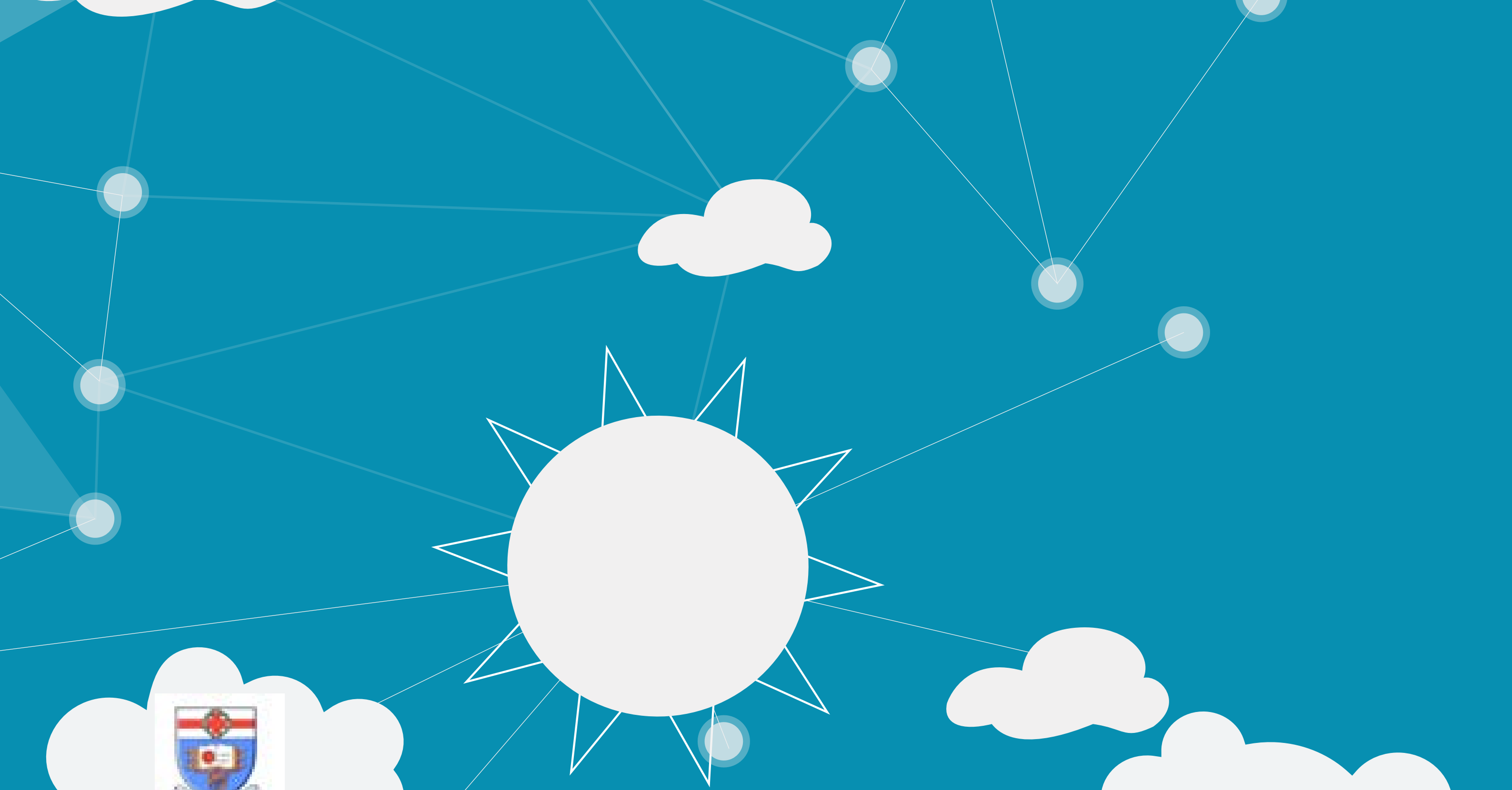
A Population-Based Profile of Prader-Willi Syndrome in Ireland



Tallaght Hospital

Trinity College Dublin
Coláiste na Tríonóide, Baile Átha Cliath
The University of Dublin

The funding for this research was provided by the Maynooth Students for Charity Galway Cycle

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PWSAI

Prader Willi Syndrome Association Ireland

Seeking a world where people with PWS, their families, and their carers are supported in all aspects of their care, education and well-being.

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Most of all we would like to thank all the people with PWS and their families and carers who participated in this research.

Foreword

"You are Not alone…Together we can do great things!"

Since the foundation of PWSAI as a charity, many committees have worked tirelessly to advocate and work towards improving services to support the needs of individuals with Prader-Willi syndrome and their families. This report would not have been possible but for the dedication and determination of those who set down the foundation of PWSAI and developed it to what it has become today.

In October 2015, following a meeting with the National Director of the HSE, it became clear to us as an association we needed to gather data to support our position that the needs of individuals with PWS of all age groups were not understood by the State and thus were not being met. A decision was taken to complete a National Survey across all age groups. This is the first national survey carried out of people with PWS and their families. We were very fortunate to have had Dr Marguerite Hughes as a committee member, as it was through her tenacity and drive this survey came to reality. PWSAI commissioned the research that formed this report and with the assistance of Prof Louise Gallagher (TCD) and her research team (Sarah Marie Feighan and Dr Hyun-Ju Kang) the survey was completed. Throughout the survey the extensive experience of Prof Edna Roche, Paediatric Endocrinologist, has been extremely valuable. Prof Roche runs a multidisciplinary Prader-Willi syndrome clinic at the Children's Hospital, Tallaght.

Without the contribution made by the 61 families who completed the survey, this report would not have become a reality. Every family who got involved had to go through the upset and frustration of remembering and reliving past and present difficulties and the individual challenges they encountered while trying to seek services for their family member with PWS, whether that related to months for those with young babies or decades for those with older family members. But everyone who participated embraced the vision of this survey and the understanding that collectively we can achieve great things. This report is your report and you should all be very proud of the accomplishment.

Currently, five or six babies are born with Prader-Willi syndrome in Ireland every year and we are acutely aware that some of the content of this survey may make difficult reading for parents of very young children with PWS. It is important to remember that all individuals born with PWS are unique individuals – just like everyone else. But what they do have in common is a PWS genetic chromosomal thumb print. As an association, we have support structures in place to help. So reach out! You are not alone!

The final report is an asset for all families with PWS members, as it documents in a scientific way what it is to have PWS and the impact that has on the family. It also highlights the uniqueness of the condition and points to the need for PWS specific services to provide appropriate care for the person with PWS throughout their lifetime. It will help each family to spread awareness and understanding of what it is to live with PWS in Ireland today.

As an association, PWSAI will use the findings of this report to advocate for improved services at national and local level with Government, the HSE, medical professionals, educational bodies, politicians, and care providers. PWS is a unique and challenging condition that requires intense, targeted and PWS specific supports.

To all those who contributed in any way to make this report a reality…
Thank You!!

Anthony Carr
Chairperson, PWSAI

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Abstract

This report is the result of collaboration between the Prader-Willi Syndrome Association Ireland (PWSAI) and Trinity College Dublin.

The aim of this report is to assess the needs of people with Prader-Willi syndrome (PWS) and their families in Ireland.

PWS, a complex multisystem genetic disorder, is characterised by developmental abnormalities leading to somatic and psychological symptoms. Symptoms of PWS include infantile hypotonia and failure to thrive followed by life-long hyperphagia, developmental delays and moderate-to-severe behavioural problems and several physical problems that impact health. The results of the report are based on a paper survey designed by experts in the field of PWS in Ireland and carers of people with PWS. The survey was completed by 61 primary carers for people with PWS across the country. This represents around 60% of all individuals with PWS living in Ireland today. The findings of this report reveal the multiple physical, developmental, and behavioural issues associated with PWS and how these issues require families and carers to devote considerable time and effort to care for a person affected by this condition.

Background

Clinical Characteristics

PWS is a multisystem genetic condition characterised by clinical manifestations which differ at each stage of life. PWS presents with severe hypotonia, failure to thrive and feeding difficulties in early infancy, followed by developmental delay and intellectual disability, a characteristic behavioural, social and psychiatric pattern, short stature and growth hormone insufficiency, hypogonadism causing small genitalia, cryptorchidism and incomplete pubertal development.

In later infancy or early childhood, people with PWS develop an insatiable appetite which if left uncontrolled, can lead to extreme obesity. It is the most common genetic syndrome associated with potentially life-threatening obesity (Mazaheri et al., 2013). PWS is also associated with an increased risk for numerous other medical and comorbid psychiatric problems including strabismus, scoliosis, hip dysplasia, seizures, gastrointestinal and skin problems, depression, anxiety and psychosis
(Cassidy et al., 2011; Sinnema et al., 2011).

Prevalence

PWS is estimated to affect 350,000-400,000 individuals worldwide. The prevalence of PWS has been reported as 1 in 10,000-30,000 (Vogels et a., 2004). Precise prevalence data in Ireland are not available; however reports from the National Centre for Medical Genetics (NCMG) in Our Lady's Hospital for Sick Children Crumlin (OLCHC), which is the only centre that tests for PWS in Ireland, indicate that 5-6 new cases of PWS are diagnosed each year. It is also possible that some individuals are tested in laboratories outside the state and that there are others who have never been genetically tested. Therefore the rate of 5-6 new cases a year is a minimum frequency. For a birth rate of 70,000 per annum (Growing up in Ireland, 2011), this amounts to a minimum frequency of 1 in 11,000 annually.

Diagnosis

Consensus diagnostic criteria for PWS developed in 1993 (Holm et al., 1993; see appendix one) have proven to be accurate in detecting new cases (Gunay-Aygun et al., 2001) and continue to be useful for clinicians. However, confirmation of the diagnosis requires molecular genetic testing, which was not widely available when the criteria were developed. Optimal diagnosis of Prader-Willi Syndrome is done via DNA methylation testing (Cassidy & Driscoll, 2009). This testing is currently provided in Ireland at the National Centre for Medical Genetics.

Genetic Underpinnings of PWS

PWS is caused by lack of expression of genes on the paternally inherited chromosome 15q11.2-q13 region. There are three main genetic subtypes in PWS: paternal 15q11-q13 deletion (65-75 % of cases), maternal uniparental disomy 15 (20-30 % of cases), and imprinting defect (1-3 %). These genetic changes occur as random events during the formation of egg and sperm in early embryonic development and thus there is typically no history of the disorder in the families of affected individuals
(Angulo, Butler & Cataletto, 2015).

Treatments

There is currently no cure for PWS. Due to the intensity and the range of clinical manifestations of PWS, input from a large variety of professionals and services is needed to ensure adequate care, prevent secondary disability and optimise quality of life for this patient population (McCandless et al, 2011). Managing the symptoms of PWS often requires modifications in the entire family’s daily routine. In infancy, special nipple or nasogastric tube feeding is used to ensure adequate nutrition while physical therapy is used to improve muscle strength and mobility.

Treatments .... continued

In childhood, around the clock supervision, locking of food cabinets and refrigerators, monitoring and regulating food intake, accommodating cognitive delays and behavioural problems, and appropriately responding to, managing and coping with emotional outbursts is required.

Beyond environmental and behavioural treatments, selective serotonin reuptake inhibitors and atypical antipsychotics have been used with children with PWS (Soni et al., 2007) to treat comorbid symptoms of depression and psychosis. While known to be effective in affective and psychotic disorders generally, research into their effectiveness in PWS is limited (Whittington & Holland 2010). No specific medication has been found to be uniquely helpful to all individuals with PWS (Butler et al., 2006).

Optimising body composition, growth and development are key therapeutic targets. Growth hormone replacement therapy is used not only for its effect on stature but also for its metabolic effects and impact on body composition (improving muscle strength, increasing lean body mass to normalize height while decreasing fat mass). However this treatment requires daily injections and frequent endocrine evaluations. Additional hormonal treatments at puberty can be considered to develop or induce secondary sexual characteristics and optimise bone health (McCandless, 2011). Speech therapy is required, particularly in infancy to support the management of feeding difficulties. Educational planning and behaviour management are often necessary to manage behavioural problems and support an appropriately structured environment.

Context for the Current Report

Prader Willi Syndrome Association Ireland (PWSAI) is an organisation founded by parents in the late 1980s as a support group for individuals with PWS and their families in Ireland. Its mission is to raise awareness and understanding of PWS and to improve the choice and quality of care, education and support for persons with PWS.

PWSAI commissioned the research that formed this report from Trinity College Dublin (TCD). Prof Louise Gallagher, Chair of Child and Adolescent Psychiatry, TCD, and Consultant Child and Adolescent Psychiatrist in the HSE was the principal investigator along with members of her group, The Autism and Rare Neurodevelopmental Disorders Research Group, in collaboration with Prof Edna Roche, TCD and Consultant Paediatric Endocrinologist, Tallaght Hospital. Professor Roche has extensive experience in PWS and leads the largest paediatric PWS service in Ireland at the National Children's Hospital, Tallaght. The research was funded by the proceeds of the PWSAI Maynooth Students for Charity Galway Cycle 2014.

Aim of the Report

The primary aim of the research contained in this report was to map the support needs of families with a person affected by PWS through the different life stages and to make recommendations regarding support provisions.

In addition, it aimed to identify the services frequently required by people with PWS and the impact of PWS on the individual and their family.

Methods

A paper survey for primary carers of people with PWS was developed to ask about their experiences of caring for an individual with PWS and their views on the support requirements. The questionnaire was designed by researchers in TCD, members of PWSAI and Prof Edna Roche, an expert in the field of PWS. The study employed two different avenues of recruitment. Members of PWSAI were informed about the study via email and asked to submit their contact details if they would like further information. In addition, individuals with PWS attending Prof Edna Roche's PWS clinic in Tallaght Hospital were informed of the study and provided with contact details for the research assistant.

Participants were informed of the study via telephone and could decide if they would like a study pack posted to them. The study pack included an information sheet, consent form and questionnaire. Participants were instructed to read through the study pack materials and consider if they would like to participate. If agreeable, the consent form was signed; the survey was completed and returned via post to the Autism Research Group Office, TCD. Each questionnaire consisted of both quantitative and qualitative questions and took 45-60 minutes to complete.

All survey data was uploaded and analysed through the use of SPSS. Means and standard deviations were reported and relevant data is presented below. Ethical approval for this project was granted by the Tallaght Hospital/St James's Hospital Joint Research Ethics Committee.

Results Section

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Section 1

Overview

A total of 61 primary carers for a person with a confirmed diagnosis of PWS participated in the study. As the clinical manifestations of PWS differ for each stage of life, participants were divided into five different groups; infants aged 0-4 years, school age children aged 5-12, teenagers aged 13-17, adults living at home (ALH) and adults living in supported accommodation (ALSA). Table one below depicts a breakdown of the age and gender for each participant group. There was a greater percentage of females represented in our sample (57% overall), however this seems to have occurred due to chance as PWS is not known to affect either sex preferentially (Angulo, Butler & Cataletto, 2015).

	Age 0-4	Age 5-12	Age 13-17	Adults Living at Home	Adults Living in Supported Accommodation	Total
No. of Participants	10	16	10	16	9	61
Age	2.1 (±1.2)	8.9 (±2.4)	14.8 (±1.2)	23.7 (±4.5)	34.1 (±9.0)	61
Age Range	6m - 4	5 – 12	13 – 17	18 – 35	23-52	6m - 52
Male (%)	70	31.3	50	31.3	44.4	43
Female (%)	30	68.8	50	68.8	55.6	57
Type of PWS						
Deletion	3 (30%)	7 (44%)	5 (50%)	9 (56%)	2 (22%)	26 (43%)
Uniparental Disomy	5 (50%)	6 (38%)	1 (10%)	3 (19%)	1 (11%)	16 (26%)
Imprinting Defect	0	0	0	1 (6%)	1 (11%)	2 (3%)
Unknown	2 (20%)	3 (19%)	4 (40%)	3 (19%)	5 (56%)	17 (28%)

Table 1: Characteristics of the person with PWS for whom participants in the survey cared. Values are given as means and standard deviations except where percentages are indicated.

Figure one illustrates a breakdown of gender for each participant group.

