

Diagnostic criteria for Joubert syndrome include the presence of a characteristic “molar tooth” sign on magnetic resonance imaging of the brainstem isthmus.² As of this publication, four loci and two genes have been implicated in disorders that share the molar tooth sign malformation.²³ The findings from the present study provide information about oromotor and communication profiles that might shed light on the disorder and provide a more complete understanding of malformed neuromotor circuits in Joubert syndrome.

In future research, these fine-grained profiles will need to be examined in various hindbrain malformation disorders to uncover molecular and structural central nervous system etiologies. Further, it is necessary to determine if the degree of apraxia in speech is related to a broader motor disturbance in Joubert syndrome, encompassing other systems, such as oculomotor and fine motor abilities. In addition, analyses of the interplay among gesture, language, cognition, and motor ability are crucial to understanding the neurobehavioral profile of Joubert syndrome.

Barbara A. Braddock, MHS
Department of Psychological Sciences
University of Missouri-Columbia
Columbia, Missouri
 Janet E. Farmer, PhD
 Kathleen M. Deidrick, PhD
Department of Health Psychology
University of Missouri-Columbia
Columbia, Missouri
 Jana M. Iverson, PhD
Department of Psychology
University of Pittsburgh
Pittsburgh, Pennsylvania
 Bernard L. Maria, MD, MBA
Department of Pediatrics
Medical University of South Carolina
Charleston, South Carolina

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Address correspondence to Dr Janet Farmer, Department of Health Psychology, University of Missouri-Columbia, One Hospital Drive, DC116.88, Columbia, MO 65212. Tel: 573-882-8847; fax: 573-884-4540; e-mail: farmerje@health.missouri.edu.

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Parenting Stress and Its Relationship to the Behavior of Children with Joubert Syndrome

ABSTRACT

This study describes the relationship between parenting stress and behavior in children with Joubert syndrome, a rare genetic neurodevelopmental disorder. Parents ($N = 43$) reported severely impaired child adaptive behaviors. Most children did not show maladaptive behaviors, but a subset of approximately 20% displayed significant problems in areas such as inattention, overactivity, social withdrawal, and atypical behaviors. Mothers (59%) and fathers (40%) reported elevated levels of parenting stress. A hierarchical regression, including demographics, adaptive behavior, and maladaptive behavior, predicted 67% of the variance in mothers’ stress and 40% of the variance in fathers’ stress. Maladaptive behaviors uniquely contributed to maternal and paternal stress. The child’s adaptive behavior level contributed significantly to parenting stress for mothers but not for fathers. Findings provide a better understanding of the impact of child behavior on parents car-

ing for a child with Joubert syndrome. (*J Child Neurol* 2006;21:163–167; DOI 10.2310/7010.2006.00019).

Joubert syndrome is a rare autosomal recessive disorder characterized by cerebellar and brainstem abnormalities, including an increase in the depth of the posterior interpeduncular fossa, thickened superior cerebellar peduncles, and vermian hypoplasia.¹ In addition to these brain abnormalities, clinical features often include hypotonia and ataxia and can sometimes include abnormal breathing, visual problems, and hepatic or renal involvement. Global developmental delay is a prominent feature of Joubert syndrome.^{1,2}

Little is known about the experiences of caregivers for children with Joubert syndrome. In a study that involved mostly mothers of children with Joubert syndrome, Luescher et al described high levels of caregiver burden and identified a subgroup of mothers with symptoms of depression.³ This finding is consistent with previous research indicating higher levels of parenting stress, psychiatric symptoms, and caregiving burden among parents of children with physical or intellectual disabilities compared with parents of children without such chronic conditions.^{4–6} Contrary to their hypotheses, Luescher and colleagues found that caregiver burden and depression were not associated with the severity of developmental delays identified on a measure of child adaptive functioning.³ They posited that caregiver burden might be more associated with family characteristics such as parental coping skills and level of family functioning than with child characteristics. Other explanations are possible, however, because a growing body of research indicates that child maladaptive behavior can be an important predictor of stress for parents of children with neurodevelopmental disabilities.^{7–9} Evidence from small sample studies of children with Joubert syndrome suggests that they are at increased risk of problem behaviors that can be stressful for parents.^{10,11}

The purpose of the current study was to investigate the relationship between child behavior and parent stress in Joubert syndrome. We hypothesized that child maladaptive behavior will predict parent stress even after the relationship between parent stress and child adaptive behavior is accounted for. The relationship between child behaviors and parenting stress was examined separately for mothers and fathers because research suggests that fathers can respond differently to caregiving demands.

