

# The Multifaceted Burden Experienced by Caregivers of Individuals With Bardet-Biedl Syndrome: Findings From the CARE-BBS Study

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## DISCLOSURE STATEMENT

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- I have the following potential conflicts of interest to report:
  - Research Contracts
  - Consulting
  - Employment in the Industry
  - Stockholder of a healthcare company
  - Owner of a healthcare company
  - Other(s) – advisory board speaker for Rhythm Pharmaceuticals, Inc.

# Caregivers of Individuals With BBS Face Substantial Burden

- BBS is a rare genetic disease associated with retinal dystrophy, polydactyly, developmental delays, renal abnormalities, hyperphagia, and early-onset obesity<sup>1</sup>
- Hyperphagia and obesity in particular place a substantial burden on those living with BBS and their families<sup>2</sup>
- The extent of this burden and its impact on the well-being of caregivers have not been well characterized

**Objective:** To quantify the extent of physical, emotional, and financial burden of BBS on caregivers, particularly as they relate to hyperphagia and obesity

BBS, Bardet-Biedl syndrome.

1. Sherafat-Kazemzadeh et al. *Pediatr Obes.* 2013;8:e64-e67. 2. Ervin et al. Presented at: Pediatric Endocrine Society Annual Meeting 2022; April 28-May 1; Virtual.

# Approach

- Cross-sectional multi-country survey of adult caregivers of individuals with a diagnosis of BBS and obesity and hyperphagia in the United States, United Kingdom, Canada, and Germany
- The survey assessed BBS-related characteristics and impacts, caregiver burden, and caregiver health status
- Inclusion and exclusion criteria
  - Adult caregivers who had cared  $\geq 6$  months for individuals with BBS who had obesity or were in the  $\geq 95$ th weight percentile and hyperphagia
  - Individuals with BBS could not be currently enrolled or have participated in a clinical trial within the past 6 months
- Data were collected between December 2021 and February 2022

## Study Questionnaires (completed by caregivers)

### Impact on Individuals With BBS

Symptoms of Hyperphagia<sup>a</sup>

Impacts of Hyperphagia<sup>a</sup>  
Caregiver Version

PROMIS<sup>b</sup> Parent Proxy Scale  
v1.0 – Global Health 7

IWQOL-Kids<sup>c</sup>: Parent Proxy

### Impact on Caregivers

Impacts of Hyperphagia<sup>a</sup>: Caregiver  
Version

PROMIS<sup>b</sup> Scale v1.2 Global Health

Revised Impact on Family Scale

Caregiver Work Productivity and  
Activity Impairment (WPAI)<sup>d</sup>

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# Demographics and Clinical Characteristics