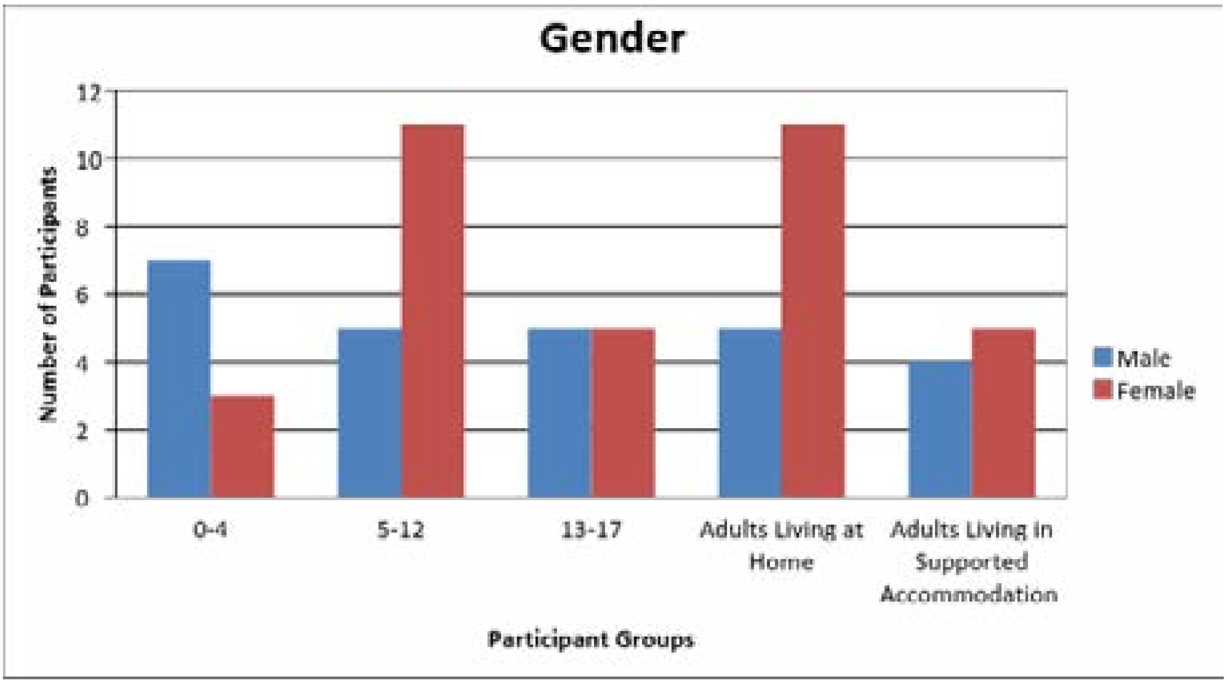


Figure 1: Graph depicting the breakdown of gender for each participant group

Figure two illustrates the relationship of the survey respondents to the person with PWS. The survey was designed to be completed by the primary carer of the person with PWS. In total, 50 of the respondents were the biological mother of the person with PWS, eight were the biological father of the person with PWS and three were siblings of the person with PWS.

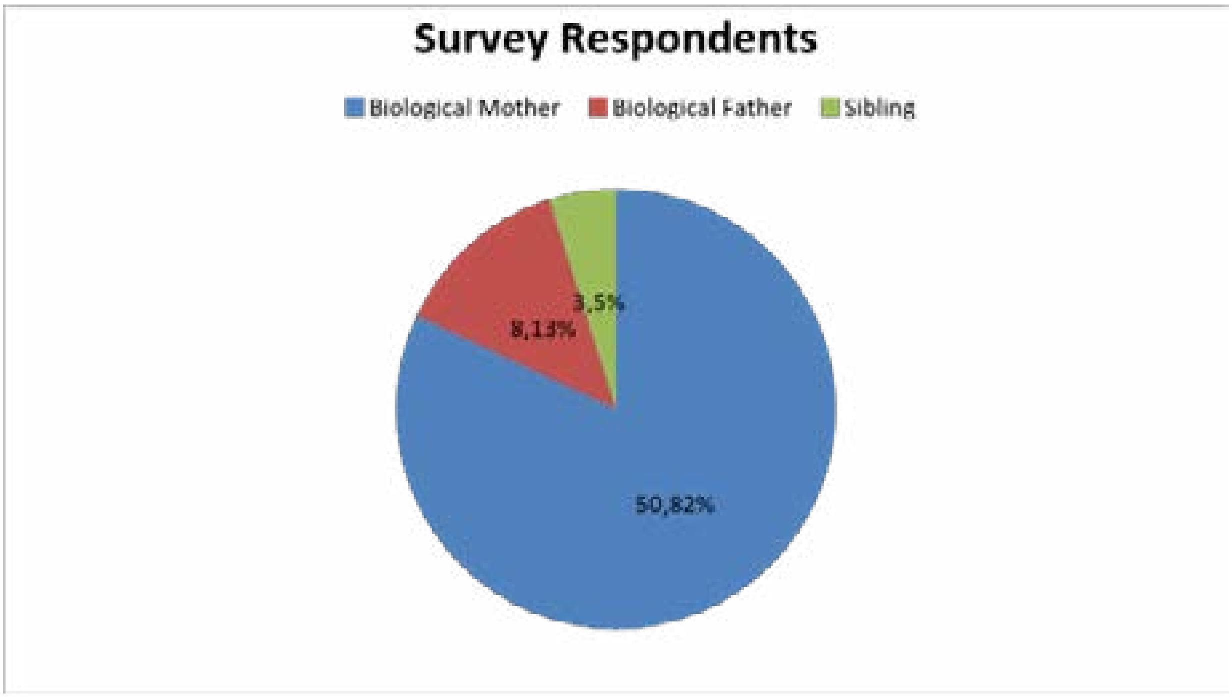


Figure 2: Graph depicting the relationship of the survey respondent to the person with PWS

In terms of type of PWS diagnosis, 26 (43%) respondents cared for a person with deletion PWS, 16 (26%) cared for a person with uniparental disomy, two (3%) cared for a person with an imprinting defect and in 17 (28%) cases the carer was unsure of the type of genetic diagnosis.

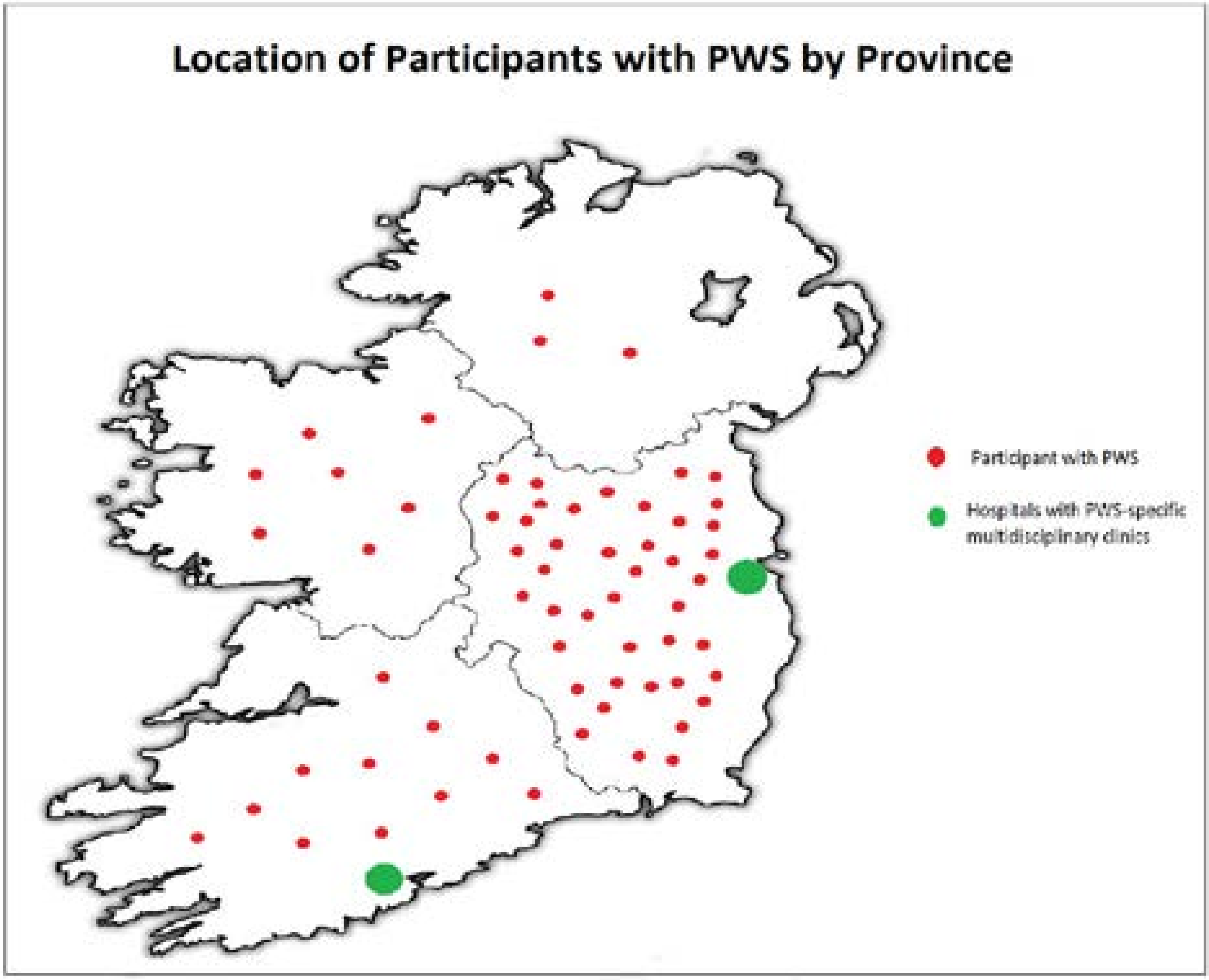


Figure 3: Map showing the location of participants with PWS by province

Section 2

Birth History

The majority of respondents (60%) reported that their relative with PWS was born “on or near due date” with the average length of pregnancy being 38 weeks which is in keeping with the national average length of pregnancy (Growing up in Ireland, 2011). The average age of the birth mother at the time of birth was 34.3 and the father was 35.9 (see table 2).

Table 2: Characteristics of PWS Birth History

	Length of Pregnancy	Father’s Age at Birth	Mother’s Age at birth	Weight at birth	ICU Placement (Days)
Average	38 weeks	35.9 years	34.3 years	2.82 Kg	26 days
Range	31-42 weeks	21-47 years	19-45 years	0.96 - 4.01 Kg	1 – 120 days

Fifty-three of the participants (87%) reported that their relative with PWS was admitted to the intensive care unit (ICU) after birth (see figure 4). The amount of time spent in ICU ranged from 1 to 120 days with the average duration of admission lasting 26 days. This is significantly higher than the average duration of admission for the general population which is four days (GUI, 2014).

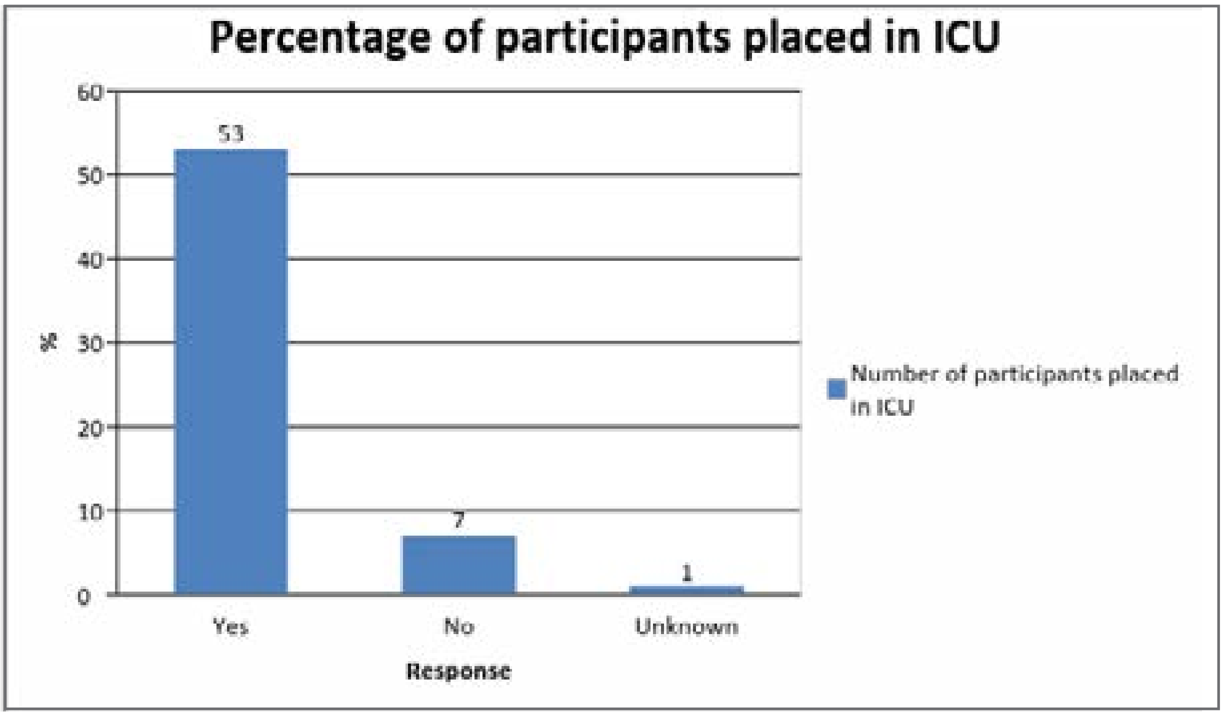


Figure 4: Graph depicting the percentage of participants with PWS admitted to the Intensive Care Unit after birth

Thirty-five participants (57%) reported that that their relative with PWS required feeding via a nasogastric tube and 12 participants (21%) reported their relative with PWS encountered difficulties when transitioning from a liquid diet to a solid diet (see figure 5).

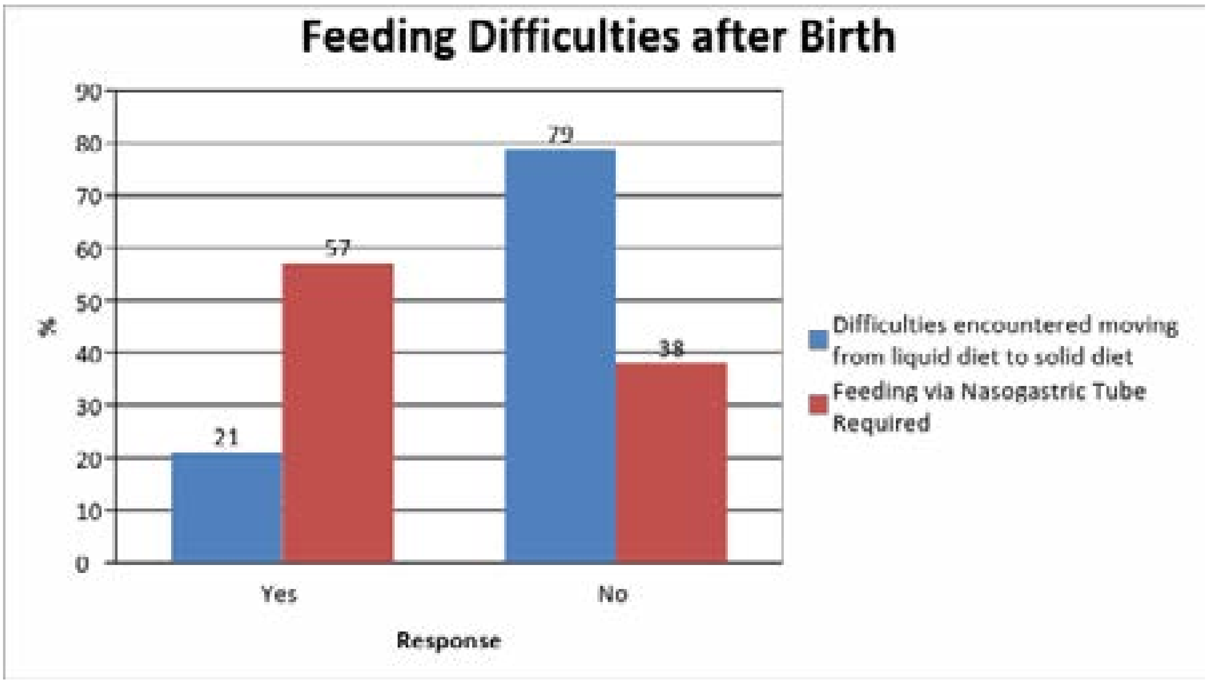


Figure 5: Graph illustrating the percentage of people with PWS who encountered feeding difficulties/required feeding via nasogastric tube after birth

Section 3

Diagnostic History

The average age of diagnosis for each age group shows that the time taken for a person to be diagnosed with PWS has significantly decreased. Children born with PWS in the last four years (2012-2016) received their diagnosis within four weeks on average, compared with an average of 6 weeks for those born between 2005 -2011 and 19 weeks for those born between 1999 and 2003 (see figure 6).

