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Health-related quality of life and family functioning in parents of children with Barth syndrome: an application of the Double ABCX model

Yoonjeong Lim[1*], Ickpyo Hong[2] and Areum Han[3]

Abstract

Background Living with children with disabilities has a significant impact on parental health-related quality of life (HRQoL) and family functioning. Barth syndrome (BTHS) is a rare, X-linked disorder that primarily affects males, presenting symptoms such as cardiomyopathy, neutropenia, muscle weakness, and growth delays. In this study, we investigated how a child's functional performance, family cohesion, and satisfaction with healthcare affect parents of children with BTHS.

Methods Thirty-three parents of children with BTHS and 31 parents of age-matched unaffected children participated in this study. The parents completed a series of questionnaires. The Double ABCX model was applied to select measurement variables for this study. An independent samples *t*-test was used to compare HRQoL and family functioning between the two groups. Regression analysis was conducted to determine how a child's functional performance, family cohesion, and satisfaction with healthcare affect HRQoL and family functioning of parents of children with BTHS.

Results The HRQoL and family functioning of parents of children with BTHS were significantly lower than those of unaffected children ($p < .05$). In the regression analysis, the child's functional performance was a significant predictor of HRQoL and family functioning ($F(3, 32) = 6.047$, $p = .003$) for parents of children with BTHS.

Conclusions This study lays the groundwork for examining the impact of raising children with BTHS on parents and families. It is crucial for health professionals to understand the clinical features of BTHS and to consider not only the child but also the family in order to address their unmet needs and provide holistic healthcare services.

Keywords Barth syndrome, BTHS, Family functioning, Functional performance, Health-related quality of life, And HRQoL

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Background

Barth syndrome (BTHS) is a rare X-linked genetic disorder that primarily affects males, resulting from a mutation in the TAFAZZIN gene (*TAZ*, G4.5) located on chromosome Xq28 [1]. Its primary symptoms include cardiomyopathy, neutropenia, muscle weakness, growth delays, and difficulty with physical exertion [1–2]. In the United States, BTHS occurs in approximately 1 in every 1,000,000 births [3]. BTHS causes problems in muscular and neurological function due to mitochondrial impairment and low levels of cardiolipin [4–5]. Mitochondria, the main energy producers in cells, require cardiolipin to maintain their structure and function; therefore, a deficiency in cardiolipin causes mitochondria to produce fewer of the essential lipids necessary for stability [6–7]. This leads to muscle weakness, limited exercise tolerance, and a quick onset of fatigue in individuals with BTHS [8]. As a result, strenuous activities can be particularly challenging for children with BTHS [8–9]. The existing literature extensively documented the genetic basis and natural history of BTHS [4–5, 8] and the physical symptoms and functional performance of individuals with BTHS [9–12]. However, there is currently no cure or targeted treatment for BTHS yet, which leads to parents and families of children with BTHS requiring continuous support to help manage the daily challenges they face, potentially impacting parental quality of life and family functioning.

Quality of life (QoL), often used interchangeably with health-related quality of life (HRQoL), is one of the key outcomes [13]. QoL reflects "an individual's perception of their position in life in the context of the culture and value systems in which they live and in relation to their goals, expectations, standards, and concerns" [14]. HRQoL is a multidimensional construct that encompasses physical, emotional, cognitive, and social functioning, all of which contribute to a person's overall health and well-being [15–16]. Several studies have shown that a child's health condition affects parental HRQoL and family functioning [17–20]. Examining family impacts, such as parental HRQoL and family functioning, can help healthcare providers better understand the unmet needs of families managing challenges related to their child's health condition [21]. However, investigating how a child's illness, its progression, and its impact on the family interrelate is a complex and multifaceted process [22].

