The Societal Burden of Bardet-Biedl Syndrome in the United States

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Summary

- **Bardet-Biedl syndrome (BBS) is associated with significant economic and quality of life burden for patients, caregivers, and society in the United States**

Background

- BBS is a rare heterogeneous genetic disorder, which may include obesity; insatiable, pathologic hunger (hyperphagia); progressive retinal degeneration; and decreased kidney function[1]
- In most of North America and Europe, the incidence rate of BBS ranges from 1 in 140,000 to 1 in 160,000 newborns[2]
- Given its lifelong and progressive nature, BBS imposes a substantial burden on patients and their families, including increased costs and quality of life decrements; however, the full burden of illness from clinical, economic, and societal factors has yet to be quantified

Objective

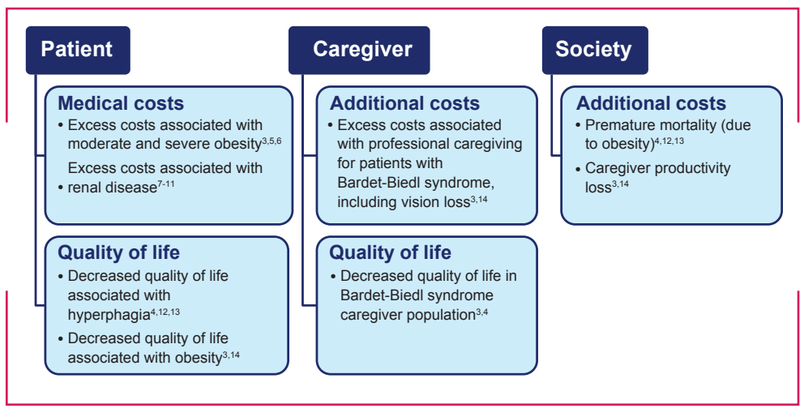
- To estimate the burden of disease in patients with BBS, their caregivers, and society in the United States

Methods

Economic model synthesizing information from multiple sources

- A prevalence-based approach was employed to estimate direct health care costs among the most prevalent comorbidities (ie, medical services due to obesity and renal disease), direct non–health care costs (ie, professional caregiving), and indirect costs (ie, premature mortality, caregiver productivity loss, quality of life decrements) (Figure 1; Table)
- Estimates were derived from scientific literature, and costs were expressed in US dollars
  - The estimated diagnosed BBS population was 555 in the United States in 2020[3]
  - The value of 1 quality-adjusted life-year (QALY) was estimated as $150,000[4]
- The incremental costs associated with BBS were calculated as the difference between costs incurred by patients with BBS and the general US population (comparison group), and thus constitute the "excess" costs attributed to BBS
- Annual and lifetime per-patient and total costs were reported
  - Lifetime costs were calculated assuming a life expectancy associated with patients with severe obesity (70.8 years)[3]; future costs were discounted to present values using an annual rate of 3%, assuming a current patient age of 27 years[3]
- Sensitivity analyses using different sources and assumptions were performed to assess the robustness of estimates and account for variability in published estimates

Figure 1. Model framework.



Methods (Cont)

Table. Calculation of Cost Components

	Component	Key inputs
Medical costs	Obesity	■ Number of pediatric and adult patients diagnosed with BBS[5] ■ Number of patients with BBS who have moderate/severe obesity[3] ■ Excess medical costs associated with moderate/severe obesity[6]
	Renal disease	■ Number of pediatric and adult patients with BBS who have ESRD[7] ■ Number of years adult patients with BBS who have ESRD receive dialysis before kidney transplant[8] ■ Number of adult patients with BBS who have ESRD and received kidney transplant[9] ■ Expected remaining life-years for adult patients who received dialysis but not kidney transplant[10] ■ Annual cost for dialysis[10] ■ Cost for kidney transplant[11]
Additional costs	Excess direct costs for professional caregiving	■ Proportion of patients with severe visual impairment due to BBS[3] ■ Caregiving costs for severe visual impairment[14]
	Premature mortality (due to obesity)	■ Estimated excess number of premature deaths in the population with obesity[12] ■ Potential life lost per death[13] ■ Value of 1 QALY[4]
	Caregiver productivity loss	■ Proportion of employed primary caregivers of patients with BBS[3] ■ Mean hourly income[15] ■ Hours of presenteeism and absenteeism[3]
Quality of life	Patient quality of life	■ Hyperphagia disutility score[16] ■ Obesity disutility score[17] ■ Number of patients with hyperphagia and obesity due to BBS[3] ■ Value of 1 QALY[4]
	Caregiver quality of life	■ Caregiver of patients with BBS disutility score[3] ■ Value of 1 QALY[4]

BBS, Bardet-Biedl syndrome; ESRD, end-stage renal disease; QALY, quality-adjusted life-year.

Results

- The societal burden associated with BBS in 2020 was estimated to be $110,172 per patient ($61 million in total), which amounts to lifetime costs of $5.2 million per patient with BBS ($2.9 billion in total) (Figures 2 and 3)
  - Reduced quality of life was the component associated with the highest annual and lifetime costs
- The main drivers of burden were reduced patient quality of life (including obesity and hyperphagia), caregiver productivity loss, and reduced caregiver quality of life, accounting for approximately 53%, 24%, and 21% of the total economic burden of disease, respectively
- Costs attributed to hyperphagia and obesity made up >50% of the total burden (Figure 4)

Figure 2. Annual total costs.