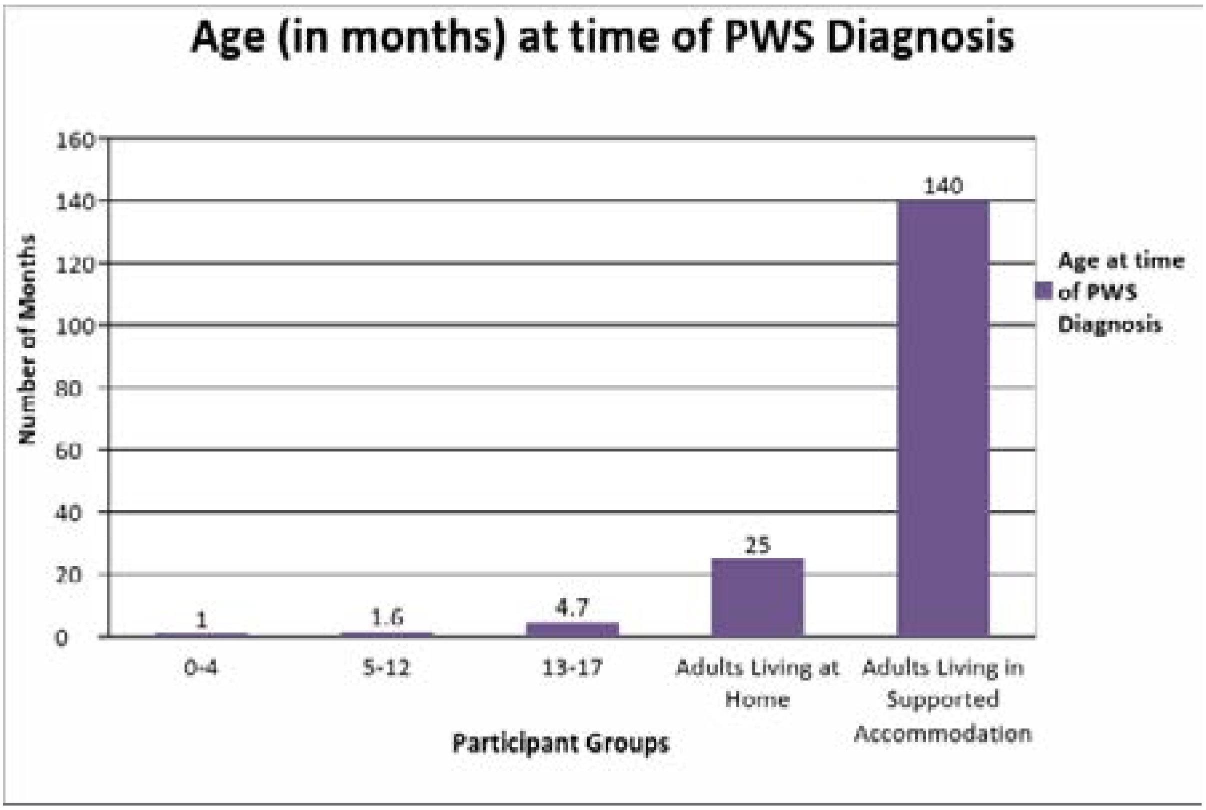


Figure 6: Graph illustrating the average age (in months) at which each group received their PWS diagnosis.

Section 4

Developmental History

Figure seven is a graph depicting the average age in months that it took people with PWS to reach their developmental milestones in comparison with typically developing children (Centres for Disease Control and Prevention [CDC], 2010). There was a significant delay in achieving developmental milestones for children with PWS compared to the typically developing population (see figure 7).

Figure 7: Graph illustrating differences between children with PWS and typically developing children in reaching milestones (CDC, 2010).

In order to analyse the level of intellectual disability seen in people with PWS in the study, only respondents who confirmed that the individual with PWS that they cared for had undergone an IQ assessment were included. Out of the 61 respondents, 37 (60.7%) confirmed that the individual with PWS had undergone an IQ assessment. The majority of the primary carers (23 cases, 65.7%) reported that their relative with PWS had received a mild learning disability diagnosis after undergoing IQ assessment. A moderate learning disability diagnosis was the next most frequent diagnosis (9 cases, 25.7%) with only 1 participant reporting that their relative with PWS had not been diagnosed with a learning disability (see figure 8).

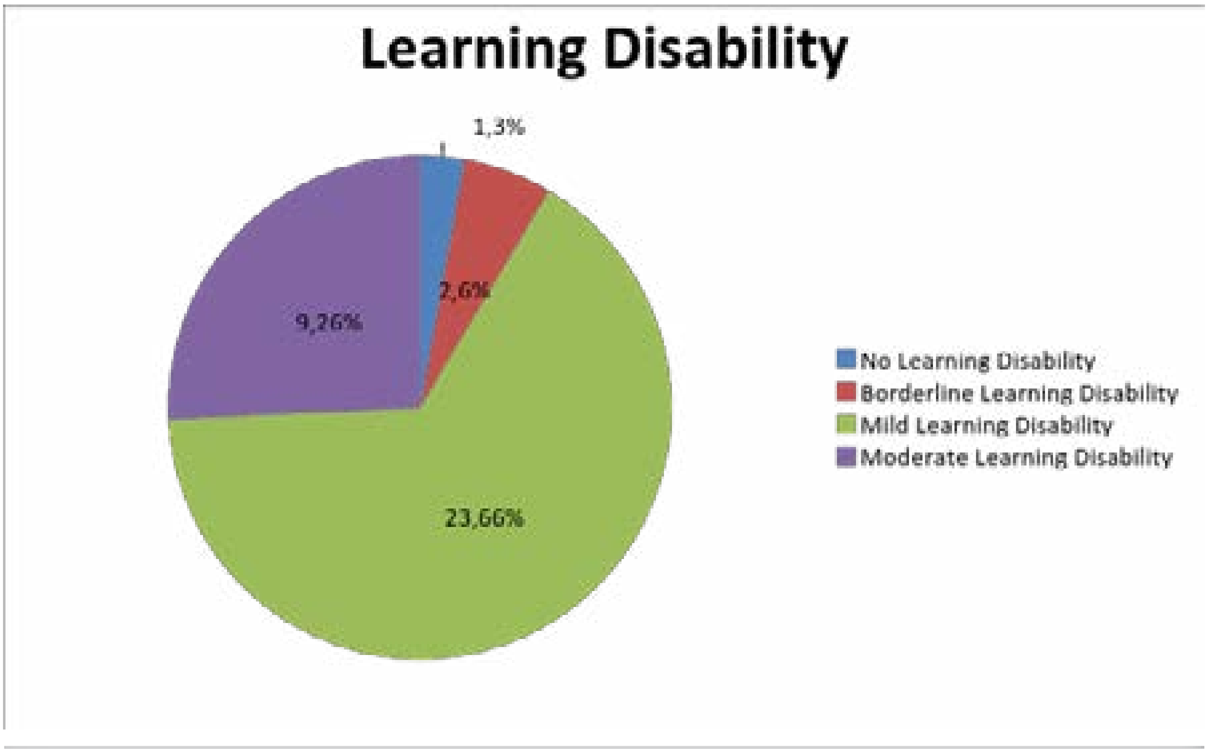


Figure 8: Graph illustrating the IQ results of people with PWS who have undergone an IQ test

Section 5

Education

Figure 9 on the following page illustrates a breakdown of the type of educational settings attended by individuals with PWS aged 5-12, aged 13-17, and adults. All participants between the ages of 5-17 were reported as currently attending an educational setting. The majority of the school aged children (ages 5-12) were attending a mainstream primary school (n = 10, 63%) with the remainder attending either a special needs primary school, a special needs class in a primary school or a special needs secondary school. In the adolescent group, the majority were attending a special needs secondary school (n = 6, 67%). In the infant group aged 0-4, two participants were attending a preschool. The average in the infant group was reported as 2.1 years which may explain low preschool attendance. In terms of adults with PWS, the majority (83%) were not currently attending an educational setting.

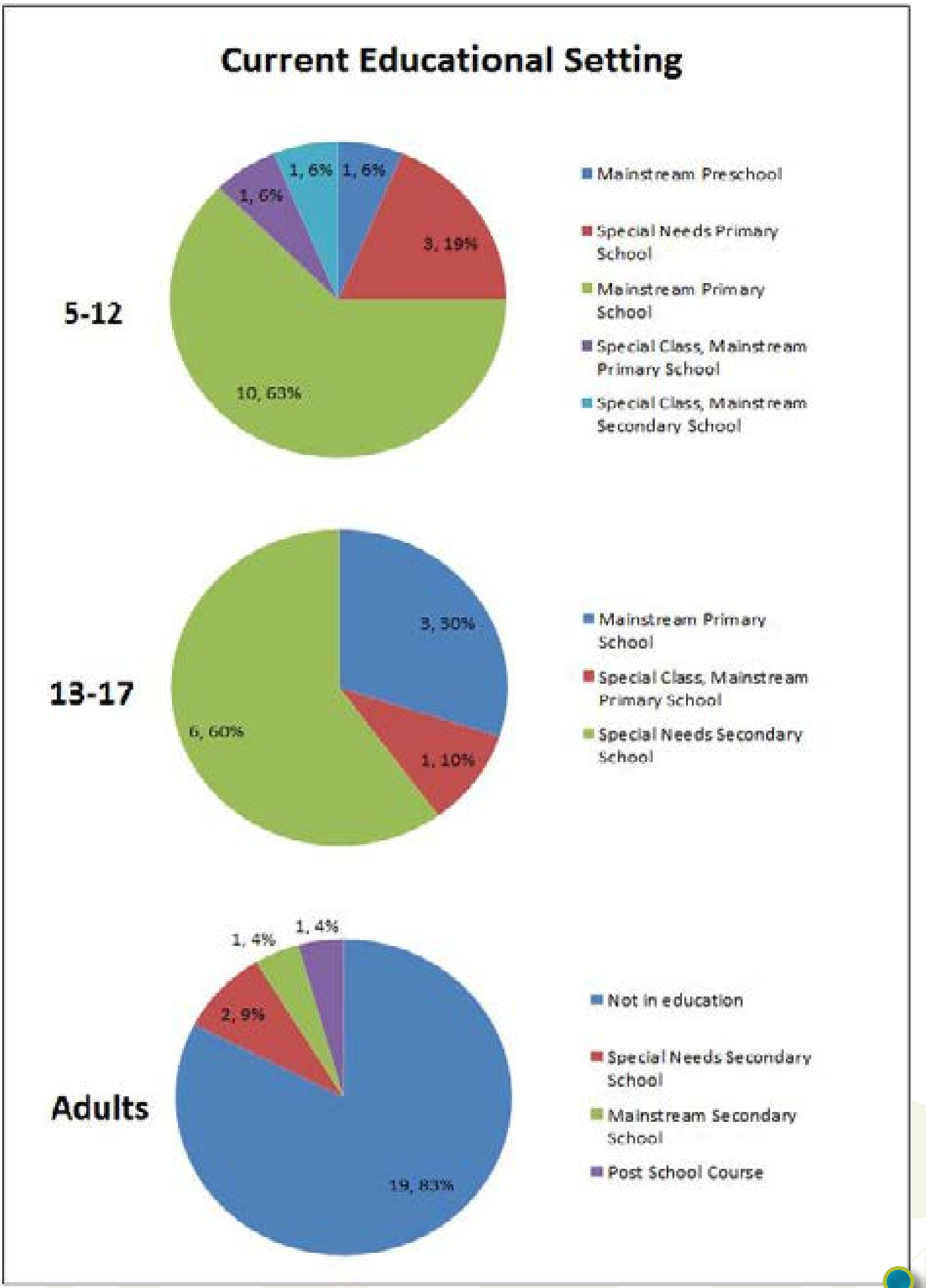


Figure 9: Pie charts illustrating the current educational setting of each participant group

Education ......continued

Figure 10 shows a breakdown of the educational supports currently being received by people with PWS in their educational setting. A higher number of participants had full-time Special Needs Assistants in the 5-12 group than the 13-17 group. This may be explained by the higher percentage of participants attending a mainstream school in the 5-12 group. The results show that a high level of individual support is required for each child with PWS when attending an educational setting.

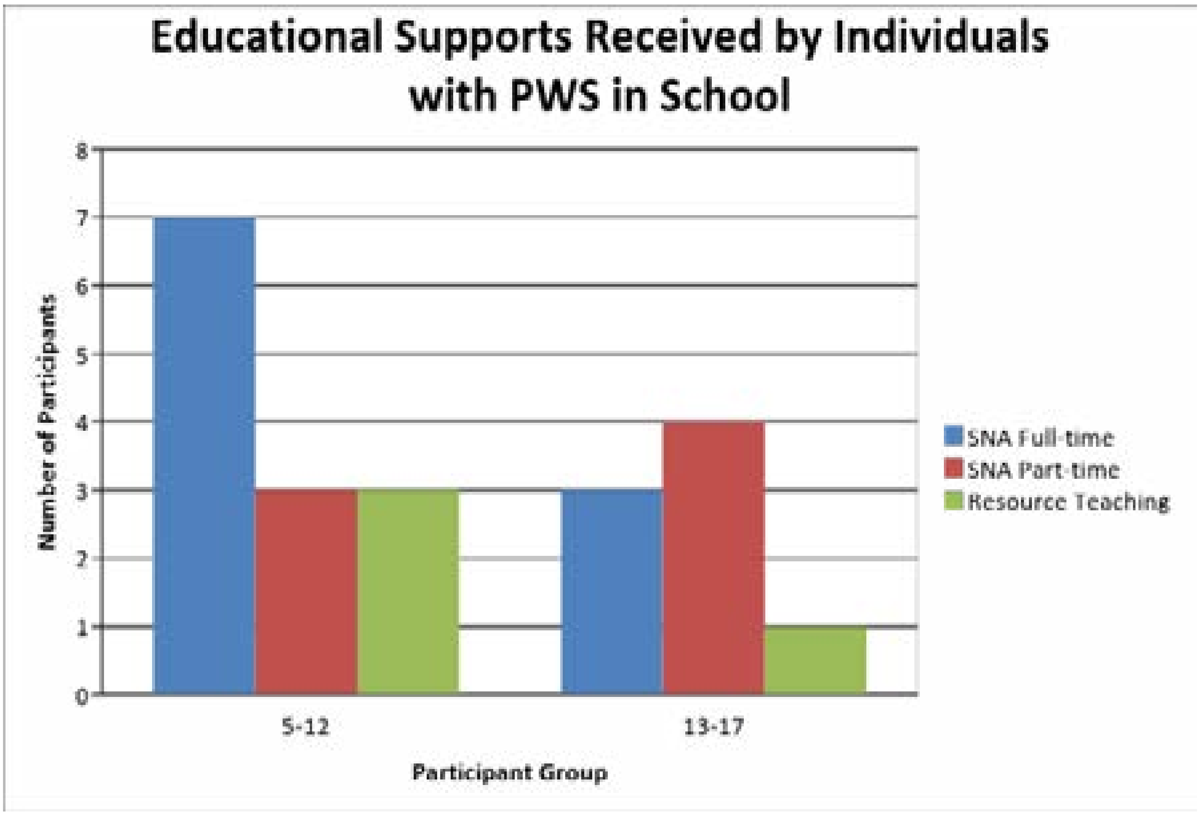


Figure 10: Types of Educational Supports being received by school-age children with PWS

Education ......continued

Participants were asked about the highest education qualification received by the person with PWS before they finished education. Figure 11 below shows at the time of the survey, 22% of adults were still attending an education setting. The majority of participants (56%) reported that the adult with PWS that they care for did not receive any education qualification at all. Thirteen per cent reported that the individual had received their Junior Certificate and 9% of participants had received their Leaving Certificate Applied.

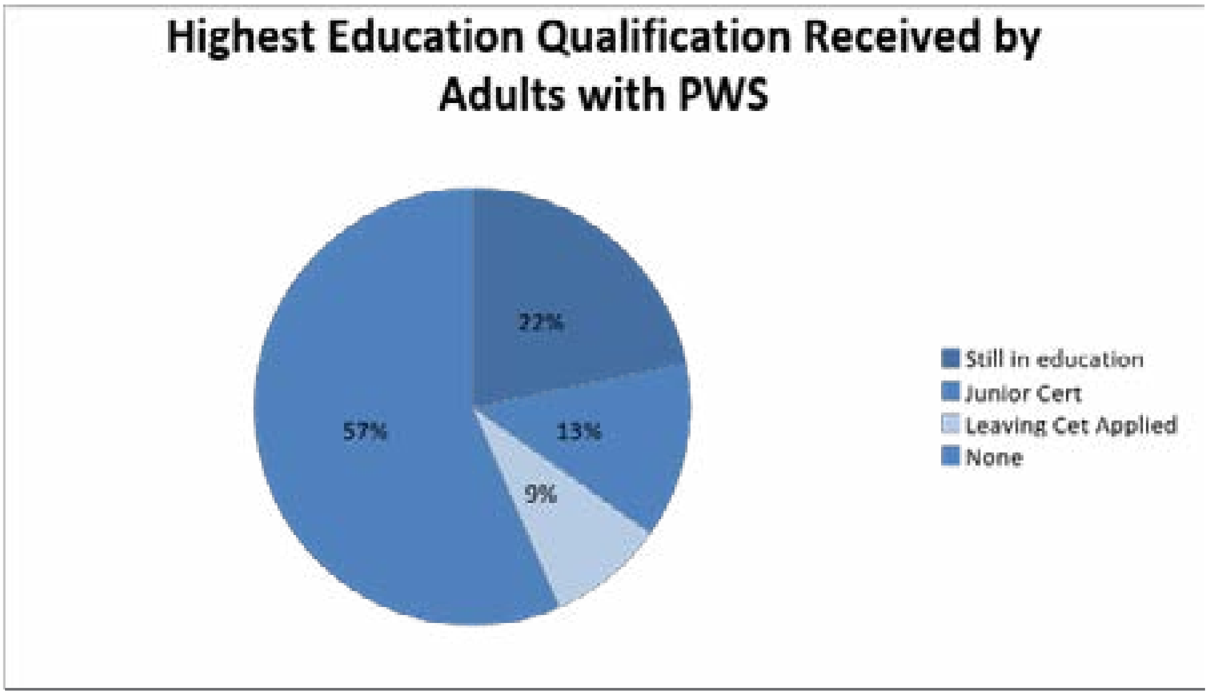


Figure 11: Type of Education Qualification Received by Adults with PWS

Education ......continued

Figure 12 below depicts the employment status of adults with PWS. Participants caring for adults with PWS were asked about the employment status of the individual with PWS. Only one individual with PWS was currently in paid employment (4%). An additional 3 (12%) were in voluntary employment. The majority of adult participants were attending day programmes.