Researchers have sought to identify factors influencing the adaptation and adjustment processes of families with children with disabilities over time. The Double ABCX model [23] describes family recovery from a major event (e.g., a child's disease) and the adaptation and adjustment processes of the families. The factors of this model include variables related to a child's disease, family characteristics, and healthcare services. In Fig. 1, the A factor is the pile-up of demands (e.g., those associated with the child's disease), and the B factor indicates adaptive resources, such as healthcare services. The C factor reflects the family's perception of A, B, and the major event (e.g., the child's disease). Adaptation (X) is the outcome of the family's responses to these factors above. The Double ABCX model has been used in research exploring

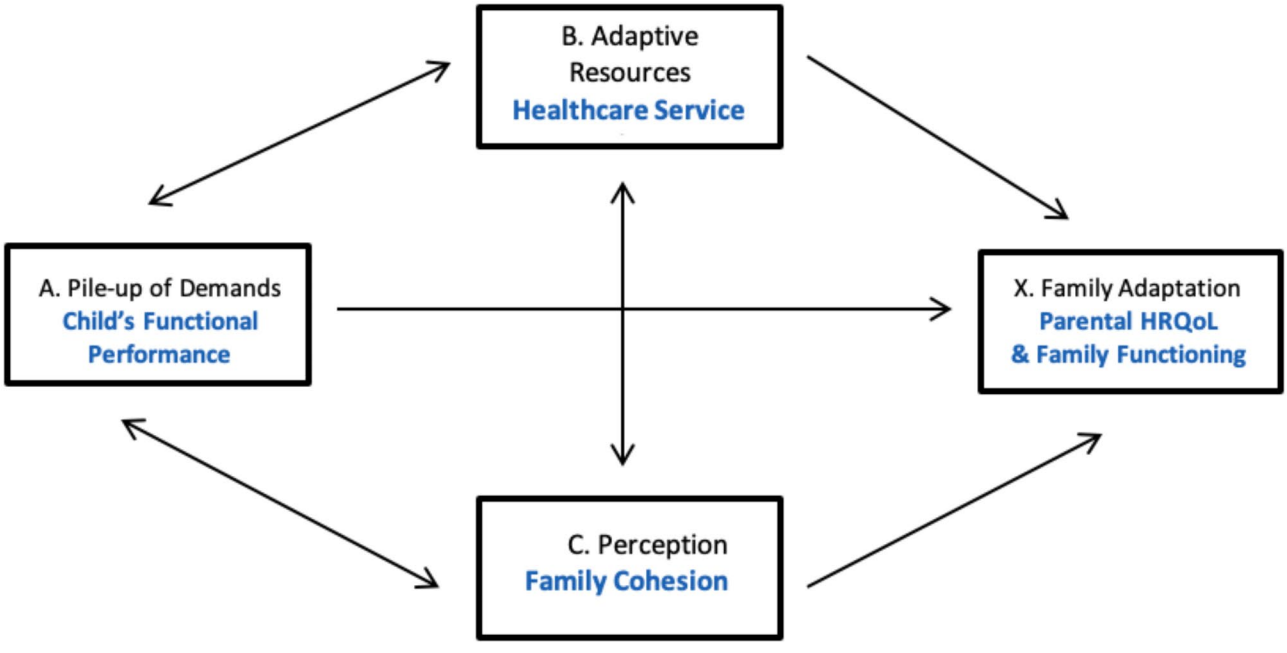

Fig. 1 The double ABCX model

factors that influence families with children with disabilities [17, 24–26].

Currently, there is little evidence about the impact of raising children with BTHS on parents and families. Providing this evidence is crucial for understanding the daily challenges these parents and families encounter and for delivering services and support to address their unmet needs. Therefore, this study applied the Double ABCX model to examine how a child's functional performance, family cohesion, and satisfaction with healthcare affect the parents of children with BTHS. Additionally, the health-related quality of life (HRQoL) and family functioning between parents of children with BTHS and those of unaffected children were compared in this study.

Methods

Participants

Thirty-three parents of children with BTHS and 31 parents of age-matched, unaffected children participated in this study. Eligibility criteria included: (1) being a parent of a child or youth diagnosed with BTHS or of an unaffected child between the ages of 4 and 19, and (2) speaking English and reading at an 8th-grade level. Parents of children with BTHS were recruited in collaboration with the Barth Syndrome Foundation, who attended the 7th and 8th Barth Syndrome Foundation International Scientific, Medical & Family Conferences. Parents of unaffected children were recruited through word of mouth, flyers posted in local areas, Facebook groups, and HealthStreet in Gainesville, FL. Both groups completed the PedsQL™ Family Impact Module and a demographic information form. Only parents of children with BTHS completed the Modified Barth Index, Family Adaptability and Cohesion Evaluation Scale-IV, and PedsQL™ Healthcare Satisfaction Generic Module. This study was approved by the University of Florida Institutional Review Board. Participating parents provided written informed consent.

