Parents' perception of health care services for girls with Rett syndrome

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Abstract

Background: Rett syndrome (RTT) is a severe neurodevelopmental disorder, implying impairment and disability across several domains.

Method: We investigated parents' perception of the caregiving process in a sample of 55 mothers and fathers of girls with RTT using the MPOC-20 questionnaire. The association of parents' satisfaction with clinical variables has also been explored.

Results: We obtained intermediate levels of satisfaction on the MPOC-20 Coordinated and Comprehensive Care and Respectful and Supportive Care scales. The performance was lower on the scales Providing General Information and Providing Specific Information. Mothers' assessment was not associated with clinical variables such as walking disability, presence of scoliosis, or epilepsy. For children with greater degree of walking impairment, fathers expressed the need of having more information available.

Conclusions: Although parents seemed satisfied of the caregiving process, clinicians should put more emphasis on their need of receiving general and specific information on RTT along the entire rehabilitation program.

KEYWORDS
caregiving process, parents, Rett syndrome, satisfaction, walking disability

INTRODUCTION

Rett syndrome (RTT) is a neurodevelopmental disorder predominantly linked to MECP2 gene mutations, affecting one female birth in 10,000/15,000 (Leonard, Cobb, & Downs, 2017). Girls with RTT may have a typical development up to 18 months, even though some prodromal signs of the disease could be identified in the first months of life. Around 6–18 months, development stops, and regression of previously acquired skills can be observed (Neul et al., 2010). While hand stereotypes, such as wringing, clapping, or washing, represent a hallmark of the syndrome, language and motor functions are progressively lost. Concomitant conditions include cardiac and respiratory disorders (hyperventilation and apneas), gastrointestinal diseases, scoliosis, seizures, metabolic, and sleep disorders (Hagberg, 1995).

Even though there is a large strand of empirical research investigating on clinical outcomes in RTT, less is known about how parents of these girls perceive the caregiving process. In this study, we take an ecological perspective (Bronfenbrenner, 1979), and we refer to a family-centred service perspective (Rosenbaum, King, Law, King, & Evans, 1998). Such model is in line with the International Classification of Functioning (World Health Organization, 2001) philosophy of not considering disease alone to determine patients’ health status; the focus is instead on the interaction between environmental and family components (Hammal, Jarvis, & Colver, 2004; Rosenbaum & Gorter, 2011). In this framework, the consequences of physical impairment are conceived in terms of participation and activity. Taking this perspective implies considering parents as a primary source of information with respect to the children needs and opportunities, as well as

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Rett syndrome (RTT) is a neurodevelopmental disorder predominantly linked to MECP2 gene mutations, affecting one female birth in 10,000/15,000 (Leonard, Cobb, & Downs, 2017). Girls with RTT may have a typical development up to 18 months, even though some prodromal signs of the disease could be identified in the first months of life. Around 6–18 months, development stops, and regression of previously acquired skills can be observed (Neul et al., 2010). While hand stereotypes, such as wringing, clapping, or washing, represent a hallmark of the syndrome, language and motor functions are progressively lost. Concomitant conditions include cardiac and respiratory disorders (hyperventilation and apneas), gastrointestinal diseases, scoliosis, seizures, metabolic, and sleep disorders (Hagberg, 1995).

Even though there is a large strand of empirical research investigating on clinical outcomes in RTT, less is known about how parents of these girls perceive the caregiving process. In this study, we take an ecological perspective (Bronfenbrenner, 1979), and we refer to a family-centred service perspective (Rosenbaum, King, Law, King, & Evans, 1998). Such model is in line with the International Classification of Functioning (World Health Organization, 2001) philosophy of not considering disease alone to determine patients’ health status; the focus is instead on the interaction between environmental and family components (Hammal, Jarvis, & Colver, 2004; Rosenbaum & Gorter, 2011). In this framework, the consequences of physical impairment are conceived in terms of participation and activity. Taking this perspective implies considering parents as a primary source of information with respect to the children needs and opportunities, as well as
a fundamental component of the "rehabilitative" intervention, which is conceived in terms of promoting child development (Rosenbaum & Gorter, 2011; Rosenbaum & Stewart, 2004). Moreover, also parents' expectations, their need of social support as well as clinical outcomes, such as those referring to their mental health, are taken into account, and appropriate interventions may be targeted by health care providers.