Methods

Participants included 43 of 47 (91%) parents of children with Joubert syndrome who attended a biannual national conference on Joubert syndrome. Children ranged in age from 5 months to 16 years 3 months. The mean age was 6 years 7 months (SD = 4 years 4 months). Sixty-one percent of children were male. With the exception of one African-American child, participants were Caucasian. Demographic information was available for a subset of 32 mothers and 31 fathers, with slight variability in the responses for each demographic variable. The mean age of mothers was 36.7 years (SD = 5.1 years) and of fathers was 39.8 years (SD = 5.5 years). More than half of the mothers (63%) and fathers (53%) had achieved a bachelor's or graduate degree. Most of the other parents had completed high school, with only a few who had not (3% of mothers, 7% of fathers). For 65% of children, the mother was the primary caregiver (father primary caregiver, 7%; caregiving shared by both parents, 26%).

As part of a larger multidisciplinary study, caregivers completed questionnaires regarding child behavior and parenting stress. Parents were invited to complete the child behavior measures together for their child but to provide independent ratings of their parenting stress. Researchers conducted the study according to a protocol approved by the Institutional Review Board. The measures administered are described below.

Scales of Independent Behavior-Revised

Parents rated the adaptive ($n = 43$) and maladaptive behavior ($n = 41$) portions of the checklist form of the Scales of Independent Behavior-Revised,¹² with 92% of forms completed by mothers. The Adaptive Behavior Scales provide standard scores (mean = 100, SD = 15) for overall functioning and across

four specific skill domains: motor, social interaction/communication, personal living, and community living. The Maladaptive Behavior Indexes require parents to rate the frequency and severity of eight behaviors in three domains (Table 1). Standard scores for the Maladaptive Behavior Indexes (mean = 0; SD = 10) were transformed to T-scores (mean = 50, SD = 10) for ease of interpretation. Normative data for both indices are available for all ages, including infants.

Behavior Assessment System for Children-Parent Rating Scale

The Behavior Assessment System for Children-Parent Rating Scale¹³ is a questionnaire that measures behavioral and emotional problems in children ages 2 years 6 months to 18 years. Parents with children in this age range ($n = 31$) rated statements about child behaviors on a 4-point Likert scale (1 = never to 4 = almost always). Scales from this standardized measure are presented in Table 1 (T-score mean = 50, SD = 10).

Parenting Stress Index-Short Form

The Parenting Stress Index-Short Form¹⁴ is a 36-item questionnaire that assesses parent stress level in relation to a specific child. Ninety-eight percent of mothers ($n = 39$) and 75% of fathers ($n = 30$) independently completed the measure on 40 children with Joubert syndrome, resulting in 69 total parent ratings. Both parents rated their stress in relation to 29 children; only the mother's report was available for 10 children, and only the father's report was available for one child. This measure yields a composite index and three subscales (T-score mean = 50, SD = 10): the Parental Distress subscale (related to parent factors such as depression or social support), the Parent-Child Dysfunctional Interaction subscale, and the Difficult Child subscale.

Results

On the Scales of Independent Behavior-Revised Adaptive Behavior Scales, overall adaptive skills were severely impaired on average (Broad Independence median = 29.00, range = 0–91). Most of the children (84%; $n = 36$) functioned in the severely impaired range compared with same-age peers, and 16% ($n = 7$) performed in the borderline to average range. Younger children were more likely to be higher functioning ($r = -.39$, $P = .01$), but three of the children who were higher functioning were between the ages of 5 and 10 years. For the entire sample, subscale ratings indicated mild impairments in social/communication skills (median = 59, range = 0–118), moderate impairments in community living skills (median = 44, range = 0–100), and severe impairments in motor (median = 23, range = 0–93) and personal living skills (median = 31, range = 0–93).

As shown in Table 1, mean ratings of maladaptive behavior problems on the Scales of Independent Behavior-Revised and the Behavior Assessment System for Children were within the average range. However, approximately 1 in 5 children were rated as showing clinically significant problems ($T \geq 65$) on the composite Maladaptive Behavior Index of the Scales of Independent Behavior-Revised, primarily owing to higher scores on the Internalizing Scale (eg, inwardly directed behaviors such as repetitive behavior, withdrawal, inattention, and self-injurious behavior). Additionally, approximately 20% of the children displayed behavior problems that were clinically significant ($T \geq 65$) in the areas of overactivity, atypical behaviors, withdrawal, and inattention on the Behavior Assessment System for Children.