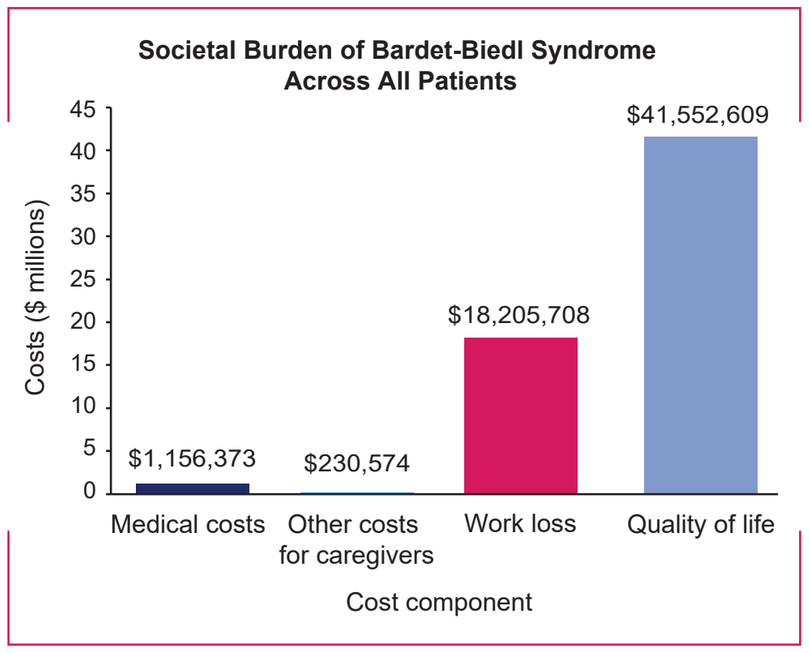


Figure 3. Lifetime total costs.

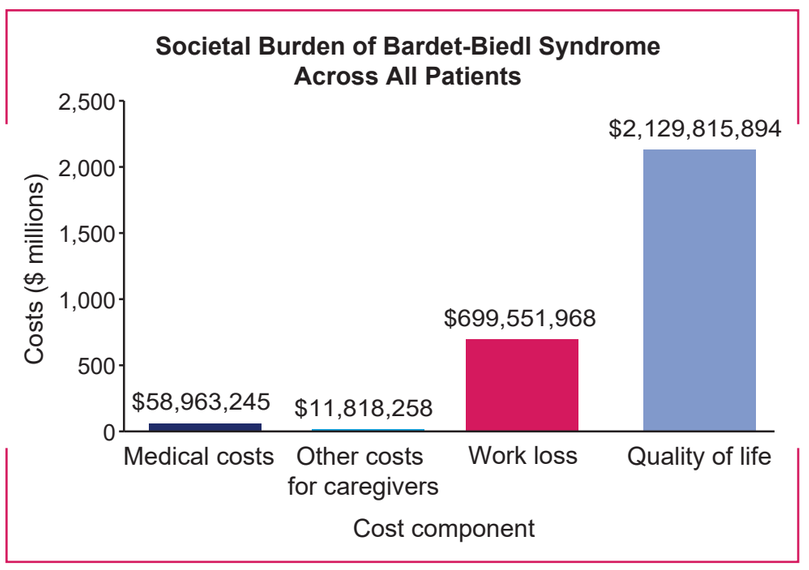
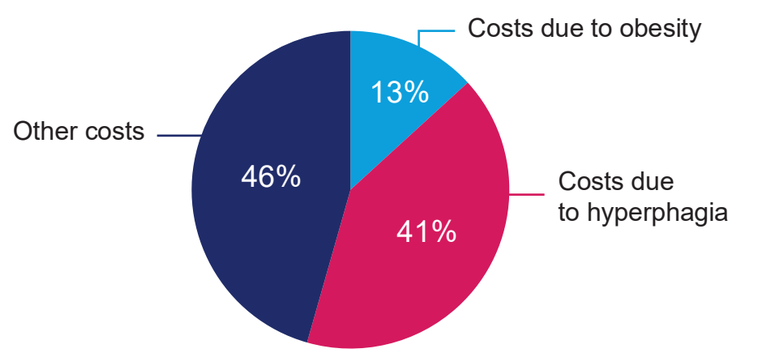


Figure 4. Obesity and hyperphagia cost distribution.[a]



[a]Costs due to obesity included medical costs associated with obesity, premature mortality due to obesity, and patient quality of life decrement associated with obesity. Costs due to hyperphagia included patient quality of life decrement associated with hyperphagia. Other costs included all other costs not in the previously mentioned categories.

Conclusions

- From our model, we conclude BBS resulted in significant economic and quality of life burden for patients, caregivers, and society
- Estimating the economic burden of BBS is limited by the small patient population size and associated challenges in estimating specific components of disease burden in a real-world setting, and further research is needed to more comprehensively estimate the economic burden of disease
- Given the considerable burden per patient, treatments that improve disease management, especially those addressing hyperphagia and obesity, carry the potential for significant economic and quality of life benefits for patients and their caregivers

Acknowledgments: This study was sponsored by Rhythm Pharmaceuticals, Inc. Medical writing and editorial assistance were provided under the direction of the authors by Kelby Killoy of MedThink SciCom and sponsored by Rhythm Pharmaceuticals, Inc.

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