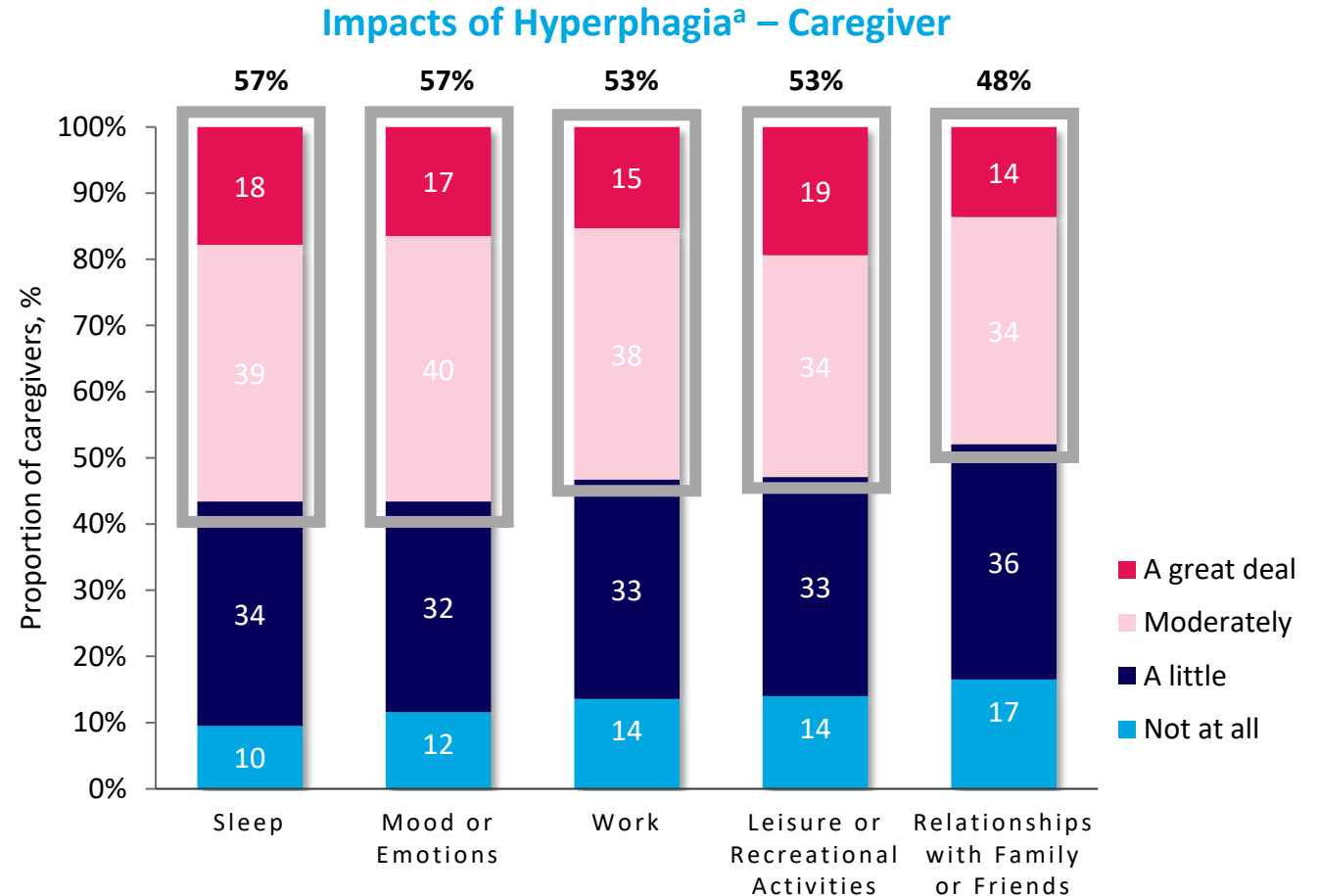
Caregivers	Total (N=242)	Individuals with BBS	Total (N=242)
<b>Age (years), mean ± SD</b>	41.9 ± 6.7	<b>Age (years), mean ± SD</b>	12 ± 3.7
<b>Sex, n (%)</b>		<b>Sex, n (%)</b>	
Male	131 (54.1)	Male	155 (64)
Female	111 (45.9)	Female	87 (36)
<b>Relationship to person with BBS, n (%)</b>		<b>Hyperphagia prominent feature at presentation, n (%)</b>	230 (95)
Mother	101 (41.7)		
Father	125 (51.7)		
Other	16 (6.6)		
<b>Sole caretaker of person with BBS, n (%)</b>	43 (17.8)	<b>Modified BMI Z score, mean ± SD</b>	4.1 ± 4.5

- Overall, caregivers' sociodemographic characteristics were similar across Canada (n=62), Germany (n=61), United Kingdom (n=59), and United States (n=60)

BBS, Bardet-Biedl syndrome; BMI, body mass index; SD, standard deviation.

# Hyperphagia in BBS Impacts Multiple Aspects of Caregivers' Lives

- Hyperphagia experienced by individuals with BBS has direct negative impacts on caregivers' daily lives
- Across the individual domains of sleep, mood, work, leisure, and relationships, nearly half of all caregivers rated hyperphagia as affecting them “moderately” to “a great deal”
- Results were generally consistent across countries



