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Psychological aspects of being a parent of an individual with Rett syndrome: A scoping review

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Abstract

Background: Rett syndrome (RTT) causes multiple disabilities with a lifelong need for substantial care, placing a tremendous lifelong responsibility on the parents. Parenting an individual with RTT can therefore be challenging. Research on the psychological aspects of parenting individuals with RTT is limited and unclear. We aimed to identify and map the existing literature on this subject.

Method: A scoping review was conducted with systematic searches in PubMed, PsycINFO and CINAHL.

Results: Eighteen studies were included. Negative and positive psychological aspects were described with the majority focusing on the negative. Three factors seemed to especially affect the parents: severity of the diagnosis, time (increasing age of parents or individual with RTT; years of caretaking), work-status of the mother.

Conclusions: Seemingly, parents are highly affected; however, the literature is scarce and has several gaps. Future research should include older parents, fathers, parents of individuals living in group homes, and positive aspects.

KEYWORDS
caregivers, mental health, parents, Rett syndrome, severe disability, well-being

1 | INTRODUCTION

Parenting an individual with multiple disabilities can be strenuous. Parents may be required to take on many different caretaking roles and also advocate for their child (Currie & Szabo, 2019; Graungaard et al., 2011; Nachshen & Jamieson, 2000). Research on the psychological aspects of parents of individuals with multiple disabilities indicates that parents experience a high level of burden (Rousseau et al., 2020); reduced well-being and quality of life (Griffith et al., 2011; Rousseau et al., 2019); and have a higher risk of developing stress, anxiety, and depression (Grant et al., 2013; Wulffar et al., 2010). Although less researched, positive aspects, such as personal growth, changes in personal values, and closer family relationships, have also been experienced (Griffith et al., 2011; Luijicau et al., 2019; Rubin & Schreiber-Divon, 2014).

There are several genetic syndromes causing multiple disabilities in the affected individual, and one of them is Rett syndrome (RTT) (Neul et al., 2010; Smeets et al., 2012). The syndrome is in most cases caused by a variant in the methyl-CpG-binding protein 2 gene (MECP2) (Amir et al., 1999) and affects mostly females with an incidence of 1:9000 (Fehr et al., 2011); males are affected in rare cases (Schönewolf-Greulich et al., 2019). Historically, other kinds of genetic syndromes with variants in the genes CDKL5 and FOXG1 have been described as variant forms of RTT. Individuals with CDKL5 variants develop early seizures and an epileptic encephalopathy and are now described as having CDKL5 deficiency disorder (CDD) (Leonard et al., 2022). Individuals with variants in FOXG1 are born with microcephaly and have developmental delay from birth. It was known as congenital RTT but is now called FOXG1 syndrome as the presentation has shown to be different from...
classical RTT (Brimble et al., 2023). However, these variants are included in the current criteria of RTT (Neul et al., 2010).

There is a considerable variation in how RTT presents from individual to individual, but overall newborns with RTT appear to develop typically in the first 6–18 months of life with only subtle signs of disease, followed by an arrest in their development. At the age of 18 months to about 4 years the children reach a phase of regression where they are more sensitive and cry more, eye contact worsens, spoken language and purposeful hand use are affected and may be lost, and they may develop characteristic hand stereotypes that can impede motor abilities (Hagberg et al., 1983; Neul et al., 2010). After the period of regression, eye contact improves, but spoken language and purposeful use of the hands are often severely reduced. Individuals with RTT may develop a range of comorbidities and clinical characteristics including episodes of laughing or crying spells (Neul et al., 2010), epilepsy (Glaze et al., 2010; Henriksen et al., 2018), sleep disturbances (Wong et al., 2015), and severe movement disorders (Singh et al., 2021). Gross motor function is also affected but to differing degrees, thus some can walk independently, some with assistance, while others are wheelchair bound. However, only a few can transfer unsupported from sitting to standing, and few are able to transfer an object from one hand to the other (Downs et al., 2008; Schonewolf-Greulich et al., 2017). Individuals with RTT are also affected intellectually, but to what degree is unknown (Smeets et al., 2012). All these severe comorbidities and disabilities require a high level of caretaking in terms of self-care tasks, medical caretaking, exercising, and advocacy for the individuals with RTT, for example, for treatment, exercise, aids, and activities, with the parents being the ones carrying out the greater part of these tasks (Lim et al., 2012, 2013; Palacios-Ceña et al., 2018).

Because individuals with RTT can live long into adulthood (Kirby et al., 2010; Schonewolf-Greulich et al., 2017), a lifelong comprehensive responsibility is placed on the parents in relation to caretaking and ensuring the quality of life of the individual with RTT. From this perspective, the quality of life of individuals with RTT is highly connected to the parents and their ability to carry out these different demanding caretaking tasks (Aran et al., 2007; Rosenbaum, 2011). To manage this lifelong responsibility, development of a mentally healthy and sustainable parenting role can be highly important. Furthermore, support and counselling from professionals working with the families and the parents of individuals with RTT might be a helpful assistance for the parents in their efforts to develop this kind of parenting role (Rosenbaum, 2011). However, to offer useful support and counselling to the parents, knowledge of their needs and different psychological aspects at stake is needed. One important step towards understanding the parents' needs is to examine the existing evidence on psychological aspects of parents of individuals with RTT.

Research on the psychological aspects of parents of individuals with RTT shows results similar to those regarding parents of individuals with multiple disabilities in general. Thus, the parents' quality of life and well-being are affected (Corchón et al., 2018; Laurvick et al., 2006), they have a higher risk of experiencing stress and depression (Byers et al., 2014; Cianfaglione et al., 2015; Pari et al., 2020), and the positive aspects have been scantily described (Cianfaglione et al., 2015, 2017). However, the evidence is limited and a need for a systematically examination of the extent and nature of research done within the field and an identification of the research gaps is warranted. In the long term this type of knowledge can contribute to the development of relevant support for these parents as well as guide future research. One review regarding parents of individuals with RTT (and their children) have been done previously, but it is only focusing on the parental quality of life (Corchón et al., 2018). Some reviews regarding parents of individuals with disabilities and/or complex care needs have also been done. However, in these studies the focus is primarily on one or two specific parts of the psychological aspects, for example, burn out (Patty, 2020), anxiety and depression (Scherer et al., 2019), or coping and quality of life (Fairfax et al., 2019).

Accordingly, the aim of this study was to conduct a scoping review to map and characterise the literature on the psychological aspects of being a parent of individuals with RTT at all ages and phases of life.

2 | METHODS

We conducted a scoping review because it is a suitable method for identifying and summarising what is known within a particular field or subject, and because we wished to identify any research gaps (Arksey & O’Malley, 2005; Levac et al., 2010). We followed the Joanna Briggs Institute (JBI) methodology for scoping reviews (Peters et al., 2020) and reported the results using the Preferred Reporting Items for Systematic Reviews and Meta-analysis for Scoping Reviews (PRISMA-ScR) (Tricco et al., 2018). A protocol for the scoping review was made and uploaded at the Open Science Framework. It can be accessed through the following link: https://osf.io/txqm/.

