

***The price we pay:* a report on the  
financial costs of raising a child with  
Prader-Willi syndrome around the world**

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## ABOUT THIS REPORT

*Marguerite Henry is a student at the London School of Economics, UK. She is working towards a BSc in International Social and Public Policy and Economics. Marguerite spent her summer 2021, researching and writing this report for the Board of IPWSO. We asked her to take an overview of the complicated financial situation facing the parents and families of people with PWS.*

*In this report Marguerite has summarised the existing research and written up the interviews she conducted. Many thanks to those of you who gave your time to support her project.*

## INTRODUCTION

Prader-Willi syndrome (PWS) is a complex and rare neurodevelopmental condition. Studies have shown that between 1 in every 15,000 and 1 in every 25,000 people are born with PWS and it affects all races and sexes equally. PWS occurs when there is a lack of expression of one or more genes in a specific region of chromosome 15.

### **Signs and symptoms of the syndrome include:**

- Low muscle tone and failure to thrive at birth.
- Short stature, if not treated with Growth Hormone.
- Learning disabilities.
- Incomplete sexual development.
- Behavioural and psychiatric challenges.
- An excessive drive to eat (hyperphagia).

PWS is usually suspected on the basis of the clinical signs and symptoms and can be confirmed by genetic testing. There is no cure for PWS but a supportive and empathetic environment with tailored care and treatment can reduce many of the challenges.

IPWSO is an international, parent-led, non-profit, membership organisation supporting national PWS associations, as well as people with PWS, their families and the professionals who work with them. Founded in 1991, the organisation provides information and support and shares best practice around the world.

### **What is a rare disease?**

- There is no global definition of rare diseases, its definition varies across countries.<sup>1</sup>

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<sup>1</sup> 'Navigating rare neurological diseases: meeting the challenge for policy makers, patients, and healthcare professionals' Economist Intelligence Unit, (2020)

- There are over 300 million people living with one or more of over 6,000 identified rare diseases around the world.<sup>2</sup>
- Each rare disease may only affect a handful of people, but taken together the number of people directly affected is considerable.<sup>3</sup>
- Rare diseases currently affect 3.5% - 5.9% of the worldwide population.<sup>4</sup>
- 72% of rare diseases are genetic whilst others are the result of infections, allergies and environmental causes.<sup>5</sup>
- 70% of those genetic rare diseases start in childhood.<sup>6</sup>
- In Europe, a rare disease is a condition that affects less than one in 2,000 people.<sup>7</sup>

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<sup>2</sup> <https://www.nature.com/articles/s41431-019-0508-0>

<sup>3</sup> <https://www.rarediseaseday.org/article/what-is-a-rare-disease>

<sup>4</sup> Ibid

<sup>5</sup> Ibid

<sup>6</sup> Ibid

<sup>7</sup> <https://www.eurordis.org/about-rare-diseases>

## **SECTION 1: Why is it important to characterise the financial cost of PWS ?**

Quantifying the economic burden of rare diseases in general, and Prader-Willi syndrome (PWS) in particular, is essential to address current health policy challenges. This report aims at reviewing what is known about costs associated with rare diseases in general and with PWS in different parts of the world. Researchers have all outlined the lack of research and data on this subject, leading to underestimating PWS economic consequences on families and society.

### **Research has a key role in raising awareness**

Costs of rare diseases are often hidden or underestimated, leading to a misunderstanding of patients' needs. Understanding the costs associated with PWS and measuring its socio-economic impact on society is necessary to provide adequate and universal support for people with the syndrome. Cost-of-illness studies are used to estimate the economic burden of diseases in society; they are defined as the "maximum amount that could potentially be saved or gained if a disease were to be eradicated."<sup>8</sup> Costs related to Prader-Willi syndrome, or a rare disease, are likely to be substantial. Importantly, scientific knowledge allows accurate advocacy and raises awareness about the economic consequences of PWS and rare diseases in general.

### **Cost estimation is critical in implementing effective policy**

Socio-economic and health-related quality of life data is essential to guide policy implementation and determine the outcomes of political interventions. PWS entails a significant hidden cost for the society that policymakers and international organisations should acknowledge. Indeed, existing research has emphasised that having a child with PWS entails many hidden costs. Apart from direct healthcare costs such as drugs, hospitalisation and medical visits, families worldwide also face indirect costs. Therefore, a proper estimation of all costs related to PWS would allow proper and adequate health-related support.

### **Meeting the needs of people with PWS and their caregivers**

As a rare disease affects a small percentage of the population, resources dedicated to research and adequate support are often limited. As a result, the community of rare disease patients and PWS has significant unmet needs. This report emphasises the direct and indirect burden of PWS for both the patient and the caregiver. People with PWS need constant monitoring. As a result, one of the parents often needs to reduce

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<sup>8</sup> Lidia García-Pérez et al., "Cost-of-Illness Studies in Rare Diseases: A Scoping Review," *Orphanet Journal of Rare Diseases* 16, no. 1 (2021), <https://doi.org/10.1186/s13023-021-01815-3>.

their time at work to dedicate time for informal care. Therefore, special attention and support need to be given to those caregivers.

## **Identifying the inequalities faced by families in different parts of the world**

The report principally aims to record differences in costs of raising a child with PWS in different parts of the world. Every region needs to respond to different challenges. Most of the research papers available were focusing on the United States or European countries. However, even within developed nations, disparities are significant.

Countries with less-developed economies have made some progress in managing rare diseases. Nevertheless, the allocation of health resources and research has been chiefly on managing acute and primary care, leading to an under-recognition of RDs' economic burden.<sup>9</sup>

Interviews with families from different countries have highlighted substantial inequalities. The report aim at determining whether people with a rare condition and with PWS in particular in different parts of the world are well supported by their healthcare system.

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<sup>9</sup> Asrul Akmal Shafie et al., "State of Rare Disease Management in Southeast Asia," *Orphanet Journal of Rare Diseases* 11, no. 1 (February 2016), <https://doi.org/10.1186/s13023-016-0460-9>.

## **SECTION 2: Overview of costs related to PWS and possible methods for their estimation**

### **How can we estimate costs related to a disease?**

Evaluating the cost of a disease is a complicated task. Diseases involve considerable expenditures dedicated to the use of health resources to improve the person's life, but there are also many non-healthcare costs to adapt the activities of daily life to the individual. A way to analyse the costs of an illness from a societal perspective as well as cost for individuals is through costs-of-illness studies.

"Cost-of-illness studies measure the socio-economic burden of a disease and can be used as a public policy tool to assist in prioritization and justification of healthcare policy."<sup>10</sup> Rare diseases are often incurable conditions that require expensive continuous monitoring and strict medical follow-up. Moreover, the lack of scientific research and data on rare disease compromises the improvement of people's lives.

There are different ways to evaluate the cost of a disease. Firstly, the perspective chosen has a significant impact on results. Most of the time researchers choose to study costs of illness from a societal perspective.<sup>11</sup> Economists often study the costs to society as a whole, looking at both national expenditures and out-of-pocket costs to individuals. Indeed, the patient or individual perspective can be informative but underestimate the total cost of a disease as a significant part of costs are paid by national healthcare systems, private institutions or international organisations. Few studies make the distinction between what is paid for by the Government and what is paid by the individual. Depending on the healthcare system, individuals won't bear the same economical burden, there will be a lot of variation.

Advocating for research on the effectiveness and coverage of different government's healthcare systems is necessary to understand how much money a person with PWS or their family actually spends. Ideally studies should include information about national expenditure and individual costs associated with a particular disease.<sup>12</sup>

Drugs and healthcare resources are in general more expensive for rare disease than for chronic diseases. Moreover as a rare disease affects a small part of the society, patients can feel isolated and marginalised. To respect medical and human rights, careful attention needs to be given to this community to make sure they can access the same rights, medical attention and health coverage as the general population.