This paper aims to assess how parents of girls with RTT experience health care services and the caregiving process. We evaluated whether services provided to girls with RTT were centred on the family itself, targeting parents' wishes and needs. We aimed to compare mothers' and fathers' assessment, and we also studied the association of the perceived caregiving process with clinical aspects of RTT.

2 | METHODS

2.1 | Participants

The caregiving process was evaluated by parents of girls with RTT. Participants were enrolled (2014–2018) for 1 day of evaluation and parental supervision. Contact with families were provided by the Italian Association for Rett Syndrome, and participants could be potentially enrolled from different regions and provinces in Northern Italy, and, to a lesser extent, from Central and Southern Italy. Fifty-five couples of parents (mothers, mean age: 45.95, mean educational level: 13.36; fathers, mean age: 47.87, mean educational level: 12.38) participated in this study. The inclusion criteria for parents to be enrolled were (a) being Italian mother tongue and (b) a minimum of 5 years of education. As to patients, in the present analysis, we only included girls with (a) diagnosis of typical RTT according to the most recent international guidelines (Neul et al., 2010), that is, regression from the standard acquisition of psychomotor functions, partial or total loss of language, partial or total loss of manual abilities; (b) age greater than 2 years; and (c) positive genetic test (molecular analysis of the MECP2 gene by sequencing the coding exons or through multiplex ligation-dependent probe amplification). We excluded girls with presence of encephalic damage secondary to perinatal trauma, neurometabolic diseases, or cerebral infection. We also excluded patients with impairment of psychomotor development in the first 6 months of life or with a clinical/genetic picture compatible with a diagnosis of variant of RTT. The mean age of girls included in the sample was 12 years (SD = 8.2). All parents were able to provide informed consent to participate in this study and to understand task instructions. The procedures followed were in accordance with the ethical standards of the Helsinki Declaration, and the study protocol was approved by the local Ethical Committee.

2.2 | Measure of processes of care

Parents were required to complete the Italian version of the Measure of processes of care (MPOC-20; King, Rosenbaum, & King, 1995), which is a validated self-report questionnaire aimed to evaluate how health care services provided to children with chronic disability are centred on the family (S. King, King, & Rosenbaum, 2004; King, Rosenbaum, & King, 1996). We specifically required both mothers and fathers to independently complete MPOC-20, because one of the aims of this study was to investigate on possible differences in their perception of the caregiving process. The questionnaire is composed of 20 items, and the focus is on the process of care (Donabedian, 1988). Each question is rated on a 1–7-Likert scale, where 1 indicates "not at all," and 7 means "to a very great extent." MPOC-20 has a well-established factorial structure, made up of five scales (King et al., 1996): (a) enabling and partnership; (b) providing general information; (c) providing specific information about the child; (d) coordinated and comprehensive care for the child and family; (e) respectful and supportive care. The MPOC-20 questionnaire could be completed the same day of the clinical evaluation, or it could be subsequently sent to clinicians (within 2 weeks from assessment) by regular mail. All responses have been considered as confidential and analyzed anonymously in all phases of investigation.

2.3 | Rett assessment rating scale

We evaluated the clinical severity of RTT using the Rett Assessment Rating Scale (RARS), which is a scale completed by parents assessing all aspects of RTT (Fabio et al., 2014). Seven subscales are provided by RARS, targeting the following domains (a) sensory; (b) cognitive; (c) motor-structural and functional; (d) emotional; (e) autonomy; (f) physical characteristics; and (g) behavioral characteristics. Higher scores would indicate a higher level of impairment. Scores from 0 to 55 would indicate mild impairment, from 56 to 81 moderate impairment, and from 82 to 124 severe impairment.

2.4 | Neurological and musculoskeletal assessment

Patients underwent a neurological exam as well as a clinical evaluation of the musculoskeletal components of the limbs (reduction of the range of motion in the main articulations, presence of heterometry...
of the lower limbs) and of the rachis. Because X-ray images were not standardized (patients could be supine, seated, or standing, depending on disease severity and on compliance during the exam), to codify the presence of scoliosis, we adopted the scale of scoliosis standardized in the RARS questionnaire (Fabio et al., 2014). To assess the normal distribution of MPOC-20 scores, we preliminarily ran the Kolmogorov–Smirnov test to verify the normal distribution of the scores. The test never rejected the null hypothesis of normality, either for mothers’ or for fathers’ responses; hence, we used a parametric approach. We used Student t test to compare mothers’ and fathers’ evaluation in each of the five scales, but no comparison proved significant, hence, indicating that mothers and fathers provided a similar assessment of caregiving services.