2.1 | Eligibility criteria

Drawing on the biopsychosocial model of health, we overall understand psychological aspects as something that stems from the interaction between biological and social/contextual circumstances and the perceptions of and reactions to these circumstances (Borrell-Carrión et al., 2011). Even though, CDD and FOXG1 syndrome today are considered their own syndromes, they are still included in the current diagnostic criteria of RTT (Neul et al., 2010). We therefore choose to include them in the review as well. Thus, studies focusing on parents of individuals with RTT caused by a variant in MECP2, CDKL5 or FOXG1 were considered eligible for this review.

Peer-reviewed observational and qualitative studies were included, with no restriction regarding language and year of publishing. Reviews were excluded to avoid a risk of double effect. As the focus was exclusively on the psychological aspects of the parents, we did not include studies focusing on intervention and intervention strategies, the individuals with RTT’s access to healthcare and social support, costs and economics, the perspective of the individuals with
TABLE 1  Eligibility criteria based on the PPC framework (Peters et al., 2020).

<table>
<thead>
<tr>
<th>Participants</th>
<th>Concept</th>
<th>Context</th>
</tr>
</thead>
<tbody>
<tr>
<td>Parents (biological parents, adoptive parents, foster parents, stepparents, etc.) of individuals with RTT</td>
<td>Psychological aspects and concepts relating to the parenting role when being a parent of an individual with RTT</td>
<td>The individual with RTT living in the family home or in a group home, with the parents having an active parenting role. The individual with RTT can be a child, an adolescent, or an adult.</td>
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</tbody>
</table>

RTT, or methodological questions of questionnaires and outcome measures. Moreover, we did not include studies focusing on the psychological aspects of the parenting role during the Covid-19 pandemic because we considered this to be an extraordinary situation that differed from the usual life of a parent of an individual with RTT. Due to time limitations, grey literature was not included in this study.

2.2  Search strategy

The search strategy was developed by the research group together with an information specialist. The complete search strategy is presented online in supplemental information Table 1. On 15 August 2022, PubMed (Medline), PsycINFO (Ebscohost), and CINAHL (Ebscohost) were searched to identify all relevant empirical studies. The reference lists from the included papers were screened for additional studies. On 12 January 2023, we updated our search.

2.3  Study selection

The result of the search was imported to Covidence (Covidence systematic review software, Veritas Health Innovation, Melbourne, Australia, 2022) and duplicates were removed. Titles and abstracts were screened by two reviewers (MS and JLL) independently, and relevant references were retrieved in full text and assessed in accordance with the inclusion criteria. This was also done independently by the two reviewers. There were only a few numbers of discrepancies in the screening process, and in any of the stages they were all resolved by consensus through face-to-face discussion between the two reviewers. Hence, the inclusion of additional reviewers to solve the conflict as stated in the protocol was not necessary.

2.4  Data extraction, data analysis and synthesis of results

Data from the included studies were extracted using a template developed for this review. The template can be accessed from the study protocol online (https://osf.io/tqym/). We extracted data on the study identifications (title, authors, year of publication, country, language), aim and methods (type of study, measuring tools/questionnaires, type of analysis), type of psychological terms or concepts used or studied, participants (number of participants, age and sex of parents, age, and sex of children—including adult children, children's place of living), and results/findings.

A consultation stage is optional in scoping reviews (Arksey & O'Malley, 2005; Peters et al., 2020). In this scoping review, we chose to present our results to two stakeholder groups: four parents of individuals with RTT of different ages, and three clinical experts from the Danish Center for Rett syndrome. The presentation of the results for the parents of individuals with RTT took place on the 28th of November 2022 as an online meeting since the stakeholders came from different parts of the country. The presentations of the results for the professionals were presented at a staff-meeting on the 16th of December 2022. Afterwards, we asked the stakeholders about their reflections on the results, what kind of knowledge they considered lacking, and what they considered important to study further based on the results of the review.

3  RESULTS

3.1  Characteristics of the studies

An overview of the included studies is presented in Table 2. We identified 174 studies; 35 references were included for full text screening from which 17 studies were excluded, leaving 18 studies eligible for inclusion in the scoping review—see flow diagram (Page et al., 2021) in Figure 1. The 18 studies were published between 1992 and 2022, with the majority (n = 14) being published after 2010. The studies were conducted in the UK (n = 4), the US (n = 4), Australia (n = 5), Germany (n = 2), Canada (n = 1), Italy (n = 1), and Serbia (n = 1). Two studies were qualitative interview studies. One study was described as a mixed-method study; however, we retrieved only the results from the qualitative interview part of the study. The remaining studies (n = 15) were quantitative questionnaire studies, of which (n = 13) used a cross-sectional design and (n = 2) used a longitudinal design.

In 15 studies, the focus was on parents of individuals with a MECP2 variant only, two studies focused on parents of individuals with a CDKL5 variant only, and one study focused on parents of individuals with a MECP2 or CDKL5 variant. No studies focused on parents of individuals with a variation in the FOXG1.

The sample sizes ranged from 29 to 398 in the quantitative studies and from 5 to 37 in the qualitative studies. The majority of participants were mothers; eight studies included only mothers, and six studies had invited both mothers and fathers to participate but it was primarily the mothers who responded. Two studies had an equal distribution of mothers and fathers. In two studies, the sex of the participants was unknown. No studies included only fathers.

The age of the individuals with RTT across the studies ranged from a few weeks to 50 years. However, in the majority (n = 14) of
Mental health was highly different between mothers of children with autism and RTT.