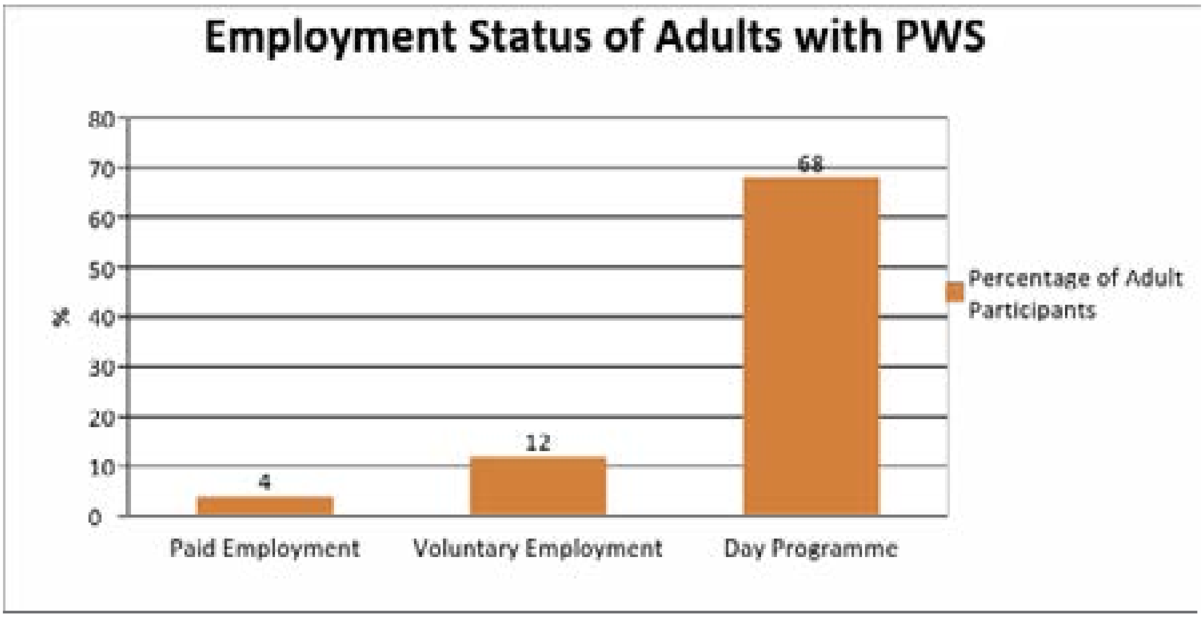


Figure 12: Graph depicting employment status of participants with PWS.

Section 6

Independence and Daily Living Skills

A section on independence and daily living skills surveyed the current amount of support that individuals with PWS required for day to day living. Respondents rated on a scale of 1-4 how much help the individual with PWS required with different daily living skills with 1 representing "no help", 2 "a little help", 3 "a significant amount of help" and 4 "maximal help". The infant group was excluded as these questions were not considered developmentally appropriate. Significant levels of personal assistance were required across all of the age groups. For example, over 40% of all adults required support with toileting, brushing teeth and showering. The adults living at home required even higher levels of support with dressing and showering. In the teenage group 80-90% of individuals required assistance with dressing, showering and brushing teeth while 50% required assistance with toileting. Similar proportions of individuals in the 5-12 year old age range also required assistance (with the exception of dressing which was 62%).

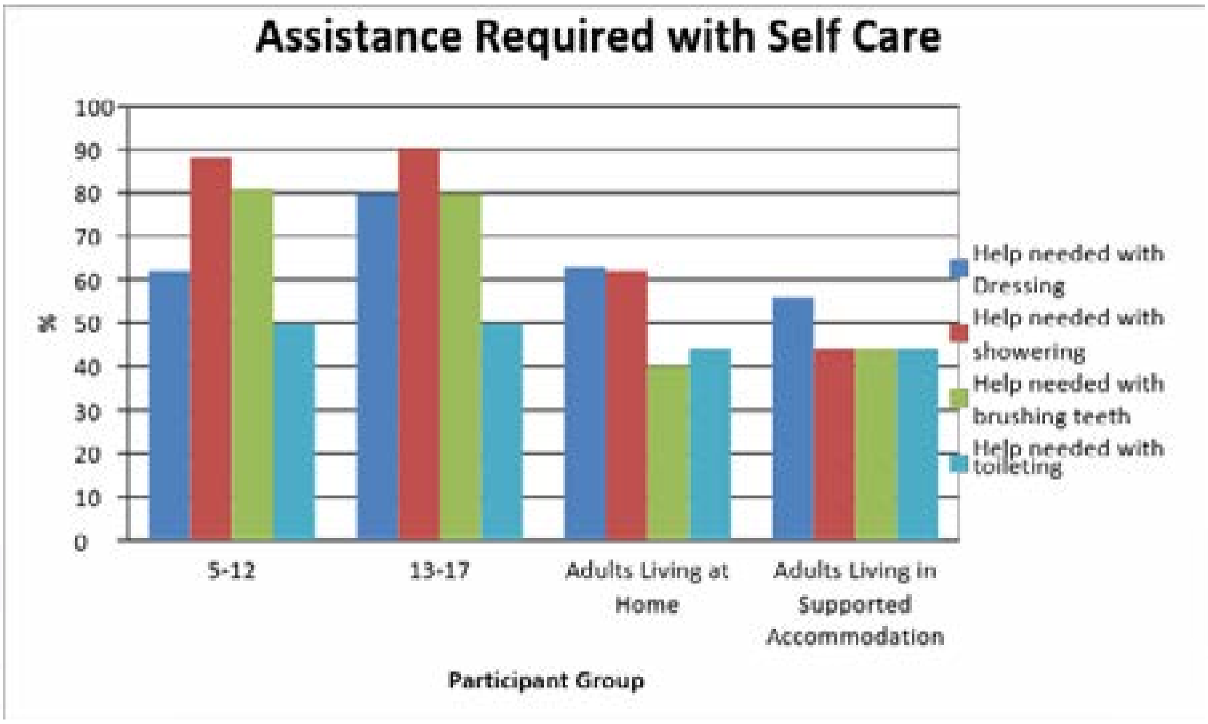


Figure 13: Percentage of carers that reported a score of 2 or higher on level of help required with each daily living skill

Independence and Daily Living Skills .... continued

Managing money and personal health were also significantly impaired for a high number of individuals. These questions were confined to adults. Ninety-three per cent of carers of the adults living at home group reported that help was required with managing health (e.g. taking medication) and 88% reported that help was required with managing money. Similarly in the adults living in supported accommodation group, 89% of the carers reported that help was required with managing health as well as managing money (see figure 14).

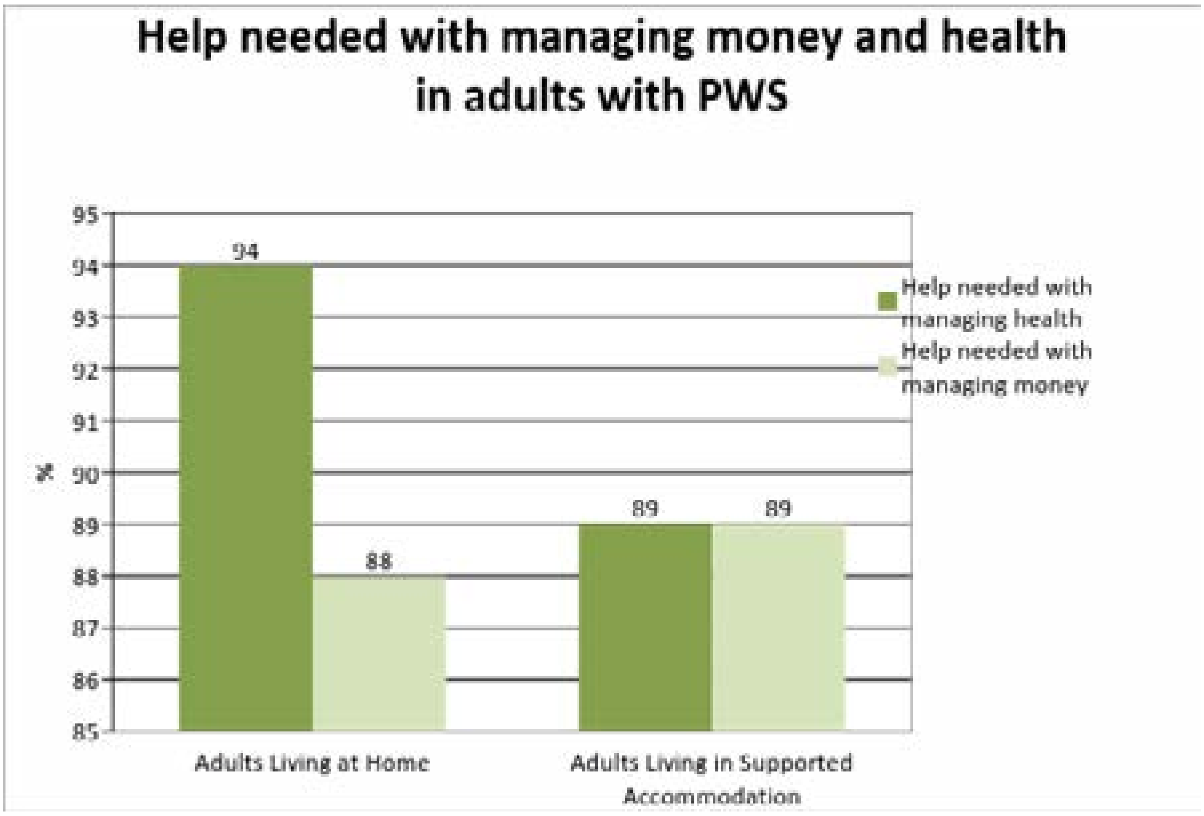


Figure 14: Percentage of carers that reported a score of 2 or higher on level of help required with managing health and finances

Section 7

Medical Needs

Hospital Admissions

Individuals with PWS experience a wide range of complex medical conditions and attend a wide range of health professionals. The complexity of PWS necessitates extensive support in clinical care throughout all life stages. Participants were asked how many times the person with PWS that they care for had to be admitted overnight to hospital as a result of symptoms of their PWS. Figure 15 below illustrates overnight hospital admissions for individuals with PWS as well as focusing in particular on the last 12 months. The adults living in supported accommodation had the highest percentage of overnight hospital admissions with 75% reporting an overnight admission due to symptoms of PWS in the last 12 months compared with the adults living at home group which was reported as 13%. Forty per cent of the 0-4 age group reported an overnight stay in the last 12 months and 47% of the 5-12 group reported an overnight stay in the last 12 months.

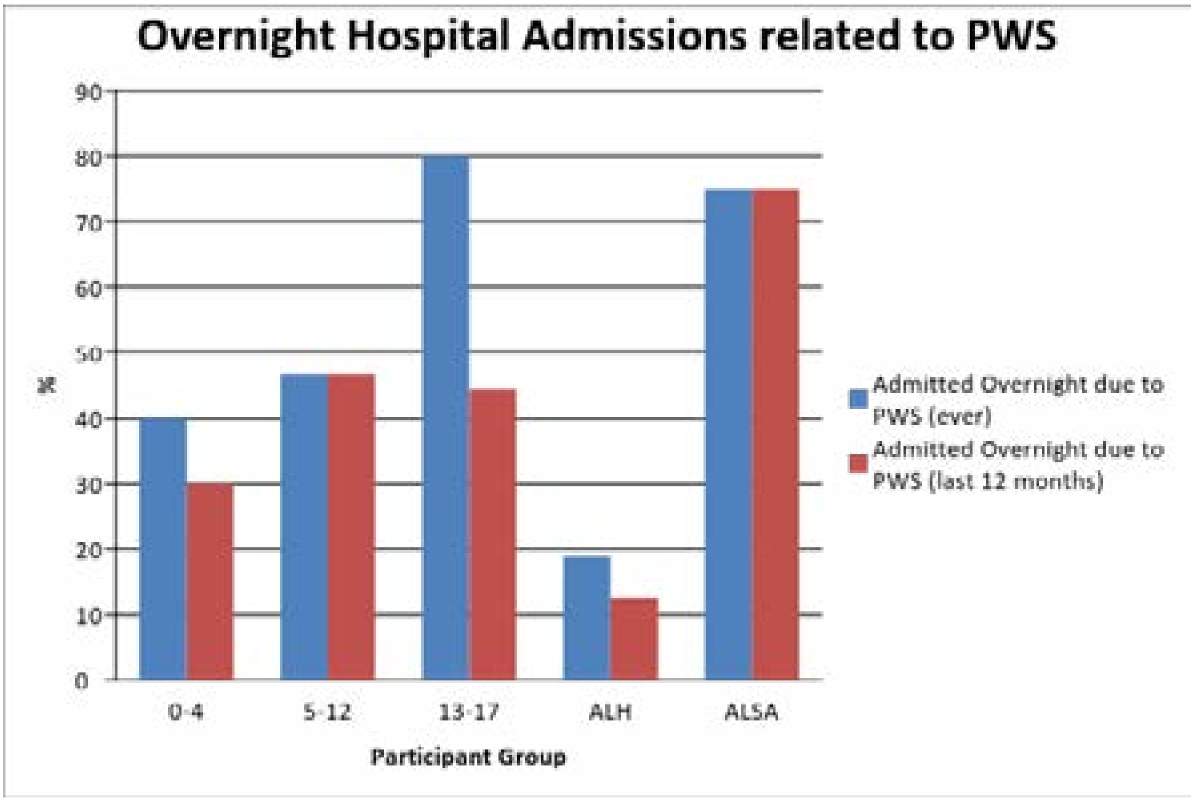


Figure 15: Graph illustrating percentage of individuals with PWS admitted overnight to hospital due to PWS

Medical Specialists

In terms of medical specialists visited by individuals with PWS, the type of medical specialist required varies across the groups. In the infant group, the top five medical specialists seen are: paediatric endocrinologist (90%), paediatrician (70%), dentist (70%), respiratory doctor (60%) and ophthalmologist (40%) (See figure 16).

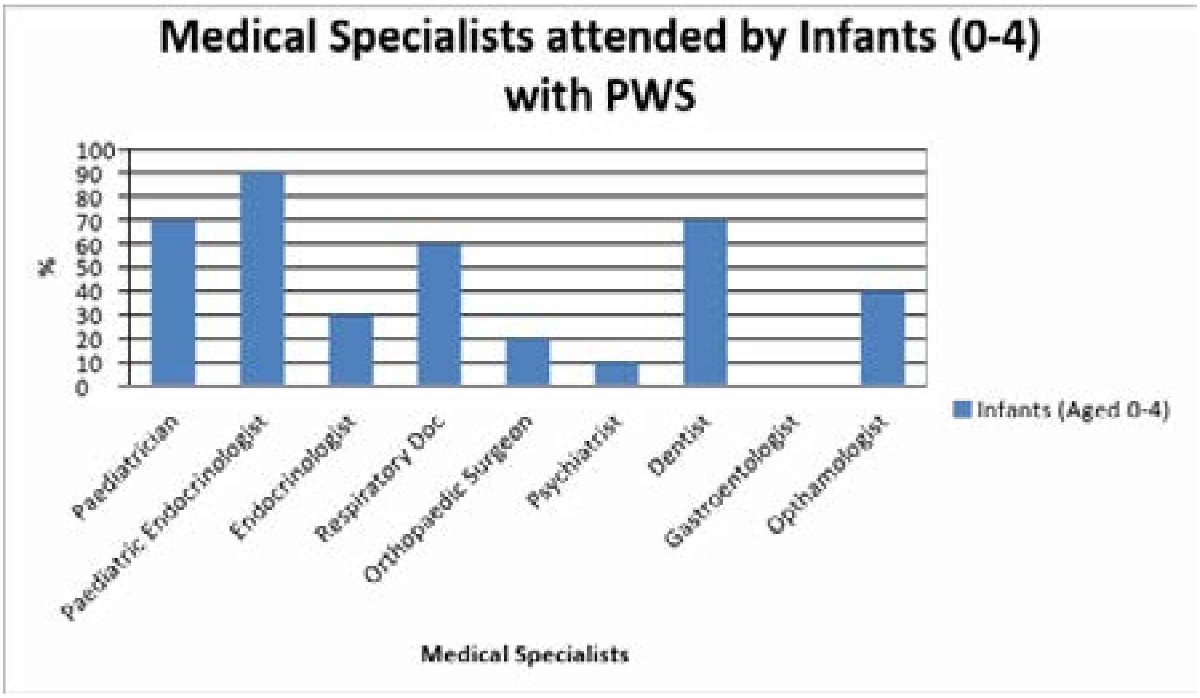


Figure 16: Graph illustrating medical professionals attended by infants with PWS

Medical Specialists ....continued

Similar medical specialists were visited by the 5-12 group with the addition of visits to an orthopaedic surgeon (44%) psychiatrist (31%) and gastroenterologist (25%) (See figure 17).