3 | STATISTICAL ANALYSIS

We analyzed data using descriptive indicators, such as mean and standard deviation. We preliminarily ran the Kolmogorov–Smirnov test to assess the normal distribution of MPOC-20 scores. We followed a parametric approach to data analysis, and we compared means of MPOC-20 scales using t tests for independent samples. We calculated Pearson’s coefficients to evaluate the correlations among subscales. Subsequently, we estimated multiple regression models inserting mothers’ responses in each of the five scales of MPOC as independent variables. The same analyses were repeated in the fathers’ subsample.

4 | RESULTS

4.1 | Descriptive indicators

MPOC-20 questionnaires were completed by 55 mothers (100%) and 41 fathers (74.5%). In Table 1, we report the average evaluation (and standard deviation) for the five scales of the MPOC-20 questionnaire. Both mothers and fathers obtained an intermediate level of satisfaction with respect to the scales (d) coordinated and comprehensive care and (e) respectful and supportive care. The performance was lower for the scales (b) providing general information and (c) providing specific information.

<table>
<thead>
<tr>
<th></th>
<th>N</th>
<th>Enabling and partnership</th>
<th>Providing general information</th>
<th>Providing specific information</th>
<th>Coordinated and comprehensive care</th>
<th>Respectful and supportive care</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mothers</td>
<td>55</td>
<td>4.23 (1.99)</td>
<td>3.88 (1.91)</td>
<td>3.78 (1.69)</td>
<td>4.65 (1.64)</td>
<td>4.69 (1.59)</td>
</tr>
<tr>
<td>Fathers</td>
<td>41</td>
<td>3.97 (1.85)</td>
<td>3.53 (1.93)</td>
<td>3.65 (1.74)</td>
<td>4.37 (1.83)</td>
<td>4.38 (1.59)</td>
</tr>
<tr>
<td>Difference</td>
<td>0.26</td>
<td>0.35</td>
<td>0.13</td>
<td>0.28</td>
<td>0.31</td>
<td></td>
</tr>
</tbody>
</table>

Note. We also report the difference between average mothers’ scores and average fathers’ scores.

4.2 | Comparison of fathers’ and mothers’ evaluation

For each of the five scales of the MPOC-20 questionnaire, we ran a Kolmogorov–Smirnov test to verify the normal distribution of the scores. The test never rejected the null hypothesis of normality, either for mothers’ or for fathers’ responses; hence, we used a parametric approach. We used Student t test to compare mothers’ and fathers’ evaluation in each of the five scales, but no comparison proved significant, hence, indicating that mothers and fathers provided a similar assessment of caregiving services.

4.3 | Correlation analysis

We calculated Pearson’s correlation coefficients between scale scores as completed by mothers and by fathers, separately (Table 2). For both mothers and fathers, the scores of the scale Enabling and Partnership were highly correlated with those of the scale Coordinated and Comprehensive Care and with those of the scale Respectful and Supportive Care. Moreover, the scores of the scale Coordinated and Comprehensive Care were highly correlated with those of the scale Respectful and Supportive Care.

4.4 | Multiple regression models

Multiple regression models were run in the mothers’ subsample for each scale of the MPOC-20 questionnaire (considered as dependent variable), inserting as regressor the Total RARS score, the Scoliosis RARS score, the Walking RARS score, and the epilepsy score. Models were also adjusted by other prognostic indicators, such as the age of the child, the age of the mothers, and the years of school attended by the mothers. No variable proved significant from this analysis, which indicates that evaluation of satisfaction of health care services would not depend on clinical or demographic factors.

We repeated the same analyses in the fathers’ subsample. We found that fathers’ evaluation in the Providing Specific Information scale was positively associated with the RARS scale of scoliosis ($\beta = 0.83(0.38)$, $t = 2.19$, $p = 0.03$), implying that fathers reported to receive good information with respect to presence of scoliosis. By contrast, the RARS walking scale was negatively associated with the Providing Specific Information scale ($\beta = -0.82(0.39)$, $t = -2.09$, $p = 0.04$), which denotes that for greater degree of walking disability,
fathers’ satisfaction about specific information provided by clinician decreases. No other significant result did emerge from these analyses.