**Characteristics of included studies.**

<table>
<thead>
<tr>
<th>Reference</th>
<th>Study design</th>
<th>Aim</th>
<th>Psychological term(s)</th>
<th>Characteristics parents</th>
<th>Characteristics individuals with RTT</th>
<th>Methods</th>
<th>Key findings</th>
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</thead>
<tbody>
<tr>
<td>Adams et al. (2018)</td>
<td>Quantitative—cross-sectional</td>
<td>To examine the profiles of positive mental health, stress and depression in mothers of children with a range of rare genetic syndromes and compare with mothers of children with autism</td>
<td>Mental health and well-being, positive gain, positive affect, depression, stress</td>
<td>N = 87</td>
<td>Age (years) (y): Mean 50.7, SD 9.2, Range unknown Sex: Females n = 87, Males n = 0, Unknown n = 0</td>
<td>N = 87</td>
<td>• The Positive Gain Scale—The Positive Affect Scale-5&lt;br&gt;• Hospital Anxiety and Depression Scale (seven depression-items)&lt;br&gt;• Questionnaire on Resources and Stress Friedrich—Short Form—Parent and Family Problems subscale&lt;br&gt;• Mental health was highly different between mothers of children with autism and RTT&lt;br&gt;• Mothers of children with RTT showed higher levels of positive gain and positive affect and lower levels of depression and stress</td>
</tr>
<tr>
<td>Byiers et al. (2014)</td>
<td>Quantitative—cross-sectional</td>
<td>To investigate parenting stress and its relation to pain and health issues experienced by individual with RTT</td>
<td>Parenting stress</td>
<td>N = 35</td>
<td>Age (y): Unknown Sex: Females n = 30, Males n = 3, Unknown n = 2</td>
<td>N = 35</td>
<td>• Parenting Stress Index—Short Form&lt;br&gt;43% of parents met the criteria for clinically significant levels of parenting stress for the total parenting stress score&lt;br&gt;Caregivers whose children had seizures had significantly higher scores on the total stress score&lt;br&gt;Caregivers reporting uncertainty about gastrointestinal pain in their child had significantly higher scores on the Parent–Child Dysfunctional Interaction subscale&lt;br&gt;General child factors (age, residential placement, adaptive behaviour) were unrelated to parenting stress score</td>
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<td>Cianfaglione et al. (2015)</td>
<td>Quantitative—cross-sectional</td>
<td>To compare maternal mental health in families of individuals with RTT to normative data, and explore associations between maternal well-being and characteristics of child with RTT</td>
<td>Well-being, stress, anxiety, depression, positive gain</td>
<td>N = 87</td>
<td>Age (y): Mean 50.66, SD unknown, Range 30–70 Sex: Female = 87, Males n = 0</td>
<td>N = 91 (children n = 43, adults n = 48) Age (y): Mean 20.5, SD Unknown, Range 4–47 Sex: Females n = 91, Males n = 0</td>
<td>• Hospital Anxiety and Depression scale&lt;br&gt;• The Positive Gain Scale&lt;br&gt;• The Parent and Family Problems Subscale— from Questionnaire on Resources and Stress Friedrich—Short Form&lt;br&gt;• Mothers of individuals with RTT were more likely to report high levels of anxiety&lt;br&gt;• Same level of depression was seen&lt;br&gt;• Increased severity of RTT behavioural phenotype was associated with higher levels of maternal stress and anxiety&lt;br&gt;• An increased level of current health problems and daughters living at home were associated with higher levels of positive gain</td>
</tr>
<tr>
<td>Reference</td>
<td>Study design</td>
<td>Aim</td>
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<tr>
<td>Cianfaglione et al. (2017)</td>
<td>Quantitative—longitudinal</td>
<td>To examine maternal well-being over a 16-months period in parents of individuals with RTT</td>
<td>Well-being, stress, anxiety, depression, positive gain</td>
<td>N = 50 Age (y): Mean 51.94, SD Unknown, Range 37–70 Sex: Females n = 50, Males n = 0</td>
<td>N = 50 Age (y): Mean 22.52, SD unknown, Range 7–48 Sex: Females n = 50, males n = 0</td>
<td>Hospital Anxiety and Depression Scale The Positive Gain Scale The Parent and Family Problems Subscale— from Questionnaire on Resources and Stress Friedrich—Short form</td>
<td>Measures of stress, positive gains, anxiety, and depression were stable over time Severity of behavioural and emotional problems in the child (at time point 1) was a significant independent predictor of later maternal anxiety and depression scores</td>
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<tr>
<td>Demarest et al. (2022)</td>
<td>Mixed method—only data from the qualitative part were extracted</td>
<td>To explore parent's lived experiences of receiving their child's diagnosis of CDKL5</td>
<td>Grief, long-term benefits of receiving the diagnosis</td>
<td>N = 35 Age (y): 25-30 n = 8 31-40 n = 18 41-60 n = 9 Sex: Females n = 31, males n = 4</td>
<td>N = 37 Age (y): &lt;1 n = 16 1-2 n = 5 2-9 n = 6 &gt;10 n = 10 Sex: Sex unknown Diagnosis: CDKL5 Place of living: Unknown</td>
<td>Phenomenological interviews</td>
<td>Grief was a universal theme expressed among the parents Parents of younger children discussed grief in the context of receiving the diagnosis Parents of older children were at different stages along on their grieving journey when they received their diagnosis Parents with poorer prognostic awareness connected their grief to receiving the diagnosis Positive emotions in relation to receiving the diagnosis: explanation for their child's disability, validation of their sense of something was wrong, having something specific to guide management and expectations (of the condition), relief of guilt Implications of having a child in the family with CDKL5: Brought them closer to their spouse, helped the siblings to be more compassionate people, more aware of differences and disabilities in general, more gratitude for what they had in life, strain with family members outside the immediate family, who did not seem to understand their situation, caretaking pulled time and resources away from other family needs.</td>
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<td>Reference</td>
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<td>Killian et al. (2016)</td>
<td>Quantitative—cross-sectional</td>
<td>To examine relationships of caretaker QoL with both caretaker and child disease-burden characteristics</td>
<td>Caretaker quality of life—Physical and mental QoL</td>
<td>$N = 727$ ($n = 220$ at follow up) Age (y) (at baseline): Mothers: Mean 38.3, SD 9.0, Range Unknown Fathers: Mean 40.5, SD 9.5, Range Unknown Sex: Distribution unknown</td>
<td>Age (y) (at baseline): Mean 9.2, SD 8.3, Range Unknown Sex: Females $n = 727$, Males $n = 0$ Diagnosis: RTT Place of living: Family home $n = 727$</td>
<td>SF-36v2 Health Survey</td>
<td>No changes in the Mental component score from baseline and to the 5 y follow-up. The following variables were associated with mental health: Child having a feeding problem; Parents stating emotional worry caused by child's emotional well-being; Parents stating limitations for their personal needs caused by child's learning disabilities; Parents stating that child's health or behaviour caused tension at home; Poor ability to get along in family</td>
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<tr>
<td>Lamb et al. (2016)</td>
<td>Quantitative—Cross-sectional</td>
<td>To investigate potential relationships among parental self-efficacy, coping methods, family functioning, and caregiver adaptation in parents of individuals with RTT.</td>
<td>Adaptation, coping, self-efficacy</td>
<td>$N = 398$ Age (y): Mean 43.28, SD 9.68, Range 22–74 Sex: Females $n = 361$, Males $n = 31$, Unknown $n = 6$</td>
<td>Age (y): Mean 12.6, SD 9.4, Range 1.6–50 Sex: Females $n = 390$, Males $n = 8$ Diagnosis: RTT Place of living: Family home $n = 398$</td>
<td>The Efficacy Component Parenting Sense of Competence Scale The Ways of Coping Checklist-Revised Family Assessment Measure III Psychological Adaptation Scale</td>
<td>Caregivers who share more caregiving responsibilities with other family members have more effective family functioning Decreasing parental self-efficacy, less problem-focused coping and more emotion-focused coping are associated with poorer family functioning Increasing parental self-efficacy, more problem-focused coping, less emotion-focused coping and better family functioning is associated with better adaptation Family functioning is a significant partial mediator of the relationships between adaptation and parental self-efficacy, problem-focused coping, and emotion-focused coping</td>
</tr>
<tr>
<td>Laurvick et al. (2006)</td>
<td>Quantitative—cross-sectional</td>
<td>To examine maternal, family, child and disability characteristics that are positively associated with the mothers’ good physical health and mental well-being</td>
<td>Physical and mental well-being, stress and coping</td>
<td>$N = 135$ Age (y): Mean 40.4, SD 5.5, Range 21–60 Sex: Females $n = 135$, Males $n = 0$</td>
<td>Age (y): Mean 12.5, SD 7.5, Range 3–27 Sex: Distribution unclear Diagnosis: RTT Place of living: living with parents $n = 135$</td>
<td>SF-12—version 1 McMaster Family Assessment (general function only) Depression Anxiety Stress scale—Short Version Abbreviated Dyadic Adjustment Scale (marital adjustment)</td>