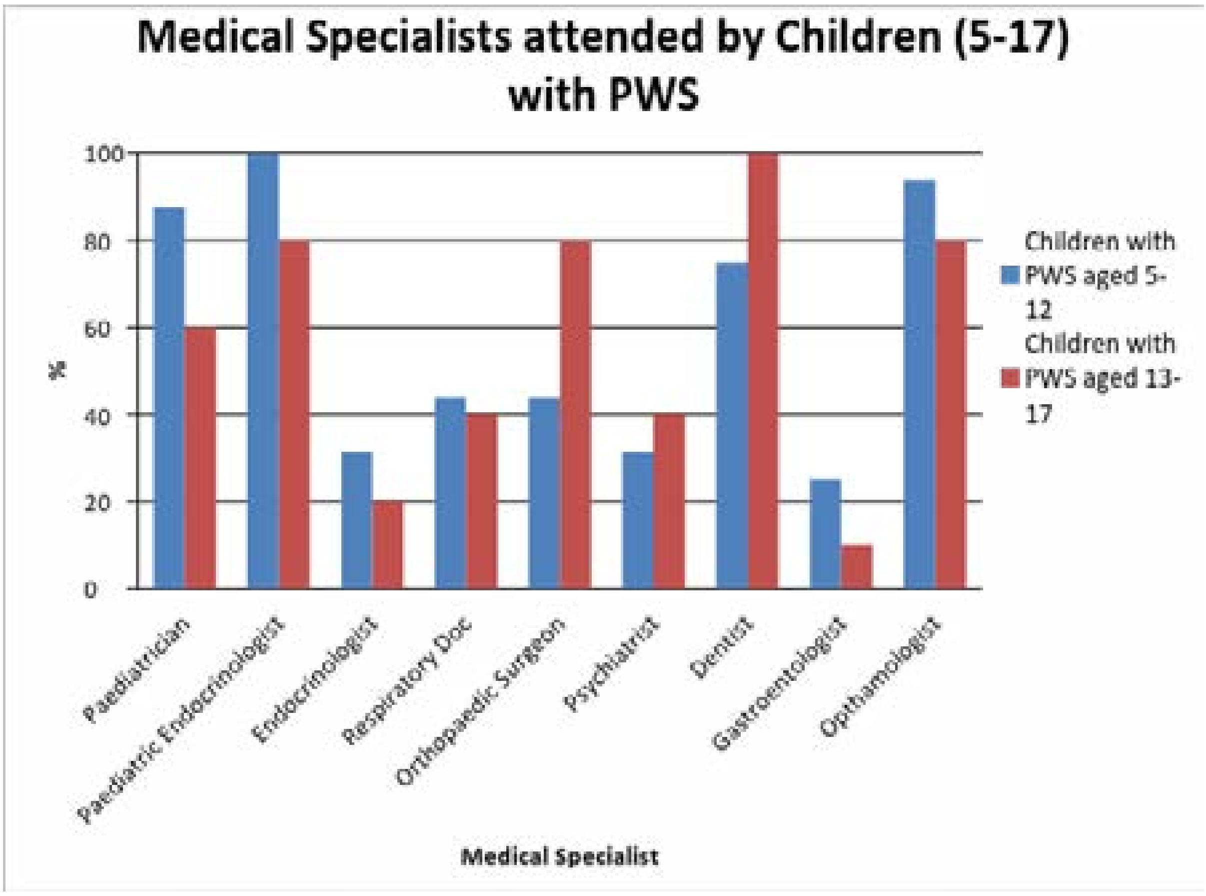


Figure 17: Graph illustrating medical professionals attended by children with PWS

Medical Specialists ....continued

In the 13-17 group, there was an increase in the percentage of participants attending a psychiatrist (40%) and orthopaedic surgeon (80%) (See figure 18). In the two adult groups, the biggest increase was seen in the percentage of participants attending a psychiatrist with 44% of the adults living at home group and 78% of the adults living in supported accommodation reporting visiting a psychiatrist (see figure 18).

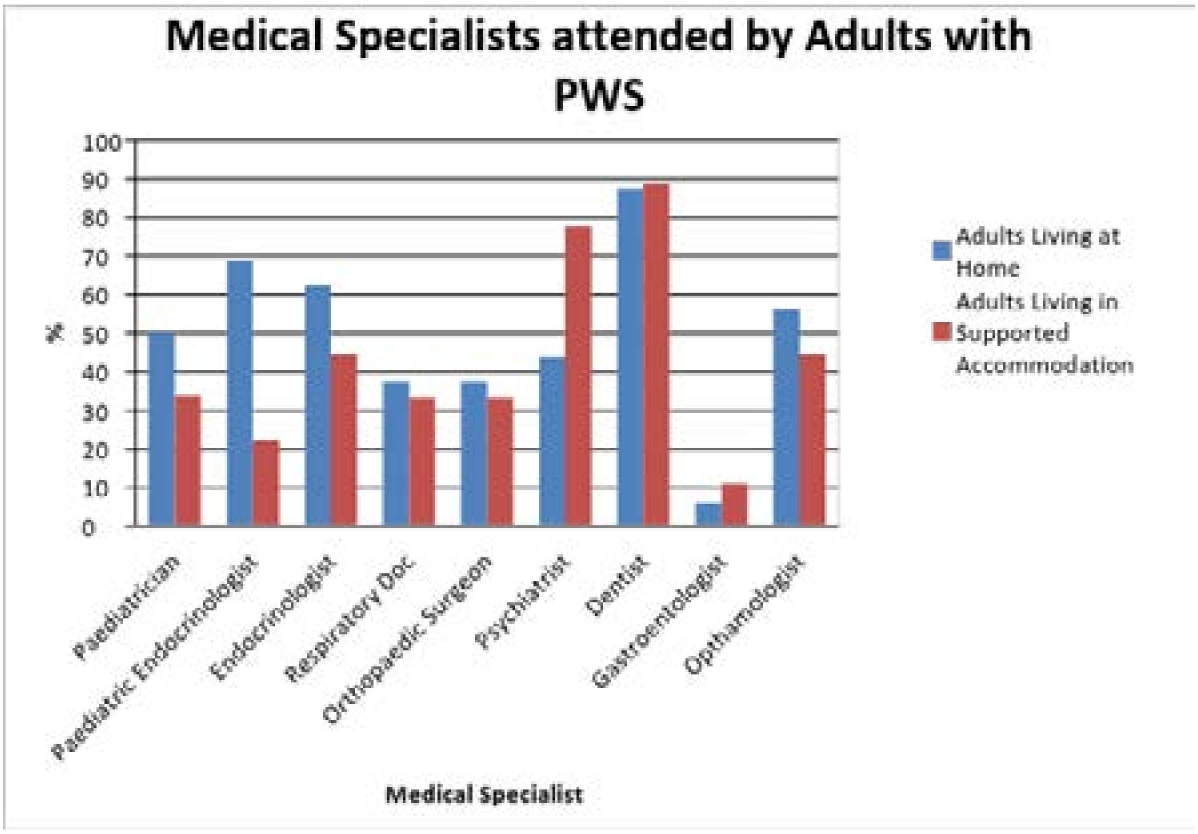


Figure 18: Graph illustrating medical professionals attended by adults with PWS

Medical Conditions

Similarly, the types of medical conditions reported by the participants changed across the groups. The most common medical conditions affecting infants were hypotonia (90%) and sleeping difficulties (80%) (see figure 19).

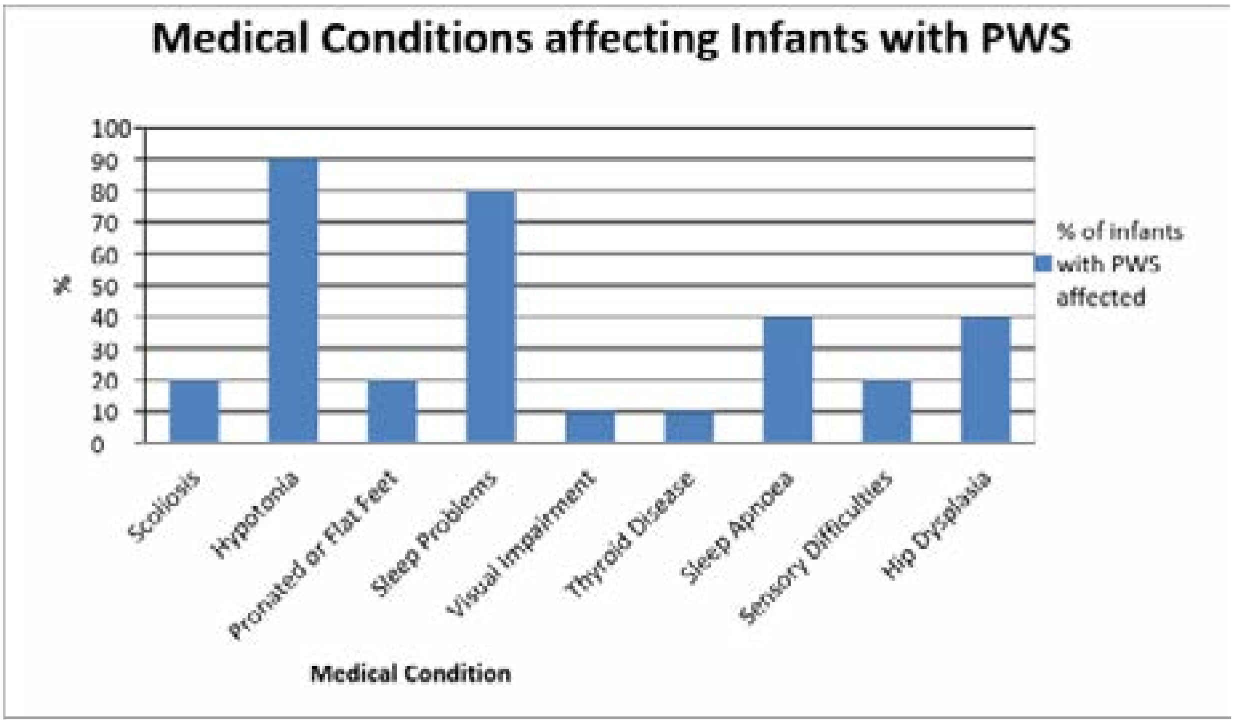


Figure 19*:* Graph illustrating medical conditions seen in infants with PWS

Medical Conditions ....continued

In the 5-12 group, new emerging conditions appeared such as scoliosis (50%), pronated or flat feet (44%), visual impairment (50%), and sensory difficulties (43.8%). Similar results were seen in the 13-17 group with the highest reported medical condition being sleep difficulties (90%) and an increase in mental health difficulties reported (23%). Forty per cent also reported a delay in pubertal development.

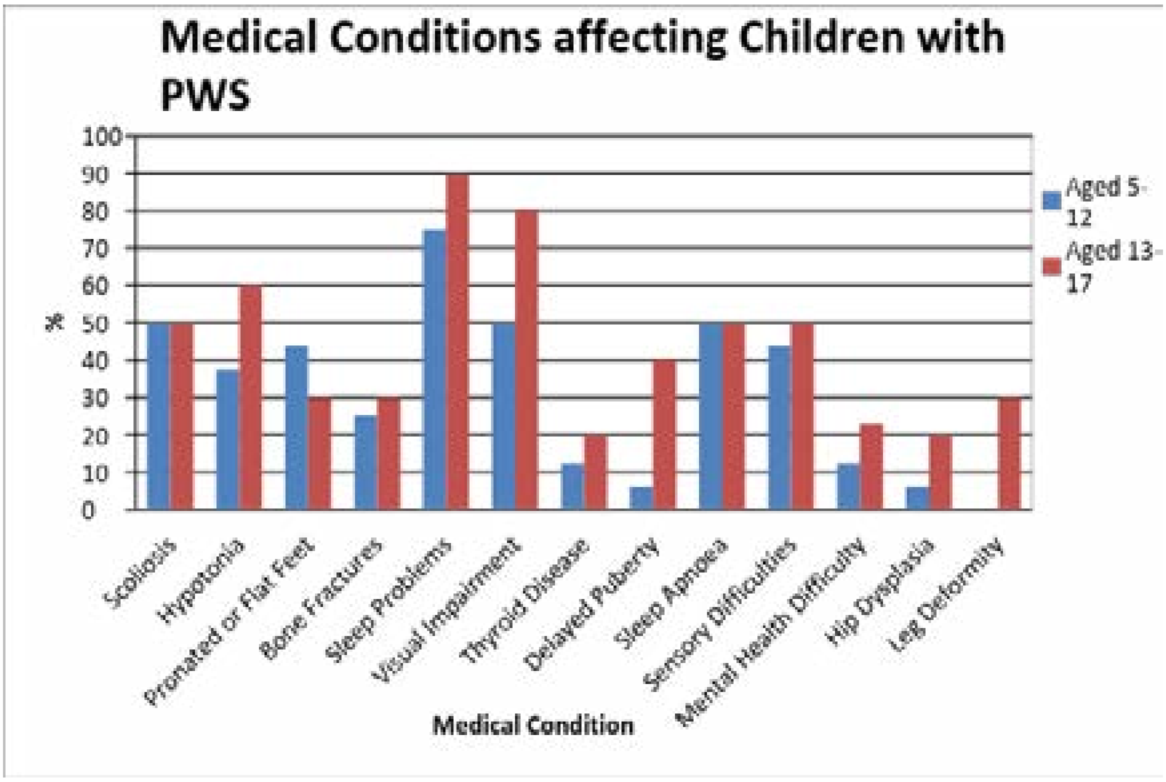


Figure 20: Graph illustrating medical conditions seen in children with PWS.

Medical Conditions ....continued

A drastic increase in mental health difficulties can be seen in the adult group with 50% of the adults living at home group and 78% of the adults living in supported accommodation group reporting a mental health difficulty. Emerging medical conditions in adulthood also included type 2 diabetes, which is a consequence of obesity (25% of adults living at home group and 56% of adults living in supported accommodation group affected); bone fractures (60% of adults living at home group and 56% of adults living in supported accommodation group affected) and osteoporosis (40% of the adults living in supported accommodation group affected).

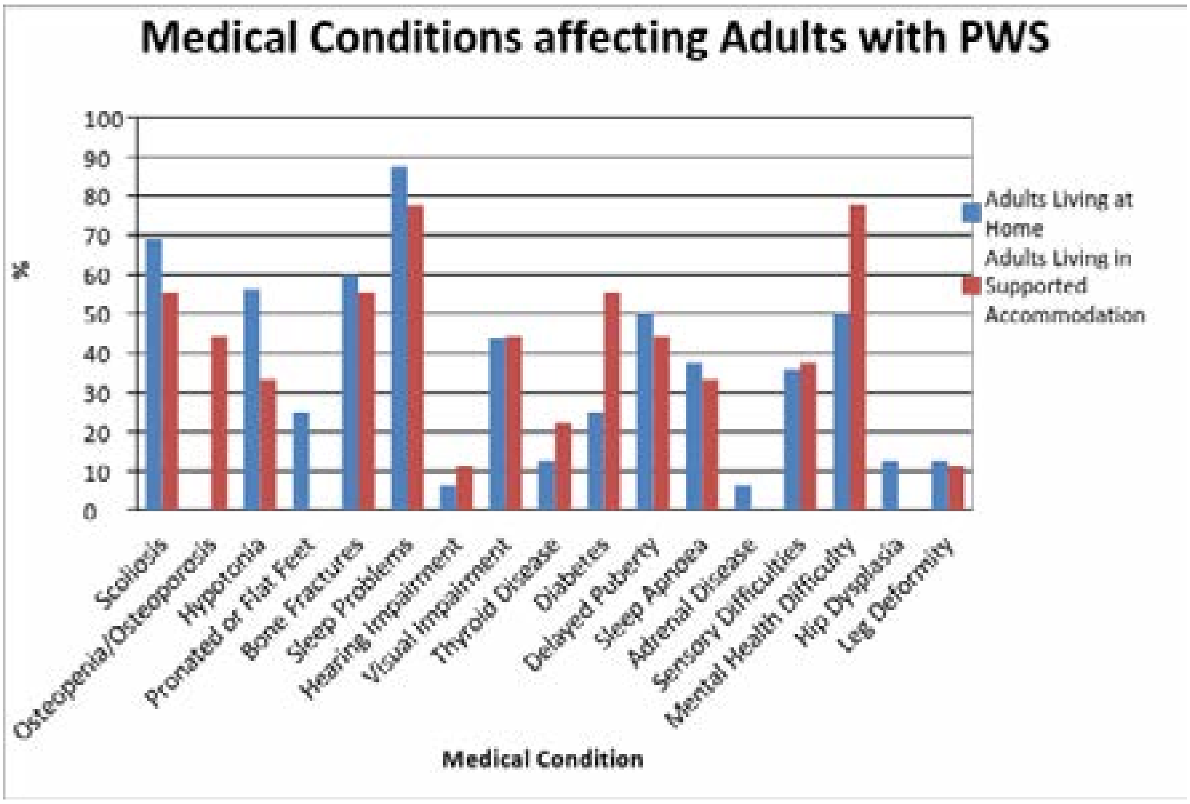


Figure 21: Graph illustrating medical conditions seen in the adults living at home group and the adults living in supported accommodation

Growth Hormone Treatment

Growth hormone treatment (GHT) refers to the use of growth hormone as a licensed indication in PWS. Growth hormone is a peptide hormone secreted by the pituitary gland that stimulates growth and cell reproduction. In addition to its effect on linear growth, growth hormone in PWS has numerous metabolic effects with a positive effect on body composition, promoting fat free mass and enhancing muscle strength and mobility. Human growth hormone may help with cognitive development (Myers et al., 2006).

