

A Mother's Experience: a report on the social and psychological experiences of mothers raising children with Prader-Willi syndrome

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About this report:

Vandana Venkat is an undergraduate student at the London School of Economics studying International Relations. As an IPWSO intern, she has been tasked to research and write a report on mothers as caregivers to children with Prader-Willi syndrome.

In this report, Vandana is working towards understanding the social and psychological experiences of mothers raising children with Prader-Willi syndrome by summarising literature and interviews she has conducted. Through this project, she hopes to provide IPWSO with practical recommendations to better support mothers around the world in their journeys raising their children with Prader-Willi syndrome.

Introduction:

Prader-Willi syndrome (PWS) is a complex and rare genetic condition that causes a wide range of physical symptoms, learning difficulties and developmental challenges. Studies place the incidence of PWS between 1 in 10,000 to 30,000 individuals in the general population and is shown to affect male and females in equal numbers across all ethnic groups and geographical regions in the world. Prader-Willi syndrome occurs when there is a lack of expression in a specific group of genes on chromosome number 15. This leads to several neurodevelopmental problems that affect part of the brain called the hypothalamus, which produces hormones and regulates growth and appetite.

Signs and symptoms of the syndrome include:

- Low muscle tone and floppiness (hypotonia). This is usually present at birth alongside the failure to thrive and the need for tube-feeding.
- Excessive appetite and overeating (hyperphagia), leading to dangerous weight gain. This usually develops at a later stage between 4 to 6 years old.
- Restricted growth and short stature
- Lack of sexual development
- Learning difficulties
- Behavioural and psychological challenges

It is particularly important to understand the nuances of Prader-Willi syndrome as a rare disorder. While the syndrome can be viewed in many different perspectives, be it from a neurodevelopmental or an intellectual disability standpoint, identifying Prader-Willi syndrome as a rare disorder specifically is critical in order to truly understand the extent of struggles experienced by various individuals and families.

Although rare diseases affect a small fraction of the population, a study by Orphanet confirms that 300 million worldwide are affected by a rare disorder¹. The co-author of the study, Yann Le Cam, aptly states that “collectively rare diseases are not rare”² as the effects of rare disorders is a public health priority that impacts millions around the world. Additionally, the impacts of rare disorders extend beyond diagnosed individuals, with families and caregivers being affected by insufficient social healthcare systems.

By recognising Prader-Willi syndrome as a rare disorder and acknowledging the public health inequality associated with it, one can internalise the challenges experienced by families and caregivers around the world. This is particularly pertinent for issues surrounding accessibility, such as to genetic testing, early diagnosis and intervention in order to prevent the escalation of Prader-Willi syndrome symptoms like obesity.

¹ Stéphanie Nguengang Wakap et al., “Estimating Cumulative Point Prevalence of Rare Diseases: Analysis of the Orphanet Database,” *European Journal of Human Genetics* 28, no. 2 (2019): pp. 165-173, <https://doi.org/10.1038/s41431-019-0508-0>.

² “EURODIS,” *EURODIS*, September 17, 2019, https://download2.eurordis.org/pressreleases/PrevalencePaper_JointStatement_170919_Final.pdf.

Section 1: Why is it important to look at the roles of mothers?

Narratives of parents with children with disabilities have remained at the edge of larger medical and social systems, with parental needs being taken for granted. When zooming in closer, parental experiences when caring for a child with a rare disease are even more unrecorded due to the lack of support in accessing complex care services.

Mothers typically are made more responsible for managing the daily medical and social care needs of children with rare diseases³. The social constructs placed on a mother's worth to raise the ideal able-bodied and able-minded child is one of the many factors that contribute to the suppression of parental struggle. Such societal expectations of the mother merge the experience of the disabled child with the mother's physical and social body as she navigates through various social structures⁴. The mother becomes an inextricable part of her disabled child's vulnerable spaces and is silenced in the process. Feminist disability theorists categorise both mothers and their children as subjects to their social, cultural and environment, in which the mother as a carer is figuratively silenced as both a 'witness and participant'⁵, experiencing the public landscape of the disabled child.

This report thus aims at understanding the social and psychological experience of mothers of children with rare diseases, specifically Prader-Willi syndrome, to gain insight into their lives as carers. Personal parent narratives about caring for children with rare diseases can offer deeper insights that exist beyond current literature.

The Personal Is Political

Qualifying a mother's personal experience into a published text can be considered political by giving mothers the agency to platform their voices. Writing and publishing the PWS motherhood story signals that her mothering experience is significant, worthy and a resistance to predetermined social expectations and predetermined roles⁶. Existing literature has identified that having a child with Prader-Willi syndrome entails many

³Currie, Genevieve, and Joanna Szabo. "It Would Be Much Easier If We Were Just Quiet and Disappeared": Parents Silenced in the Experience of Caring for Children with Rare Diseases." *Health Expectations* 22, no. 6 (2019): 1251–59. <https://doi.org/10.1111/hex.12958>.

⁴Robertson, Rachel. "Misfitting' mothers: Feminism, Disability and Mothering." Other Journal Article, *JOUR. Hecate* 40, no. 1 (2014): 7–19. <https://search.informit.org/doi/10.3316/informit.286336999740490>.

⁵ Ibid

⁶ Garlick, Lynn. "Contemporary Australian Motherhood Memoir As Personal, Creative, and Political Narrative". *Journal of the Motherhood Initiative for Research and Community Involvement* 7 no.)(2017). <https://jarm.journals.yorku.ca/index.php/jarm/article/view/40327>.

challenges in parenting. Apart from the direct healthcare costs such as hospitalisations, medicine and support services, mothers face indirect social costs. Therefore, allowing mothers to share their experiences and struggles through this report is critical to raise awareness about raising a child with Prader-Willi syndrome and the effects on the caregiver.

Identifying Barriers in Accessing Prader-Willi Syndrome Resources

Very often, people with rare diseases are not recognised as potential users of the social welfare system. Additionally, many countries lack a classificatory process within their healthcare systems in which a lack of specific social care services results in inadequate social care. This renders a lot of families living with Prader-Willi syndrome invisible as a result of professionals' lack of knowledge and leads to limited support. Mothers thus have to overcome care challenges within their respective healthcare systems and manage such gaps in social care to provide for their children⁷. Identifying these core challenges in accessing Prader-Willi syndrome resources can help improve understanding of what support they require to better meet their needs.

Meeting the Needs of Caregivers of People with PWS

Since Prader-Willi syndrome only affects a small percentage of people, there is only a limited amount of research and resources available to assess the needs of caregivers. Hence, many of the needs of caregivers, including mothers, are significantly unmet due to the lack of sufficient resources. Many mothers sacrifice their careers to dedicate more time to their children, as caring for someone with Prader-Willi syndrome requires consistency monitoring. Alongside the loss of labour productivity, many mothers experience psychological impacts from dedicating the majority of their time to informal care. Thus, the needs of caregivers must be spotlighted and monitored to reduce the struggles faced in parenting a child with Prader-Willi syndrome.

Reducing Inequality

Past research has analysed the inequalities in different regions in the allocation of health resources to support rare diseases. Not only is there disparity in research regarding the challenges in different regions, there is very little research surrounding the social stigma and inequality surrounding parenting and rare diseases. This report principally aims to map such systematic and social inequalities faced by mothers of children with Prader-Willi syndrome and provide solutions on how to better support them.

⁷ Currie and Szabo. "It Would Be Much Easier If We Were Just Quiet and Disappeared" 1251–59.

Section 2: Overview on Maternal Struggles Surrounding Diagnosis, Early Intervention and Access to Healthcare

Understanding the immediate reactions and challenges faced by a mother upon learning about her child's Prader-Willi syndrome diagnosis is a nuanced task. A considerable amount of emotions are involved, specifically surrounding the Prader-Willi syndrome diagnosis, early intervention and issues surrounding access to healthcare and resources. Employing social research to understand the specific impacts of a child's rare disease diagnosis is thus pertinent.

Clarifying how mothers raise children with rare diseases is essential for medical and other professionals to be better equipped to provide more efficient support in the future⁸. Rare disease research is in need of a greater understanding of mothers and their longitudinal struggles towards in order to shape better policies.