Instruments

The Pedsql™ Family Impact Module

The PedsQL™ Family Impact Module (PedsQL™ FIM) assesses the impact of pediatric chronic health conditions on parental HRQoL and family functioning [27]. This self-report questionnaire consists of 8 subscales with 36 items: (1) Physical Functioning, (2) Emotional Functioning, (3) Social Functioning, (4) Cognitive Functioning, (5) Communication, (6) Worry, (7) Daily Activities, and (8) Family Relationships. Each item is scored on a 5-point scale (never a problem = 0, almost never = 1, sometimes = 2, often = 3, almost always = 4) and transformed to a 0 to 100 scale (never a problem = 100, almost never = 75, sometimes = 50, often = 25, almost always = 0). The total score of the PedsQL™ Family Impact Module is calculated by averaging the 36 items. Within this module, the Parental HRQoL score is calculated by averaging items from the Physical Functioning, Emotional Functioning, Social Functioning, and Cognitive Functioning subscales, while the Family Functioning score is derived from the Daily Activities and Family Relationships subscales. Higher scores indicate better parental HRQoL and family functioning.

The Modified Barthel Index

The Modified Barthel Index (MBI) is a 10-item questionnaire that measures a child's performance in activities of daily living (i.e., the child's functional ability) [28]. The MBI consists of 10 subscales: (1) Personal Hygiene, (2) Bathing Self, (3) Feeding, (4) Toileting, (5) Stair Climbing, (6) Dressing, (7) Bowel Control, (8) Bladder Control, (9) Ambulation/Wheelchair, and (10) Chair/Bed Transfers. The MBI is administered by a therapist or family member/friend based on their observations of the child's daily life. Each item is scored from "unable to perform task" to "fully independent" (0–5 for Personal Hygiene and Bathing Self; 0–10 for Feeding, Toilet Transfer, Dressing, Bladder Control, Bowel Control, and Stair Climbing; 0–15 for Chair/Bed Transfer and Ambulation/Wheelchair). The total score is calculated by summing the 10 items and ranges from 0 to 100. Higher scores indicate greater functional ability or independence in activities of daily living.

The Family Adaptability and Cohesion Evaluation Scale-IV

The Family Adaptability and Cohesion Evaluation Scale-IV (FACES-IV) is a 42-item questionnaire that measures the dimensions of family cohesion and family flexibility [29]. It consists of 6 subscales: (1) Balanced Cohesion, (2) Balanced Flexibility, (3) Disengaged, (4) Enmeshed, (5) Rigid, (6) Chaotic. Each item is rated on a 5-point scale (strongly disagree = 1, generally disagree = 2, undecided = 3, generally agree = 4, strongly agree = 5). Raw subscale scores are converted to percentiles using a percentile conversion chart. The total Circumplex Ratio is computed from the 6 subscales as follows, with values above 1 indicating a more balanced/healthy family dynamic.

Cohesion Ratio = Balanced Cohesion / ((Disengaged + Enmeshment) / 2).

Flexibility Ratio = Balanced Flexibility / ((Rigid + Chaotic) / 2).

Total Circumplex Ratio = (Cohesion Ratio + Flexibility Ratio) / 2.

The Pedsql™ Healthcare Satisfaction Generic Module

The PedsQL™ Healthcare Satisfaction Generic Module (PedsQL™ HS) measures healthcare satisfaction [30]. It consists of 6 subscales with 27 items: (1) Information (5 items), (2) Inclusion of Family (4 items), (3) Communication (5 items), (4) Technical Skills (3 items), (5) Emotional Needs (4 items), and (6) Overall Satisfaction (3 items). Each item is scored on a 5-point scale (never = 100, sometimes = 75, often = 50, always = 25, not applicable = 0). The total PedsQL™ HS score is calculated by averaging the six subscales. Higher scores indicate greater satisfaction with healthcare.

The demographic information form

The demographic information form, generated by the investigators of this study, consisted of (1) the child's date of birth and age, (2) the parent's age, (3) marital status, (4) race, (5) employment, (6) level of education, and (7) family income.