The blue line in graph 22 below illustrates the percentage of participants who have received GHT. The red bar represents the percentage of people who received GHT but stated that they experienced a delay in starting the treatment. The green bar represents the percentage of people who received GHT but had to end treatment early for medical reasons. There is no set age to commence growth hormone therapy and the commencement of GHT is individualised as certain conditions may require stabilisation and treatment before GHT may be commenced. However, there is increasing consensus to support an earlier age at initiation of GH therapy where possible ideally prior to the onset of obesity which can occur as early as two years.

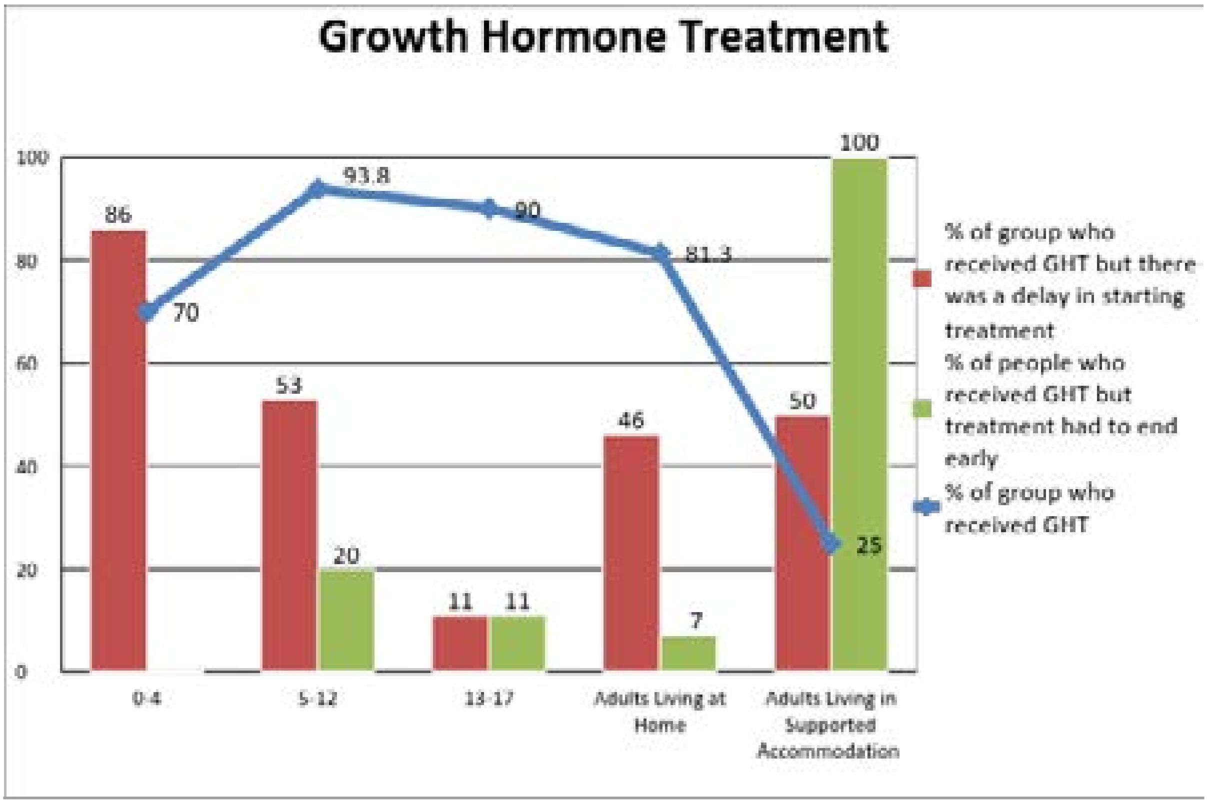


Figure 22: Line graph depicting the total percentage of participants who received GH treatment across groups. Bar chart representing the percentage of participants in each group who underwent treatment and had a reported delay in treatment or stopped treatment early

In the 0-4 group, 7 (70%) had received GHT; however, 6 of these 7 (86%) participants experienced a delay in beginning treatment. A delay in beginning treatment was seen across all groups. There were two main reasons for a delay in commencing GHT reported by participants. The first reported reason was a previous medical condition that had to be treated. The second reported reason was a delay in undergoing a sleep study assessment which is a prerequisite assessment for starting treatment. Apart from the 0-4 group, each group had participants who had to end treatment early. The average age at the start of GHT for the 0-4 group was 1.7 years. This increased to 3.06 years and 3.7 years in the 5-12 and 13-17 groups respectively. In the two adult groups, the average age of starting GHT was 9.7 years. It should be noted that GHT in PWS was first licensed for use in Ireland in 2001 which may explain the increase in average age for the adult groups (see table 3).

Table 3: Table displaying age of Growth Hormone Treatment commencement for each group

Beginning of Treatment	0-4	5-12	13-17	Adults Living at Home	Adults Living in Supported Accommodation
Minimum Age (years)	1	1.5	1	2	2
Maximum Age (years)	3	5	8	25	15
Average Age (years)	1.7	3.06	3.7	10.08	9.33

Body Mass Index

PWS is the most common genetic cause of extreme obesity in children. Access to food must be strictly supervised and limited, usually by installing locks on all food-storage places including refrigerators. BMI for children changes with age and should be plotted on growth charts. A BMI of 25kg/m2 is overweight and BMI >30 is obese. BMI median scores were reported in the normal range for the 0-4 group and 5-12 group and slightly higher in the 13-17 group according to World Health Organisation (2000) body mass index (BMI) categories. The median BMI scores for both adult groups were obese. However, the large standard deviation (SD) values indicate a wide variation of BMI scores within all age groups.

Table 4: Body Mass Index of Participants

Participant Group	0-4	5-12	13-17	Adults Living at Home	Adults Living in Supported Accommodation
BMI Median	16.6	17.9	23.2	31.3	33.0
BMI SD	7.4	7.6	6.2	20.7	16.7
BMI Min	15.8	13.4	17.6	19.2	23.9
BMI Max	33.4	39.7	32.8	84.1	56.3

Section 8

Mental Health and Challenging Behaviours

PWS has a well described behavioural and mental health phenotype. The most widely described characteristic is hyperphagia associated with an intense interest in food. In addition people with PWS may have obsessive traits, mood lability, temper tantrums, impulsivity, low activity levels, repetitive speech and weak social skills and adaptive behaviour. In addition people with PWS have an increased risk of mental health disorders which appear to be related to the underlying genetic anomaly. Individuals with maternal uniparental disomy are at increased risk of mental health conditions such as mood disorders, including bipolar illness and psychosis (Sinnema et al., 2011).

Mental Health Difficulties

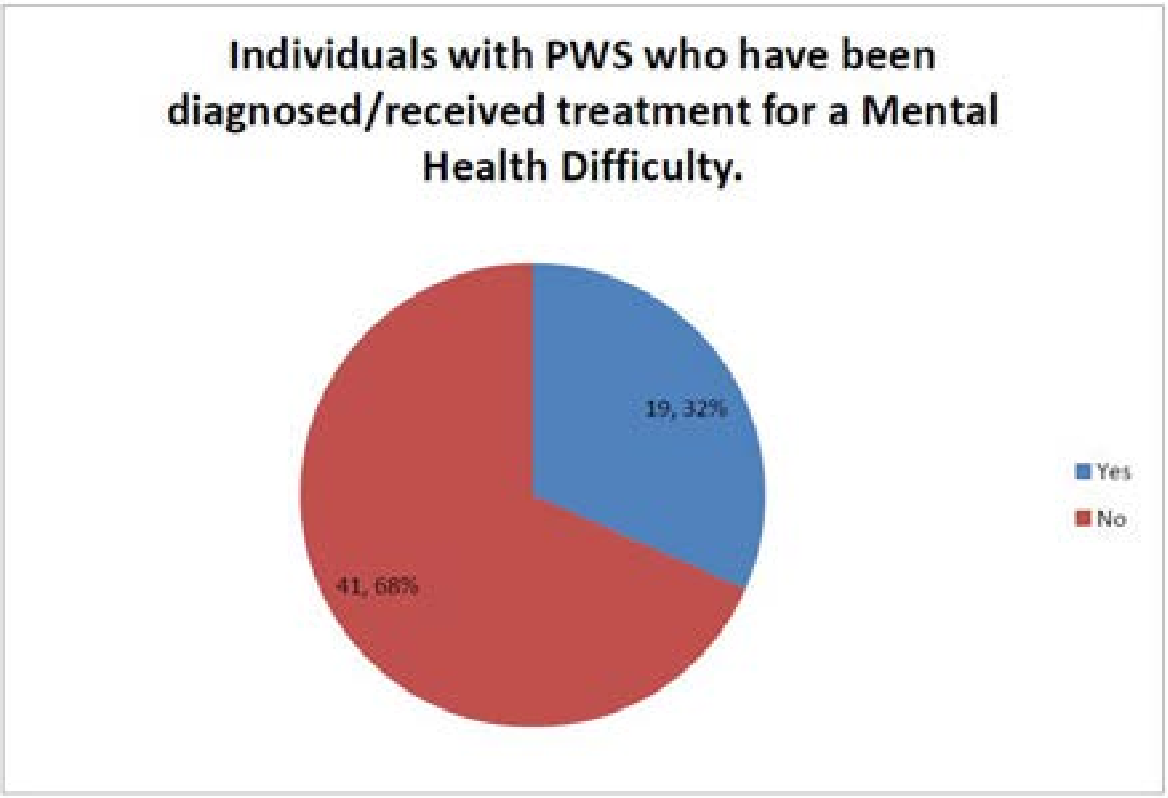


Figure 23: Pie chart illustrating the percentage of participants diagnosed with /receiving treatment for a mental health difficulty.

Sixty of the participants responded to the questions "Has the person you cared for ever been diagnosed with/or received treatment for a mental health difficulty?" (see figure 23). In total, 32% (n = 19) responded "yes".

Figure 24 below shows a breakdown of the percentage of participants diagnosed/ receiving treatment for a mental health difficulty by age group. The graph depicts an increase in the occurrence of mental health difficulties as individuals with PWS enter adolescence and adulthood. Thirteen per cent in the 8-12 group reported a diagnosis of a mental health difficulty which increased to 22% in the 13-17 age. Sixty per cent of adults were reported to be diagnosed with/receiving treatment for a mental health difficulty.

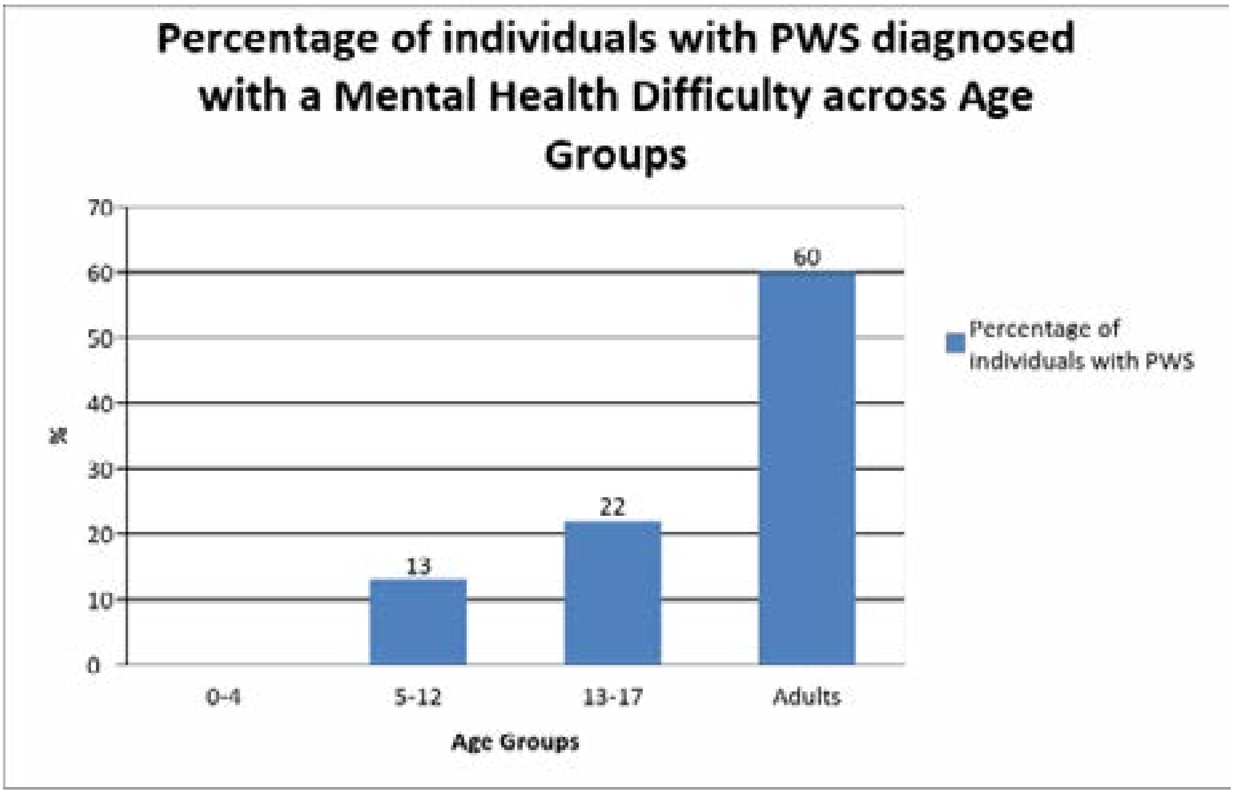


Figure 24: Figure illustrating the percentage of mental health diagnoses across age groups

Mental Health Difficulties ......continued

Figure 25 below shows a break-down of the specific type of mental health diagnosis reported. Of the adults who were diagnosed with a mental health difficulty, the most common disorders reported were anxiety (76%), depression (40%), psychosis (24%) and bipolar disorder (12%). Anxiety and Autism Spectrum Disorders (ASD) were also reported in both the 5-12 and 13-17 groups.

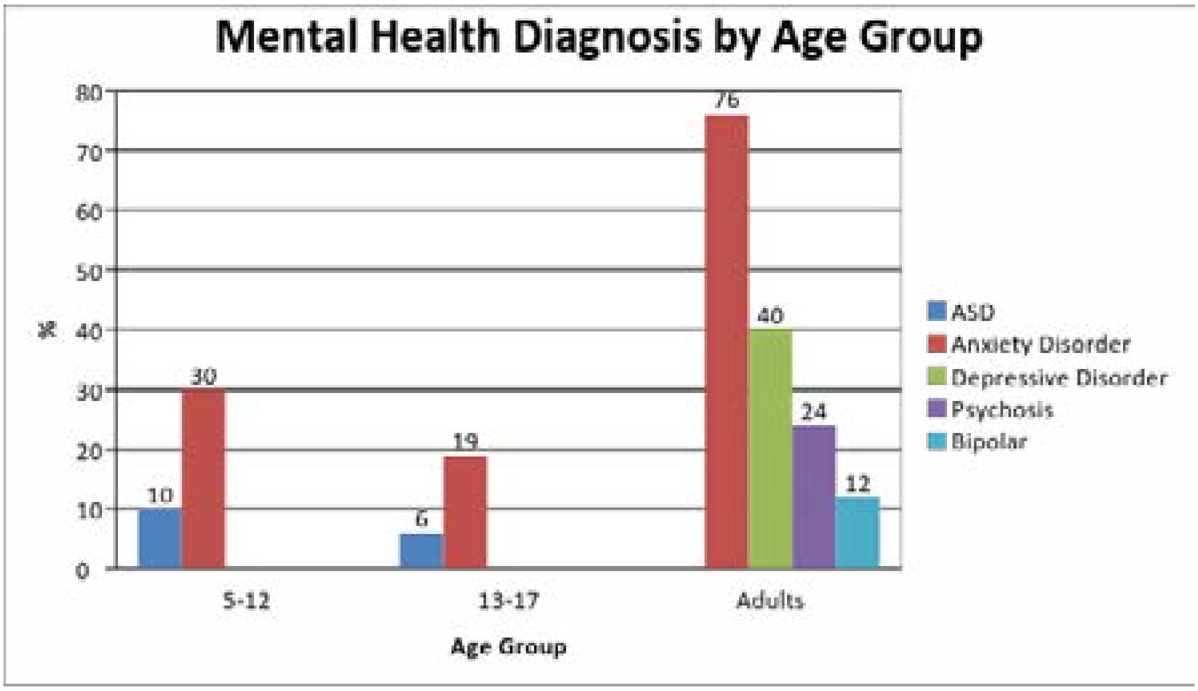


Figure 25: Graph illustrating the category of mental health diagnoses seen across age groups.

Challenging Behaviour

We adapted a behavioural questionnaire relevant to typical PWS related maladaptive behaviours. Parents were asked to rate on a scale of 0-3, with 0 representing "no problem", 1 representing "a mild problem", 2 representing "a moderate problem" and 3 representing "a severe problem" on a range of challenging behaviours associated with PWS. Figure 26 below represents a graph of the challenging behaviours most commonly reported as being mild, moderate or severe. Repetitive questioning, self-scratching, obsessions/compulsions and non-compliance were the most commonly reported challenging behaviours.