<td>Mental component score was significantly lower compared to norms. The following maternal and child variables were positively associated with better mental health: the mother working full time or part time; well-adjusted marriage; the child not having fracture in the last 2 years; lesser reporting of facial stereotypes and involuntary facial movements; having low stress score</td>
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<td>Mori et al. (2017)</td>
<td>Quantitative cross-sectional</td>
<td>To examine primary caregivers’ well-being and family QoL among families with a child living with the CDKL5</td>
<td>Caregiver well-being</td>
<td>N = 192 (158 in analysis)</td>
<td>N = 192 Age (y): Mean 7.1, SD 6.1, Range 0.2–34.1</td>
<td>SF-12, Version 2</td>
<td>The Beach Center Family Quality of Life Scale</td>
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<td>Country: Australia</td>
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<td>Age (y): Mean 38.2, SD 7.0, Range 24.6–63.7</td>
<td>Sex: Females n = 134, Males n = 28 Diagnosis: CDKL5 Place of living: Unknown</td>
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<td>Sex: Females n = 142, Males n = 15, Unknown n = 1</td>
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<tr>
<td>Mori et al. (2018)</td>
<td>Quantitative–cross-sectional</td>
<td>To examine parental well-being among those raising a child with one of three genetic disorders associated with intellectual disability; Down syndrome, RTT, CDKL5 disorder.</td>
<td>Well-being—physical and emotional well-being</td>
<td>RTT N = 187, Age (y): Mean Unknown Median 46, SD Unknown, Range 28.5–722 CDKL5 N = 168 Age (y): Mean Unknown Median 38 SD Unknown, Range 24.6–63.7 Sex (Whole group): Females n = 593, Males n = 42, Unknown n = 8</td>
<td>RTT N = 187 Age (y): Mean Unknown, Median 16.7, SD Unknown, Range 26.0–35.7 Sex: Females n = 187, Males n = 0 Diagnosis: RTT Place of living: Family home n = 167, Group home n = 20 CDKL5 N = 168 Age (y): Mean Unknown, Median 5.9, SD Unknown, Range 0.0–34.7 Sex: Females n = 143, Males n = 25 Diagnosis: CDKL5 Place of living: Family home n = 164, Group home n = 4</td>
<td>SF-12, Version 2</td>
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<td>Country: Australia</td>
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<tr>
<td>Mori et al. (2019)</td>
<td>Quantitative–longitudinal</td>
<td>To examine the longitudinal well-being of parent of individuals with RTT over a period of 9 years.</td>
<td>Well-being—physical and emotional wellbeing</td>
<td>N = 198 Sex: Females n = 183, Males n = 15 Baseline: Age (y): Mean unknown, Median 41.2, SD Unknown, Range 26.9–63.9 Follow up</td>
<td>Baseline: N = 132 Age: &lt;17 y n = 148, +17 y n = 50 Sex: Females n = 132, Males n = 0 Diagnosis: RTT</td>
<td>SF-12, Version 2</td>
<td>McMaster Family Assessment Device</td>
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<td>Country: Australia</td>
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<tr>
<td>Pari et al. (2020)</td>
<td>Quantitative—</td>
<td>To investigate perceived levels of stress in parents of children with RTT</td>
<td>Perceived level of stress—emotional distress, stress burden</td>
<td>N = 141&lt;br&gt;Age (y):&lt;br&gt;Mothers: Mean 43.73, SD 9.57, Range Unknown&lt;br&gt;Fathers: Mean 46.43, SD 9.19, Range Unknown&lt;br&gt;Sex: Females n = 71, Males n = 70</td>
<td>N = 79&lt;br&gt;Age (y): Mean 12.0, SD 8.4, Range Unknown&lt;br&gt;Sex: Females n = 79, Males n = 0&lt;br&gt;Diagnosis: RTT&lt;br&gt;Place of living: Family home n = 79</td>
<td>Parenting Stress Index, Short Form—Italian Version</td>
<td>• 38.6% of fathers and 43.6% of mothers reported clinical levels of stress (total stress score)&lt;br&gt;• The Parental Distress score and Defence Levels score were lower in fathers&lt;br&gt;• A cumulative effect of caring was seen&lt;br&gt;Stress levels were high when parents had spent more years taking care of a child with RTT&lt;br&gt;This effect was stronger in mothers who showed high levels of stress when taking care of a teenager aged 15+&lt;br&gt;• Severity as measured by RARS was associated with total stress scores, Parent–Child Dysfunctional Interaction Scores and Difficult Child Scores in fathers&lt;br&gt;• Severity was associated with Parent–Child Dysfunctional Interaction scores in mothers</td>
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<td>Reference</td>
<td>Study design</td>
<td>Aim</td>
<td>Psychological term(s)</td>
<td>Characteristics parents</td>
<td>Characteristics individuals with RTT</td>
<td>Methods</td>
<td>Key findings</td>
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| Retzlaff (2007)    | Qualitative  | To explore key factors contributing to the resilience of families of children with a RTT | Resilience, coherence | N = 12 Age (y): Mothers: Mean 38.8, SD unknown, Range unknown | N = 6 Age (y): Mean 7.5, SD Unknown, Range Unknown | Narrative interviews       | • To types of narratives/resilience stories. Type 1 = story of the re-found balance. Type 2 = story of the long tedious walk  
• Ensuing emotional crisis (after diagnosis) is overcome very slowly  
• Extended family does not react positive and supportive way  
• Social environment is openly rejecting.  
• Professional helping system takes additional energy  
• Mentioning specific positive aspects of the child, family life has changed in a positive way, family has developed meaningful values  
• Confidence in challenges can be mastered.  
• Couples have flexible roles, balancing marital relationship as well as individual interests |
| Sarajlija et al. (2013) | Quantitative—cross-sectional | To investigate health related QoL and depression in mothers caring for children with RTT in Serbia | Health-related quality of life, depression | N = 49 Age (y): Mean 37.5, SD 7.5, Range 22—55 Sex: Females n = 49, Males n = 0 | N = 49 Age (y): Mean 12.2, SD 6.7, Range 3—29 Sex: Females n = 49, Males n = 0 | SF-36, Beck Depression Inventory II | • Mild/moderate and severe depression was observed in 16.4% and 30.6% of the mothers, respectively  
• Significantly lower values of the total composite SF-36 score and higher depression scores were seen in mothers of children with clinic severity scores >20  
• Increasing age of mothers and higher clinical severity were associated with lower total composite SF-36 score  
• Increasing age of mothers, higher clinical severity and unemployment in mothers were associated with higher depression scores |
<p>| Sarimski (2003)    | Quantitative, cross-sectional | To examine the variation in behaviour and communication skills in girls with RTT and psychosocial burden in their | Psychosocial burden | N = 83 Age (y): Mean Unknown, SD unknown, Range unknown Sex: Females n = 83, Males n = 0 | N = 83 Age (y): Mean 8.4, SD 3.7, Range Unknown Sex: Female n = 83, male n = 0 | Handicap-related Problems for Parents Inventory | • The daily care of the child, time for personal interests and time for work and housework were reported as a weekly burden with no immediate solution by more than half of the mothers (68.7%, 60.2% and 55.4%, respectively) |</p>
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<th>Reference</th>
<th>Study design</th>
<th>Aim</th>
<th>Psychological term(s)</th>
<th>Characteristics parents</th>
<th>Characteristics individuals with RTT</th>
<th>Methods</th>
<th>Key findings</th>
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<td>Urbanowicz et al. (2011)</td>
<td>Country: Australia</td>
<td>Qualitative—cross-sectional</td>
<td>To examine the relationships between use of resources and later caregiver health in parents of girls/women with Rett syndrome (second aim)</td>
<td>Caregiver health—health related QoL</td>
<td>$N = 119$&lt;br&gt;Age (y): Mean Unknown, SD unknown, Range unknown&lt;br&gt;Sex: Females $n = 119$, Males $n = 0$</td>
<td>SF-12</td>
<td>• The burdens expressed by mothers did not differ with child age, or classical versus atypical RTT &lt;br&gt;• The psychosocial burden was higher in mothers of children with lower general mood, more fear/anxiety and more night-time behaviours</td>
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<tr>
<td>Williamson (2019)</td>
<td>Country: UK</td>
<td>Qualitative</td>
<td>To explore the experiences of single mothers caring for a daughter with RTT</td>
<td>Psychological aspects of caregiving, self-sacrifice, no meaningful roles beyond caregiving, caregiver burnout, day-to-day burden of caregiving, existential level of suffering, lost sense of themselves, loss of identity, despondency, exhaustion, sense of self-esteem, coherence, pride, meaningfulness from devotion to child</td>
<td>$N = 5$&lt;br&gt;Age (y): Mean 43.4, Median 44, SD unknown, Range 35–47&lt;br&gt;Sex: Females $n = 5$, Males $n = 0$</td>
<td>Visual documentation of experiences using photographs and interviews (with interpretative phenomenological analysis)</td>
<td>Two interconnected themes were derived: &lt;br&gt;1. Committing to ‘Total Caregiving’ where caregiving is central to the mothers’ lives, and they experience a need to anticipate and respond to their daughters’ needs&lt;br&gt;2. Self-abnegation and existential Crisis shows that caregiving comes with a considerable cost to the mothers themselves. The mothers’ experiences of psychological challenges in relation to their sense of self and personhood</td>
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studies the mean age of the individuals with RTT was lower than 18 years and in none of the studies was the mean age of the individuals with RTT more than 25 years.