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<sup>10</sup> Lidia García-Pérez et al., "Cost-of-Illness Studies in Rare Diseases: A Scoping Review," *Orphanet Journal of Rare Diseases* 16, no. 1 (2021), <https://doi.org/10.1186/s13023-021-01815-3>.

<sup>11</sup> Ibid

<sup>12</sup> Ibid

There is a huge lack of rare disease data and research. Scientific knowledge is the key to guiding effective policy implementation and cost-of-illness studies are a key element in targeting patients' needs.

## **Direct healthcare costs**

**Direct healthcare costs are expenditure related to:**

- Hospital admissions
- Emergency visits
- Outpatient care (rehabilitation, medical tests, medical visits and home medical care)
- Use of devices
- Treatment and drugs

**For PWS in particular:**

There is no one treatment or cure available however various supportive treatments exist.

- Growth Hormone is generally used to normalise height.
- Hospitalisations
- Neonatal care
- Speech therapies
- Regular exercise
- Physiotherapies
- Psychological therapies

Direct healthcare costs include all expenditures related to outpatient and inpatient care.

## **Direct non-healthcare costs**

Direct non-healthcare costs are costs associated with managing a disease which are not related to medical resources used. PWS entails many direct non-healthcare costs.

- Special equipment at home
- Social services
- Professional caregivers
- Special education
- Transportation costs
- Home adaptation
- Food management resources

These costs can be substantial and often hidden, thus less well supported by state or insurance funding . Generally these costs represent the major burden for families as they are paid for by out-of-pocket money and not by healthcare coverage.



## Loss of labour productivity

Costs associated with loss of labour productivity are also significant. In certain circumstances, caring for a child with PWS requires constant monitoring. Parents or relatives may be obligated to dedicate more time to the child or adult with PWS, especially when living at home and not in an adapted residential setting. Loss of labour productivity can impact both the carer and the person with PWS.

Loss of labour productivity is often estimated with the human capital-based approach. "The value of the **human capital** is approximated by the value of an average individual's future earning. The entire period of absence from work due to illness is considered and valued by the achievable gross income."<sup>13</sup>

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<sup>13</sup> (2008) Human Capital Approach. In: Kirch W. (eds) Encyclopedia of Public Health. Springer, Dordrecht. [https://doi.org/10.1007/978-1-4020-5614-7\\_1583](https://doi.org/10.1007/978-1-4020-5614-7_1583)

## SECTION 3: Quantifying the socio-economic burden of rare diseases

Researchers from different countries have attempted to evaluate costs associated with rare diseases. This part of the report gives an overview of this literature. Researchers have conducted an estimation of total cost from a societal perspective of rare diseases. They do not give precise information about the economic burden on families but rather emphasise the need for more research and for policies targeting the rare diseases community and their caregivers, who are at risk of financial disaster if they are not well supported by the state.

### Case study 1: European Union, The BURQOL-RD project

The BURQOL-RD project is an EU founded programme aimed at creating a common model to evaluate the socio-economic burden of rare diseases on society and to assess health-related quality of life of patients and their caregivers<sup>14</sup>. This project gathers research about 10 rare diseases (Cystic Fibrosis, Prader-Willi syndrome, Haemophilia, Duchenne Muscular Dystrophy, Epidermolysis Bullosa, Fragile X syndrome, Scleroderma, Mucopolysaccharidosis, Juvenile Idiopathic Arthritis and Histiocytosis) in eight different countries (Bulgaria, France, Germany, Hungary, Italy, Spain, Sweden and the UK.)

The “Social Economic Burden and Health-Related Quality of Life in Patients with Rare diseases in Europe” ( BURQOL-RD) was conducted over three years and began in April 2010. EURORDIS (Rare Diseases Europe) was an associated partner in this project amongst 12 others<sup>15</sup> plus many individual researchers.

#### Key findings:

This project is the largest study aimed at quantifying the economic burden of rare diseases in Europe<sup>16</sup>. The project aimed to help in the prioritisation and justification of preventive healthcare policies.<sup>17</sup> Knowing what costs rare disease patients face is important to produce adequate support.

These findings show that, although evaluating the direct healthcare costs incurred by rare diseases is relevant, the social costs are even higher, due to the loss of labour productivity and formal or informal care involved.

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<sup>14</sup> <http://www.burqol-rd.com>

<sup>15</sup> <https://www.eurordis.org/content/burqol-rd-project>

<sup>16</sup> <http://www.burqol-rd.com/index.html>

<sup>17</sup> <https://link-springer-com.gate3.library.lse.ac.uk/content/pdf/10.1007/s10198-016-0780-7.pdf>

There are important differences between the countries studied:

- Differences in the level of development of formal care provided by formal services across countries entails differences in costs and health related quality of life.
- However, in most countries and for most diseases the main cost was informal care and the associated loss of labour productivity specially in southern European countries.
- Differences in costs related to a rare disease between countries can be explained by the difference in costs of resources used as well as differences in intensity of their utilisation.
- Countries have different healthcare systems and all countries do not have the same level of achievement in terms of recognition of healthcare services needed by patients.
- The use of formal care services depends significantly on the economic situation of a country and its social policies.
- The self-perceived quality of life depended more on the disease than the country's healthcare system.

**Conclusion and recommendation:**

- Quantifying the socio-economic burden of rare disease is essential.
- More studies in different parts of the world should be conducted in order to have a better understanding of the needs of individuals with rare diseases.
- Research is essential to implement targeted policy and to prioritise allocation of state resources.
- The main issue that has come up is the lack of available care services.
- This network of researchers has emphasised the need to increase resources dedicated to social support for families (reinforcement of social care).
- It is important to advocate for more social protection for informal caregivers.
- Increasing those services and raising awareness about the burden of care would relieve families by enabling them to dedicate more time to work and feel less isolated and overwhelmed.
- It is important that policymakers and the general population recognise the work of family carers and support them in an effective manner.

## Case study 2: The United States, The national economic burden of rare diseases

*The national economic burden of rare disease study*, conducted in 2019 in the United States, had the objective of estimating the costs of rare diseases to society<sup>18</sup>. Such studies aim at a greater understanding of costs for people with a rare disease. The Every Life Foundation supported this project to empower the rare disease community in filling the knowledge gap.<sup>19</sup> In addition, this global economic evaluation allows effective policy implementation and greater support for caregivers and families of people with PWS. The research “The national economic burden of rare diseases” focused on the costs associated with 379 rare diseases. Approximately 15.5 million people took part in the study. The estimated total cost of rare diseases in the US in 2019 was \$966 billion.

### Direct medical costs:

The study includes outpatient and inpatient care and treatment. The study also provides an estimation of the overall impact of rare diseases on health and wellbeing by comparing rare disease patients’ healthcare expenditure with a matched sample without a rare disease. The research gathered data on direct medical costs across different insurance coverages, ages, and type of healthcare services. By comparing healthcare expenditure of rare disease patients with similar patients (same age, insurance, use of healthcare resources) with patients without a rare disease, researchers have estimated the excess costs directly linked with the rare condition.