Table 1 Summary statistics for child and parent demographic variables

Characteristics	BTHS ($n=33$)	Unaffected ($n=31$)	*p*-value
Child Age	11.53 (4.65)	11.44 (3.94)	0.934
Age range	4–19	5–19	
Median	11.75	11.11	
Child Sex			
Male	33	31	
Parent Age	42.45 (6.97)	42.90 (6.60)	0.793
Age range	28–54	30–54	
Median	44	44	
Parent Race			0.0002
Non-Hispanic white	30 (90.91)	15 (48.39)	
Other	3 (9.09)	16 (51.61)	
Parent Education			0.0002
Graduate or professional degree	9 (27.27)	23 (74.19)	
Less than graduate or professional degree	24 (72.73)	8 (25.81)	
Marital Status			0.9247
Married	29 (87.88)	27 (87.10)	
Other	4 (12.12)	4 (12.90)	
Employment Status			0.1016
Full-time	18 (54.55)	23 (74.19)	
Less than full-time	15 (45.45)	8 (25.81)	
Family Income			0.9938
$80,000 or above	16 (48.48)	15 (48.39)	
Less than $80,0000	17 (51.52)	16 (51.61)	

Note. Mean (standard deviation) for child age; *n* (%) for the others (e.g., child sex, parent race, etc.); categorical variables were analyzed by Chi-square test. Continuous variables were analyzed by independent *t*-test

Data analysis

The Shapiro-Wilk was used to test the normality of the data. For child and parent demographic variables, an independent samples *t*-test was used for continuous variables (e.g., child's age), and the Chi-square test was used for categorical variables (e.g., parent education). The PedsQL™ FIM scores were compared between the parents of children with BTHS and parents of unaffected children using an independent samples *t*-test and an adjusted method. In the adjusted method, propensity score matching with inverse probability of treatment weighting (PS-IPTW) [31–32] was applied to control for demographic differences between the two groups when comparing the mean scores of the PedsQL™ FIM. The correlation between parental HRQoL and family functioning (PedsQL™ FIM) with a child's functional performance (MBI), family cohesion (FACES-IV), and satisfaction with healthcare (PedsQL™ HS) was analyzed using the Spearman correlation coefficient. Regression analysis was conducted to examine variables that significantly predict parental HRQoL and family functioning in the parents of children with BTHS. The independent variables were the child's functional performance (MBI), family cohesion (FACES-IV), and satisfaction with healthcare (PedsQL™ HS). The dependent variable was parental HRQoL and family functioning (PedsQL™ FIM). Statistical analyses were performed using SAS statistical software version 9.4 (SAS Institute, Inc.). The alpha level was set at 0.05.

Results

Summary statistics for child and parent demographic variables

Sixty-four parents participated in this study, including 33 parents of children with BTHS and 31 parents of unaffected children. Table 1 displays summary statistics for child and parent demographic variables. Parent race ($p=.0002$) and parent education ($p=.0002$) were significantly different between the two groups. Parent race differed significantly, with 30 (90.91%) parents being non-Hispanic White in the BTHS group compared to 15 (48.39%) in the unaffected group. In addition, 23 (74.19%) parents in the unaffected group had a graduate or professional degree, compared to 9 (27.27%) in the BTHS group.

Parental HRQoL and family functioning between the two groups

In the PedsQL™ FIM, all subscale scores, parental HRQoL, family functioning, and the total score between the parents of children with BTHS and parents of unaffected children were significantly different (Table 2). The Worry subscale demonstrated the highest mean difference between the two groups, followed by the Communication subscale. After controlling for the significantly

Table 2 Mean score differences of the pedsql™ family impact module between the unaffected and BTHS groups