Some studies \((n = 11)\) reported the place of living for the individuals with RTT. In 7 of these studies, all individuals with RTT lived in the family home; in 4 of the 11 studies some individuals with RTT lived in a group home, but with the majority living in the family home.

Many different psychological concepts and terms were used to describe the psychological aspects of being a parent of an individual with RTT. Some overlapped, for example, health-related quality of life \((\text{Sarajlija et al., 2013})\) and emotional wellbeing \((\text{Mori et al., 2018; Mori et al., 2019})\), while others clearly differed, for example, psychosocial burden \((\text{Sarimski, 2003})\) and depression \((\text{Cianfaglione et al., 2015, 2017})\). Figure 2 shows the different psychological concepts and terms related to the parents that were used in the included studies. The many different factors affecting the parents according to the included studies are also shown in the figure.

### 3.2 Parental stress

The level of parental stress was measured in six studies \((33\%)\) \((\text{Adams et al., 2018; Byiers et al., 2014; Cianfaglione et al., 2015, 2017; Pari et al., 2020; Perry et al., 1992})\). One study found that the parents reported higher levels of stress compared with the norm \((\text{Perry et al., 1992})\). Similarly, two other studies reported a clinical level of stress according to the scoring of the questionnaire in approximately 40\% of parents \((\text{Byiers et al., 2014; Pari et al., 2020})\). One longitudinal study lasting 16 months found that the level of stress was stable over this period \((\text{Cianfaglione et al., 2017})\). The relationship between stress and child-related factors, such as age and epilepsy, showed differing
results. One study found that the level of stress was high when the parents had spent more years on caretaking and that this effect was stronger in mothers caring for an individual with RTT aged 15 years or older (Pari et al., 2020). However, other studies found no relationship between parental stress and the age of the individual with RTT (Byiers et al., 2014; Perry et al., 1992).

One study found that parents of individuals with RTT who suffered from epilepsy had a significantly higher total stress score (Byiers et al., 2014), while another found no associations between seizures and the parental level of stress (Pari et al., 2020). Gastrointestinal pain is another factor found to affect parental stress (Byiers et al., 2014). Parents who were unsure whether their child experienced gastrointestinal pain reported a higher level of stress than did those who knew their child experienced the pain (Byiers et al., 2014). Overall, the studies showed an increased level of parental stress but more knowledge about associated factors is needed.

Despite it has been found that the parents reported an elevated level of stress, one study compared the parental level of stress between parents of individuals with RTT (and 12 other genetic rare syndromes) and parents of individuals with autism spectrum disorders and found a lower level of stress in the parents of individuals with RTT compared to the parents of individuals with autism spectrum disorders (Adams et al., 2018).

3.3 | Depression

Five studies (28%) focused on parental depression, and the results differed (Adams et al., 2018; Cianfaglione et al., 2015, 2017; Sarajlija et al., 2013). One study found mild/moderate depression in 16.4% and severe depression in 30.6% of Serbian mothers (Sarajlija et al., 2013), whereas another study found no differences in the level of depression between British mothers and the norm (Cianfaglione et al., 2015). The latter findings persisted over 16 months (Cianfaglione et al., 2017).

An association was found between higher depression scores and increasing maternal age, unemployment of mothers, and higher clinical severity in the individual with RTT (Sarajlija et al., 2013). Severity of the behavioural and emotional problems in the individual with RTT was found to be an independent predictor of later maternal depression (Cianfaglione et al., 2017).

Due to a low number of studies and differences in methods and assessments between the studies more research is needed to better understand whether elevated levels of depression are present in the parents.

Compared with parents of individuals with autism spectrum disorders, parents of individuals with RTT showed lower levels of depression (Adams et al., 2018).

3.4 | Anxiety

Two connecting studies (11%) measured anxiety in mothers (Cianfaglione et al., 2015, 2017). In these studies, the mothers reported a higher level of anxiety compared with the British norms and the measures were stable over 16 months. In the first study, there was an association between the severity of RTT behavioural phenotype and anxiety (Cianfaglione et al., 2015); in the second, the severity of behavioural and emotional problems in the child showed as a significant independent predictor of later anxiety in the mothers (Cianfaglione et al., 2017).
As such, it might seem that behavioural problems in the individual with RTT can increase the risk of the development of anxiety in the mother.

### 3.5 SF-12 and SF-36 mental health

Six studies (33%) measured and reported the parents’ mental health (Killian et al., 2016; Laurvick et al., 2006; Mori et al., 2017, 2018, 2019; Urbanowicz et al., 2011) using either the SF-12 or the SF-36 which is a generic tool to measure general health (Ware et al., 1993, 1996, 1998, 2000; Ware & Sherbourne, 1992). The questionnaire has a physical health score, a mental health score and a total health score. Due to the scope of this review, we chose only to report the mental health scores from the included studies. The mental health score measures the general mental health, including psychological distress and psychological wellbeing, and does not focus on specific pathological states like stress, anxiety and depression (Ware & Sherbourne, 1992). All studies found that the parents had lower mental health compared with the norm.