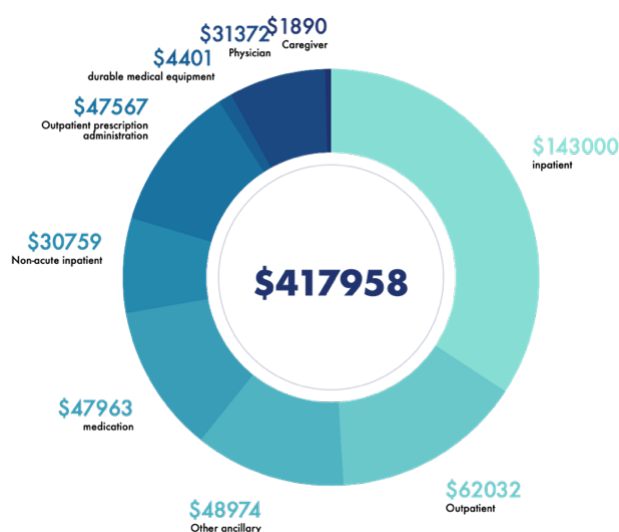
- More expensive components of direct healthcare costs were: inpatient and outpatient care, prescription medication and other ancillary care costs.
- An average person with a rare disease has an annual medical cost that is \$26,887 more than a person without a rare disease.
- Approximately 70% of US citizens are privately insured. It raises some concerns for the rare disease community as the total excess medical cost due to a rare disease is higher when the patient is privately insured compared to a patient under Medicare or Medicaid. (Most American are privately insured as state insurances Medicaid and Medicare concerns only people with specific characteristics (age / or low income))
- Excess medical costs due to a rare disease per capita are higher for children than for adults.

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<sup>18</sup> The National economic Burden of rare disease study

<sup>19</sup> <https://everylifefoundation.org/about-us/>

## Excess direct healthcare costs of rare diseases (in \$ million)



Source: Figures from Lewin analyses of rare disease prevalence calculated from the 2018 dNHI claims, 2019 Medicare SAF 5% sample claims, and 2016 Medicaid claims combined with the Census population projection for 2019; direct medical cost estimates were obtained using 2018 dNHI claims, 2019 Medicare SAF 5% sample claims, and 2016 Medicaid claims. Other ancillary services for example include telehealth, ambulance transportation via land, air or water, mobile unit services, etc.

### Non-medical costs:

These include the price of hiring a professional caregiver to assist with daily living, access to special education, home modifications, and/or increased transportation costs.

There are many costs that can be associated with a rare disease. Non-medical costs are often directly borne by families and individuals. Those costs are substantial and often not recognised and not well evaluated. As their estimation is more complicated than direct healthcare costs evaluation tends to underestimate their impact on families.

### Indirect costs:

The American paper attempted to estimate the costs associated with reduced employment. Patients with rare diseases often require continuous monitoring. As each rare disease affects a small population, trained professional caregivers specific to the condition are rare, and family members or relatives often have to take part in crucial daily care activities. This leads to a decrease in time dedicated to working. Loss of income for the caregiver is common but not well acknowledged. Unpaid care provided by a relative, informal care, entails a sizeable economic burden on families. Diagrams below only report indirect costs associated with RDs for the primary caregiver. However, the study also informs us about the loss of income of the adult patient and the secondary caregiver. Loss of income for caregivers is crucial in understanding a rare disease's financial impact on a household. Moreover, the costs are often hidden

and thus less well supported by institutions. A strong understanding and estimation are needed to bring the most universal and accurate support to families.

#### Transfer payment associated with rare disease to the Person with RD:

- Financial assistance from disability income, is approximately 115\$ billion.
- We can observe **that financial assistance from charitable programs** was one of the major sources of transfer payment specially for adults.

Exhibit 8. Transfer Payments Associated with Rare Disease to the Persons with RD (in 2019)

	Age <18		Age ≥18	
	Financial Assistance (in \$ Millions)	Per Capita (\$)	Financial Assistance (in \$ Millions)	Per Capita (\$)
Financial assistance from charitable programs	\$319	\$241	\$19,381	\$1,363
Supplemental security income (SSI)	\$452	\$342	\$16,770	\$1,179
Social security disability insurance (SSDI)	NA	NA	\$46,896	\$3,297
Other disability benefits	\$924	\$698	\$49,532	\$3,483
Total	\$1,694	\$1,281	\$132, 578	\$9,322

Sources: Lewin analyses of RD prevalence calculated from the 2018 dNHI claims, 2019 Medicare SAF 5% sample claims, and 2016 Medicaid claims combined with the Census population projection for 2019; financial assistance is estimated using the Lewin's analyses of the RD Impact Survey data.

#### Conclusion:

- Overall, the American rare disease community faces inequalities compared to other American patients. **Treatments are generally more expensive; they face reimbursement issues related to private insurance, Medicare, and Medicaid. They struggle to access medical, social, or financial services because decision-makers are not familiar with the disease. Non-medical and indirect costs (costs bear by the family) were greater than direct healthcare costs generally bear by the state or insurance.**
- Findings have demonstrated that the economic burden associated with rare diseases is substantial and by far **surpasses the costs of chronic diseases.**
- Even if a single rare disease affects a small proportion of the population, together, they affect approximately 1 in 10 Americans.
- Indirect costs are a key element in the evaluation of the economic burden of a disease . They are often hidden and underestimated.

#### Recommendations:

- Raising awareness about indirect costs related to rare diseases.
- Advocate for more information about how to care for an individual with a rare disease.

- Advocate for more research for more adequate state support.

## Case study 3: Asia, Early stage of developing rare disease policies

In Asia, no studies looking at the economic burden of PWS exists and very few studies on costs of rare disease have been conducted.

### Analysis of economic burden and its associated factors of twenty-three rare diseases in Shanghai <sup>20</sup>

The research provides with an analysis of the economic burden and its associated factors of rare disease in Shanghai from January 2013 to December 2016. The researchers' objective was to fill the knowledge gap on the economic impact of rare diseases in Shanghai, China. Asia is not at the same stage of development compared to the United States or Europe in terms of healthcare policies and scientific knowledge on rare diseases. This study aspires to raise awareness on the economic burden of rare diseases in Shanghai and influence incentives specific to the Chinese context.

#### Summary of results:

It has been estimated that there are about 10 million rare disease patients in China. Although significant policy progress can be observed in the field of rare disease management, some key challenges remain to be addressed. A major knowledge gap still needs to be filled. There is little research on rare diseases and their economic impact, which leads to an underestimation of their consequences on individuals and society.

This research aimed at evaluating annual direct medical cost and analysed its associated factors. 23 rare diseases were studied with 16,933 people diagnosed. Analysed effect of age, gender, disease type and payment type on inpatient and outpatient costs. **Overall, outpatient costs were greater than inpatient costs**, because of the smaller number of patients admitted in hospital for a rare disease. However, the individuals who were not hospitalised had a greater average cost per person. **Inpatient direct costs are predicted to be much higher than outpatient costs.**

#### Conclusion and recommendations:

**Direct costs and treatment affordability remains a key challenge to address in Shanghai and in China.** Even if significant progress can be observed more research is needed to raise awareness and to influence a health policy shift. This study only emphasized direct inpatient and outpatient costs. As other studies in different countries have illustrated, many costs non directly linked to medical care can be

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<sup>20</sup> Xiaoshu Cai et al., "Analysis of Economic Burden and Its Associated Factors of Twenty-Three Rare Diseases in Shanghai," *Orphanet Journal of Rare Diseases* 14, no. 1 (2019), <https://doi.org/10.1186/s13023-019-1168-4>.

substantial and are often borne by individuals. Researchers have reported costs of treatment for inpatient care with the annual disposable income of different areas of China. **The treatment costs represented a large proportion of the annual disposable income of urban area inhabitants and almost half of annual disposable income for rural inhabitants of Shanghai.** Thus, the direct cost of rare diseases is a heavy burden for a family, especially one from the countryside and rural areas. A global effort is needed to achieve an universal access of rare disease treatment when it exists. “Management of rare disease is a complex and multifaceted problem requiring increased awareness, patient tracking, and cost control”<sup>21</sup>. Some steps have been achieved toward universal coverages of rare diseases treatments: The National Rare Diseases Registry System of China launched in 2016<sup>22</sup> has enabled some people in Chinese provinces to have access to treatment, financially supported by the state. Treatment affordability remains a challenge to health policy makers, patients and society in general.