Across a range of studies, the lack of knowledge and provision of care and support by medical professionals has been identified as a major concern amongst mothers - especially in regards to the immediate Prader-Willi syndrome diagnosis. According to a 2015 rare disease study on the importance of training and provision of information by physicians, most participants were shown to have not received support during the early years of their child's diagnosis⁹. Mothers generally report being dissatisfied with health professionals regarding their level of knowledge surrounding their child's disease, the process of gaining a formal diagnosis and feeling supported by the care-team provided - with various surveys pointing towards a majority of parents feeling insufficiently supported by their medical care teams. Despite parents of children with chronic health conditions encountering similar issues, mothers of children with rare diseases face many additional problems such as: delayed or undetermined diagnosis, limited access to health information and resources and limited support groups and services (if existing) that are geographically scattered and feeling socially isolated.

Studies on the equity of care provision for mothers of children with rare diseases is thus critical to determine what is missing in early intervention support for mothers and to

⁸ Saeko Kutsunugi et al., "Mothers' Experiences of Parenting a Child with Chromosomal Structural Abnormalities: The Journey to Acceptance," *Japan Journal of Nursing Science* 18, no. 2 (June 2020): 1-13, <https://doi.org/10.1111/jjns.12387>, 3.

⁹ Picci, Rocco Luigi, Francesco Oliva, Federica Trivelli, Claudio Carezana, Marco Zuffranieri, Luca Ostacoli, Pier Maria Furlan, and Roberto Lala. "Emotional Burden and Coping Strategies of Parents of Children with Rare Diseases." *Journal of Child and Family Studies* 24, no. 2 (2013): 514-22. <https://doi.org/10.1007/s10826-013-9864-5>.

improve overall maternal satisfaction. Ideally, such studies should include information on diagnostic processes, efficiency, practical support provision and individual costs associated with a particular disease¹⁰.

Another key factor to look at as an impact of a rare disease diagnosis, would be the isolation experienced by mothers. To respect equal medical and human rights, focus should be given to mothers to ensure that they have equitable access to medical attention and health coverage as the general population to ease the struggles they face. Scientific knowledge is thus critical to guiding effective policy implementation and understanding maternal struggles surrounding rare diseases diagnosis, early intervention and social care provision is critical in targeting parental needs.

In general, key concerns amongst mothers of children with Prader-Willi syndrome include:

- Provision of immediate medical support and early intervention,
- Insufficient access to knowledge of health professionals regarding their child's disease and with receiving familial support,
- Availability of support groups,
- Information on available social services,
- Special educational support for their children.

¹⁰ García-Pérez, Lidia, Renata Linertová, Cristina Valcárcel-Nazco, Manuel Posada, Inigo Gorostiza, and Pedro Serrano-Aguilar. "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>.

Section 3: Prader-Willi Syndrome, Family and Relationships

Another key element to look at would be the familial impact of Prader-Willi syndrome on a mother's experience and lifestyle. This part of the report gives an overview of this literature, looking at how a child's Prader-Willi syndrome diagnosis affects familial and social relationships. Researchers have conducted studies on such social relationships, looking into areas such as a child's Prader-Willi syndrome diagnosis on a mother's psychological wellbeing, her relationship with her family and the impact of societal stigma.

Exclusion and Maternal Psychological Well-Being

Social research has shown that mothers to children with rare diseases experience greater medical, psychological and familial dysfunction due to the additive nature of the child's rare disease and symptom manifestations, leading to a long-term commitment to caring responsibilities¹¹. With rare diseases like Prader-Willi syndrome, mothers typically provide most of the caregiving to their children, including managing the provision of care with physicians, community-based and social care and navigating interpersonal relationships, which contributes to an increase in stress and burnout¹². Social and Anthropological research also points towards the impact of cultural norms and standards of the good child and good mother¹³, with experiences of mothers to children with rare disorders disrupting the popular narrative of mothering. Often, their atypical experiences of motherhood are pushed to the side-lines, causing feelings of shame and marginalisation as a result of having different childbearing responsibilities and experiences¹⁴.

There are key points that need to be taken away on a mother's psychological well-being from a range of studies:

- In a Qualitative Semi-Structured set of interviews on the parenting experience of caring for children with rare diseases, mothers commonly divulged a sense of desperation of never being able to be off-duty when caring for their child as there was a sense of responsibility associated with making mistakes¹⁵. Mothers further relayed stories of having to push back against ineffective social care models and

¹¹ Miodrag, Nancy, and Robert M Hodapp. "Chronic Stress and Health among Parents of Children with Intellectual and Developmental Disabilities." *Current Opinion in Psychiatry* 23, no. 5 (2010): 407–11. <https://doi.org/10.1097/ycp.0b013e32833a8796>.

¹² Lafferty A, O'Sullivan D, O'Mahoney P, Taggart L, van Bavel B "Family carers' experiences of caring for a person with intellectual disability". (2016) Dublin: University College Dublin

¹³ Robertson, Rachel. "'Misfitting' mothers:." 7–19.

¹⁴ Carpenter, Lorelei, and Helena Austin. "Silenced, Silence, Silent." *Qualitative Inquiry* 13, no. 5 (2007): 660–74. <https://doi.org/10.1177/1077800407301179>.

¹⁵ Currie and Szabo. "It Would Be Much Easier If We Were Just Quiet and Disappeared" 1251–59

being made to become fighters for their children.

- In a study on social isolation and exclusion on the parenting experience of caring for children with rare diseases, one of the key takeaways was that parents were excluded in and in-between a liminal space where they could neither access normalcy or have their experiences of “abnormality” validated.¹⁶ Many mothers have expressed feeling uncomfortable in disclosing feelings of being overwhelmed or struggling to access care due to societal pressures of being a strong mother. This is an experience that is largely unaddressed in research into the maternal experience surrounding Prader-Willi syndrome - especially since exclusion is a large part of the experience.
- Mothers also struggled navigating within dominant hierarchical systems and power imbalances in regards to health-care and government disability support providers. In particular, mothers describe being excluded because of insufficient care modalities not addressing medical and behavioural manifestations associated with rare diseases.

Conclusions and Recommendations

- **Qualifying the socio-psychological impact of rare diseases on a mother** is essential through recording the unique experiences of such mothers.
- **Research on the impact of societal expectations of motherhood** can provide insight into the individual stresses and pressures experienced by mothers of children with rare diseases.
- A common issue of the **lack of available care and medical services** is present. There is thus a need to increase accessibility of resources available for social support to families to reduce feelings of exclusion for mothers.
- At a policymaking level, the **recognition of mothers as caregivers** is essential especially around the rare-disease network so that they can feel better supported and acknowledged.

Familial Dynamics and Social Relationships

Research into the impact of rare diseases on individual familial dynamics and social relationships is critical in understanding the social impacts of raising a child with Prader-Willi syndrome. In an Oceanic online study on the supportive care needs of parents caring for a child with a rare disease, the specific familial dynamics and social relationships were recorded to better understand the unmet needs of mothers¹⁷. An online survey was

¹⁶Currie, Genevieve, and Joanna Szabo. “Social Isolation and Exclusion: The Parents' Experience of Caring for Children with Rare Neurodevelopmental Disorders.” *International Journal of Qualitative Studies on Health and Well-being* 15, no. 1 (2020): 1725362. <https://doi.org/10.1080/17482631.2020.1725362>.

¹⁷ Pelentsov, Lemuel J., Andrea L. Fielder, Thomas A. Laws, and Adrian J. Esterman. “The Supportive Care Needs of Parents with a Child with a Rare Disease: Results of an Online Survey.” *BMC Family Practice* 17, no. 1 (2016). <https://doi.org/10.1186/s12875-016-0488-x>.

developed consisting of 45 questions which was answered by 230 parents, with questions on the satisfaction with care, experiences with daily care, the impacts of disease on relationships, the emotional and psychological burdens of disease, and parents' overall satisfaction with the support received. The last three domains were particularly pertinent, with the findings below:

- Majority of respondents said having a child with a rare disease has impacted the relationship with their partner, with open-ended comments from mothers suggesting both positive and negative experiences.
- Parents in the survey also reported having a child with a rare disease has impacted on their friendships, with 58% parents stating that their number of friends had reduced since the birth of their child with a rare disease.
- 75% of the parents expressed that they have not come into contact with other parents with a child with a similar condition to them - leading to increased feelings of isolation.
- A majority of the mothers expressed how their child's needs have impacted their employment status, either by reducing working hours or quitting entirely. Their partner's employment statuses were largely affected as well - either through reducing working hours to commit more to parenting responsibilities or by increasing working hours to cope financially.
- For parents with multiple children, 54% expressed that they were unable to provide enough time and attention for their other children due to the caregiving responsibilities they had to the child with a rare disease.