The specific child-related factors that impacted mental health negatively were clinical severity of the child (Mori et al., 2018), sleep disturbances (Mori et al., 2017), feeding problems and gastroesophageal reflux (Killian et al., 2016). Factors that were positively associated with mental health were fewer facial stereotypes, no fractures in the individual with RTT during the preceding 2 years, increasing age of the individual with RTT, and a tube-fed child (Laurvick et al., 2006; Mori et al., 2017). Parent-related factors affecting their mental health negatively were concerns about the individual with RTT’s emotional wellbeing, and limitations in fulfilling personal needs due to the individual with RTT’s learning disabilities (Killian et al., 2016). Parent-related factors that were positively associated with mental health were presence of a well-adjusted relationship, low stress scores, increasing maternal age, and mothers working full- or part-time (Killian et al., 2016; Laurvick et al., 2006). Structural factors affecting mental health negatively were financial hardship, use of both formal and informal respite, presence of siblings and the child not attending a day activity (Mori et al., 2017, 2018).

In an Australian longitudinal study with up to 9 years’ follow up, it was found that mental health was negatively associated with the individual with RTT being partly or fully tube-fed and with the individual with RTT’s increasing age, especially if the child was between 11 and 17 years at baseline. The mental health was positively associated with living in the outer regions or remote regions of Australia or with the child being in care (Mori et al., 2019). To sum up, many different factors seem to affect the mental health measured with the SF-12/36 underlining the complexity of how and by what the parents are affected.

One study compared the mental health of parents of children with RTT, CDD, and Downs syndrome. The parents of children with CDD had the lowest quality of mental health, although all three groups scored below the norm (Mori et al., 2018).

### 3.6 Burden

Burden was only addressed in two studies (11%). One study investigated what they referred to as the psychosocial burden in mothers, without further definition of the concept. This study found that the burden was reported as being higher in mothers of individuals with RTT who had a lower general mood, expressed more fear/anxiety and had more night-time behaviours (Sarimski, 2003). Factors reported as being weekly burdens for which there was no immediate solution were the daily care of the child (reported by 68.7%), no time for personal interests (reported by 60.2%) and no time for work and housework (reported by 55.4%) (Sarimski, 2003). No associations were found between the expressed burden and the age of the individual with RTT (Sarimski, 2003).

In a qualitative study investigating five single mothers of individuals with RTT, the mothers described a burdensome parenting role with profound and comprehensive self-sacrifice and self-abnegation (Williamson, 2019). They experienced a life of total caregiving with time-poverty and time-pressure when having to use all their time caring for the individual with RTT. They also described how they had various roles that resulted in a loss of identity and sense of self, and a feeling of not having other meaningful roles (Williamson, 2019). Based on these two studies, mothers do experience weekly burdens relating to the extensive caretaking tasks.

### 3.7 Positive aspects

Five studies (28%) included positive aspects in their study. A quantitative study measured positive gains reported by parents of individuals with RTT, for example, personal growth (Cianfaglione et al., 2015). It was found that the reported positive gains were higher when the individual with RTT resided with the mother rather than living in a group home and if the individual with RTT had an increased level of current health problems (Cianfaglione et al., 2015). The positive gains were stable over 16 months (Cianfaglione et al., 2017). Another quantitative study showed that the mothers of individuals with RTT had higher levels of positive gains than did mothers of individuals with autism spectrum disorders (Adams et al., 2018).

Two qualitative studies included a focus on the positive aspects alongside the focus on the hardship and negative aspects of parenting an individual with RTT. In the first study, single mothers expressed that being able to care well for their child positively affected their self-esteem and made them feel proud. These feelings occurred despite the mothers simultaneously experiencing a burdensome parenting role (Williamson, 2019). In the other study, some parents often mentioned specific positive aspects about the individual with RTT, and they felt the family had changed in a positive way and had developed meaningful values through the experience of being a family where one of the members has RTT (Retzlaff, 2007). As such, positive gains and positive feelings have been described in studies focusing primarily on mothers.
3.8 | Adaptation

In all, three studies (17%)—one quantitative (Lamb et al., 2016) and two qualitative studies (Demarest et al., 2022; Retzlaff, 2007)—focused on the adaptation process and the feelings after receiving their child’s diagnosis. The quantitative study found that better adaptation was associated with increasing parental self-efficacy, less emotional coping, and better family functioning (Lamb et al., 2016). In one of the qualitative studies, different feelings were described when receiving the diagnosis of CDD. These feelings were shock, anger, sadness, disbelief, isolation and very often grief (Demarest et al., 2022). Acute onset of grief when receiving the diagnosis was particularly seen when the individual was a very young child. In contrast, for the parents with older children, receiving the diagnosis of CDD was often associated with a sense of relief because they got an explanation of the symptoms they were experiencing in their child (Demarest et al., 2022). However, these parents described another type of grief: episodic grief, which often related to life’s milestones not being achieved (Demarest et al., 2022).

In the other qualitative study, there were two overarching narratives about the parents’ re-found balance after the emotional crisis they experienced when receiving their child’s diagnosis. In the first narrative, the parents talked about a re-found balance that occurred when there was a clear acceptance of the diagnosis. Parents tended to mention specific positive aspects of the individual with RTT, had a feeling that the family had changed in a positive way (which was also described earlier under positive gains) and had a good relationship with flexible roles and time for individual interests (Retzlaff, 2007). The other group of parents in that study described a narrative of a slow and difficult way for them to overcome the severe shock of receiving the diagnosis. In this narrative of adaptation, the parents focused more on the ongoing stressors and burdens than on their daughter as a resource (Retzlaff, 2007).

From this it might seem that the parents’ feelings, their way of coping and their views on the diagnosis and/or the individual with RTT influences how they adapt to their parenting role.

3.9 | Stakeholder perspectives

There was a general recognisability of the results by both stakeholder groups. However, they also wondered about how little research has been done when considering how much RTT affects the parents and the families, and how important the parents are to ensure the well-being of the individual with RTT. They were particularly critical about the lack of inclusion of fathers in the studies, and how little research that has been done about older parents, because they considered the parenting role and responsibility to be lifelong. Mental health was considered an important focus, including negative and positive perspectives of it. The parents found grief an interesting term since being parents of an individual with RTT is a meaningless condition of life that from time to time causes feelings of grief. Worries was mentioned by the parents several times. Worries about the future, for example, about transitions from child to adult life, if the individual with RTT outlives the parents, or changes in the life circumstances were present for the parents, and research about when it is particular demanding or difficult being a parent of an individual with RTT was wanted. In extension, it was stressed that having access to a competent team of health care professionals and their ability to meet the parents’ concerns and worries about the individual with RTT was considered important.

Furthermore, the parent stakeholder group found it interesting that mothers’ working was found to be a positive factor. All the mothers in the stakeholder group did find it important for them to be working, even though it is often necessary to have reduced hours, when the individual with RTT is living at home, due to the amount of required care.

4 | DISCUSSION

This scoping review showed that the literature described both negative and positive psychological aspects of being a parent of a child with RTT, although there was a predominance of studies focusing on the negative aspects. The results also revealed a heterogeneous use of terms, concepts, and outcome measures to describe and assess the psychological aspects. Overall, a higher risk of developing parental stress and poorer mental health was described. The parents experienced several weekly burdens and high caregiving demands and some felt a loss of identity and feelings of self-sacrifice. Feelings of grief could persist and be episodic during the individual’s childhood. The parents described positive gains such as feelings of personal growth, pride, and self-esteem together with the development of meaningful values and a re-found balance in their mental wellbeing. Many different factors relating to the diagnosis, the parent, and the structural conditions were found as associated with the psychological aspects. However, three overarching factors recurred across the studies: (1) severity of the diagnosis of the RTT (the clinical severity score, feeding problems/individual with RTT being tube-fed, emotional or behavioural problems); (2) time (increasing age of the parent, increasing age of the individual with RTT, years of caretaking); and (3) work-status of the mother (positive when working full/part time, negative when being a full-time homemaker).