### **Rare Disease management in Asia <sup>23</sup>**

This study aimed at reviewing the state of rare disease management strategies in Southeast Asian countries (Philippines, Singapore, Malaysia, Indonesia, Vietnam and Thailand). Compared to other studies mentioned above it did not attempt to estimate the costs of rare diseases but rather to draw a picture of the overall rare disease management. Researchers analysed different aspects: healthcare systems, governance, patient advocacy and rare disease management, clinical expertise and patient management, funding and new-born screening of rare diseases.

Rather than having an estimation of costs of rare disease for society and individuals, this study is useful to understand in what conditions rare disease patient live in. This study has concluded, that despite major progress in this region, Southeast Asian countries are still at an early stage of developing a healthcare system capable of supporting the rare disease community.

Rare diseases are characterised by their low prevalence, which increases the isolation of patients and limits the resources available to provide adequate care. However, overall, about 9% of the population in Southeast Asia is affected by rare diseases. This means that a significant part of the population is at risk of marginalisation and is not well supported by regional governments.

### **Key challenges**

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<sup>21</sup> Xiaoshu Cai et al., “Analysis of Economic Burden and Its Associated Factors of Twenty-Three Rare Diseases in Shanghai,” *Orphanet Journal of Rare Diseases* 14, no. 1 (2019), <https://doi.org/10.1186/s13023-019-1168-4>.

<sup>22</sup> Feng S, et al. National Rare Diseases Registry System of China and Related Cohort Studies: Vision and Roadmap. *Hum Gene Ther.* 2018;29(2):128–35.

<sup>23</sup> Asrul Akmal Shafie et al., “State of Rare Disease Management in Southeast Asia,” *Orphanet Journal of Rare Diseases* 11, no. 1 (February 2016), <https://doi.org/10.1186/s13023-016-0460-9>.



**Rare diseases are not seen as a priority:**

- Though a large amount of progress has been made in rare disease management in southeast Asian countries, key challenges regarding funding still need to be addressed.
- Research and effective supervision of rare diseases is not a priority in Southeast Asia.
- The focus has been on primary preventive care and acute care; funding for research and treatment of rare diseases remains a major challenge.
- Lack of financial incentive for research and improvement of financial support for patients with rare diseases.
- The six targeted regions showed significant differences in health capacity and financing, largely due to the heterogeneity of the economic systems in the region.
- Because of the high cost of treatment for a relatively small number of individuals, there is often a concern that rare disease coverage will jeopardise the financial viability the universal coverage of health care in a country.
- In the Southeast Asian region, about 45 million people are estimated to live with rare diseases, however countries have many other critical health challenges to address.
- Other health challenges are often prioritised as they are more common, thus awareness about their consequences is greater.

**Healthcare systems and funding:**

- Even if significant progress can be observed toward an effective healthcare system in Southeast Asia, there is a considerable lack of funding dedicated to support patients with rare diseases.
- Treatment costs for rare diseases are generally extremely expensive and affect a small share of the population, thus they are often not reimbursed by public insurance.
- The lack of knowledge and understanding of rare conditions strengthens inequalities faced by rare disease patients in terms of healthcare coverage.
- Lack of resources and inadequate funding remains a central challenge that need to be addressed.
- Patients with rare diseases are often marginalised as funding for treatment often comes from out-of-pocket money or charitable organisations.
- Only a small number of patients have access to treatment.
- As funding is limited, although government subsidies exist in some countries (such as Malaysia), patients are still subject to strict eligibility criteria.
- In other countries, most of the rare disease financing is through industry subsidisation, employer benefits, charitable work or out-of-pocket payment.

- Only a small percentage of total health expenditure is allocated to rare diseases compared to European countries or the United States.<sup>24</sup>

### **Conclusions:**

Families with a child with a rare disease in Southeast Asia face significant health expenditures. This region is at the early stage of implementing efficient health coverage. Economic prosperity may be a key element in establishing effective support for individuals with rare diseases. However, even if a country is not economically stable, it can still implement a policy to improve health support for its citizens.

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<sup>24</sup> Schey C, Milanova T, Hutchings A. Estimating the budget impact of orphan medicines in Europe: 2010–2020. *Orphanet J Rare Dis.* 2011;6:62.

Divino V, et al. The budget impact of orphan drugs in the US: A 2007–2013 MIDAS sales data analysis. In: 56th ASH Annual Meeting and Exposition. 2014.  
[https://www.imshealth.com/deployedfiles/imshealth/Global/Content/Home%20Page%20Content/Real-World%20insights/IMS-Celgene\\_Orphan\\_Drugs\\_ASH\\_Presentation\\_Slides.pdf](https://www.imshealth.com/deployedfiles/imshealth/Global/Content/Home%20Page%20Content/Real-World%20insights/IMS-Celgene_Orphan_Drugs_ASH_Presentation_Slides.pdf). Accessed 26 Aug 2015.

## **SECTION 4: Socio-economic consequences of PWS in particular**

### **Case study 4: European comparison: An evaluation of the economic burden and health-related quality of life of patients with Prader-Willi syndrome**

This study's objective was to estimate the economic burden and health-related quality of life of patients with the Prader Willi syndrome. This research is part of the BURQOL-RD research network mentioned previously. The method used was a cross-sectional study of patients from Spain, Bulgaria, Hungary, France, the UK, Italy, Sweden and Germany. Data used for this estimation came from: healthcare resources utilisation, data on informal care, labour productivity losses and demographic characteristics of different countries. Health-related quality of life was estimated through questionnaires (EQ-5D questionnaire), collecting information about mobility, self-care, everyday activities, pain, discomfort, anxiety and depression. A total of 261 patients participated. This study, attempted to evaluate costs of PWS for society in 8 different countries. It does not exactly give us information of PWS economic burden for individuals and their caregiver. However, assessing costs from a societal perspective, guides policymakers of different countries in implementing health-related measures. By having an idea of total costs of a disease and the weight of different type of costs, policymakers can adapt their allocation of resources to provide the most effective support to the PWS community.

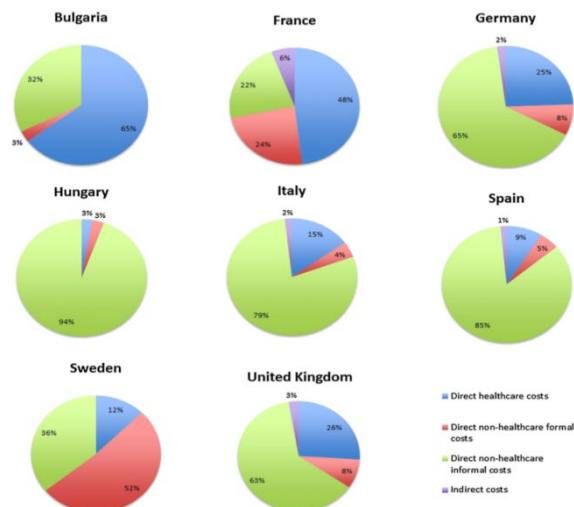
**The conclusion of the study was that PWS patients incur substantial costs and experience a significant deterioration of HRQOL (Health-related quality of life).**

The research estimated direct healthcare costs, direct non-healthcare costs and indirect costs associated with loss of labour productivity.

## Summary of results: <sup>25</sup>

Social/economic costs and health-related quality of life in patients with Prader-Willi...

**Fig. 1** Cost components per country



### The burden of informal care:

Germany and Sweden are the two countries where average annual costs per patient are the highest. Their largest cost component was direct non-healthcare. Overall, the largest cost component was direct non-healthcare for 6 of the 8 countries (Germany, Sweden, Italy, Hungary, the UK and Spain). Those high costs are driven by informal care. Indeed, the study estimated the cost of informal care by evaluating the potential cost of hiring a professional caregiver to replace the main informal caregiver. The question is: "How much would it cost to replace the main informal caregiver by a professional (formal care)?"