Conclusions and Recommendations

- Having peer support with other parents who share similar circumstances and receiving mutual support and encouragement is of central importance to mothers due to the provision of shared social identities and a sense of belonging.
- Mothers commonly find it difficult to maintain relationships and emotionally invest in new ones - becoming withdrawn or limiting their social interactions. Looking into providing psychological and emotional support to mothers to ensure the maintenance of healthy relationships is thus a useful area to explore.
- In regards to mothers with multiple children, looking into the impact of rare diseases on siblings is worth exploring as mothers often experience struggles balancing responsibilities amongst her children.

Societal Stigma

Stigma is a prevalent concern and struggle experienced by mothers with children with rare diseases. Qualitative research into stigma at both an internalised and external level shows how social taboos contribute to decreased psychological wellbeing within mothers.

- At an internalised level, cultural expectations surrounding genetic diseases and childrearing may cause mothers of rare diseases to withdraw and avoid interactions with their peers due to the guilt they experience¹⁸.
- At a cultural level, specific cultural stigmas and taboos may have an effect on a mother's caregiving experience to her child with a rare disease. For example, in Japan genetic diseases have been considered to "contaminate the blood"¹⁹ in the family, and many people still have a sense of "fear, pity, and aversion" against genetic diseases²⁰. Though governmental and policy efforts to eliminate discrimination based on disability does help, the lack of awareness surrounding rare diseases still has prevalent effects on parents with children with rare diseases due to the societal judgement they receive.
- At a communal level, mothers in surveys and studies often spoke of difference and lack of understanding within common social settings such as playgrounds, shopping centres, and schools when their children exhibited difference from other children²¹. Though not always malicious or ill-intentioned, mothers share feelings of being bothered or feeling unwelcome in public domains when their children exhibit certain behaviours such as outbursts.

Conclusions and Recommendations

- **Regional studies on Prader-Willi syndrome** are critical in understanding different societal and cultural expectations surrounding rare diseases and support as this drastically contributes to a mother's individual caregiving experience.
- **Offering psychological support for mothers with Prader-Willi syndrome** is essential to help them confront their own personal stigmas surrounding raising children with rare diseases.
- **Increasing awareness at a larger public level on Prader-Willi syndrome** may be helpful to increase empathy amongst the general public. This can be

¹⁸ Kutsunugi, Saeko, et.al. "The Journey to Acceptance" : 1–13..

¹⁹ Kato, Masae, and Margaret Sleeboom-Faulkner. "Cultures of Marriage, Reproduction and Genetic Testing in Japan." *BioSocieties* 4, no. 2-3 (2009): 115–27. <https://doi.org/10.1017/s174585520999010x>.

²⁰ Hasegawa, T., & Igarashi, T. Popular image of the term "genetic disease". *Journal of Tokyo Women's Medical University*, no.63 (1993), 94–101 (in Japanese).

²¹ Currie, Genevieve, and Joanna Szabo. "Social Isolation and Exclusion."

done through conducting talks and lectures at schools and local community centres, especially in neighbourhoods where mothers of children with **Prader-Willi syndrome** are present.

- **Policy has an impact on societal cultures**, in which countries with strong rare disease infrastructure report to do better culturally when it comes to reduced stigma and increased acceptance.

Section 4: Measuring Maternal Satisfaction - Literature Review

Researchers from different parts of the world have attempted to evaluate maternal satisfaction associated with rare diseases in general and Prader-Willi syndrome. This part of the report gives an overview of this literature. Researchers have measured the challenges within specific social care systems to reflect the struggle of caregiving on parents. However, there is not as much information available on the social and psychological impact of caregiving on parents of children with rare diseases, especially mothers. The few studies that measure maternal satisfaction give insight into the specific challenges and stress factors for mothers with children with Prader-Willi syndrome - emphasising the need for more unique individualised support services and policies catered to caregivers and their needs.

Maternal Stress

Three studies have been conducted on parenting stress in regards to Prader-Willi syndrome, with all studies finding that parents displayed high stress levels²². Additionally, Prader-Willi syndrome parenting stress seems to be related to the development of specific behavioural problems as opposed to other factors such as gender, age, IQ or obesity levels of the child²³.

Key investigative factors that more recent research has focused on include testing the hypothesis that Prader-Willi syndrome is correlated to high parenting stress and that certain child characteristics contribute to the increase in maternal stress. Such scientific studies are critical in expanding knowledge about child characteristics and their specific relevance to the maternal caregiving experience as it contributes to the development of more specific support services for families.

Case Study 1: Hodapp et.al Study - Families of Children with Prader-Willi Syndrome: Stress-Support and Relations to Child Characteristics

This study's objective was to look at the relationship of a child's associated maladaptive behaviours and familial stress levels²⁴. By analysing the nature of familial stressors for families raising children with Prader-Willi syndrome, this study attempted to provide an in-depth examination on offspring characteristics on familial stress and support - looking at

²² Hodapp, Robert M., Elisabeth M. Dykens, and Linda L. Masino. "Families of Children with Prader-Willi Syndrome: Stress-Support and Relations to Child Characteristics." *Journal of Autism and Developmental Disorders* 27, no. 1 (February 1997): 11–24. <https://doi.org/10.1023/a:1025865004299>.

²³ Ibid

²⁴ Ibid

factors such as obesity and maladaptive behaviours specific to the syndrome. The study was conducted on 42 children diagnosed with Prader-Willi syndrome who were recruited through parent groups such as the Prader-Willi Syndrome Association | USA, the Prader-Willi California Foundation, and the Prader-Willi Alliance (New York-New England). Familial Stress was measured through questionnaires which collected demographic information on familial structure. The questionnaire also adapted the “Questionnaire on Resources and Stress - Friedrich Edition”²⁵, the “Family Support Questionnaire” and the “Child Behaviour Checklist” to measure areas such as parental pessimism, efficacy of existing support and extent of their child’s behavioural conditions.

Summary of Results

The examination of QRS-F findings revealed that families of children with Prader-Willi syndrome showed greater stress and parental pessimism than others. Though factors such as age, gender and IQ had no significant relationship with familial stress, there was a correlation to the QRS-F overall and specific child characteristics. Higher rates of five out of nine child maladaptive behaviours were associated with increased levels of overall stress and familial issues.

In terms of support, lower IQ was associated with more familial support while increasingly obese children were associated with fewer numbers and lesser percentages of support from neighbours and surrounding social environments. Overall, professionals only played a small role in the support received by all families - adding up to 8% of all supports and with families not listing more than 3 professionals.

This study highlights that families of children with Prader-Willi syndrome experience specific types of stress and support and that these stress levels are correlated to the extent of maladaptive behaviours displayed by a child. Though there is sufficient research surrounding the effects of specific demographic considerations such as age, gender and IQ levels in a child - more attention needs to be paid to maladaptive behaviour due to it being an important association to higher stress levels in families of children with PWS.

Conclusions and Recommendations

- **Provision of specialised support:** IPWSO and its members could look into providing more specialised and individualised support services for mothers who

²⁵ Friedrich, William N., Mark T. Greenberg, and Keith Crnic. “Questionnaire on Resources and Stress--Friedrich.” *PsycTESTS Dataset*, 1983. <https://doi.org/10.1037/t10563-000>.

struggle more with specific maladaptive behaviours i.e. workshops and/or support groups with other mothers.