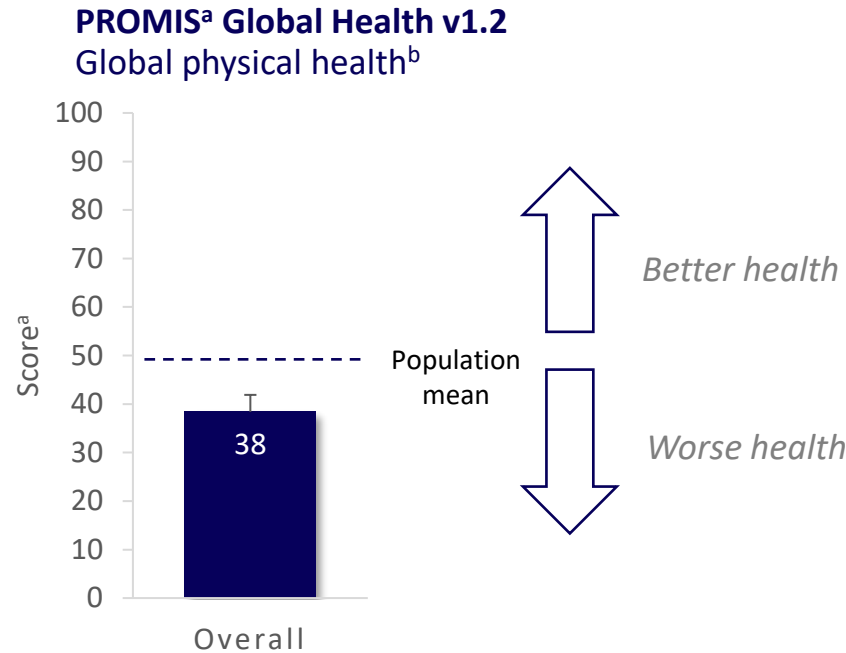
Overall mean (standard deviation), 7.8 (3.2)<sup>b</sup>

BBS, Bardet-Biedl syndrome.

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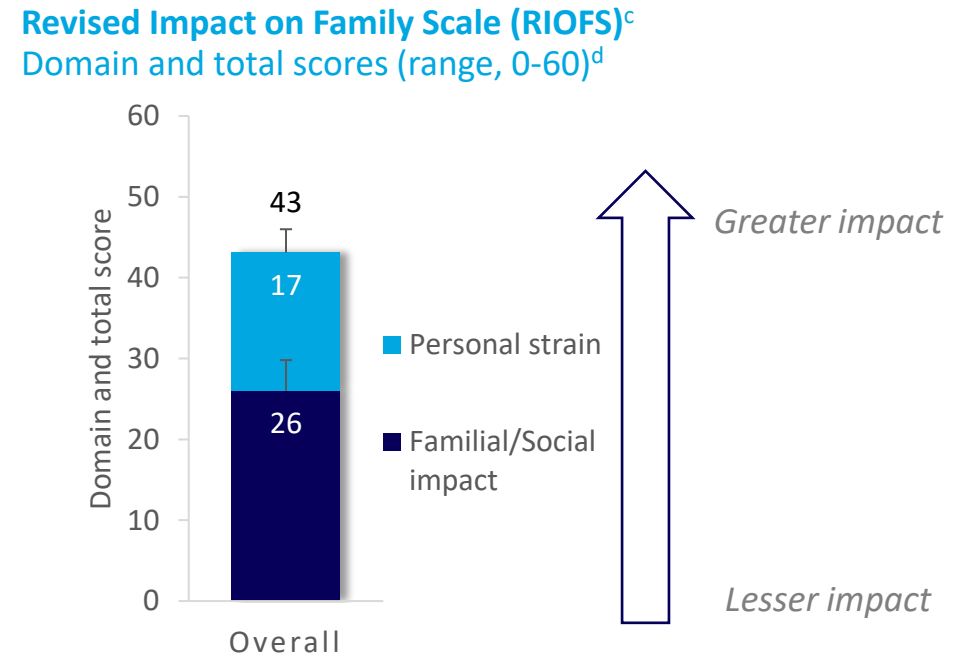
# Caregivers Report a Negative Impact on Health and Increased Burden on Families

Caregivers experience poor physical health



Results were similar across countries

Caregivers reported substantial personal strain and impacts on family dynamics



Perceived burden was slightly greater in the United States and slightly lower in Canada and Germany