Recommendation: interviewees highlighted the fact that there is no trained professional caregivers who could be able to provide continuous care or monitoring for a person with PWS. As such, family members even if they would want to hire a professional caregiver, are not able to due to the lack of service available. Advocating for the formation of professional caregivers for PWS and the coverage of those services by the state or insurance would relieve family members from a significant burden. Even in developed economies, informal care represents a substantial economic burden. Moreover, if those services were available, informal caregivers could dedicate more time to work, limiting indirect costs (in particular the loss of income).

<sup>25</sup> López-Bastida, Julio, Renata Linertová, Juan Oliva-Moreno, Manuel Posada-de-la-Paz, Pedro Serrano-Aguilar, Panos Kanavos, Domenica Taruscio, et al. "Social/Economic Costs and Health-Related Quality of Life in Patients With Prader-Willi Syndrome in Europe." *The European Journal of Health Economics* 17, no. S1 (2016): 99–108. <https://doi.org/10.1007/s10198-016-0788-z>.

## **Case study 5: Ireland, A population-based profile: the financial impact of caring for an individual with PWS across groups**

The Prader-Willi Syndrome Association Ireland (PWSAI) and Trinity College, Dublin collaborated to publish a report aimed at assessing the needs of people with PWS and their families in Ireland. Findings were collected through a survey given to caregivers asking about their experience of caring for an individual with PWS as well as their point of view about available support. This report mapped all aspects of PWS patients and their caregivers. Results on the financial aspect of supporting a person with PWS in Ireland are summarised below.

### **Perceived economic burden of PWS in Ireland**

Participants in Ireland were asked to think about the financial consequences of raising a child or supporting an adult with PWS. They were asked to estimate whether PWS has a small negative impact, a significant negative impact, an extreme negative impact or no impact at all. Results were then compared between different age groups. It has therefore been shown that in Ireland, the **financial burden** of PWS for families is **greater when the patient with PWS is older**. The majority of people who selected “extreme financial impact” were families supporting an adult living at home.

### **Loss of income**

Overall we can see that PWS has a huge impact on time dedicated to work in Ireland. It is important to note that the majority of respondents were mothers. The financial burden tended to fall unequally on women underlining strong gender inequalities. Caregivers of adults in supported accommodation were less likely to experience reduced working hours.

### **Conclusion:**

It is difficult to assess the financial burden caused by the impact of PWS on families. However, we can see that many caregivers across all age groups experienced significantly reduced hours dedicated to the labour market. Apart from the main caregivers supporting an adult in an adapted home, loss of income can be assumed to be highly significant. Most respondents had access to some entitlements and financial benefits in respect of their relative with PWS. However, given the high number of respondents who reported being denied access to a medical card, it can be assumed that some parents may have had to finance health services themselves.

### **Recommendations:**

- More research on costs associated with PWS to target carers needed.
- A review of access to welfare services.
- Advocate for universal and equitable access to entitlements and benefits.
- Increase the number of adapted homes and improve access to reduce loss of income because of time dedicated to informal care activities.

## **SECTION 5: Inequalities faced by families in different parts of the world, Interviews**

### **Introduction**

Interviews with parents of a child with PWS across different countries have emphasised strong inequalities in healthcare coverage. Parents from Denmark, Morocco, Brazil, Romania, Austria, the United States, Ireland and Qatar responded to several questions regarding costs associated with the needs of their child and to what extent they are publicly supported. Our questions focused on:

- Direct medical costs and whether they were provided by the state or reimbursed by insurance or borne by the family.
- The use of social services and state-funded measures to support these costs.
- Education costs associated with specialised education.
- Home adaptation costs to meet the needs of the child / adult with PWS.
- Transportation costs to attend medical appointments.
- Loss of income.
- Change in costs over time, relating to the age of the child.

Depending on the scope and nature of a country's healthcare system, answers varied considerably. Only the mother from Denmark felt that raising a child with PWS did not involve extra costs. Overall, most families faced significant out-of-pocket costs even if their country had an established healthcare system, generally covering basic direct healthcare expenditure.

In most cases, families outlined the lack of awareness of these costs and important lack of services available.

- Interviews raised a common theme concerning therapies and early intervention programs for children with PWS. Several therapies are needed regularly. In several countries, families reported that the state covered or provided insufficient therapies, forcing families who could afford it to pay for privately provided healthcare.
- The interviews raised a concern about the lack of social services available. Expenditure related to social services did not appear to be significant as even in developed countries, few or no professional carers were trained to manage PWS, so employing them was not an option.
- Very few families had access in their country to specialised homes or centres for their child.
- Loss of income was also significant cost for families across the world, unequally affecting women. Very few countries offered a financial compensation for this loss of income.
- Not all families had access to financial support or grants from the state to support families with a disabled child. Often, even when those kinds of financial benefits existed, it was very little compared to the costs faced by the family.

- Education costs were substantial in Qatar. Most families paid for private education, but it was a rather a personal choice not linked with PWS, rather than an obligation due to the need of specialised schooling.

## Denmark:

The situation in Denmark is perhaps unique. The mother of an adult with PWS was the only one of the families interviewed who said that raising a child with PWS did not entail any additional costs. The state had established and comprehensive health coverage measures to ensure that an individual with a disability would never face economic instability.

Firstly, all **necessary direct health care costs** are covered by the state in Denmark. Families do not pay for Growth Hormone, for medical appointments or medical diagnosis. But the Danish health system does not only cover medical care. The state ensures that all the needs of disabled people are met. Adults over 18yrs who are not able to work because of their condition, receive a sum of money from the state, allowing them to pay for adapted houses, food, and spending money.

Raising a child or supporting an adult with PWS in Denmark does not entail any additional costs compared to raising a child without the disease. It may even be cheaper. A child without a disability will participate in many extracurricular activities, which are quite expensive, that a child with PWS may be unable to take part in.

### Loss of income:

In the traditional Danish family model, both parents work 37 hours per week. Very few women or men choose to stop working. When a couple has a disabled child, one of the parents (often the mother) may in some cases decide to resign. In this situation, **the parent receives compensation from the state to cover part of the loss of income.** In addition, to relieve the family of the burden of informal care, the municipalities have set up a system where the disabled child can spend some weekends with another family (respite). This measure is paid for by the municipalities.

### Social services:

This was not a cost either for the mother from Denmark, she did not spend on social services. She received considerable help from her parents to care for her son. However even if many families find solutions to manage daily care as she did, for some who don't have access to family support or respite, daily care could be more challenging.

### Transport costs:

The mother lived near the hospital, but sometimes she had to go to Copenhagen to see specialists. Transportation costs were not paid by the state. However, low income families could ask municipalities for financial support for this purpose.

The mother had **to adapt her home** due to the condition of her son. However **she did not pay for home adaption costs.** She was living in a hospital flat and she had to

remove steps in front of the doors because her son couldn't use the stairs. The municipality paid for those costs. When you have to adapt your home in Denmark because your child is in wheelchair or unable to move around the house, **the municipality covers the costs.**

### **Education:**

The family did not have any **costs related to education**. Both mainstream and special school **are paid for by the state**. Unless parents choose a private setting for education, schooling is generally provided by the state.

The mother even added that **raising a child without PWS could be more expensive than raising a child with PWS**. What is costly in Denmark are **extracurricular activities**. For her child with PWS, some sports activities were arranged by the municipality for disabled children. **She did not have to pay for those extracurricular activities.**

Raising a child or supporting an adult does not appear to entail any direct costs to the family in Denmark. The state covers and provides for healthcare services but also provides excellent support to caregivers and other non-medical aspects of the care of a person with a disability. The state is able to provide all those services for the Danish population because of its taxation system. Indeed 50% of the mother's income goes to taxation which is used to fund this universal and broad health system.