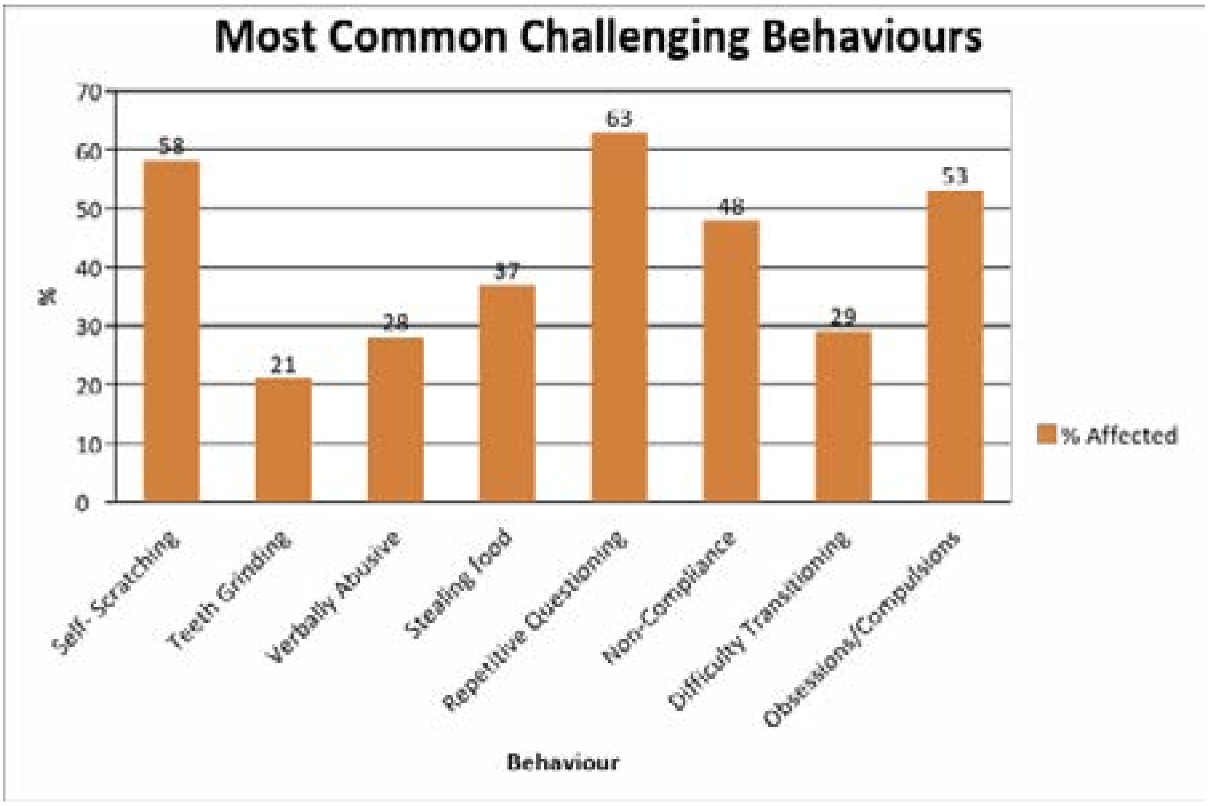


Figure 26: Graph illustrating the most common reported challenging behaviours

Challenging Behaviour......continued

Participants were also asked to rate on a scale of 1-7 the level of difficulty they felt in dealing with the challenging behaviours presented by the individual with PWS that they care for, with 1 representing "very difficult" and 7 representing "not very difficult at all". Of the 57 participants who answered this question, 12% reported a score of 1 "very difficult". Over 50% of the participants reported a score of 4 or less when rating the level of difficulty experienced when dealing with challenging behaviour.

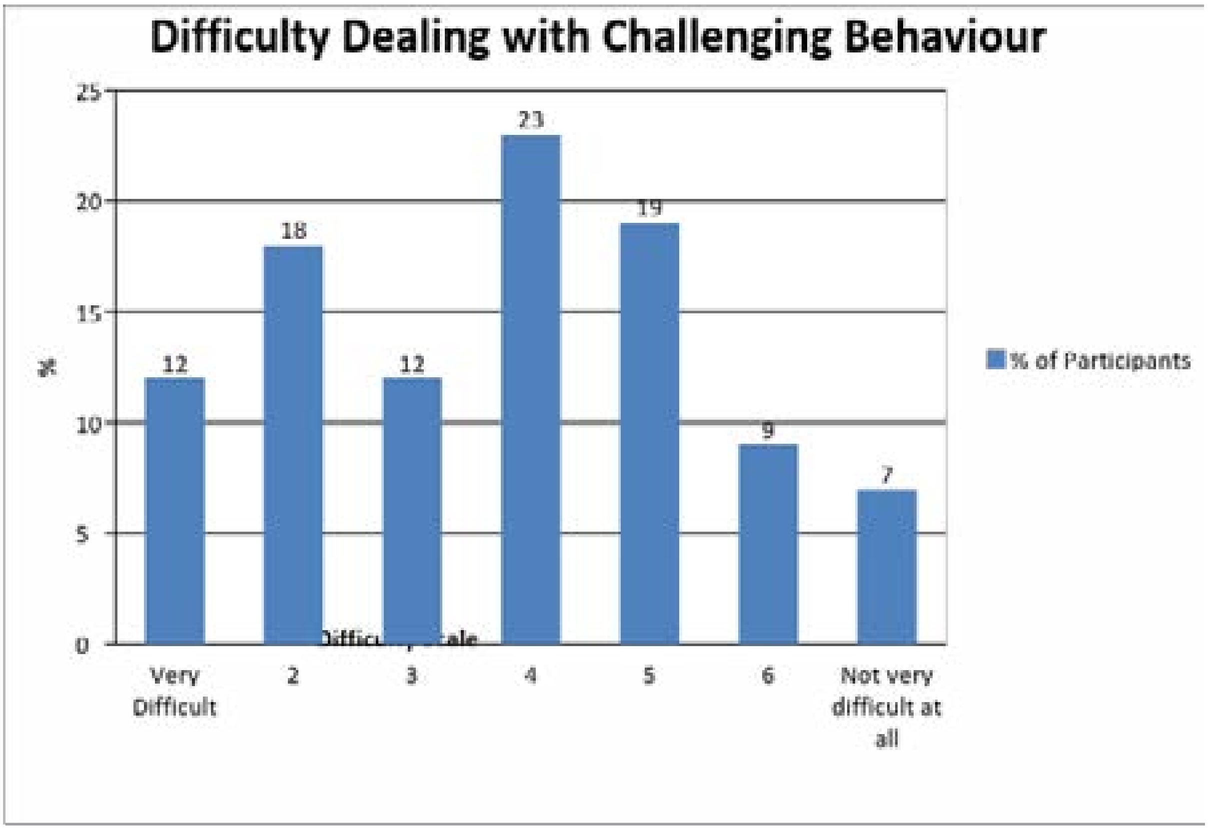


Figure 27: Graph illustrating the level of difficulty primary caregivers felt in dealing with the person with PWS's challenging behaviours.

Emotion Regulation

Participants were also asked about emotion regulation. Figure 28 highlights that across all groups help was needed in managing emotions and feelings. In particular, the most significant amount of help was required by the 5-12 and 13-17 age groups.

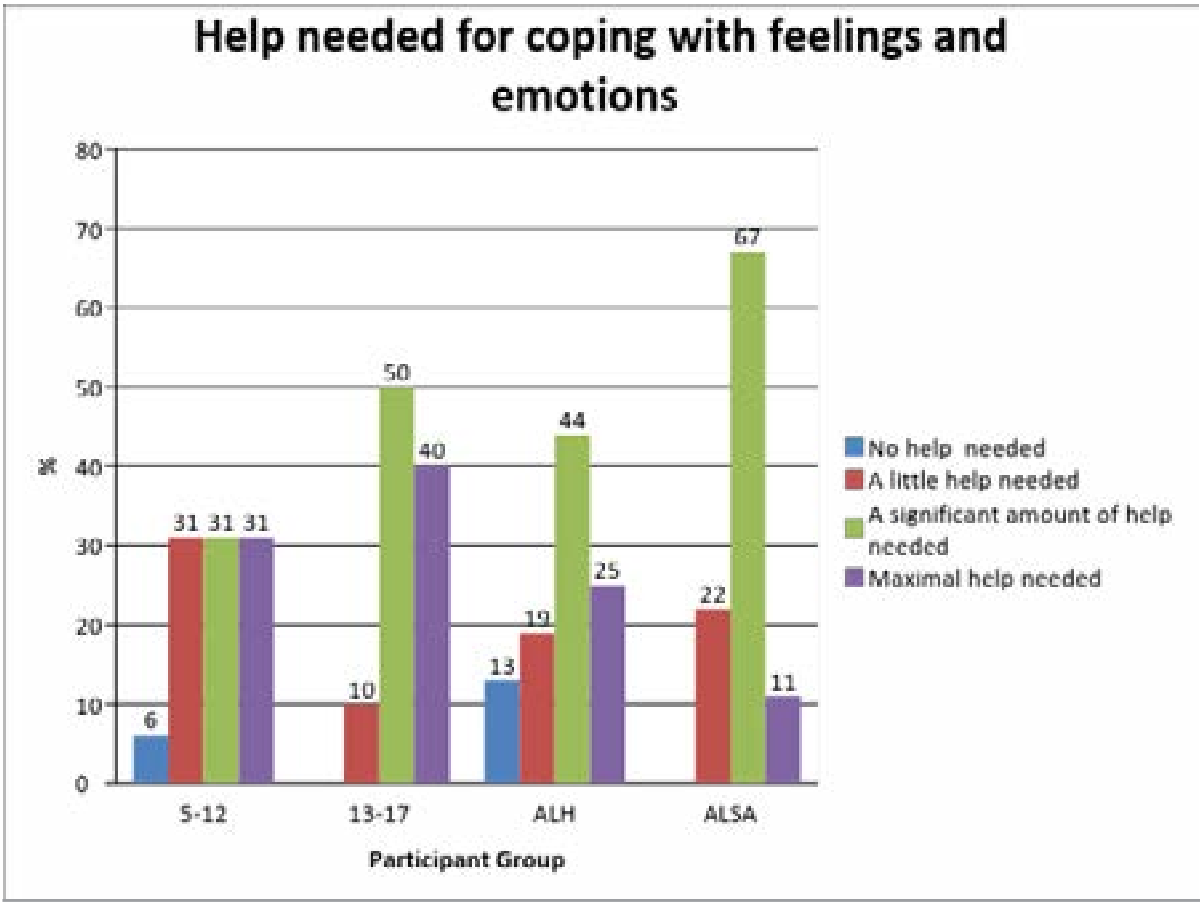


Figure 28: Graph depicting the amount of help needed by the individual with PWS in order to be able to cope with their emotions and feelings.

Section 9

Hyperphagia

Hyperphagia, as may be expected, was a significant challenge for the majority of participants. Over 80% of the 5-12 years and 13-17 years age-groups and the adults in supported accommodation had challenges in this area. 67% of adults living at home had hyperphagia (see figure 29).

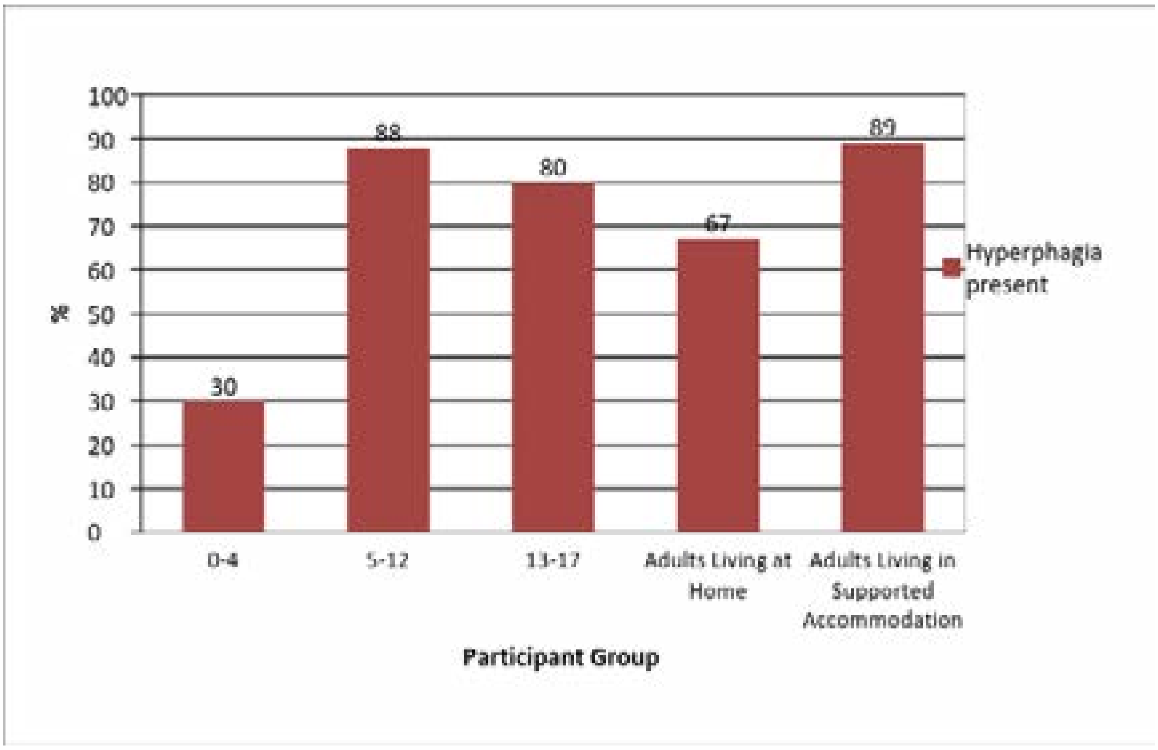


Figure 29: Graph depicting percentage of participants with hyperphagia across participant groups

Hyperphagia, which can result in gastric rupture and extreme obesity, is potentially life threatening and therefore is likely to precipitate individuals into care arrangements in adult life - possibly explaining the higher proportion of individuals with this challenge amongst those in residential care. This is also reflected by a high proportion in residential care reporting that this was of moderate severity (50%) compared with adults still residing at home (7%) (see figure 30).

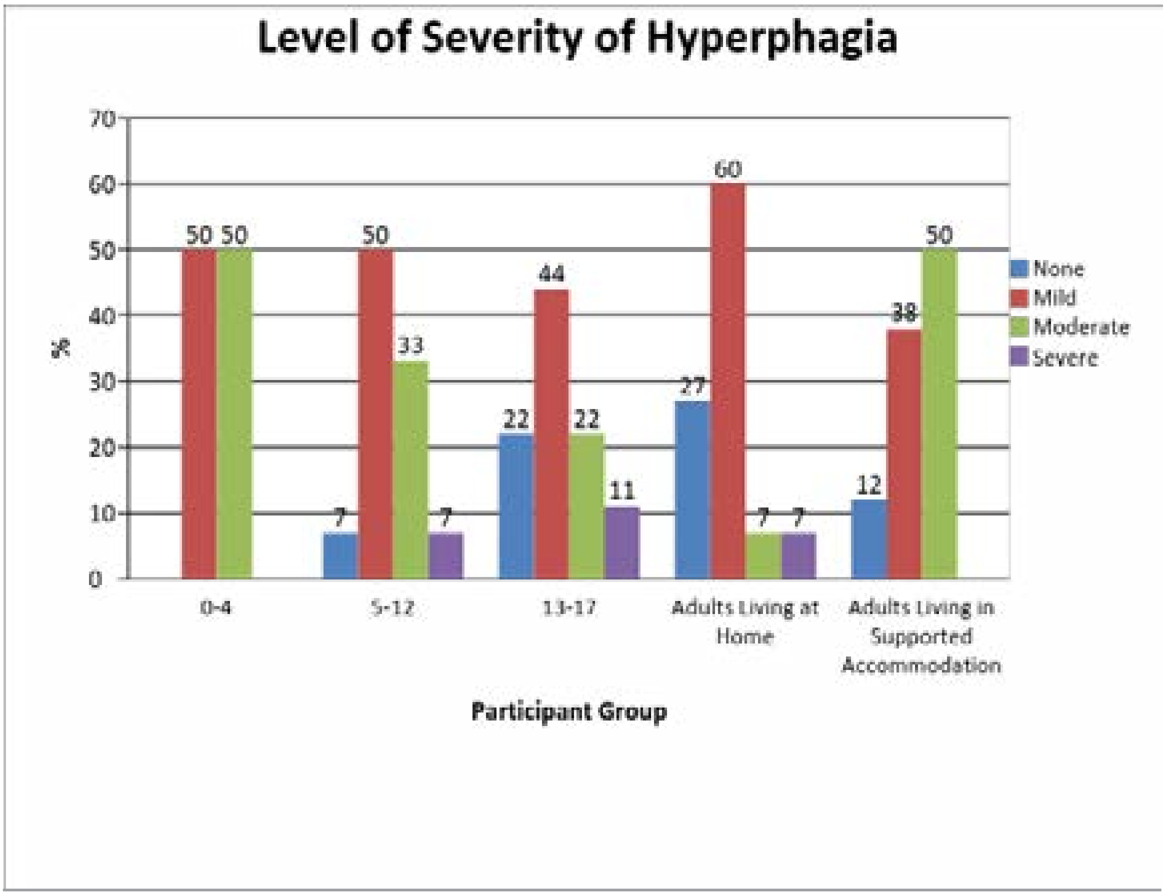


Figure 30: Graph depicting the reported severity of hyperphagia across participant groups

Section 10

Residential and Respite Needs

Residential Needs

Participants were asked about the long-term accommodation needs of the person with PWS for whom they care. For this particular analysis, responses were only included from the following groups; children aged 5-12, adolescents aged 13-17 and Adults Living at Home. Primary caregivers whose child with PWS was under the age of four and primary caregivers of adults with PWS who already were living in assisted long-term accommodation were not asked this question. The results show that according to participants, two people with PWS will require long-term accommodation within 2 years and an additional 14 will require long-term accommodation within the next five years. Of note only three primary caregivers reported not requiring long-term accommodation while 15 primary caregivers said they were unsure (see figure 31).

