and was then asked to complete the item with either the correct missing piece (target) or with a first distractor piece. The participant was asked to look at each piece and to indicate, either with the gaze or with an eye-hand coordinated movement, which piece would correctly complete the item. Then the participant was shown the same item, the same target but with a second distractor and in the last phase with a third distractor. Each item was presented three times in succession with the target in a randomized right-left order and with a new distractor each time in order to control casual choices. The randomization of right-left order reduces the possibility that a choice was made always in the same position, and so reduces casual choices. Successful completion of the task consisted of correctly completing at least two out of the three trials. Each item was therefore scored as either a success (1) or a failure (0). The items were consecutive and if three consecutive failures occurred, the test was stopped. The sum of the correct items provided the raw score for the CPM.

3. We used the false-belief task of the Unexpected Transfer Test (Wimmer & Perner, 1983) acted out in the condition of “let’s play a joke on mum” specifically devised for this research in order to make it doable by RS girls. The girl (G), her Mum (M) and the experimenter (E) were sitting at a table
and were listening to music (RS girls are fond of music and this motivated them to attend to the task). At a certain point, M with an excuse placed the tape recorder (TR) in a box (box A) and left the room, saying that when she came back she would use it again. During M.’s absence, E told G that they were going to play a joke on M and so she changed the location of the object (box B). Before M came back, G was asked three questions:

Q1: 1st order false belief question: “Where will M look first for the TR once she will be back in the room?”; the correct answer is eye-gaze on box A or eye-gaze and hand coordination on box A;

Q2: 1st order emotion false-belief question: “What emotion will M express when she will open box A?” (E points to box A, i.e., the one that is now empty but where M believes the TR is); the correct answer is eye-gaze on the picture of a smile with a sad expression or eye-gaze and hand coordination on the picture of a smile with a sad expression;

Q3: control question about reality: “Where is the TR actually?”; the correct answer is eye-gaze on box B or eye-gaze and hand coordination on box B.

The false belief task was acted out in the same way as that above described with autistic participants.

For each of the three tasks, the procedure was always the following: the participant was sitting at a table and had the target on his/her right and the other stimulus on his/her left. The participant was asked to look at them and to choose the target by looking at it and touching it (eye-hand coordination). The position of the stimuli was changed and the question was repeated (second presentation) and once again the position of the stimuli was changed and the question was repeated (third presentation). In each task, each item or each question was posed three times consecutively, in order to control for chance responding. In each presentation, the participant’s performance was judged as follows:

- He/she looks at the target and touches it with his/her hand: +
- He/she looks at the distractor and touches it with his/her hand: -
- He/she looks at the target (l+ = look+) but touches the distractor (t- = touch-): l+t-
- He/she is given a little help (i.e. the head is kept still so that he/she can concentrate gaze and hand movement): +.
- He/she needs considerable help: +-.

The above performances were then scored with the following scoring system:

- positive answers (+, +., l+t-) = 1 point
- answers that need help (+-) = 0.5 point
- wrong answers (-) = 0 point
We scored l+t- as a positive answer because, given the strong motor impairment in RS, we were aware of the difficulty that these girls have moving their hand purposefully and/or coordinate eye-gaze and hand movement. What was important was the correct use of eye pointing as a form of non-verbal communication.

Given that each item or question was repeated three times, the final score of each item or question could range from 0 (no correct answer out of three or only one correct answer out of three) to 1 (at least two correct answers out of three or all three answers correct).

All experimental sessions were videotaped and were coded by two independent raters who were blind to the goals of the research but were experts in the assessment of cognitive abilities of both RS and autistic patients using the method of evaluation previously described. The few cases of judge uncertainty in participants’ answers were solved by a third independent rater who was similarly blind to the research goals as well as an expert in assessing the cognitive abilities of RS and autistic patients.

Data analysis inclusion required participants to fulfil the following requisites: 1) possess the pre-requisites of intentional choice and 2) answer correctly the control question of the false-belief task (Q3).

3. Results

All nine RS girls were described as having the pre-requisites for intentional choice, and therefore they were all tested with the CPM and the false-belief task. Four girls (age range 5-12 yrs.) were removed from the data as they were unable to answer the false-belief task control question, leaving the remaining five girls (age range 12-19 yrs.) for data analysis. None of the autistic participants failed to correctly answer the false-belief task control question.