### **Ireland:**

For the mother in Ireland, costs associated with raising a child with PWS were significant. The greatest cost was her loss of income. Indeed even if the state provided a grant for social services, no professional is trained for PWS daily care. Therefore she had to first reduce her work time and eventually stopped working to care for her son. Indirect loss of income was her greatest cost and was not compensated by the state as her husband did not meet the low-income criteria. Overall the costs were greater when her son was a child (he is now 17 years old). As in many other countries, only direct healthcare costs are well supported by the state. She also faced significant expenses for home adaptation and transportation costs. She had witnessed and experienced a strong lack of awareness by the state of the costs associated with raising a child with PWS.

Most **direct healthcare services** are provided by the state. However, the mother experienced some extra costs associated with healthcare services for her child with PWS. The mother used to spend €144 per month for her son's Growth Hormone for a period of about 6 years. The full cost of Growth Hormone is much higher but, even with the reduced price, the cost of prescription medicines is still very significant. Recently, new measures were implemented for people with significant disabilities and **Growth Hormone is now free of charge for individuals.**

Inpatient care at birth were paid for by the state. The family would only pay for extra costs, for example for a private room in hospital.



### **Direct non healthcare costs:**

- **Transportation costs** are significant. The family often need to travel across the country to see specialists. It cost about €100 per trip. Petrol and parking expenditures to go to medical appointments are a big expense for the family.
- **Home modification:** children with PWS have hyperphagia, an abnormally increased appetite for consumption of food. To manage their son's access to food, the family had to build a brand new kitchen as it was in the middle of the house before. There are as well a lot of costs associated with repairing items broken because of behavioural issues of their son. Moreover, there are some costs associated with special equipment needed at home and some items to deal with her son's anxiety. Those costs are not covered by the government and are substantial.
- **There were no special costs dedicated to education:** apart from extracurricular activities costs, essential to develop the motor skills of a child with PWS.

### **Social costs:**

The state provides a social services grant of €1,850 per year that is supposed to help with carers costs. However, as mentioned by other families, there is no one trained to care for a child with PWS. So even if the family wanted to spend money on social costs, those services are not available. The mother emphasised that she would rather have access to social care services than receiving a grant.

### **Loss of income:**

**Loss of income has overall been the greatest cost associated with raising a son with PWS.** Before her son was born, the mother used to work full-time. When he was born after his stay at hospital, she reduced her work to 40% (2 days per week) , she then stopped working for 11 years and she has recently started working again 15 hours per week. Overall, her loss of income has been considerable. Her husband has not been impacted in terms of loss of income. **She did not receive any compensation from the state** for that loss of income. Grants provided to informal carers exist in Ireland but are means-tested. **Only very low-income families are eligible for these grants.** Although her career was significantly affected by her son's disability, her husband's income was above the limit for compensation.

### **State financial support:**

One other state financial support is tax credits. As the mother stopped working to care for her son, her husband had a slight tax reduction. **However, this tax reduction is minimal compared to the loss of income experienced by the family.**

Finally in Ireland when a family has a child with a significant disability, they receive €309 per month to spend as they need. When the child reaches 16yrs, this financial support stops and is replaced by a new payment of €209 given directly to the person with the disability.

## Austria:

For the mother from Austria, the costs related to raising a child with PWS depend, as with any other child, on the standard of living one wants to achieve. But overall there are extra costs associated with PWS. In Austria, financial supports exist for people with PWS and their caregivers. Her daughter can meet her own needs and is living in an adapted house in Germany, her place is funded by the Austrian state. However, the mother outlined the lack of social services available and potential loss of income for families.

### Direct healthcare costs:

In Austria nearly all medical costs are covered by the state. Only some extra therapies that parents found useful were not covered. **The diagnosis played an important role.** Once the disability is known it is a lot easier to benefit from state financial support. When you have a child with a disability, **families benefit from increased family allowance.**

### Direct non healthcare costs:

- **Transport costs** are not covered unless it is an emergency. **Social services** are not covered, but few families use professional caregivers as there are very few people trained to care for people with PWS. The mother's daughter is now living in a supported home in Germany. Supported homes are quite different in Austria compared to Germany. In Austria, patients have no salary, only some pocket money, whereas in Germany people living in supported group homes have access to social security and a salary, and when they are not able to work anymore they have a pension.
- **Education costs:** The mother paid for education but this was because she chose a private school, the education costs were not related to PWS.

### Loss of labour income:

As in many families, the time spent caring for children with PWS is substantial. The mother had a fairly flexible job, allowing her to make up the time not worked during the day during the evening. Her husband retired early. **Overall, the loss of income is quite a big problem for families. There are no public measures to compensate for this.**

Her ongoing costs are all the costs associated with 'extras' such as holidays, travel, mobile phones, etc. - costs that a young person of her daughter's age would normally be able to afford herself. Saving for the future is also an important ongoing cost for the family. **However, her daughter, thanks to her small income and state financial support can meet her own needs.**

Economic support from the state exists in Austria for people with PWS. However, accepting the disability of a child can be very hard for a family, so they often don't claim financial help. For example, there is a card which gives some financial advantages for disabled people. This card can for instance give reduction to the swimming pool or

travel discount. Many families don't ask for this card as they do not wish to accept or acknowledge the disability of their child.

## **Romania:**

**The father interviewed assumed that raising a child with PWS entails more costs than raising a child without a disability.** Even if the states provide some healthcare services, it is only at a basic level and of a lower quality than private facilities. A family can not only rely on public services to ensure the best for their child. There are no policies broad enough to compensate for the range of costs associated with raising a child with PWS .

### **Direct healthcare costs:**

In Romania, as is most European countries, every citizen benefits from a state healthcare system. **However, as in many other countries, public provision of healthcare services is not broad and of a high standard.** As children with PWS require intensive therapies and medical care, families often need to **go to private facilities to complement public services.** The father had to pay for the diagnosis which was around €200. Growth Hormone is paid for by the state. But only a small part of the therapies needed to ensure the best possible development of the child, is covered by the state. **Public facilities are limited.** If you want your child to have a special therapy every week, parents need to go to private clinics and **cover the costs themselves.**

### **Financial support:**

The state offers **small financial supports** to families with disabled children, however it is insignificant compared to the costs associated with PWS. The family have **received a grant from a charitable association.**

### **Loss of income:**

The father did not experience any loss of income personally, **but his wife did.** She had to reduce her hours of work to take care of their children and felt that her career was impacted. The state did not provide any financial compensation for this loss.

The father felt that there was not enough awareness about the economic burden of raising a child with PWS as this condition was so rare. As with many other parents we hear from, he would want a respite centre or a residential home in Romania as seen in Germany.

## **The United States:**

In the United States, raising a child or supporting an adult with PWS is extremely costly. The situation is quite different from the European model. The US has a liberal welfare system, which involves a minimal role for the state in the provision of social services

and is based on means-tested benefits.<sup>26</sup> Low and middle-income families are often not able to afford sufficient, good quality care for their child. Individuals often need to attend private facilities to receive health services. The main issue in the United States is the substantial costs of direct healthcare services.

### **Direct healthcare costs:**

In the United States, the major part of the population is privately insured. This insurance covers 80% of each cost related to health services and medication. Managing PWS, especially at a young age, is very costly. A child with PWS requires many special therapies and constant medical monitoring. Many children are also on Growth Hormone. Even if the private insurance covers most of costs, the remaining 20% is paid for by out-of-pocket money. **Adding up those costs, many families are not able to afford vital care.**

As an example, the mother from the United States told us she had to spend \$700 per month for Growth Hormone. The mother, then applied for a state-funded insurance, which can be accessed by low-income families. Since her child turned 7 years old, she has paid \$11,000 a year for this public insurance which covers all medical costs. Without this, the mother estimated that she would have spent about half a million dollars a year for healthcare services.

The issue **is this insurance is only available for families under a certain income level.** However some families may be above this line, not eligible for public funded insurance, but are still not able to afford quality health services. Costs associated with PWS healthcare services therefore represent a huge problem for families. Many families can't afford essential resources for the health and development of their child.