Case Study 2: Behavioural Phenotypes and Family Stress

As part of a research project at the "Kinderzentrum Minchen", this study attempted to utilise data on children with Prader-Willi syndrome to analyse behavioural characteristics more frequently associated with the syndrome. The study was conducted amongst parents of 35 people with PWS, using questionnaires such as the "Society for The Study of Behavioural Phenotypes Postal Questionnaire", the "Parenting Stress Index" and the "Family Functioning Style Scale". More specifically, the parents of the preschool age group completed the "Verhaltensfragebogen ftir Vorschulkinder" on the social competence and behavioural problems in preschool children, while the parents of the school age children completed the "Child Behaviour Checklist".

Summary of Results

Parenting stress in the child domain was extremely high - with mothers reporting a lower sense of parenting competence than mothers in a comparative sample²⁶. Results also confirmed that hyperphagia was a significant characteristic behaviour in PWS children that contributed to overall stress levels. Overall, the degree of parenting stress depends less on the type of disability; instead, individual coping resources and perceived social support seem to mediate interactional and emotional stress and determine parental satisfaction in families with children with intellectual disabilities.

Conclusions and Recommendations

- **Diversifying support materials on specific Prader-Willi syndrome symptoms** i.e. hyperphagia, mood outbursts.
- A mother's individual coping method contributes towards the correlation between specific behavioural characteristics and parenting stress as feelings of discouragement from struggling contributes to increased stress. **More psychological research into mothers and their welfare** could be useful to build nuance around such correlations.

²⁶ Sarimski, K. "Behavioural Phenotypes and Family Stress in Three Mental Retardation Syndromes." *European Child & Adolescent Psychiatry* 6, no. 1 (1997): 26–31. <https://doi.org/10.1007/bf00573637>.

Case Study 3: Dutch Prader-Willi Syndrome Parent Support Group Study on Maternal Stress

This study's objective was to measure the correlation between specific Prader-Willi syndrome characteristics in a child and maternal stress levels. This research was conducted by the Department of Clinical Child and Adolescent Studies at Leiden University in the Netherlands²⁷. The study was conducted amongst members of the Dutch Prader-Willi syndrome parent support group, with a total of 23 children with a definite Prader-Willi syndrome diagnosis and their mothers participating. Maternal stress was measured through questionnaires - using the "Parenting Stress Index" by Abidin and the "Developmental Behaviour Checklist - Primary Carer" to measure parenting stress in families with children between 2 to 13 years and the emotional and behavioural problems exhibited in the recent 6 months respectively. A combination of both these questionnaires were used alongside the "Vineland Screener" to measure adaptive functioning of children up to age 12 or older persons with comparable levels of functioning.

Summary of Results

Common to the trends present in other research on families with children with intellectual disabilities, higher parenting stress is present when more behavioural problems are expressed. Maternal parenting stress was not related to a Prader-Willi syndrome child's gender or age. This conclusion is in line with Hodapp's 1997 research on Prader-Willi syndrome. Likewise, the level of intellectual disability, based on adaptive functioning, was not related to maternal parenting stress in Prader-Willi syndrome. This once again asserts the conclusion of Hodapp et.al's study, who found no relationship between parenting stress and IQ.

However, the study concluded that there were no significant correlations between maladaptive behavioural problems and maternal stress, which is at odds with takeaways from past research. There are several possible explanations for this anomaly, such as the study using an instrument specifically developed for children with intellectual disability. Since a proportion of the participants functioned in the borderline range to normal functioning, it is possible that many characteristic behavioural problems were not measured by this particular questionnaire as opposed to participants that did display intellectual disabilities. Moreover, this particular study was limited to an age cohort of 2-13, which could limit the set of behaviours expressed as many maladaptive behaviours

²⁷ Wulffaert, Josette, Evert M. Scholte, and Ina A. Van Berckelaer-Onnes. "Maternal Parenting Stress in Families with a Child with Angelman Syndrome or Prader-Willi Syndrome." *Journal of Intellectual & Developmental Disability* 35, no. 3 (2010): 165-74. <https://doi.org/10.3109/13668250.2010.499101>.

tend to develop amongst children with PWS in their teen years. Thus, more prominent behavioural problems in adolescents could give rise to the different results. However, further studies with different age cohorts are needed to confirm this hypothesis.

Conclusions and Recommendations

- **Increased age-based studies on Prader-Willi syndrome maladaptive behaviours:** To understand the trend in the emergence of specific characteristics with age and to capture a broader range of age cohorts.

Life Satisfaction

Life Satisfaction is one of the key factors that make up the subjective facet of one's quality of life and the cognitive aspect of well-being. It is an important construct to examine amongst mothers of children with rare diseases, as it offers *personalised* insight into how mothers feel about their own personal life situations as opposed to factors others may deem as important. Shin and Johnson's model (1978) states that the factors that determine an individual's judgement of their quality of life may be unique to each individual, with some placing more emphasis on certain factors compared to others²⁸. The flexibility of what constitutes life satisfaction allows mothers to determine what is important to them individually rather than looking into predetermined and quantifiable measures such as income.

Comparative research on life satisfaction in mothers of children with or without rare diseases, yet alone Prader-Willi syndrome, is largely limited, with results being largely mixed. Such mixed findings highlight the complexity of factors that contribute to life satisfaction among mothers of children with rare diseases, which may include characteristics of the child (e.g., behaviour problems, level or type of disability), as well as characteristics of the mother (e.g., age, coping style).

The extent of existing Prader-Willi syndrome research on mothers has focused on negative psychological outcomes, with studies showing that mothers report higher levels of stress and an increased set of negative emotional outcomes than mothers with other intellectual developmental disabilities²⁹. Studies on life satisfaction as a measure of a

²⁸ Shin, D. C., and D. M. Johnson. "Avowed Happiness as an Overall Assessment of the Quality of Life." *Social Indicators Research* 5, no. 1-4 (1978): 475–92. <https://doi.org/10.1007/bf00352944>.

²⁹ Mazaheri, M. M., R. D. Rae-Seebach, H. E. Preston, M. Schmidt, S. Kountz-Edwards, N. Field, S. Cassidy, and W. Packman. "The Impact of Prader-Willi Syndrome on the Family's Quality of Life and Caregiving, and the Unaffected Siblings' Psychosocial Adjustment." *Journal of Intellectual Disability Research* 57, no. 9 (2012): 861–73. <https://doi.org/10.1111/j.1365-2788.2012.01634.x>.

maternal well-being may thus be a useful research mechanism to tap more into to better understand:

- What Prader-Willi syndrome factors present in a child are associated with life satisfaction of a mother i.e. genetic subtype, BMI, hyperphagia severity, maladaptive behaviours etc.
- How do maternal factors relate to life satisfaction of a mother i.e. marital status, coping mechanism, psychological well-being etc.
- Does maternal satisfaction change over time and what factors contribute to such changes?

Case Study 4: Vanderbilt University Study on Life Satisfaction Among Mothers of Individuals with Prader-Willi Syndrome

The study was conducted with 196 participants enrolled in a longitudinal study of individuals with Prader-Willi syndrome and their families at Vanderbilt University, where recurring visits for each family were scheduled every 2 years and data was collected from both mother and child³⁰. Maternal Life Satisfaction was measured by the satisfaction with life scale³¹, which used a Likert-type scale to assess certain statements on quality of life - creating a total life satisfaction score from 5 to 35, with higher scores indicating greater satisfaction with life. Maternal Stress and Coping Styles was also measured using a short form of the "Parenting Scale Index" and the "COPE questionnaire"³². Demographic Prader-Willi syndrome factors were also measured such as age, weight, IQ, adaptive behavioural skills using the Vineland behavioural scale and hyperphagia severity using Dyken's "Hyperphagia Questionnaire"³³.

Summary of Results

In terms of significant correlations, the study concluded that maternal stress was negatively related to life satisfaction. Similarly, maternal coping style was related to life satisfaction, with mothers with more avoidant coping styles displaying lower levels of initial life satisfaction. The study showed there were no statistically significant relationships between maternal life satisfaction and maternal age, marital status,

³⁰ Shivers, Carolyn M., Caroline L. Leonczyk, and Elisabeth M. Dykens. "Life Satisfaction among Mothers of Individuals with Prader-Willi Syndrome." *Journal of Autism and Developmental Disorders* 46, no. 6 (2016): 2126–37. <https://doi.org/10.1007/s10803-016-2741-5>.