	Unadjusted[a]				PS-IPTW adjustment[b]	
	BTHS (*n* = 33)	Unaffected (*n* = 31)	Mean difference (SE)	*p*	Mean difference (SE)	*p*
Parental HRQoL	61.40 (21.59)	76.23 (15.14)	14.82 (4.68)	0.002	12.92 (4.40)	0.005
Physical Functioning	60.61 (26.29)	75.27 (19.36)	14.66 (5.80)	0.014	9.70 (4.98)	0.057
Emotional Functioning	59.39 (21.50)	73.87 (18.65)	14.47 (5.04)	0.005	15.93 (5.43)	0.005
Social Functioning	61.10 (27.12)	78.02 (19.76)	16.92 (5.96)	0.006	16.07 (5.79)	0.007
Cognitive Functioning	63.79 (25.28)	77.74 (16.42)	13.95 (5.29)	0.011	10.27 (5.17)	0.052
Communication	59.09 (17.23)	88.17 (12.69)	29.08 (3.80)	< 0.001	27.49 (4.05)	< 0.001
Worry	47.73 (19.33)	86.13 (21.16)	38.40 (5.06)	< 0.001	42.41 (4.19)	< 0.001
Family Functioning	57.23 (21.46)	73.79 (19.46)	16.56 (5.10)	0.001	16.58 (5.28)	0.002
Daily Activities	49.75 (31.56)	71.77 (21.70)	22.02 (6.73)	0.002	23.62 (6.87)	0.001
Family Relationships	62.54 (20.72)	75.81 (19.28)	13.26 (5.01)	0.01	11.23 (5.24)	0.037
Total Score	58.37 (18.03)	78.35 (14.75)	19.97 (4.13)	< 0.001	19.46 (3.92)	< 0.001

Note. *M* (*SD*) for unaffected and BTHS; propensity score matching with inverse probability of treatment weighting (PS-IPTW); standard error (*SE*)

[a] Independent-samples *t*-test between the unaffected and BTHS groups

[b] After applying propensity score matching with inverse probability of treatment weighting, all demographics were balanced between the unaffected and BTHS groups

Table 3 Correlation analysis in the BTHS group

	PedsQL™ Family Impact Module		
	Parental HRQoL	Family Functioning	Total Score
MBI	0.561**	0.342	0.547**
FACES-IV	0.303	0.414*	0.296
PedsQL™ Healthcare Satisfaction	0.175	0.314	0.264

*$p < .05$

**$p < .01$

Table 4 Regression analysis in the BTHS group

Variable	Parental HRQoL and family functioning		
	B	*SE*	*p*
Child's Functional Ability	0.586	0.167	0.0008*
Family cohesion	2.745	2.809	0.1589
Satisfaction with healthcare	0.157	0.145	0.8028
R^2	0.385		
F	6.047*		

SE = standard error

*$p < .05$

different demographic variables (parent race and education) using the PS-IPTW adjusted method, the Physical Functioning and Cognitive Functioning subscales were not significantly different between the two groups.

Correlation analysis in the BTHS groups

The MBI and PedsQL™ HS did not meet the normality test ($p < .05$); therefore, the Spearman correlation coefficient, a non-parametric approach, was used in this study (Table 3). In the correlation analysis, the child's functional performance showed a positive moderate correlation with parental HRQoL ($r(33) = 0.561$, $p < .001$) and the total score ($r(33) = 0.547$, $p < .001$). Family cohesion demonstrated a positive moderate relationship with family functioning ($r(33) = 0.414$, $p = .016$).

Regression analysis in the BTHS group

Multiple regression analysis was conducted to determine the predictors of parental HRQoL and family functioning in parents of children with BTHS (Table 4). In this model, 38.5% of parental HRQoL and family functioning in parents of children with BTHS was significantly explained by the three independent variables ($F(3, 32) = 6.047$, $p = .003$). The child's functional performance factor significantly predicted parental HRQoL and family functioning in the parents of children with BTHS ($p = .0008$). However, family cohesion and satisfaction with healthcare variables were not significant predictors in this model.

Discussion

By applying the Double ABCX model, this study investigated the impact of a child's functional performance, family cohesion, and satisfaction with healthcare on parents of children with BTHS. The HRQoL and family functioning of parents of children with BTHS were compared to those of parents of unaffected children. Parents of children with BTHS demonstrated significantly lower HRQoL and family functioning than those of unaffected children, which is consistent with previous studies conducted with other clinical populations [17–18, 21, 33–34]. Among the PedsQL™ FIM scales, the Worry and Communication subscales showed the highest mean differences between the two groups. These differences reflect worries about their child's future and the impact of others on the child's condition, as well as feelings that others do not understand the family's situation and difficulties talking to others about their child's health.