### **Social services:**

The son of the mother interviewed lives in an adapted house, so the mother doesn't have costs related to the use of professional caregivers at home. This home is state-funded because the mother is publicly insured. Overall it costs around \$200,000 to \$300,000 a year to provide food, shelter and staffing for his placement there. Without this service the mother would not have been able to work and the loss of income would have been considerable.

The mother considered that her experience is far from being typical of other families, rather she is the exception to the rule. **Most families in the United States and around the world, don't have housing or medical care for their children,** they're fighting with insurance companies for Growth Hormone. Accessing public insurance and finding good quality services to support a relative with PWS is very difficult. There is not enough awareness about the economic burden of PWS in the United States.

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• <sup>26</sup> Esping-Andersen, The Three Worlds of Welfare Capitalism.

**Additionally, in the US there is hesitancy to use a state-funded programme. People who apply for state support can sometimes be seen as overly relying on the state or taking advantage of the system.**

## **Morocco:**

A mother living in Morocco shared her experiences regarding the costs associated with raising a child with PWS in her country. **As with many of the people we spoke to, she felt that raising a child with PWS entailed extra costs.** Costs directly related to healthcare represented the most significant part of the additional costs associated with the PWS. Families in Morocco lack the resources that would allow their child to achieve a certain degree of independence. Medical appointments such as, physiotherapy, speech therapy and psychological therapies, are extremely important for a child with PWS. Thanks to those therapies, a child with this syndrome can achieve a better quality of life. There is a lack of awareness of the effectiveness of the therapies which are often perceived as a luxury even though they are in reality essential.

### **Direct healthcare costs:**

Medical costs mainly related to the numerous medical appointments during childhood to support the child's development. Therapeutic sessions were recurrent and essential. Her son had about 2 to 3 appointments per week which costed €20 each during his childhood. The costs of the diagnosis and medical analysis and MRI were also significant expenditures for the family. The diagnosis test for PWS, cost €350 and was not available in Morocco. The price of an MRI scan was €750 at the time, but now it is less expensive. Analysis scan expenditure associated with medical analysis cost about €200 to €300 per month.

Care in public hospitals is paid for by the state, however appointments with specialists as mentioned above, as well as various analyses such as MRIs, are not covered. Although extremely important for the development of a child with PWS, those services are perceived in Morocco as "luxuries". Moreover, in theory, families have the opportunity to have their child treated in public medical facilities, paid by the government. Unfortunately, as PWS is a rare condition, hospitals often lack the resources and knowledge to provide the most appropriate healthcare. The best quality services are often provided by private clinics, which are more costly.

### **Insurance coverage of healthcare services:**

There are several types of health insurance. Individuals working in the public sector benefit from state-provided health coverage, those working in the private sector are insured by private health insurers. Some professions do not have health insurance coverage.

### **Direct non-healthcare costs:**

**Social services costs:** this was not a major burden. The mother had someone to help her at home in general with domestic tasks but not particularly in relation to the PWS. Her son is very autonomous.

**Education costs:** the mother paid for her son's education, but she did not face extra educational costs associated with the PWS. She did not experience a difference in costs related with education between her son with PWS and his siblings.

**Transport costs:** the use of the car was vital to attend numerous medical appointments. Petrol was therefore a cost, but not seen as a significant one.

**Loss of income:** even though she has left her job, she does not associate it with her child's disability, but more with her family life in general

Overall, the mother's costs were greater when her son was a child, her greatest expenses were for direct healthcare services: the numerous appointments with different specialists. Her experience is not typical of that of other families in her country. She had access to private insurance and was able to afford the medical services her son required. Moreover, her son is very autonomous, he was able to have a normal education and didn't need constant monitoring.

## **Qatar:**

Raising a child with PWS in Qatar entails substantial costs. The mother of a daughter with PWS living in Qatar shared her experiences. Even if Qatar's government covers all basic healthcare costs of everyone (even visitors), expenses related to early development programmes can be significantly high. Moreover, education costs present a huge challenge for families. The few inclusive schools for children with disabilities are private. In addition, expatriate children (90% of the population is expatriate) do not have access to public schools. This is a huge issue as most children with PWS do not have access to education.

### **Direct healthcare costs:**

In Qatar, all residents have health cards which allow them to access public provision of healthcare services. With this card, families only pay for administrative fees which are about £10 per consultation. Growth Hormone treatment is highly subsidised. You don't have to be a citizen of the country for your treatment to be covered. Her daughter is covered under the government's exemption programme for people with disabilities. She can access the best care, free of charge. This recent program allows individuals with disabilities to pay reduced health care costs.

However, in Qatar, information about these kind of measures is very difficult to get. A person who has just moved to this country will not know about this programme, especially if they do not speak Arabic. A typical family pays about £10 for the administrative costs of health services. However, under the programme for people with disabilities, the government will cover more of these costs. For example, the mother from Qatar will only pay £2 to £5 for the administrative costs of her daughter's medical care. All her daughter's basic medical care is provided in the same hospital and covered by the state.

A child with PWS needs early intervention programmes to improve cognitive, academic and social outcomes. These therapies are quite specific and ongoing during childhood.

**Early intervention programmes can be provided in theory by the state free of**

**charge.** However, the need for therapies continues to grow as more and more children are getting diagnosed, but support structures have not grown despite the growing demand. **The state is unable to provide enough of this care for children with PWS.** Therefore, there is an obvious need to complement the care with private sector provision. The price of private provision of medical care is not regulated by the state. The price is set by market forces and is therefore extremely sensitive to considerable increases. **Low-income families earning about £1,000 a month are not able to access special therapies because the public provision is limited and the private provision unaffordable.**

### **Education:**

Education also represents a considerable cost for families raising a child with PWS in Qatar. The education system is divided between the public and private sectors. Public schools offer a Middle Eastern curriculum and public schools an international curriculum. Qatari citizens with Arabic as their first language have access to public schools. These schools are in Arabic only. **However, about 90% of the population of Qatar is made up of expatriates,** who do not necessarily speak Arabic. **Expatriate parents are therefore often forced to enrol their children in British schools, which are privately run and very expensive.** In addition, the higher-level schools are often those with a British or American curriculum.

Middle and lower-level schools (Middle Eastern programmes) are affordable. **However, they are not inclusive of children with disabilities.** A child with Prader-Willi syndrome has very little chance of getting into a highly competitive Indian school, for example, even if the tuition fees are more affordable. **In Qatar, only 5% of schools are inclusive.** Most inclusive schools are British or American schools, which are expensive. Tuition fees are around £8,000 per year. In addition, children with Prader-Willi syndrome often need a learning assistant and additional teacher, which adds up to about £3,600 and £12,500 per year respectively. **British education is therefore unaffordable for the vast majority of families in Qatar. Most families are unable to enrol their child in school, and the child is placed in a therapy centre or remains at home because education is unaffordable.**

### **Brazil:**

In Brazil, citizens, residents and visitors have access to a universal public health care system funded through tax revenues. This health systems provides for free services including outpatient care, prescription drugs, and outpatient specialty services. Approximately 75% of Brazilian citizens rely solely on this universal health care system, however more and more middle and high-income families have sought private care to complement public provision of health services.

Private health insurance is voluntary and complementary to the SUS (universal public health system) and regulated by the National Agency for Complementary Health. In 2018, 23% of Brazilians had private medical insurance<sup>27</sup>. Most individuals benefit from a private health insurance through their employment benefits. Out-of-pocket

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<sup>27</sup> <https://www.commonwealthfund.org/international-health-policy-center/countries/brazil>

expenditures for health however still account for considerable expenses for households and unequally affect low-income families. In 2014, 5.3% of households experienced such high health costs that they had to forego paying for non-health related goods. The cost of medicines was one of the main contributors to this, as only certain medicines are available free of charge under the SUS.<sup>28</sup>

Two Brazilian families were interviewed. A father with an 8 year old boy and a mother with a 9 year old daughter.