³¹ Diener, Edward. "Satisfaction with Life Scale." *PsycTESTS Dataset*, 1985. <https://doi.org/10.1037/t01069-000>.

³² Carver, Charles S., Michael F. Scheier, and Jagdish K. Weintraub. "Assessing Coping Strategies: A Theoretically Based Approach." *Journal of Personality and Social Psychology* 56, no. 2 (1989): 267–83. <https://doi.org/10.1037/0022-3514.56.2.267>.

³³ Dykens, Elisabeth M., Melissa A. Maxwell, Elizabeth Pantino, Rebecca Kossler, and Elizabeth Roof. "Prader-Willi Syndrome Hyperphagia Questionnaire." *PsycTESTS Dataset*, 2007. <https://doi.org/10.1037/t64391-000>.

household income. In regards to maternal satisfaction over time, time itself was not a significant factor but a significant covariance was present between satisfaction and the extent of maternal stress - suggesting that mothers with greater initial reports of life satisfaction show greater change in satisfaction related to stress. The results also show differences surrounding Prader-Willi syndrome characteristics, with hyperphagia having a significant correlation with maternal life satisfaction as opposed to BMI.

Conclusions and Recommendations

- **Measuring Change over Time:** To better measure change over time, a more precise consideration of life events (e.g., Prader-Willi syndrome diagnosis, onset of hyperphagia, transition to adulthood) may reflect more pronounced changes in maternal life satisfaction.
- **Family Factors:** Further research on how family factors such as family coping styles, sibling factors, marital satisfaction and sources of support influence parents' well-being is needed to build a stronger comparison of experiences of mothers to children with Prader-Willi syndrome, compared to other families of children with rare diseases or intellectual development disabilities
- More Prader-Willi syndrome-specific support services for mothers surrounding **coping styles.**
- **Extending Prader-Willi syndrome support services to families** i.e. supporting the roles of siblings and grandparents can help alleviate maternal stress and improve maternal life satisfaction.

Section 5: The Maternal Experience Around the World - Interviews with Mothers

Introduction

Interviews with mothers of children with Prader-Willi syndrome across different countries have highlighted common shared experiences and struggles surrounding caregiving and parenting. Mothers from **Germany, Norway, Ireland, the United States, Chile, Singapore and India** responded to several questions surrounding their experiences as parents and the challenges they experienced. Our questions focused on:

- Challenges and changes in family dynamic over time to raise a child with PWS.
- Home adaptation changes to meet the needs of the child / adult with PWS.
- Use of social services, support networks and resources when parenting a child with PWS.
- Experience of a mother of a child of PWS: roles, struggles, sacrifices and strategies.
- Influence of social culture and stigmas in raising a child with PWS.

The overall goal of the interviews was to really capture the individualised experience of a mother of a child with Prader-Willi syndrome. Depending on the nature of a country's healthcare system and the age of the child, the answers were unique and personal to each mother's experience. Nevertheless, there were common themes that emerged throughout the interviews - underlining a shared struggle experienced by most mothers to children with PWS.

In most cases, mothers outlined challenges in areas such as:

- **Therapies and early intervention programs for children with Prader-Willi syndrome:** Many mothers highlighted feeling overwhelmed upon the initial Prader-Willi syndrome diagnosis, having been provided with limited informational support by medical practitioners - leading mothers having to seek out early support on their own.
- **Social services:** The interviews raised a concern about the lack of social services available, especially as the children grew up. Many mothers expressed concern surrounding the lack of specialised homes or centres for their child.
- **Isolation:** Many mothers expressed feelings of isolation when describing their parenting experiences - alluding to feelings of uncertainty surrounding the future.

- **Siblings:** For mothers with more than one child, many highlighted challenges surrounding distributing equal attention to their children alongside struggles between siblings, such as exclusion, due to Prader-Willi syndrome.
- **Family:** Though subjective to each mother's experiences, a few mothers shared struggles surrounding familial support i.e. their partners, their in-laws, extended family etc. when it came to accepting their child with Prader-Willi syndrome. The relationship mothers had with their family seemed to contribute to the extent of support and the sharing of responsibilities within a household.

Norway

The situation in Norway was quite unique compared to other mothers. The mother of a daughter with Prader-Willi syndrome was one of the few mothers interviewed who lived in a country in which Prader-Willi syndrome social care treatment costs were completely subsidised by the state.

Social Services

Overall, the mother had extremely positive experiences with social services, with the state having comprehensive and established health coverage measures to ensure that an individual with a disability was supported adequately. She even shared how *Frambu*, the national resource centre for people with rare disorders, came for a visit in her area to better understand Prader-Willi syndrome and expand on their expertise, knowledge and services surrounding the specific disorder. She also highlighted the impact of Norway's local Prader-Willi syndrome association in providing her a sense of community and relief in being able to talk to other parents with shared experiences. This points towards how the availability of resources within a state helps alleviate the stress experienced by a mother raising a child with Prader-Willi syndrome

Responsibilities as a Mother

The mother highlighted how upon finding out about her daughter's Prader-Willi syndrome diagnosis, there was an immediate understanding that things needed to be done differently. The major adjustment she made to her working life was sending her daughter with Prader-Willi syndrome and her older daughter to Kindergarten. Before the birth of her daughter with Prader-Willi syndrome, she had intended to transition from working night shifts as a nurse to being a full-time stay-at-home mother. However, she needed to take up more shifts as her children progressed through school to be able to support her daughters - contributing to increased stress as she had to work flexibly around her daughter's schedules. She shares how she attempted to work part-time but

still felt an overwhelming sense of exhaustion from juggling her career and caregiving relationships - often feeling stuck in a limbo. "Sometimes I got tired of everything and thought it was easier to be at home and stick to my ordinary routine", she shares - highlighting the impact of drastic adjustments on herself.

PWS, Family and Relationships

- **Home Lifestyle Modification:** Children with Prader-Willi syndrome have hyperphagia - an abnormally increased appetite for food. To manage her daughter's access to food, the mother had to establish strict rules within the household surrounding portion control and the prohibition of food-sharing. She highlighted having to have separate meal-times to prevent sharing of food.
- **Relationship between Siblings:** She shared how her oldest daughter experienced exclusion by her relatives and friends compared to her younger PWS daughter. While the younger daughter received tremendous support and encouragement from extended family, the older daughter often was unrecognised. The oldest daughter also had to make sacrifices growing up, such as having to give up on Saturday night desserts with her cousins so as to not entice her younger sister with sugar. Though these changes may seem small, the mother shares how they had subtle impacts on her oldest daughter as the differences between both children were clear.
- **Social Circles:** The mother shares how she had to educate her friends on Prader-Willi syndrome and household rules surrounding food. By establishing such rules and sticking to regiments, she was able to have successful social experiences with friends and families such as holidays and picnics.

Personal Identity and Stigmas

"Having a life with a lot of challenges does not take away from the happiness", she shares - opening up about how having a daughter with Prader-Willi syndrome has allowed her to grow as a person and expand her perspectives as a mother. She highlighted how the stigma she experienced was more internal rather than external, specifically surrounding mothers with disabled children. Initially, her assessment of mothers with disabled children was that they would "look unhealthy, pale, tired with greying hair, not focusing on themselves". However, as she raised her daughter with Prader-Willi syndrome through adolescence to adulthood, she worked on reversing that internalised stigma by finding

happiness within her personal experience of motherhood. She shares how her daughter helped her emotionally with how to deal with life and seeing growth in the little things. She does point towards the exhaustion as a challenge she experiences, with many of her peers not knowing the extent of her tiredness due to her ability to camouflage her struggles. Nevertheless, she highlights how having a child with Prader-Willi syndrome has shaped her identity and perspectives towards motherhood.

Recommendations

- **Psychological Support:** IPWSO and its members could look into providing specific support to equip mothers with social and psychological skills and strategies to handle exhaustion.
- **Raising awareness on the prioritization of individual needs:** especially for mothers who spend a lot of their time and energy caring for children with PWS.