According to the correlation and regression analyses, the child's functional performance was a significant factor affecting parental HRQoL and family functioning of parents of children with BTHS. This finding is consistent with previous studies that the severity of a child's function significantly predicted parental HRQoL [17, 35]. This is not unexpected, as BTHS represents muscle weakness, which could affect overall physical function. Due to their physical symptoms, children with BTHS face challenges in participating in strenuous physical activities [8, 9]. These physical symptoms may restrict their participation in important and meaningful age-related activities, as well as in everyday activities across home, school, and community settings. Therefore, an in-depth exploration of the level of participation in everyday activities for children with BTHS and its impact on their families is crucial.

Olson [29] noted that most FACES-IV scores, which measure family cohesion, typically range from 0 to 2. However, in this study, the majority of parents of children with BTHS scored higher than 2 on the FACES-IV. This indicates that these parents maintained a high level of family cohesion, resulting in less variability in the data for the correlation and regression analyses. Since the parents in this study had high levels of education and middle to upper-middle-class incomes, this might contribute to their heightened capacity to cope with their child's diseases [36]. It is reported that raising children with disabilities may strengthen parents' inner resilience and improve family cohesion [37]. By gaining a deeper understanding of their child's disability, parents may experience positive personal growth and improved relationships [38]. Therefore, the findings from this study may indicate that parents of children with BTHS have adjusted to managing their child's disease conditions.

For families providing lifelong care for their children, the caregiving burden often increases as the disease progresses [39]. As such, ongoing disease management support, updated information, and education are essential. Additionally, families may benefit from counseling to improve their adaptive skills for managing the growing burden and responsibilities [18]. Health professionals should consider offering family education, coaching on caregiving techniques and strategies, encouraging participation in activities outside the home, and connecting them with others caring for children with disabilities for additional support [40–41]. Most importantly, to offer the most effective services to families of children with rare diseases, health professionals must understand both the symptoms of pediatric rare diseases and the experiences and unmet needs of the families, ensuring timely and necessary support.

This study has several limitations. Participants were recruited through the Barth Syndrome Foundation (BSF), which could potentially result in a selection bias. The BSF offers parent support groups and valuable up-to-date resources, which are crucially important to patient care and parental advocacy. Parents who were more engaged with the BSF may be more resilient and better prepared to handle caregiving demands, creating a sample that may not fully reflect all parents of children with BTHS. Additionally, most participants were non-Hispanic White, with middle to upper-middle-class incomes and high education levels, restricting the diversity of perspectives and experiences. Families from various racial, ethnic, and socioeconomic backgrounds may have unique caregiving experiences, which could affect their HRQoL and family functioning.

Future research should aim to include a more diverse sample representing a broader range of income, education, socioeconomic status, and racial/ethnic groups to ensure that findings are applicable to a broader population of parents of children with BTHS. Moreover, it is important to consider a longitudinal approach to examine caregiving impacts on the family over time, such as challenges as the child ages or the disease progresses. Lastly, future research might consider incorporating self-reported data with objective measures or multiple informants (e.g., healthcare providers) to provide a more comprehensive view of the caregiving experience.

Conclusion

The parents of children with BTHS reported lower HRQoL and family functioning compared to those of unaffected children, and a child's functional performance was a significant factor affecting parents and families. The findings of this study lay the groundwork for exploring the impact of raising children with BTHS on parental HRQoL and family functioning. It is crucial for health

professionals to understand the clinical features of BTHS and to consider not only the child but also the family in order to address their unmet needs and provide holistic healthcare services. They should implement practical strategies to support positive family functioning and well-being in daily life.

Abbreviations

BTHS	Barth Syndrome
HRQoL	Health-related quality of life
QoL	Quality of life

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Author contributions
YL designed and collected data for this study. IH and YL analyzed and interpreted the data. AH and YL wrote the manuscript, and YL was a major contributor in writing the manuscript. All authors read and approved the final manuscript.

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Data availability
The authors confirm that all data generated or analyzed during this study. The datasets used for this study are available from the corresponding author upon reasonable request.

Declarations

Ethics approval and consent to participate
This study was approved by the University of Florida Institutional Review Board. Participating parents provided written informed consent.

Consent for publication
Not applicable.

Competing interests
The authors declare that they have no competing interests.

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