4.5 | Comparison with other chronic neurodevelopmental conditions

In Table 3, we reported the average MPOC evaluation obtained in other four studies. Two of these studies targeted a wide range of neurodevelopment disorders (Kertoy et al., 2013; King, Rosenbaum, & King, 1997), one study considered a sample of children with traumatic head injury (Swaine, Pless, Friedman, & Montes, 1999) and another study a sample of children with cerebral palsy (Dyke, Buttigieg, Blackmore, & Ghose, 2006).

An important message provided by Table 3 is that in all studies, the area of Providing General Information attained lower scores. We used t tests for independent samples, separately for mothers and fathers, to compare these scores with those obtained in the present investigation and reported in Table 2. We obtained a significant result in 25/30 (83.3%) of the comparisons in the case of mothers’ evaluations and in 29/30 (96.6%) of the comparisons in the case of fathers’ evaluations.

5 | DISCUSSION

RTT is a severe neurodevelopmental condition, for which it is required rehabilitative support along the life cycle. In this regard, parents can provide useful information to properly identify specific objectives and strategies of the rehabilitation program (Wilkins et al., 2010). The family-centred approach views parents as experts of the children abilities and of their possibilities of development. Nevertheless, parents of girls with RTT are also exposed to experience high levels of stress (Perry, Sarlo-McGarvey, & Factor, 1992), and they should be carefully monitored by clinicians. This study examined parents’ perception of the caregiving process using the MPOC-20 questionnaire, which was proposed to a sample of 55 parents (mothers and fathers). MPOC provides useful clinical information, but it may also be considered in terms of a satisfaction and quality evaluation questionnaire (Camden, Swaine, Tétreault, & Brodeur, 2010). In this study, we found that both mothers and fathers attained an intermediate level of satisfaction with respect to the MPOC-20 Coordinated and Comprehensive Care and Respectful and Supportive Care scales. However, the performance was lower with the Providing General Information scale, and with the Providing Specific Information scale. We did not find systematic differences between mothers’ and fathers’ responses, but the

<table>
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<tr>
<th>TABLE 2</th>
<th>Pearson’s correlation coefficients between scale scores marked by mothers (above) and fathers (below)</th>
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<tbody>
<tr>
<td>Mothers</td>
<td>(a) 1.00 (b) 0.30 (c) 0.69 (d) 0.80 (e) 0.74</td>
</tr>
<tr>
<td>(a) 1.00</td>
<td>(b) 0.30 1.00 (c) 0.41 0.58 1.00 (d) 0.82</td>
</tr>
<tr>
<td>(b) 0.35</td>
<td>(c) 0.71 0.30 1.00 (d) 0.84 0.59 0.82 1.00</td>
</tr>
<tr>
<td>(c) 0.86</td>
<td>(d) 0.34 0.56 1.00 (e) 0.84 0.59 0.82 1.00</td>
</tr>
<tr>
<td>(d) 0.34</td>
<td>(e) 0.84 0.59 0.82 1.00</td>
</tr>
</tbody>
</table>

Note. (a) enabling and partnership; (b) providing general information; (c) providing specific information; (d) coordinated and comprehensive care; (e) respectful and supportive care.

<table>
<thead>
<tr>
<th>TABLE 3</th>
<th>Average evaluation (and standard deviation) in the five scales of the Measure of Processes of Care questionnaire in other four studies</th>
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<tbody>
<tr>
<td>Study</td>
<td>MPOC version</td>
</tr>
<tr>
<td>---------</td>
<td>--------------</td>
</tr>
<tr>
<td>King et al., 1997</td>
<td>56</td>
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<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Swaine et al., 1999</td>
<td>56</td>
</tr>
<tr>
<td>Dyke et al., 2006</td>
<td>56</td>
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<tr>
<td></td>
<td></td>
</tr>
<tr>
<td>Kertoy et al., 2013</td>
<td>20</td>
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Note. a indicates a significant difference (Student’s t test) with scores provided by mothers in the present investigation; b indicates a significant difference with scores provided by fathers in the present investigation.
latter were influenced by clinical variables such as presence of scoliosis or degree of walking impairment. MPOC results reported herein for RTT are generally lower than those reported in other studies investigating other conditions, such as cerebral palsy or pediatric traumatic brain injury.

The caregiving process provided to girls with RTT may depend upon the theoretical framework chosen and adopted to conceptualize the rehabilitative intervention. Families are the primary environment, a microsystem in Bronfenbrenner’s words (Bronfenbrenner, 1979), where girls with RTT grow up and develop, thus their involvement in the rehabilitation process represents a crucial condition to foster participation and social involvement.