Table 1 - RS girls’ scores in the CPM and in the false belief task

<table>
<thead>
<tr>
<th>Participant</th>
<th>Chronological Age (expressed in months)</th>
<th>CPM (rough score)</th>
<th>False Belief Task</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td>Q1</td>
</tr>
<tr>
<td>1</td>
<td>144</td>
<td>8</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>149</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>3</td>
<td>160</td>
<td>6</td>
<td>1</td>
</tr>
<tr>
<td>4</td>
<td>217</td>
<td>7</td>
<td>1</td>
</tr>
<tr>
<td>5</td>
<td>228</td>
<td>6</td>
<td>1</td>
</tr>
</tbody>
</table>
Results can be considered using two different criteria, correctly answering the false-belief question (Q1) or correctly answering Q1 as well as the emotion false-belief question (Q2). First, if only correct answers to Q1 are considered, all five RS girls succeeded in the task. Second, if responses to both Q1 and Q2 had to be correct, then one RS girl (age 19 yrs.) did not meet this criterion. The second criterion is obviously more restrictive, because it not only requires predicting M’s behaviour on the basis of her false-belief, but also to figure out the relationship between M’s false belief and her emotional state.

Table 2 - Autistic children’s scores in the CPM and in the false belief task

<table>
<thead>
<tr>
<th>Participant</th>
<th>Gender</th>
<th>Chronological Age (expressed in months)</th>
<th>CPM (rough score)</th>
<th>False Belief Task</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Q1</td>
</tr>
<tr>
<td>1</td>
<td>F</td>
<td>114</td>
<td>31</td>
<td>1</td>
</tr>
<tr>
<td>2</td>
<td>M</td>
<td>121</td>
<td>35</td>
<td>0</td>
</tr>
<tr>
<td>3</td>
<td>M</td>
<td>124</td>
<td>27</td>
<td>0</td>
</tr>
<tr>
<td>4</td>
<td>M</td>
<td>124</td>
<td>30</td>
<td>0</td>
</tr>
<tr>
<td>5</td>
<td>M</td>
<td>125</td>
<td>34</td>
<td>1</td>
</tr>
</tbody>
</table>

If correct answers to Q1 are only considered, only two autistic children succeeded in the task, while if both answers to Q1 and Q2 need to be correct, only one autistic child met this criterion. The data from the autistic children confirm the evidence of a specific ToM deficit in autistic children (Korkmaz, 2011).

Data of the two clinical groups inversely mirror each other. The fact that RS girls have a significantly better performance than autistic children is statistically supported by the non-parametric test of Wald-Wolfowitz ($z = -1.67; p = .04$). It is worth noticing the nonverbal mental age assessed with the raw score at the CPM: RS girls showed very low CPM raw scores (range: 6-8), whereas autistic children had higher ones (range: 27-35).

4. Discussion and conclusions

It may be interesting to consider if RS girls’ success on the first-order false-belief task makes them socially smarter than autistic children despite RS girls’ deeper cognitive impairments. These results are especially intriguing because RS girls in our research did not complete any cognitive rehabilitation prior to participating in our study.

Several cognitive factors might help explain these results. First, the “pure”, nonverbal cognitive level - as measured by Raven CPM - may not represent a
good example to explain RS girls’ relatively good ToM performance. Despite the fact that Raven CPM scores were higher in autistic children than in RS girls, these higher scores did not predict false-belief task performance. The false-belief task, considered in ToM literature as the “litmus test” of mental ability, seems in this case to be independent of cognitive factors. Future research should look for other factors that do not necessarily belong to the cognitive domain that could be associated with false-belief task performance, namely socio-relational and developmental-clinical factors, which are interconnected in the development.

Regarding socio-relational factors, RS girls may have benefited from the possibility of joining social interactions actively – especially with their caregivers – more than autistic children can do, given autistic children’s lack of joint-attention and pointing. During these social interactions there might have been opportunities for jokes, surprises and so on, so that these experiences may have somehow supplied to RS low cognitive level in solving the ToM task. As for developmental-clinical factors, the general regression typical of RS usually starts at 18 months of age and, therefore, the fact that this regression starts at 18 months of age may have occurred late enough to have not affected the basic cognitive abilities (i.e. joint attention and pointing) necessary for ToM development. In fact, research mentioned in the introduction (Antonietti, et al., 2005) showed the presence of joint-attention and pointing in RS girls, whereas the absence of the same ToM precursors in autistic children is well-documented in literature (Korkmaz, 2011) as well as their presence in typical pre-18 months of age development (Camaioni, 1993; Baron-Cohen & Ring, 1994).

Because two different patterns seem to characterise the social and emotional life of RS and autism, research about ToM abilities in RS girls may prove useful to evaluate better how these girls engage in everyday social interactions, which are relevant and vital for their relationships with their caregivers.

Future research should consider RS girl’s joining in social interactions actively in order to better understand RS girls’ internal mental world. Our ability to understand RS girls’ mental life is made possible despite the communicative deficits of RS girls by simply adapting the tasks to allow for nonverbal ways of communicating their answers. Future research should also include a larger sample and a control group matching each subject on the basis of the sensorimotor developmental stage, which weakens the generalizability of the current findings.

References

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