On the Parenting Stress Index-Short Form, mothers reported overall parenting stress in the borderline clinically significant range on average (mean = 61.6, SD = 14.7). Fifty-nine percent of mothers described levels of total parenting stress that were above the cutoff for clinical significance (≥ 85 th percentile). Subscale analysis showed that this elevation was primarily due to the percentage of mothers with significant concerns related to child behaviors and perceived problems in the parent-child interaction (parent stress = 28%, difficult child = 63%, parent-child dysfunctional interaction = 55%). Fathers, on average, did not indicate clinically significant total parenting stress (mean = 57.8, SD = 11.2). However, 40% of fathers described levels of total stress above the clinical cutoff (≥ 85 th percentile), primarily owing to the percentage of fathers with elevations on the child behavior and parent-child interaction subscales (parent stress = 19%, difficult child = 40%,

Table 1. Mean Parent Ratings of Maladaptive Behaviors (T-Score Mean = 50, SD = 10) and Percentage of Children With Clinically Significant Behavior Problems

Assessments	Mean (SD)	T ≥ 60 (%)	T ≥ 65 (%)
Scales of Independent Behavior-Revised (n = 41)			
Maladaptive Behavior Index	57.44 (10.59)	37	20
Internalizing	55.68 (9.34)	37	22
Asocial	52.32 (11.34)	24	15
Externalizing	50.54 (11.83)	20	12
Behavior Assessment System for Children-Parent Rating Scale (n = 31)			
Externalizing problems	46.35 (12.49)	13	10
Hyperactivity	50.23 (13.09)	19	19
Aggression	44.71 (12.54)	19	10
Internalizing problems	45.97 (10.33)	10	10
Anxiety	40.77 (9.20)	3	3
Depression	48.19 (13.90)	19	13
Somatization	51.81 (10.11)	13	10
Atypicality	55.94 (11.49)	40	19
Withdrawal	52.74 (13.99)	26	23
Attention problems	55.87 (9.68)	29	19
Behavioral Symptoms Index	48.87 (12.01)	26	6

parent-child interaction = 41%). Maternal and paternal reports did not differ significantly for mean total parenting stress ($t [29] = .72, P = .48$) or for the likelihood of significant total parenting stress (Fisher's exact $P = .15$).

To examine predictors of maternal and paternal parenting stress, hierarchical multiple regression analyses were conducted in two phases. First, to reduce the number of predictor variables included in each regression analysis, the relationship between total parenting stress and predictor variables was examined using Spearman correlation coefficients for continuous variables (child and parent age, Scales of Independent Behavior-Revised Adaptive and Maladaptive Behavior Indexes) and Kruskal-Wallis tests for categorical variables (child gender, parent education). Significant correlations are shown in Table 2. Higher maternal stress was associated with older child age, lower levels of adaptive behaviors, and higher ratings of maladaptive behaviors. Fathers endorsed higher levels of stress for children with higher ratings of maladaptive behaviors and for girls compared with boys (Kruskal-Wallis $\chi^2 [1, n = 30] = 3.97, P = .05$). There was a trend toward a significant correlation between paternal stress and child age, with older age associated with more stress ($P < .06$). The Behavior Assessment System for Children Behavioral Symptoms Index was not included in these analyses owing to the lower number of parents who completed the measure ($n = 31$) and to its high correlation with the Scales of Independent Behavior-Revised Maladaptive Behavior Index ($r = .68, P = .0001$).

Next, two separate hierarchical regression analyses were conducted to examine predictors of parenting stress for mothers and fathers (Table

3). Predictor variables were entered in three steps: (1) demographics, (2) adaptive behaviors, and (3) maladaptive behaviors. For mothers, the final regression model accounted for 67% of variance in maternal parenting stress ($F(11,63) = 3.72, P = .0004$). For fathers, the regression model accounted for 40% of variance in paternal stress ($F(11,63) = 3.72, P = .0004$). Demographic variables together accounted for a significant percentage of variance in maternal and paternal stress in the first step, and child age was a unique predictor of stress at this step. Adaptive behaviors, entered in the second step, added significantly to the prediction of maternal stress but not paternal stress. Finally, the child's maladaptive behaviors contributed significantly to the prediction of stress for both mothers and fathers after demographics and adaptive behaviors were taken into account.