Germany

Two interviews were conducted with German mothers, one with a 25-year-old son with PWS, and one with a 7-year-old daughter with PWS. Interviewing both parents gave deep insight into the different experiences and challenges experienced by mothers while raising children across different ages. It also gave insight into the development of resources available within Germany and how support has evolved over time.

Early Intervention Support

The mother of the 25-year-old received her child's diagnosis when he was 2-months-old. Immediately upon the diagnosis, she received 2 pamphlets in English from a medical book and was left to find resources by herself. "I never thought of such a possibility" she shared, highlighting feeling stunned and devastated upon finding out about the initial diagnosis. It was only after a physiotherapist started coming to her home twice a week that she was able to get specific advice and the right contacts to the German Prader-Willi syndrome association. Being able to talk to professionals with specialised experience was a lot more helpful compared to the immediate advice at the hospital as the mother was able to feel a lot more hope compared to being overwhelmed with technical information. Connecting with her local Prader-Willi syndrome association gave her hope as a mother, both in regards to emotional support and technical support in regards to accessing disability cards and insurance services.

The mother of the 7-year-old received her child's diagnosis immediately upon birth. Upon the diagnosis, she shared experiencing feelings of devastation due to the awareness of the sacrifices she would have to make as a mother surrounding her social life and travel. She did receive psychological support at the hospital but did not find it the most helpful

since she was still processing new information. Instead, she and her partner arranged psychological help after being discharged. She also shared how reaching out to the German PWS association and IPWSO acted as a helpful source of advice to “look out into the world and seek help”. In general, Prader-Willi syndrome associations and support networks seem to help relieve fears experienced from mothers, especially after being bombarded with new medical information by medical professionals that lack specialised understanding of PWS.

PWS, Family and Relationships

- **Home lifestyle changes:**
 - **Mother to 25-year-old:** She highlighted seeking external therapies for her son, such as finding physiotherapists and speech therapists.
 - **Mother to 7-year old:** She shared how she built a feeding regimen every 3 hours while receiving nutritional support surrounding building healthy diets.
- **Family Dynamics:**
 - **Mother to 25-year-old:** Only some of her extended family was able to truly understand the challenges her and her son experienced with PWS. Many family members were avoidant with addressing the challenges faced by both her and her child, with very few actively engaging with them. As a single mother, she shared how her son was her priority. “I needed every bit of energy for my son”. “He was the most important man in my life”.
 - **Mother to 7-year-old:** As a working mother who is passionate about her career, she shared equal responsibility with her partner in caregiving. Her parents are also involved in providing help with caregiving, specifically helping with transport to her child’s therapies so she can work longer. Having these shared responsibilities allows her to have more flexibility in her professional life.
- **Social Circles:**
 - **Mother to 25-year-old:** Upon her child’s PWS diagnosis, her social circle changed as she lost friends and gained new ones. She shares how the people she met through Prader-Willi syndrome associations were highly impactful additions to her social circle that provided her with a deep sense of empowerment. “I met so many people from Prader-Willi syndrome associations, national and international”. “They are such great people with such strengths fighting for their children and I feel so much respect for these people”.
 - **Mother to 7-year-old:** Most of her social circle was supportive to her child, with friends being accommodating to her at parties by letting her bring

different cakes etc. She shared how it was endearing for people to enquire about her child and her wellbeing and being empathetic to her experiences. PWS associations were also valuable additions to her social circles, and such networks helped her feel less alone.

Personal Identity and Challenges

For the mother of the 25-year-old, her child shaped a primary part of her personal identity. "I had to be flexible and creative", she says - pointing out how she and her son shared many fond moments together, both bitter and sweet. There were challenges she experienced regarding feelings of isolation and being overwhelmed compared to other parents. Nevertheless, every sign of progress was so immense and great for her to experience as a mother. "I have a special child so I have a special life".

For the mother of the 7-year-old, she shares how Prader-Willi syndrome has infiltrated into other aspects of her life including her career. As an architect of hospitals, having an intimate understanding of raising a child with a rare disease has provided her with a positive additional input in her design work - showing how PWS becomes an integral part of many mothers' identities over time. In terms of challenges, she points towards the uncertainty towards the future of her child. As a mother to a daughter still in her adolescence, there is a large amount of uncertainty regarding the future and the development of specific Prader-Willi syndrome behaviours. Additionally, she shares how there are struggles with the bulk of responsibility applying for disabled cards and accessing therapy as her child grows older. "As a mother you always have to fight for getting your therapies and your right to support your child".

Recommendations

- **Parental support and psychological help for parents:** In Germany, *DIOKOVERE* acts as a non-profit company in the health, social and educational sectors based in Hanover which provides specific psychological support for parents. However, this service is limited to parents of younger children. IPWSO and its members could look into providing or connecting mothers with online psychological services in order to allow parents to express their struggles in a safe space.
- **Legal Support:** Knowing the struggles mothers have with accessing insurance and disability services, researching more into the provision of pro-bono services for parents may be helpful to help mother navigate around different systems

Ireland

The mother in Ireland has four children, her youngest son was diagnosed with PWS.

Diagnosis and Early Intervention

After multiple hospital visits and overnight stays, the mother received her child's diagnosis when her son was 12-weeks-old. She shares how she received little support from her doctors due to the informational scarcity surrounding Prader-Willi syndrome but was given a couple of pages in medical textbooks to read.

Eventually, she began to seek support surrounding disabilities, such as seeking out a long-term illness card to help cover the costs of oxygen support and technical machines needed by her son. As a stay-at-home mother whose husband was the main breadwinner at the time, she shares how it was quite a challenge to be able to balance financial costs alongside going through the lengthy process of finding a medical social worker. This points towards how social costs and loss of income is a key factor in maternal stress.

PWS, Family and Relationships

- **Home Lifestyle Modifications:** The mother shares how rules around food were established very early on amongst the household and siblings. Before the age came where locks were necessary to hide food, the mother shared how rules around eating were strictly ingrained into each family members' heads. She shares an anecdote of overhearing her then five-year-old third child telling his friend about his Prader-Willi syndrome sister's disorder - "That's my baby sister. You are not to feed her anything. Not even a crumb". She expresses how hearing her young child be able to effectively communicate such ideas to his friends shows how inculcated such routines and regimes were within her home when it came to Prader-Willi syndrome adaptations around food.
- **Relationships with Siblings:** The mother shares how there were feelings of struggle and trauma amongst her older siblings having been exposed to their youngest sister's Prader-Willi syndrome experiences from a young age. She highlights how her third son specifically "carries the burden of being around (his sister) the most", as he had to tend to his sister during developing episodes of self-harm as she grew older. This was particularly a challenge as his two older siblings had grown up and moved out by then, leaving him to have to care for his sister with PWS the most as he was in his teens. The mother shares how as much as the siblings have love for their little sister with Prader-Willi syndrome, they do harbour

stress from memories of growing up, particularly due to the intensity of specific experiences.

Identity

Like many mothers of children with Prader-Willi syndrome, the mother shared how she transferred her experiences of caregiving into her career. As a teacher with a Classics background, she developed an interest in special education - which helped to build a second income within her family as her daughter with Prader-Willi syndrome grew older. She shares how her experience as a mother to a child with Prader-Willi syndrome was integral and largely transferred into her job as a special needs teacher as she had an integrated understanding of the challenges experienced as a parent and the empathy needed to interact with her students.

Recommendations

- **Psychological Support for Siblings:** IPWSO and its members can look into developing psychological support networks for siblings of individuals with PWS to provide them with a safe space to share their experiences.

India

The mother to an 18-year-old son in India had a very unique experience with learning about her child's Prader-Willi syndrome diagnosis. After six months of difficulty and consistent tests in India, she sought medical support in the United States, through which she was able to get her child's Prader-Willi syndrome diagnosis. This interview was very useful to understand the specific struggles in accessing resources and medical expertise in the Global South as compared to the West - a key point for IPWSO to note to build more catered support services for Parents in Asia.