Comparing MPOC scores obtained in RTT with those obtained in other clinical samples refers to a noncategorical diagnostic framework, highlighting the similarities shared by different neurodevelopmental disorders, as well as the high variability degree within each syndrome (Perrin et al., 1993). A previous investigation involving a large sample of children with different neurobehavioral disorders highlighted how family-centred caregiving predicted parents’ perception of services and their well-being (King, King, Rosenbaum, & Goffin, 1999). In this study, we found only intermediate scores associated with satisfaction of health care services provided to girls with RTT. One of the possible reasons of this result refers to the fact that children enrolled in our study came from different rehabilitation services from different Italian regions. Notwithstanding the family-centred approach is well known in Italy, not all clinical centres may specifically focus on family as a partner of rehabilitation, and this may explain the lower scores obtained in this study than those reported in other studies (Dyke et al., 2006; Kertoy et al., 2013; King et al., 1997; Swaine et al., 1999). In addition, we remark that results on parents’ satisfaction presented herein are correlational only; there is no proof of a causal link between providing a family-centred approach in rehabilitation and parents’ satisfaction, because unmeasured variables could intervene in mediating this relation.

We found higher scores on the dimensions Coordinated and Comprehensive Care, and Respectful and Supportive Care than on other dimensions. Parents did not complete a scale of emotional well-being or mental health conditions, but we may hypothesize that, also in the case of RTT, higher levels of satisfaction linked to the process of care may represent a protective factor with respect to developing clinical conditions such as depression or anxiety or with respect to psychological well-being (Cadman, Rosenbaum, Boyle, & Offord, 1991; Wallander, Varni, Babani, Banis, & Wilcox, 1989).

A challenging result of the present study is that both mothers and fathers provided lower scores for the MPOC-20 Providing General Information scale and for the Providing Specific Information scale than for other scales. This should be carefully considered because information exchange represents one of the key components of the family-centred approach to rehabilitation (Rosenbaum et al., 1998). Our result is in agreement with a comprehensive literature review on MPOC (Cunningham & Rosenbaum, 2013), which highlighted how the dimension of providing information represents an important limitation of several family-centred services across neurodevelopmental disorders. We suggest that such need of information could be also satisfied using apps or improving the quality of information provided by RTT associations’ websites, as well as parental training activities.

Because in the Italian context we may assume that mothers are more involved than fathers in the caregiving process, especially in late infancy and early adolescence (Scabini, Lanz, & Marta, 1999), we would have expected that fathers, more than mothers, could have expressed the need for more information. By contrast, we did not find any significant difference between mothers’ and fathers’ scores. Even in absolute value, for all scales, the difference between mothers’ and fathers’ scores was below the threshold of 0.5, which would be considered as clinically relevant. This indicates that, independently of the parental roles or involvement, the rehabilitative intervention should specifically target the need of the parents of being more aware of the characteristics of RTT, as well as of the progresses and limitations demonstrated by patients along the life cycle.

One of the strengths of this paper is that we had excellent sample coverage (100%) in the mothers’ subsample, whereas a lesser proportion (74.5%) of fathers completed the questionnaires. The latter proportion is similar to that obtained in a large-sample investigation on children with several neurodevelopmental disorders (74.8%) and is higher than that obtained in a study on cleft lip and palate (62.3%; King et al., 1997). A first limitation of this study is that we used the brief version (20 items) of the MPOC questionnaire, instead of the 56-items version. Notwithstanding MPOC-20 presents excellent psychometric properties, the full version of the questionnaire would be still recommended for research purposes (Cunningham & Rosenbaum, 2013). Second, even though the Italian version of the MPOC is officially available from the CanChild website,1 there is still no formal psychometric replication of validity/reliability properties of the questionnaire in the Italian translation. Third, we did not explore specific parental outcomes, such as stress or mental illness (Brehaut et al., 2004), which may be associated with perception of health care services. All these issues are open for future research.

In conclusion, we highlight the importance of using questionnaires such as MPOC in evaluating health care services for children with severe neurodevelopmental disorders, such as RTT. This study strengthens the positive role played by family-centred services for girls with RTT and demonstrates the need of improving these services, especially in terms of general and specific information provided to parents. Even though parents showed an intermediate level of satisfaction with respect to the quality of the caregiving process, a demand of having more information available on the critical area of walking did emerge from fathers’ responses.

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1https://www.canchild.ca/en/shop/1-mpec-20


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