Discussion

This is the first study to examine parenting stress in both mothers and fathers of children with Joubert syndrome, extending previous research and adding to the sparse and mixed literature on fathers of children with developmental disabilities.¹⁵ Although many parents adjusted adequately to raising a child with Joubert syndrome, both mothers and fathers were at risk for high levels of parenting stress (59% of mothers and 40% of fathers). In addition, child maladaptive behavior problems emerged as a key determinant of parenting stress for both parents, even after accounting for child age, gender, and developmental level. These findings are consistent with previous

Table 2. Mean Maternal and Paternal Ratings and Intercorrelations for Parenting Stress Inventory and Predictor Variables

	Mean	Median	SD	1	2	3
Maternal correlations (n = 39)						
Maternal total parenting stress [†]	61.6	62.3	14.7	.48***	-.60****	.69****
Child age (mo)	76.5	61.1	50.8		-.36**	.50***
Broad Independence [‡]	29.9	29.0	29.9			-.23
Maladaptive Behavior Index [†]	57.4	56.0	10.4			
Paternal correlations (n = 30)						
Paternal total parenting stress [†]	57.8	58.1	11.2	.35*	-.28	.41**
Child age (mo)	73.5	62.6	49.3		-.48***	.48***
Broad Independence [‡]	37.2	35.0	26.7			-.22
Maladaptive Behavior Index [†]	56.5	56.0	9.8			
Child gender [§]						
Male (n = 18)	54.7	55.8	10.2			
Female (n = 12)	62.3	62.3	11.5			

* $P < .10$; ** $P < .05$; *** $P < .01$; **** $P < .001$.

[†]T-score mean = 50, SD = 10; higher scores represent worse ratings.

[‡]Standard score mean = 100, SD = 15; higher scores represent better ratings.

[§]There was a significant difference in total paternal stress by gender ($P = .05$).

Table 3. Hierarchical Regression Models Predicting Total Parenting Stress

Step and Predictor Variables	Maternal Parenting Stress				Paternal Parenting Stress			
	β	R ²	ΔR^2	ΔF	β	R ²	ΔR^2	ΔF
Step 1: Demographics		.33**				.22*		
Age	.58***				.35*			
Gender	-.07				.29			
Step 2: Adaptive behavior		.52***	.19**	13.08**		.29*	.07	2.45
SIB-R Broad Independence	-.46**				-.30			
Step 3: Maladaptive behavior		.67***	.15**	14.73**		.40*	.11*	4.36*
SIB-R Maladaptive Behavior Index	.49**				.39*			

SIB-R = Scales of Independent Behavior-Revised.

 β = standardized regression coefficient; R² = amount of variance accounted for; Δ = change.* $P < .05$; ** $P < .01$; *** $P < .001$.

research suggesting increased vulnerability to psychological stress in mothers and fathers of children with chronic conditions, especially when the child exhibits maladaptive behaviors.^{6-9,16} This relationship is likely bidirectional, with stressed parents experiencing more difficulty in managing behavior and child behavior problems increasing challenges to the caregiver.⁹

Parents of older children reported higher levels of stress, a finding receiving mixed support in other research on neurodevelopmental disorders.^{7,17} Older children with Joubert syndrome exhibited higher levels of behavior problems and poorer adaptive functioning relative to peers, perhaps mediating the relationship between child age and parent stress. It is also possible that higher levels of stress resulted from the cumulative demands of parenting a child with a disability over several years.

Despite similarities in their stress level, mothers and fathers differed somewhat in the pattern of predictors of parenting stress. Child adaptive behaviors only predicted maternal stress. Mothers were the primary caregivers for the majority of children in this study, so developmental delays and functional skills deficits likely had a greater impact on mothers compared with fathers.¹⁸⁻²⁰ Potentially important predictors of fathers' stress were not included in the current study, as indicated by the fact that 40% of the variance in paternal stress was accounted for by the regression model in contrast to 67% of maternal stress. A review of other investigations of paternal stress suggests that financial worries, time spent at work, marital functioning, and coping skills might be important in predicting the impact of parenting a child with developmental disabilities on fathers.⁷ Continued study of factors related to caregiver stress and distress will allow for a more complete understanding of family members' experiences and should lead to improvements in family supports.

This study replicates and extends previous findings about the neurobehavioral characteristics of children with Joubert syndrome using the largest sample studied to date and well-standardized assessment instruments. Children with Joubert syndrome typically showed severe global delays in development.^{3,10,21} However, a small group of children in this study (16%) demonstrated less severe delays, which is consistent with previous reports that there might be a distinct subgroup of children with greater functional potential.^{3,22} In the area of maladaptive behaviors, children with Joubert syndrome were well adjusted on average compared with same-age peers. Yet approximately 1 in 5 children showed significant behavioral concerns related to inattention, overactivity, social withdrawal, and atypical behaviors (eg, repetitive behaviors, self-injury). The type and degree of problems exhibited by children with Joubert syndrome are similar to those observed in samples of children diagnosed with mental retardation of mixed etiology.^{12,13,23,24}

The reason for the behavioral heterogeneity in this sample is unclear, but it is likely determined by multiple factors. For example, the profile of child adaptive and maladaptive behaviors might be related to the degree of cerebellar dysfunction²⁵⁻²⁹ and/or cognitive impairment. Variability in the sample might also be due to the difficulty of distinguishing classic Joubert syndrome from other closely related hindbrain malformation syndromes³⁰ or to concomitant medical or neurologic conditions. In addition, different presentations might be due to contextual factors, including family environment, parent management strategies, and receipt of appropriate educational, therapeutic, psychopharmacologic, and behavioral interventions (eg, applied behavior analysis).