Social Services

After receiving her child's PWS diagnosis in the United States, the mother returned to India to share the results with the doctors at her local hospital. The doctor that provided her the greatest amount of support through her child's initial treatments then arranged a continuing medical education session to raise awareness on Prader-Willi syndrome, using her child as a case study. As Prader-Willi syndrome was quite unknown in India, the majority of the mother's social support came from the American Prader-Willi syndrome association. "The idea of meeting people on the same boat as me was a godsend", she shares - highlighting the sense of relief she felt to be able to talk to other parents who had similar caregiving experiences with Prader-Willi syndrome. As one of the pioneers of the

Indian Prader-Willi Syndrome Association, she emphasises the need to build sustainable and self-sufficient resources as a local organisation as there is stark difference between resources available in the Western world as opposed to the Global South.

PWS, Family and Relationships

- **Familial Dynamics:** Having initially lived in a joint-family household, the mother shared how there were key difficulties in the early years of making her son's grandparents understand the harms of excessive food. The Indian socio-cultural expectation of associating chubby babies with comfortable and well-to-do families³⁴ contributed to the grandparents' push in sneaking in food for her son. This was particularly a challenge in his earlier years when he had a low body weight and as he began to develop symptoms of hyperphagia. The mother highlights how reversing her in-laws' cultural expectations is still a challenge and shares how moving out of a joint-family household and into a separate flat in the same apartment complex has provided her with a lot more freedom in managing her son's diet.
- **Relationships with Sibling:** The mother shared challenges she experienced in raising a younger child alongside her son with Prader-Willi syndrome - alluding to feelings of jealousy surrounding dividing attention between both children. With COVID-19 and the pandemic, there was increased difficulty in ensuring that both her children received undivided attention as she had a responsibility to care for her son with PWS and her baby.
- **Social Circles and Stigma:** Stigma seems to have a larger prevalence in India, with many lacking openness to talk about special needs and rare diseases. Many children also lack empathy when interacting with kids with rare diseases, with the mother sharing how her son was bullied by his neighbours during cultural festivals. Stigma amongst children in the same age-group of an individual with Prader-Willi syndrome is something to be researched into more as education plays a key role in reversing negative perspectives.

³⁴ "This World, India's Supersize Kids - India's Supersize Kids." BBC Two. BBC. Accessed August 18, 2022. <https://www.bbc.co.uk/programmes/articles/2BcMW01VVrf3TZTnT7Jhl2b/indias-supersize-kids#:~:text=Chubby%20kids%20in%20India%20are,feed%20him%20on%20the%20sly>.

Recommendations

- **Increasing Medical Awareness:** Continuing medical education is essential amongst medical practitioners in order to expand their vocabulary surrounding Rare Diseases and PWS. The building of globally accessible medical programmes and courses for doctors is thus a possible online tool to develop.
- **Developing Globally Available Therapies:** to bridge the East vs West Gap.
- **Policy Research:** In the future, IPWSO could engage in more regional investigations to understand the impact of policy (or the lack thereof) in the availability of resources.

Singapore

In this interview, the mother in Singapore shared a lot about the lack of awareness and the cultural stigma surrounding her experience in raising her 11-year-old son with Prader-Willi syndrome.

Diagnosis and Early Intervention Support

The mother received her son's diagnosis when he was 9-months-old. Upon the diagnosis, she was given a long list of medical information on Prader-Willi syndrome to read up on and was sent home right after without any immediate support services. This resulted in immense feelings of being overwhelmed as she had a lot of information to process within a very limited time.

After conducting her own research, she eventually found resources and other useful articles through IPWSO. She was also able to join a support group at the KK Women's and Children's Hospital.

PWS, Family and Relationships

- **Home Lifestyle Modifications:** As her son's hyperphagia began developing between 2 to 3-years-old, the mother began devising meal plans and eating schedules. She shared how inculcating this from a young age proved to be helpful as her son grew older as they were used to following routines and regimes surrounding food.
- **Family Dynamics:** While she describes herself as the main caregiver to her son, she also shares the impact of her mother's role in raising her child. She shares how her mother has provided huge emotional and practical support, highlighting how it has become a routine for her son to spend time over the weekends with his grandmother. She expresses still having tension from her in-laws surrounding her

son's eating habits - alluding to the Asian norms and struggles to adapt around the cultural importance of food. Understanding the cultural norms surrounding food in different countries is perhaps something to research more into in regards to the relationship and impact it has on individuals with Prader-Willi syndrome.

- **Relationships with Siblings:** The mother shared how her younger children have been impacted by having an older sibling with Prader-Willi syndrome. As much as their relationship is loving, she expresses how her younger 2.5-year-old daughter and 8-year-old son are impacted by the meltdowns experienced by her eldest son with Prader-Willi syndrome, which is a common behavioural symptom of Prader-Willi syndrome that can emerge with age. She shares how being exposed to anger and aggression during such meltdowns pushed away the younger siblings out of fear - which is a challenge that she is currently working on overcoming as a mother.

Stigmas

When talking about stigmas she has experienced as a mother to a child with Prader-Willi syndrome, she shares how it was overwhelming at first to notice reactions, stares and glares in public while with her son. She also shares how there is still a large societal stigma in Asian societies surrounding special needs, with both her and members of her families losing close friendships after her son's diagnosis. These stigmas and feelings of denial sometimes manifest amongst mothers of children with Prader-Willi syndrome themselves even in support groups, with some mothers expressing disdain and judgement towards special education schools. This is an interesting point to note in future studies in understanding how societal influence can impact, and perhaps, hinder the identities and beliefs of mothers of children with rare diseases.

Culturally, she expresses how she experiences challenges during reunion and festival seasons with her Chinese family members. In Chinese culture, dishes are placed in the middle of the table for people to share and members have to wait to eat until the whole family is seated³⁵. However, this is not an ideal situation for children with Prader-Willi syndrome to be in, especially when hyperphagia is present. The mother thus expresses how the lack of supervision at such reunion dinners can sometimes result in her son overeating, which is a temporary challenge that they still experience.

³⁵Ma, Guansheng. "Food, Eating Behavior, and Culture in Chinese Society." *Journal of Ethnic Foods* 2, no. 4 (2015): 195–99. <https://doi.org/10.1016/j.jef.2015.11.004>.

Identity and Challenges

When talking about the changes she made to her social life and career, she says “Would I call it a sacrifice? No, because I love my children”. She highlights how her son with Prader-Willi syndrome has allowed her to become a better person, giving her the opportunity to develop new perspectives and learn valuable lessons on patience and maintaining calm.

Nevertheless, there are still challenges along the way that contribute to maternal stress she experiences. The mother shares how she has fallen into bouts of depression in periods when her son’s behaviours and emotions were changing. She also highlights uncertainty surrounding the future such as the lack of adequate care homes for individuals with Prader-Willi syndrome. Though her son is currently enrolled in a special education school, she also expresses how the vocational skills taught are prevalent in the Food and Beverage industry - which is not an ideal setting for individuals with Prader-Willi syndrome.

Recommendations

- **Vocational Skills:** IPWSO and its members could build skill-based programmes that provide workshops for older individuals with Prader-Willi syndrome who hope to work.

Chile

The Chilean mother has three daughters, out of which her second child was diagnosed with Prader-Willi syndrome.

Diagnosis and Early Intervention

The mother’s daughter was diagnosed at 2-years-old after Chilean doctors returned from London after completing their Continuing Medical Education. After understanding the symptoms better, such as the presence of Hypotonia, the doctors were able to run sufficient tests and accurately provide the Prader-Willi syndrome diagnosis for the mother’s child. Similar to other mothers interviewed, the importance of doctors having global exposure to rare disease information and infrastructure proves to be critical in improving care surrounding Prader-Willi syndrome.

In regards to early intervention, the mother shared how her daughter’s nursery had to make adaptation towards her child’s Prader-Willi syndrome behaviours, such as by building special cabinets to ensure that snacks were not easily accessible. As her daughter

grew older, she was able to access special education and work with a speech therapist to help her read and write.