Most of the factors affecting the parents of individuals with RTT did not specifically relate to RTT, for example, years of caretaking, increasing age of the parents, or the mothers’ working-status. Moreover, many of the factors related to the diagnosis of RTT are factors also seen generally in individuals with multiple disabilities, for example, feeding problems/tube-feeding (Carter et al., 2013), epilepsy (Jakobsen et al., 2020; Nolan et al., 2006), emotional and behavioural problems (Scherer et al., 2019). Therefore, we would argue that the majority of the results from this review does also apply to parents of individuals with other diagnoses causing multiple disabilities. If this is the case, that could be of great importance for the field in order to develop useful support and counselling to all parents of individuals with multiple disabilities.
In this review, we defined ‘psychological aspects’ as any emotion, belief, perception, adaptation or mental reaction. Thus, it encompasses many different aspects and concepts of the human psyche and behaviour. Therefore, when investigating some of the psychological aspects, it is important to define the terms and concepts in focus and/or to make a clear description of them. However, apart from the heterogeneous use of terms and concepts, the majority of the included studies in this review had no clear definition of the psychological terms and concepts they investigated. This affects the results and the validity of the studies. Further, it makes it difficult to compare the results across the different studies. In future research it is therefore important to be aware of what type of psychological aspect that are being investigated, and how these terms or concepts are being defined and/or understood by the research team. Many of the quantitative studies had a pathological focus in their outcomes, such as stress, anxiety, and depression. This is not unusual in this field of research (Grein & Glidden, 2015) but can be problematic for several reasons. First, when investigating a pathological concept, such as depression, it is important to be aware of what is measured, that is, to distinguish between clinical depression and depressive symptoms (Bailey et al., 2007). Clinical depression is a pathological and debilitating state that needs a certain type of treatment and intervention, whereas depressive symptoms are feelings of sadness that are disruptive and unpleasant and arise from the parents’ circumstances. The tools used to measure depression in the studies included in this review, the Beck Depression Inventory (Beck et al., 1996) and the Hospital Anxiety and Depression Scale (Zigmond & Snaith, 1983), are screening tools and not clinical diagnostic tools (Bailey et al., 2007). From this perspective, when one of the studies reports severe depression in 30.6% of the mothers (Sarajlija et al., 2013) it should be interpreted as the mothers displaying severe depressive symptoms.

Second, with a pathological focus, a dichotomous understanding of a psychological concept emerges. From this perspective the parents are either suffering from the pathological diagnosis in focus (e.g., depression), or they are not. As such, when a scale of depressive symptoms is used and no symptoms are reported by the parents, it is often concluded that the parents have good mental health (Fitzgerald & Gallagher, 2021). However, if we look at the results from the qualitative studies in this review, this is not the case, and the psychological aspects of the parents and the experiences they are describing are much more complex. From our perspective when investigating parents of individuals with RTT it is therefore necessary to use a non-dichotomous and non-pathological approach and use a concept that can include the whole group of parents. We will argue that such an approach will improve the possibility of achieving a broader and more nuanced insight to the different psychological aspects that are represented in the whole group of parents, and not just a part of it, which is important if we want to offer support and counselling to all the parents in their effort to develop a sustainable parenting role.

An example of a model that uses this approach is Keyes mental health model. The model disputes the assumption that the absence of a pathological mental diagnosis is the same as having good mental health (Keyes, 2002, 2005). Instead, this model suggests that mental health and mental illness should be seen as two different but related concepts that belong on two different continua. The continuum of mental illness ranges from absence to presence of mental illness, whereas the continuum of mental health ranges from a minimal level of mental health (called languishing) to an optimal level of mental health (called flourishing). In this model a parent can have a mental illness but still flourish if the illness is well managed. Similarly, the absence of a mental pathological diagnosis does not necessarily mean the parent is thriving and is mentally healthy (Keyes, 2002, 2005). The mental health model could be used to provide a more nuanced understanding of mental health in parents of individuals with RTT. Accordingly, the entire group of parents would be included, regardless of whether or not they were displaying symptoms of or suffering from mental illness. To obtain scientific knowledge that can enhance the support and counselling of the entire group of parents, we will therefore recommend that future research and developments focus on mental health from a more salutogenic perspective instead of a pathological perspective, including the development and maintenance of mental health for the entire group of parents and not only for the parents experiencing mental illness or mental illness symptoms.

The studies included in this review clearly showed that understanding what negatively and positively affects the parents is complex. One study focusing on the positive psychological aspects of the parents found that those parents who reported the highest level of positive gains were those who also reported an increased level of health problems in the individual with RTT and those who resided with the individual with RTT (Cianfaglione et al., 2015). According to the result of the review, those parents were also the ones who were experiencing more negative aspects. Other studies confirm that higher caregiving demands are positively related to personal growth (Beighton & Wills, 2019; Rubin & Schreiber-Divon, 2014), and that it is the severity of behavioural and emotional problems in the child that makes it difficult for the parents to identify positive aspects (Beighton & Wills, 2019; Lloyd & Hastings, 2009). In one qualitative study, the mothers described a high level of burden and caretaking and found it challenging to be in this parenting role; however, they also expressed that this burden and demanding caretaking was also what constituted the positive personal growth and pride they felt (Williamson, 2019). Seemingly, the negative and positive psychological aspects do not rule each other out; on the contrary, they appear to co-exist, which has also been found in several other studies (e.g., Hastings & Taunt, 2002; Horsley & Oliver, 2015; Lloyd & Hastings, 2008). As such, there is an apparent paradox within the parenting role regarding individuals with disabilities and a high level of caregiving demands, including in parents of individuals with RTT: what affects the parents negatively is also what affects them positively.

From this perspective it becomes important to also focus on the positive perspectives in the research. By including this, a more complete insight to the parenting role and the complexity of it can be provided, and more nuanced perspectives and narratives can be offered.
to the parents (Green, 2007), which can be important in their process of developing a sustainable parenting role.

The knowledge obtained in this review is a first step towards understanding how the parents are affected by their parenting role and what their needs for support might be, which is useful knowledge when choosing or developing support and intervention programs. To our knowledge, no intervention programs have been developed or been used solely on parents of individuals with RTT. However, several interventions and support programs for parents of children with various disabilities exist such as cognitive-behavioural therapy, problem-solving therapy, group-based parent training and peer-to-peer programs (Barlow et al., 2014; Law et al., 2019). Some of these parent intervention programs primarily focus on outcomes related to the functioning of the child (e.g., behaviour, communication) with indirect benefits of the parents’ wellbeing (Irwin et al., 2019). Many of the interventions hold a potential to reduce depressive symptoms, anxiety and stress and to improve relationship satisfaction and parent competence (Barlow et al., 2006; Irwin et al., 2019; Law et al., 2019). However, evidence of long-term improvements in the mental health and wellbeing of the parents of individuals with disabilities are not clear (Barlow et al., 2014; Law et al., 2019). The knowledge gained through this review and additional studies focusing on psychological aspects on parents of individuals with multiple disabilities could aid professionals and researchers to choose and/or develop relevant support and intervention programs in the future that focus on mental health from a salutogenetic point of view, with the aim of developing a long-lasting sustainable parenting role.