### **Direct healthcare costs:**

The mother interviewed was a member of a corporate health plan which covered inpatient care. Costs associated with Growth Hormone were paid for by the public health department. However, the costs of therapies are borne by the family. Her child is 9 years old, and didn't need all the therapies that he used to need. The family only pays for psychotherapy which is around \$320 per month.

A father from Brazil reported that Growth Hormone prescriptions were publicly provided, thus free of charge for the family.

However, for medical appointment and therapies, public provision is not good quality. For the father, accessing public services would have been more complicated and his child would not have been able to have all medical appointments and healthcare that he needed. The father has insurance that partially covers this costs (about 70%), however the remaining part is a significant economic burden for the family. For his child, the diagnosis was reimbursed by this insurance. The insurance is an employee benefit.

### **Direct non-healthcare costs:**

**Transportation:** the mother faced significant costs associated with transportation. They travel to the United States every two years to see a specialist. These costs are not covered by the state.

For the father interviewed, he stated that transport costs were not significant, as all services were available in the city he lives in.

**Education:** education is one of the greatest costs associated with PWS for the first family interviewed. The woman's daughter with PWS has a private tutor to help with academic progress. The tutor costs about \$380 a month and none of this is reimbursed by the state or by any insurance.

For the father of the 8 year old boy, education is costly as public provision of education is not good in Brazil. However, this is not related to PWS as his son is currently enrolled in a regular school.

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<sup>28</sup> Ibid



**The first family does not have any costs related to loss of income or social services.**

**The family of the boy, experienced significant loss of income. The mother does not work in the morning to go to several therapies with her son.**

Neither of the families interviewed hired **professional caregivers** to support the child with PWS. The father mentioned that social services costs would not be supported by the state and costs would be totally borne by the family.

**Financial support from the state:**

The father mentioned that the government did not provide any subsidies or financial support to families with a disabled child. Extremely poor families might receive some financial assistance, but it will remain low compared to the costs associated with the condition.

The mother emphasised that her experience is far from being typical of other families. Indeed, the family interviewed is socially advantaged. They could afford private education and good healthcare services.

However, as in some other countries, many families have little access to early development programmes and special therapies. Access to good quality healthcare services in Brazil is an ongoing issue for many families caring for a child with PWS.

The father also perceived himself as very lucky. His experience is far from being typical as he has an advantageous economic situation. He mentioned that he could manage the disease of his son thanks to his situation, and that it is much more challenging for families with lower income.

He would want more specialised centres to support families with a child with PWS and for these to be free of charge.

## CONCLUSION

Overall, the experiences of parents were extremely different around the world. Only one parent from Denmark felt she did not have extra costs. Interviewees emphasised that parents over the world, even living in economically developed countries, face substantial costs.

In all countries basic healthcare costs were provided by the state. However, the care and management of PWS entails many other healthcare services, that are not considered “basic”. Overall, in many countries the state may cover part of direct healthcare costs such as Growth Hormone, inpatient costs and the cost of the diagnosis, however it does not entirely support the therapies needed during childhood. Many parents interviewed have highlighted how those therapies: physiotherapy, speech therapy, and psychological therapies were life-changing for their child.

According to most parents interviewed, the majority of families in their country are not able to pay for those health services even though they are essential for their child. In many countries such as in Morocco, Qatar and Brazil, public provision of healthcare is not developed enough. To access good quality health services, parents from advantaged backgrounds need to go to private facilities to have adequate care and low income families are not able to access those services. Private insurance, often helps cover those costs but not everyone has access to this.

### **Apart from direct healthcare costs, interviews highlighted many hidden costs related to raising a child or supporting an adult with PWS.**

The main indirect cost was in most countries, reduction of income. Especially for children, the time dedicated to managing PWS can be substantial. Of course each case of PWS varies considerably. Some children will need more monitoring and intervention than others. However, many parents have experienced significant loss of income. Often the mother, needs to reduce her time at work to manage the needs of the child with PWS. As there is a lack of trained professional caregivers and lack of knowledge about the condition, even if parents would have wanted to hire a professional this is simply not an option.

Apart from Denmark and Ireland (subject to a low income criteria) there is often no financial compensation from the state to support informal caregivers.

### **Direct non healthcare costs were also a significant burden on families.**

Costs related to specialised **education** were particularly high in Qatar, where inclusive education was only available in private settings, leading to the exclusion of many disabled children from a full education.

Overall families interviewed did not especially consider education as a major cost linked with PWS. Even if their child was enrolled in private settings, it was because of personal choice unrelated to PWS.

Home modification costs can be also quite significant for families. The mother from Ireland spent a significant amount of money to rebuilt the kitchen to meet the needs of her child. At a basic level families will have the cost of locks and similar modifications to manage access to food in their home.

Transportation costs can also be quite significant. As knowledge about PWS is limited, specialists are often located in big cities. Families may travel long distances, even abroad, to see a specialist.

Social services did not appear to be significant as all families reported the lack services available due to the rarity of the condition.

Some financial benefits exist in many countries for families with a disabled child. However, these grants are often inadequate or do not solve the issue (for example in Ireland where a grant is offered for social services but there are no social services available) or very low compared to the costs experienced.

Overall, it seemed that families living in countries with advanced healthcare systems, tended to have more expenses related to loss of income. Very few countries offered state compensation for that loss. Except for Denmark, no countries offered financial compensation for direct non-healthcare costs where there are often substantial hidden costs for families.

Countries, less economically advanced or developing, tended to have a weaker healthcare system. Public provision of welfare services such as education or health services are often neither good quality or available to all. Families must often go to private facilities to complement or to access health services. In those countries, huge inequalities exist as low-income families are not accessing the care each child or adult with PWS needs. For families living in countries where the healthcare system is less developed, greater costs are linked to accessing education or therapies not provided by the state.

## Summary

**Estimating overall costs related to rare diseases from a societal point of view is needed for adequate health policy implementation.**

There are few research papers aimed at evaluating costs associated with rare diseases. Cost of illness papers are nevertheless extremely important in identifying patients' needs. Research on costs allows for prioritisation and justification of healthcare policies. Most research papers we found estimated costs of rare disease focused on the United States or the European Unions. There is a significant gap of research elsewhere

which needs to be filled. Advocating for more research, collecting data and finding contacts and families in other part of the world is extremely important to have a greater sense of the economic burden of families associated with a rare disease and PWS in particular.

Costs associated with PWS are likely to be substantial and much greater than costs associated with chronic diseases. The researchers' objective was often to estimate costs from a societal perspective. Those figures are important for health policymakers. Knowing what the costs are is necessary to allocate sufficient resources to manage PWS and rare diseases in general.

### **The burden of informal care**

Both interviews and research papers have outlined the burden of informal care activities. People with PWS often require constant monitoring. As it is a rare condition, very few professionals are trained for daily caring activities. When there is no formal care available, relatives must take on substantial caring activities. Parents or relatives raising a child with PWS are likely to experience significant loss of income. This indirect burden often falls unequally on women.

### **Direct non-medical costs**

Direct non-medical costs can be substantial. These are often not well recognised and are publicly supported in very few countries. Knowledge about those costs is key to raise awareness and adequacy support families.

### **Limited public healthcare system**

In many countries, especially **in less advanced economies**, public healthcare systems are not broad enough to provide enough services for the entire population. This leads to huge inequalities. Low-income families can't access the care they need because they can't afford private services. As health services related to managing PWS are quite specific, public services are not enough developed to provide effective support to all individuals with PWS.

### **Lack of support and information**

There is an overall lack of awareness of the economic consequences of PWS. Families are generally not well supported by public services. Information about insurance, health supports and benefits are hard to find. Building a worldwide community is extremely important for families to share knowledge and tackle common challenges.

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