PWS, Family and Relationships

- **Home Lifestyle Modifications:** Like many families, the mother shared about needing to place locks in kitchens to reduce risk of excessive food consumption.
- **Familial Support:** The mother shared how her father was a big support financially when it came to out-of-pocket special treatments for her daughter with Prader-Willi syndrome.
- **Relationships with Siblings:** Growing up, the mother shared how there were quarrels amongst her oldest and youngest daughters surrounding the sharing of desserts and the need to lock food, as her children grew up struggling to understand why they had to follow such rules and regimens. She also highlights how her daughter with Prader-Willi syndrome experienced feelings of envy towards her younger sister being taken off as a toddler since attention needed to be divided between both children.

Challenges and Identity

As a mother to an adult with Prader-Willi syndrome, the mother shares how the scarcity in medical awareness was a significant challenge in raising her child. “42 years ago nobody really knew about Prader-Willi syndrome” she shares, expressing how many mothers feel a sense of isolation upon knowing about their child’s diagnosis

However, “We made a positive experience out of an (initially) sad experience” she shares - alluding to how her daughter’s Prader-Willi syndrome shaped her identity over the years. Like many mothers, Prader-Willi syndrome became a large part of her life, with the mother engaging deeply with the Chilean and Latin American communities. Alongside developing a vested interest in special education, she also became one of the Founders of Corporación Señales, a non-profit organisation to provide schooling, training and labour opportunities for people with special needs.

Recommendations

- **Age-Based Resources:** Based on the Chilean mother’s suggestion, dividing parenting resources by the Prader-Willi syndrome individual’s age is a useful tool to have as the responsibilities of parenting and caregiving evolve over time as one transitions from adolescence to adulthood.

United States

The American mother interviewed has 5 children, of which the youngest has Prader-Willi syndrome.

Diagnosis and Early-Intervention Support

The mother received her son's Prader-Willi syndrome diagnosis at 2-days-old from geneticists and was immediately given pamphlets to read on the syndrome. However, she expressed initially not wanting to read the pamphlets due to the immediate sense of "grief" she experienced towards the loss of a "normal child". These overwhelming feelings of isolation and grief are a common experience amongst mothers of children with rare diseases, especially at the initial stages of diagnosis.

In terms of early-intervention support, the mother shared how the Birth-To-Three programme allowed for her to have in-home services with nurses, physical, speech and occupational therapists for her son till he turned 3 - highlighting how pivotal such services were for her at the early stages.

As her son grew older, her local Prader-Willi syndrome association chapters were helpful in offering her solace - especially through seeing older children with Prader-Willi syndrome and being able to envision what a potential future could look like for her child. Like many mothers, finding a community within Prader-Willi syndrome associations helps provide a sense of comfort and hope surrounding the future.

PWS, Family and Relationships

- **Home Lifestyle Modifications:** The biggest change in her at-home life was surrounding food. Structured mealtimes were implemented and locks and alarms were installed around the house to keep the family aware of when food was being accessed. The mother also shares how the older siblings had to have secret stashes of food locked in their rooms that had to be hidden from the son with Prader-Willi syndrome.
- **Relationships with Siblings:** As a single mother for most of her youngest son's life, the mother shares how her and her 4 older children functioned as an efficient family unit to support her Prader-Willi syndrome child. The older siblings grew up intrinsically aware of their responsibilities towards their youngest brother and were prepared with specific plans when he exhibited tantrums and outbursts. Though the relationship was loving, the mother highlights how her older children had to

parent themselves to some extent and grow up much faster because of their youngest sibling with Prader-Willi syndrome. She acknowledges how this had an impact on their growing up - showing how Prader-Willi syndrome affects familial dynamics.

Identity and Challenges

“Prader-Willi syndrome becomes your life - it is 24 hours a day, 7 days a week”. The mother shares how Prader-Willi syndrome drastically changed the trajectory of her life, both as an individual and as a parent. As an individual, she had to make changes in her career from working 12-hour-shifts to working from home. As a parent, she was aware of the need to switch focus due to her responsibility of caring for a child with a rare disease. Nevertheless, she shares how she regards her responsibilities as a mother to be the same - owing to how mothers will give anything up to ensure the best for their children

Recommendations

- **Improving access to information online:** Building stronger presence online presence and to allow mothers to find more resources that lead to more in-person support quickly and more efficiently.

Conclusions

Overall, the experiences of mothers around the world were quite varied, nuanced and personal. Multiple factors played into each individual mother's experience such as their country's available rare disease support, financial status, family societal culture and more. Nevertheless, conducting interviews with mothers across the world point towards how there is a shared narrative amongst mothers of children with Prader-Willi syndrome. The presence of repeated themes, concerns and feelings show how there are specific experiences unique to the Prader-Willi syndrome parenting experience.

Interviewees highlighted the importance and the necessity of psychological support for parents of children with Prader-Willi syndrome.

Almost all mothers emphasised how the time dedicated to managing Prader-Willi syndrome is substantial, especially when caregiving for a child. Though each individual instance of Prader-Willi syndrome is different, with some children needing more monitoring and intervention than others, mothers share how raising a child with Prader-Willi syndrome becomes a huge part of their lives and identities. This does take an emotional and psychological toll on a mother, with many sharing feelings of being overwhelmed, tired and depressed when experiencing caregiving challenges.

The mothers also shared how raising a child with PWS impacted the dynamic familial relationships, especially with siblings.

Though subjective, with mothers expressing a range of positive and negative experiences, there still exists a common theme surrounding familial dynamics - particularly amongst and between siblings. Mothers with more than one child share how siblings were affected growing up. Many siblings grew up having a role in caring for their brothers or sisters with Prader-Willi syndrome and were exposed to major home adjustments surrounding food and behavioural outbursts from a young age. Though mothers share that the siblings have love for their brothers or sisters with Prader-Willi syndrome, they also share that there is a psychological impact of having to grow up faster. This is a noteworthy area to research more into - with the possibility of psychological support being extended to siblings and younger family members.

Societal Culture also has an impact on a mother's individual experiences with caregiving.

Cultural practices and beliefs around parenting and food seem to have an effect on a mother's experience of caregiving for a child with Prader-Willi syndrome. Interviewees from Asian countries have shared how certain beliefs surrounding food, dining habits and appearances have affected their experiences or caused certain struggles. This is an interesting factor to look into and points towards how necessary national Prader-Willi syndrome support is in helping mothers from specific countries with provision of support.

Standardised Global Responses are integral to improving mother's individual experiences.

Mothers point towards how globally accessible information is critical for improving their experiences in caregiving. These include resources on psychological support, food and age-specific resources. Many mothers shared how they were bombarded with information upon their child's PWS diagnosis and were struggling to find apt information. Ensuring that key information is easily accessible online to mothers around the world is an important area for IPWSO to consider.

Summary of Recommendations

Psychological Support

- Increasing access to psychological support for mothers such as online therapies or support groups that can be easily accessible around the world.
- Offering personalised emotional support for mothers with children who exhibit specific or unique Prader-Willi syndrome symptoms.
- Extending psychological support to the siblings and extended families of people who have Prader-Willi syndrome.
- Spotlighting individual stories and experiences of mothers around the world to offer a sense of encouragement for new mothers.
- Developing international peer support programmes for new mothers by partnering them or connecting them with experienced mothers.

Increasing Accessibility to Resources

- Working with regional organisations to increasing public awareness, by conducting talks at local community centres, hospitals and schools.
- Improving IPWSO's status on online searches so that mothers around the world can easily find the right resources and information fast.
- Categorising resources based on a child's age.
- Working with regional medical professionals to use the experiences of mothers of children with Prader-Willi syndrome in continuing medical education sessions.
- Providing legal and pro-bono advice for mothers navigating around insurance and benefits access for their children.

Cultural Sensitivity and Regional Support

- Develop papers on the experiences of mothers around the world and the unique cultural challenges they face surrounding food, exclusion and societal attitudes.
- Develop resources specific to social stigmas experienced by mothers of children with Prader-Willi syndrome and the challenges they face i.e. shame, exclusion, isolation etc.
- Keep in regular contact with mothers from each region to understand their specific cultural challenges - especially Asia, Latin America and Africa.
- Working with national organisations to develop regionally specific Prader-Willi syndrome resources.

Expanding Research

- Developing age-based Prader-Willi syndrome research, tracking the development of maladaptive behaviours with maternal stress.

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