4.1 Research gaps in evidence base and implication for further research

This review points to several research gaps. In most studies, the mean age of the individuals with RTT was below 18 years; few studies included those above 18 years. This is not unusual in the field of disability research (Beighton & Wills, 2019; Rydzewska et al., 2021). However, the longevity of people with intellectual and developmental disabilities is increasing (Freilinger et al., 2010; WHO, 2000). In the Danish cohort of individuals with RTT, two thirds are above 18 years of age (Bisgaard et al., 2021). This could also be the case in other countries. The increasing longevity implies an extended parenting role, and knowledge is lacking about this important aspect. Further, few studies included parents of individuals with RTT who lived in a group home, and when they were included, it was only a small number. However, it is important to have knowledge of the parenting role when the individual with RTT is not living with the parents, regardless of whether the individual is over or under the age of 18 years.

Fathers as participants were also underrepresented in the studies. Nevertheless, fathers play a central role in the family systems (Rankin et al., 2019) and their perceptions and experiences may differ from those of the mothers (Fitzgerald & Gallagher, 2021; Rowbotham et al., 2011; Trute et al., 2007). If we are to support the parents in developing strong and effective family systems, we need to know how the parenting role is experienced by both mothers and fathers. Therefore, future research should make an effort to include fathers in the studies, regardless of the fathers being the primary caretaker or not.

Few studies included the positive psychological aspects of being a parent of an individual with RTT. Those positive aspects of being a parent of an individual with RTT—or with other intellectual or developmental impairments or multiple disabilities—are vital to consider because they might buffer poor wellbeing (Horsley & Oliver, 2015) and provide a more nuanced understanding of this type of parenting role (Green, 2007). Nonetheless, in disability research the focus is often on negative psychological (and pathological) aspects, such as stress, anxiety, and depression (Horsley & Oliver, 2015), which was also the case in this review. Several researchers have also pointed out that the positive aspects in the field of research about parents of individuals with disabilities in general have been neglected and that more research into these aspects needs to be done (e.g., Hastings & Taunt, 2002; Horsley & Oliver, 2015). We think this also applies to parents of individuals with RTT.

In relation to the methodological designs, we identified two substantial research gaps. First, only two studies had a longitudinal design, and one of those studies had a maximum of only 16 months between the measurements. The lack of longitudinal research makes it difficult to draw conclusions on the trajectory of the parenting role and how the negative and positive psychological aspects and experiences change over the years, including whether there are any vulnerable situations or time points in the trajectory of the life of a parent of an individual with RTT.

Second, we found that only two studies had a qualitative design, and one had a mixed method design from which we extracted only the results of the qualitative results. A qualitative study usually has an explorative approach that investigates the experiences of the parents and offers a first-person perspective about the parenting role. Knowledge obtained through first-person perspective is crucial for this field because this kind of knowledge will provide a more precise insight to the parents’ experiences, including their obstacles, challenges and positive gains, which is important when supporting and counselling the parents about their parenting role.

In this review we included two stakeholder groups which turned out to be valuable to our understanding and interpretation of the results. Furthermore, their reflections on what kind of knowledge they considered lacking made some interesting perspectives on possible future research. Based on these experiences we would very much encourage to make collaborations with stakeholders in future research.

4.2 Strengths and limitations

To obtain transparent results, we adhered to the JBI methodology for scoping reviews, including developing and using an a priori protocol, and reported the results following the PRISMA-ScR. The search was developed in collaboration with an experienced information specialist,
and we had no limitations regarding language and year of publishing. We used two independent screeners who were able to resolve all conflicts through discussions. Arksey and O’Malley recommend a consultation stage (Arksey & O’Malley, 2005) where practitioners and consumers are consulted. We presented and discussed the results from this scoping review with four parents of individuals with RTT of different ages and with three clinical experts in RTT.

However, some limitations exist. First, we did not include grey literature, which could have expanded our results. Second, the results should be interpreted with caution due to the low number of participants in many of the included quantitative studies, the heterogeneity of some results, and the use of many different terms and concepts of psychological aspects and their unclear definition which makes it difficult to draw clear conclusions.

The search generated only 174 references for title-abstract screening after the removal of duplicates. This is an unusually low number of references, which might often indicate a weak search strategy. However, RTT is a rare disease and when using this in the search strategy it automatically limits the number of references. The finding of only one additional reference (in German) in the screening of the reference lists of the included studies also confirms the strength of the current search strategy.

Before developing the search strategy for this review, we developed another search strategy with the intention of investigating the psychological aspects of parents of individuals with multiple disabilities in general, including RTT, because we wanted to attain a more broad and nuanced assessment of the literature (the protocol can be accessed through the following link: https://osf.io/p3dsq). This search resulted in a much higher number of references. However, multiple disabilities is not a clearly defined term, and we found a very divergent use of the term which made the results unclear and therefore unreliable. Accordingly, we decided not to continue with that review and instead create a new protocol where we changed the scope to parents of individuals with RTT and to include parents of individuals with either a variation in MECP2, CDKL5 or FOXG as an example of diagnoses that cause multiple disabilities. When doing so, we found that the results became much more precise. To make the results useful in relation to parents of individuals with other diagnoses causing multiple disabilities, we tried to explicate the different type of factors affecting the parents, so it is possible to see which factors were related to the diagnosis and the comorbidities, and which factors were related to the parents, the family or to structural circumstances that might imply for parents of individuals with multiple disabilities in general. Some of the factors relating to the diagnosis that affects the psychological aspects of the parents is not unique for RTT, for example epilepsy or sleeping disturbances, and therefore the effect of these factors might apply for parents of individuals with other types of diagnosis causing multiple disabilities as well. For future research, inclusion of other genetic syndromes and other types of diagnosis that causes multiple disabilities would provide even more knowledge in the field.

In our review, we included only studies that focused solely on the parents, rather than the whole family. However, research regarding the families of individuals with RTT does exist (Bolbocean et al., 2022; Müller et al., 2007; Rozensztrauch et al., 2021), and there might not be a clear distinction between studies focusing on the parents and studies focusing on the families. Furthermore, some studies reported the physical wellbeing of the parents; due to the scope of this study we did not include these results. Nevertheless, it may be argued that physical health and mental health can affect each other.

5 | CONCLUSION

This scoping review was the first to map the literature regarding the psychological aspects of being a parent of an individual with RTT. The review revealed that the literature is limited and has many research gaps. Nonetheless, the literature shows that the parents are psychologically affected in different ways and by many different factors. The knowledge gained from this review can contribute to the development of relevant support and interventions for parents of individuals with RTT. However, for a more comprehensive picture of the parenting role, future research should include positive aspects, fathers and older parents as participants, parents of individuals living in group homes and exploration of parental experiences.

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DATA AVAILABILITY STATEMENT

My protocol—including my search strategy—is available on Open Science Framework.

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SUPPORTING INFORMATION
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