Several limitations to this study suggest future research directions. First, owing to the study's correlational nature, it is not possible to understand the direction of the relationship between parent stress and child behavior. Longitudinal research is necessary to examine the direction of this relationship and to provide more information about child development and parent adjustment over time. Second, the study relies on parent report measures to assess both child behaviors and parent stress. Although parental perceptions are important, using only parent reports can result in shared method variance, inflating the relationship between parental reports of stress and child behavior. Furthermore, because mothers completed behavioral questionnaires for most of the children, the stronger relationship between child behavior and parent stress for mothers than for fathers could be explained by a relatively higher degree of shared method variance. The study could be further strengthened by adding other measures of child behavior (eg, teacher reports) and/or direct observation of parent-child interactions in addition to parent report. Third, the current convenience sample consisted of a highly educated group of middle class, Caucasian families who were able to attend the Joubert syndrome conference. This approach to participant recruitment might limit the generalizability of study findings, but it is a common method used in research on rare conditions, and it allowed us to identify a sizable number of children with Joubert syndrome.

To promote continuing research advances, a national registry for children with Joubert syndrome and related hindbrain disorders is currently undergoing development with support from the National Institute of Neurologic Disease and Stroke and the Joubert Syndrome Foundation. The registry will promote the collection of a uniform set of medical, cognitive, and behavioral data for children diagnosed with Joubert syndrome. It also will encourage families who are unable to travel to the biannual conference to participate in research, enhancing the generalizability of studies conducted on this rare disorder. Improved understanding of relationships among neurologic, behavioral, and contextual factors will enhance outcomes for children with Joubert syndrome and will serve as a model to advance knowledge about brain-behavior relationships in other neurodevelopmental disorders.

Janet E. Farmer, PhD
 Kathleen M. Deidrick, PhD
Department of Health Psychology
University of Missouri-Columbia
Columbia, Missouri
 Jill C. Gitten, PhD
Department of Psychology
Brown University
Providence, Rhode Island
 Eileen B. Fennell, PhD
Department of Clinical and Health Psychology
University of Florida, Gainesville
Gainesville, Florida
 Bernard L. Maria, MD
Department of Pediatrics
Medical University of South Carolina
Charleston, South Carolina

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Address correspondence to Dr Janet Farmer, Department of Health Psychology, University of Missouri-Columbia, One Hospital Drive, DC116.88, Columbia, MO 65212. Tel: 573-882-8847; fax: 573-884-4540; e-mail: farmerje@health.missouri.edu.

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Fat Intolerance in Developmentally Impaired Children With Severe Feeding Intolerance

ABSTRACT

Children with a variety of genetic, metabolic, and neurologic disorders can suffer from severe feeding intolerance that is unresponsive to medical, surgical, and nutritional therapy. Developmentally disabled tube-fed children with severe upper gastrointestinal symptoms that persisted after fundoplication who were unresponsive to all medical, surgical, and nutritional interventions underwent a thorough gastrointestinal evaluation, including gastroscopy, pH-metry, upper gastrointestinal barium series, and gastric emptying studies. They were placed on a low-fat diet, and the symptoms before and after the diet were compared. The patients were then rechallenged with incremental increases in fat until the symptoms recurred or the patients reached their former fat concentration. Six children meeting the study criteria were evaluated. Four of these patients had a significant improvement in symptoms, oral intake and feeding tolerance with a decrease in fat intake, and relapse of symptoms when fat calories were increased. Improvement occurred in children who had been intolerant to duodenal feeding. We were subsequently able to wean two children from tube feeding. Dietary fat can provoke upper gastrointestinal symptoms in children with gastric and intestinal dysmotility. Short-term manipulation of dietary fat intake can improve tolerance to feeding. (*J Child Neurol* 2006;21:167–170; DOI 10.2310/7010.2006.00018).

Vomiting and feeding intolerance are common problems in infancy. These symptoms are often related to gastroesophageal reflux and usually improve

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