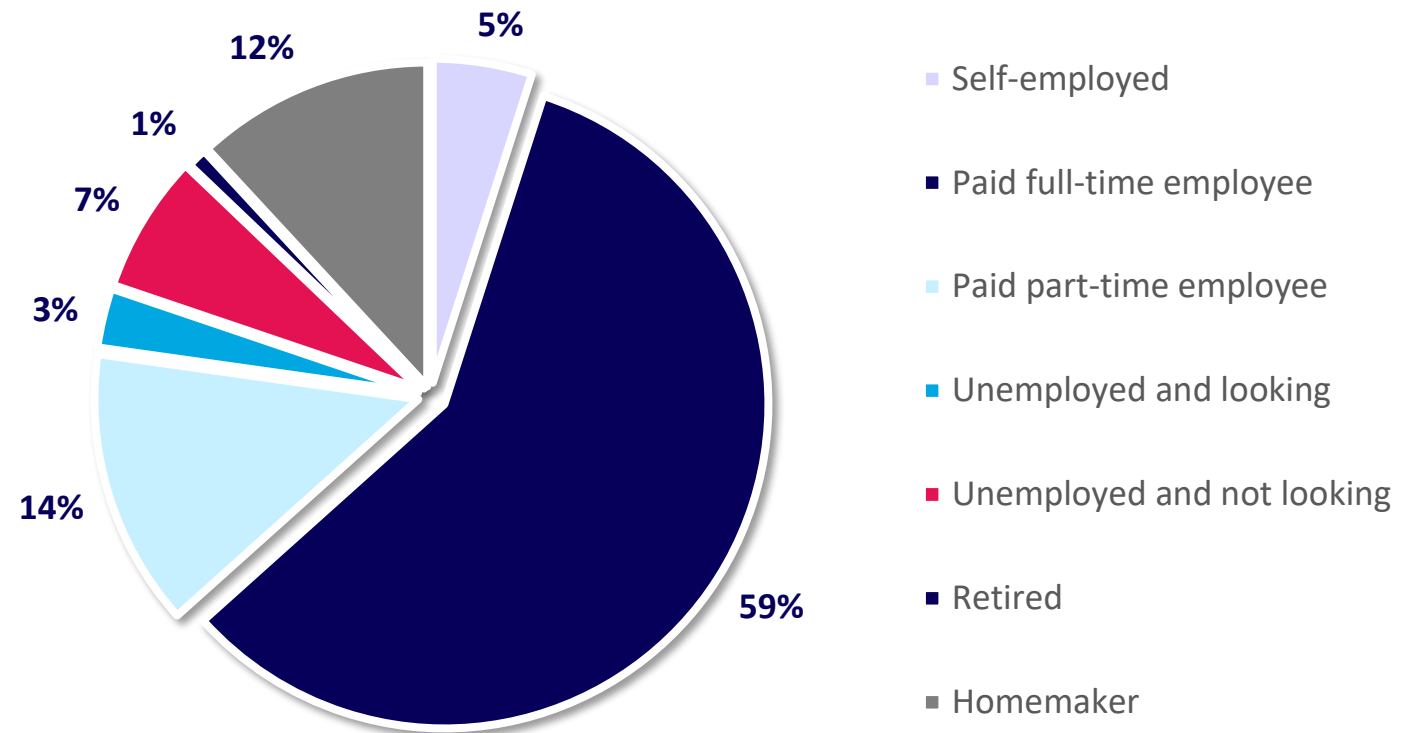
BBS, Bardet-Biedl syndrome.

<sup>a</sup>© 2022 PROMIS Health Organization. All Rights Reserved. <sup>b</sup>Scores are standardized to a mean of 50 and a standard deviation of 10 (range, 0-100). Higher scores indicate better health. Blue line denotes population mean. Error bars represent the standard deviation. <sup>c</sup>© 1978 by Ruth E. K. Stein, M.D., and Catherine Kohler Riessman, PhD. ALBERT EINSTEIN COLLEGE OF MEDICINE. DEPARTMENT OF PEDIATRICS. <sup>d</sup>The Revised Impact on Family Scale consists of 15 questions each with 4 response options (range, 0-15). Higher scores indicate that BBS has a greater impact on the family (personal strain maximum=24; Family/Social maximum=36). Error bars represent the standard deviation.

# Caregivers Experienced Increased Employment Events and Reduced Productivity

- Total productivity among employed caregivers was substantially impaired because of BBS (mean [SD], 60.9% [21.4%])<sup>a,b</sup>
- Active employment of caregivers was 75.6% at the time of the survey
  - A change in employment was experienced by 61% of caregivers after BBS diagnosis<sup>c</sup>
- Total activity across all caregivers was highly impaired because of BBS (mean [SD], 56.6% [22.8%])<sup>b</sup>

Current employment status of caregiver participants



BBS, Bardet-Biedl syndrome; SD, standard deviation; WPAI, Work Productivity and Activity Impairment.

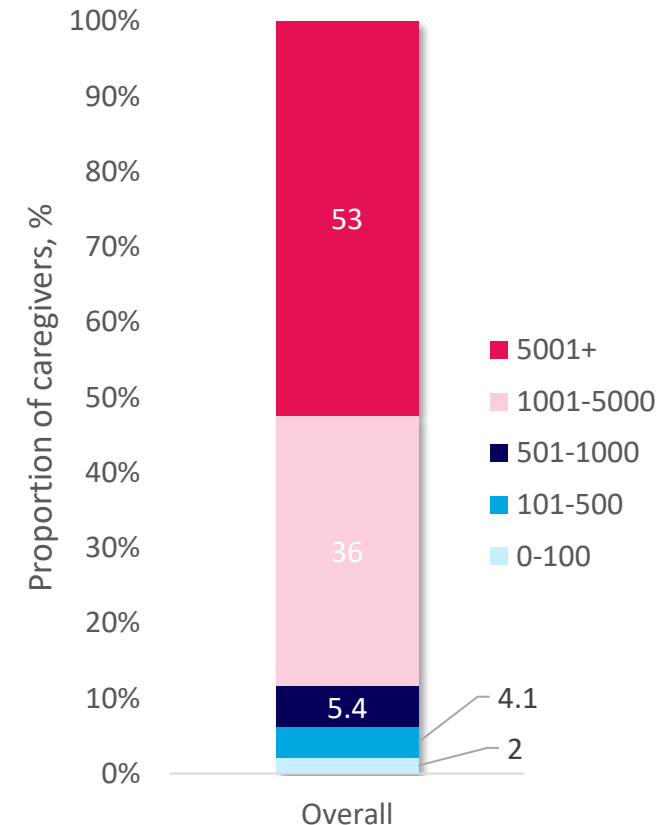
<sup>a</sup>Only caregivers who indicated that they are engaged in part-time, full-time, or self-employed work are included in this analysis. <sup>b</sup>Range 0%-100%, with higher values indicating greater absenteeism, presenteeism, or impairment. <sup>c</sup>Respondents who indicated that questions about employment changes were "not applicable" (n=42) were assumed to have not been employed and were not considered in this analysis.



# Caregivers Reported Challenges With Treatment Management

- Caregivers reported **out-of-pocket medical expenses** of \$1,883, which were **5-10 times greater than average** per capita medical estimates<sup>1,a</sup>
- The majority of caregivers perceived the **cost of BBS-related care** to be **at least a moderate financial burden**
- Caregivers reported currently using an average of **8 different weight management approaches** for the individual with BBS
- Caregivers placed a high importance on having new effective approaches to weight management (mean [SD], 7.4 [1.4])<sup>b</sup>

## Total out-of-pocket medical expenses for individuals with obesity due to BBS in the past 12 months (local currency)<sup>a</sup>



BBS, Bardet-Biedl syndrome.

<sup>a</sup>Questions with categorical responses were recorded in local currency; questions with continuous responses were converted to USD 2022 with a conversion rate of 1 USD = 0.74 Pound Sterling (UK), 1.27 CAD (Canada), and 0.88 Euro (Germany).

<sup>b</sup>Overall satisfaction/importance score range: 0 (low) to 10 (high). Error bars represent the standard deviation.

1. OECD.Stat. 2019 household out-of-pocket payments at the per capita level. <https://stats.oecd.org/Index.aspx?DataSetCode=SHA>. Accessed August 11, 2022.

# Summary and Conclusions

- This novel multi-country survey confirms and quantifies the burden of BBS on caregivers, highlighting its considerable and multifaceted nature, particularly as it relates to obesity and hyperphagia
- The negative impact of hyperphagia on caregivers affects family dynamics, emotional well-being, and caregiver employment while also causing increased out-of-pocket medical expenses

**The observed caregiver health-related burden highlights the need for new strategies and interventions to reduce the burden sustained from obesity and hyperphagia when caring for individuals with BBS**