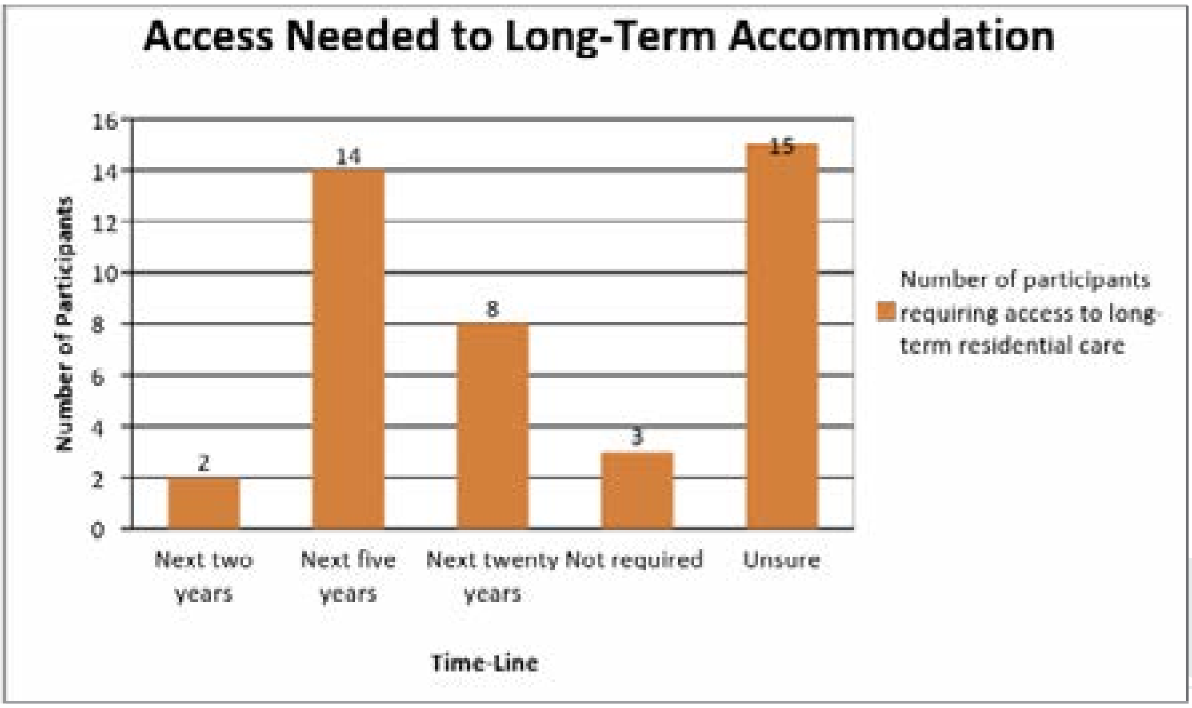


Figure 31: Bar chart depicting the timeline of long-term accommodation needs for individuals with PWS

Participants were also asked what type of accommodation they would prefer for their relative with PWS. The majority (59%) of the responses indicated that a PWS specific type group home was the preferred type of long-term accommodation (see figure 32).

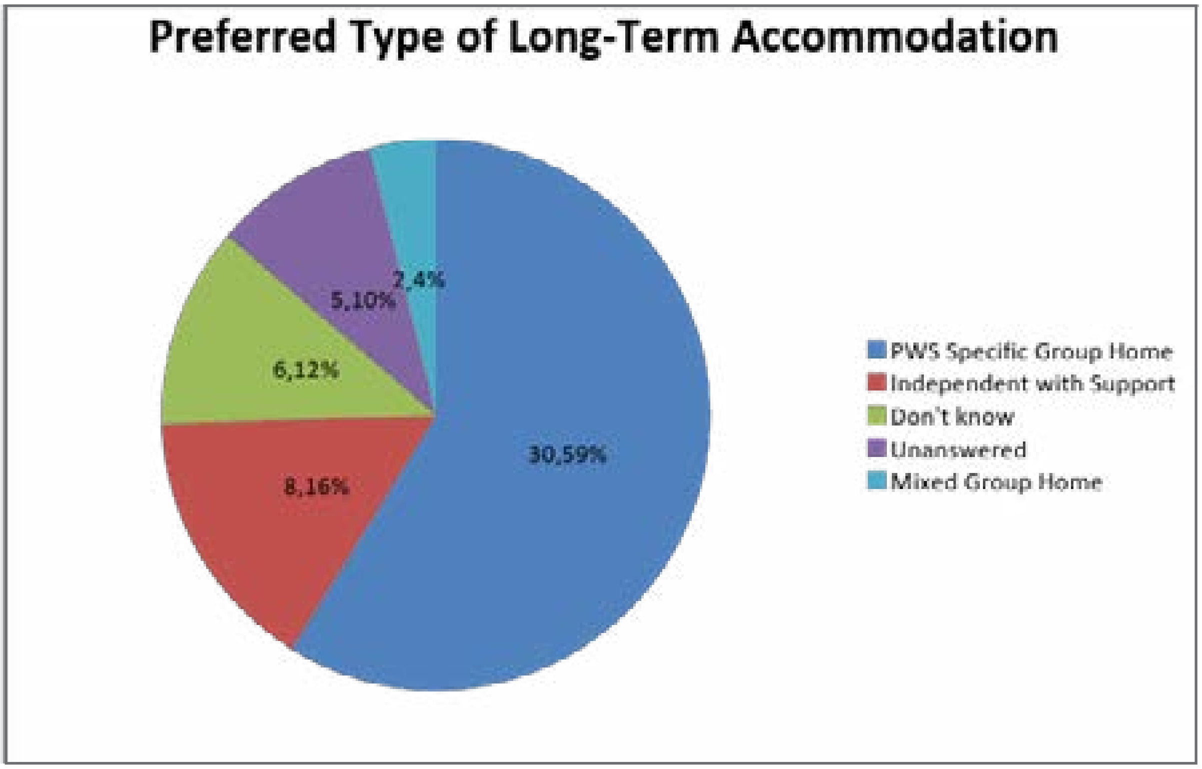


Figure 32: Pie chart showing primary caregivers' preferred type of accommodation for the individual with PWS that they care for.

Respite

Figure 33 below shows a pie chart for each participant group's response to the question "Does the individual with PWS that you care for have access to respite?". Similarly to the residential access analyses, the 0-4 age group and the 'Adult Living in Supported Accommodation' groups were excluded. The results showed that only 6% of children with PWS between the ages of 5 and 12 have access to respite. There was an increase in the adolescent group with 40% having access to respite. However this percentage decreased again with only 25% of adults with PWS who live at home currently having access to any type of respite.

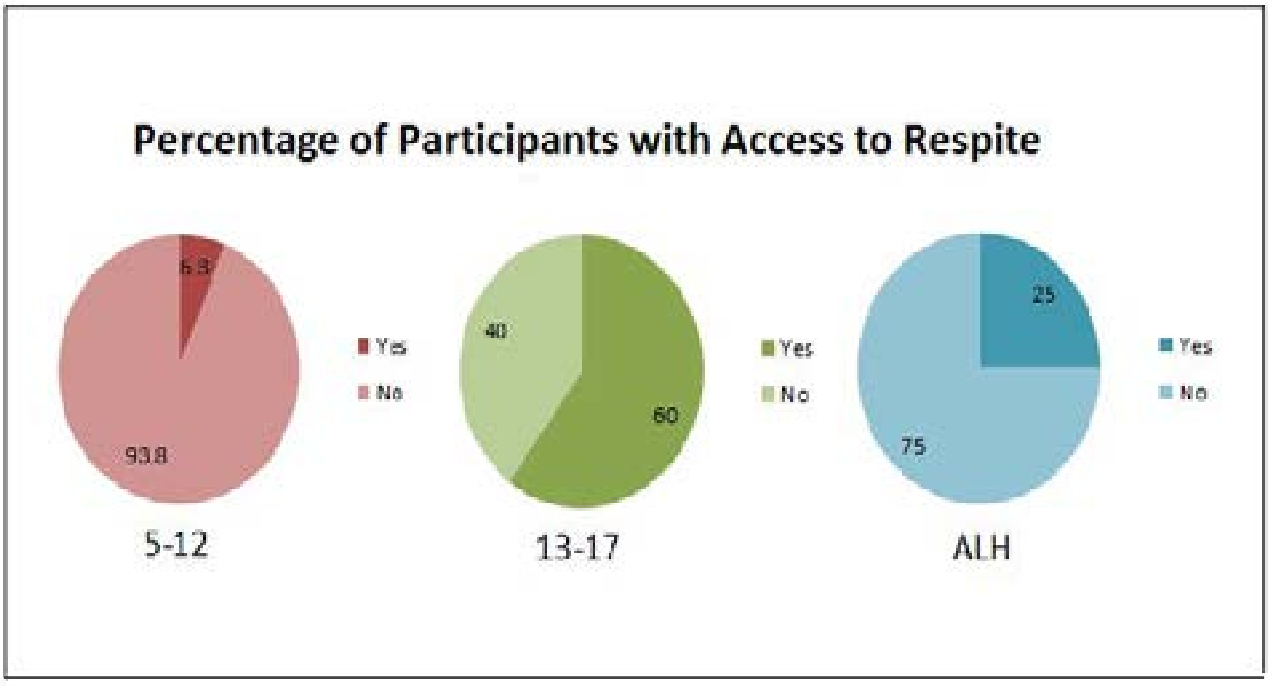


Figure 33: Pie charts showing percentage of each group category that currently have access respite

In order to assess which groups needed respite, participants were also asked if they currently felt they needed access to day respite and night respite. The majority of all groups reported that they needed access to both day and night respite. There was not a significant difference between each group suggesting that the burden of caring for an individual with PWS presents its own significant challenges and need for respite for caregivers at all life stages (see figure 34).

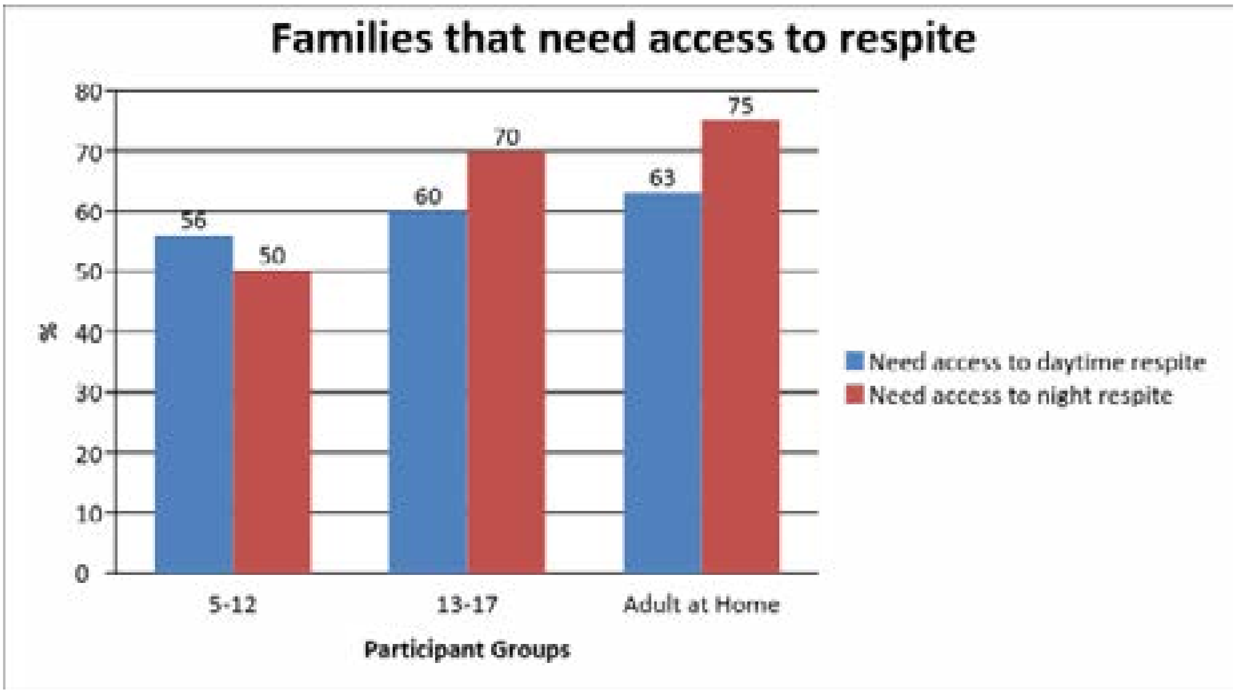


Figure 34: Graph illustrating percentage of families that currently require access to respite

Section 11

Financial Impact

As caring for an individual with PWS is associated with numerous costs (home modifications, equipment, medication, travelling to appointments, orthotics, specialised prams) and a significant financial burden, participants were asked about the financial impact as well as the financial supports available to them. In terms of perceived financial impact, there was a notable increase between groups in the percentage of participants reporting "an extreme negative impact" with 10% in the 0-4 group, 13% in the 5-12 group, 20% in the 13-27 age group and 29% in the adults living at home group suggesting an increase in financial burden as the individual with PWS ages. This reduced back to 25% in the adults living in supported accommodation group (see figure 35).

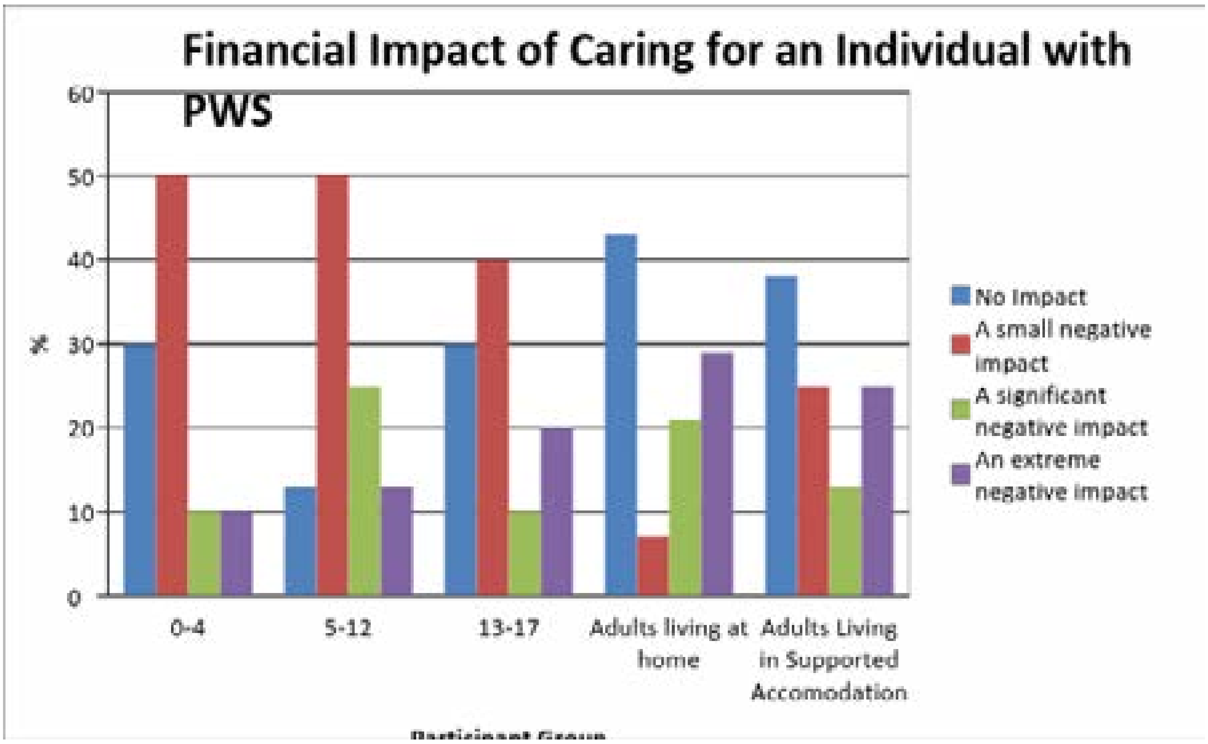


Figure 35: Graph illustrating financial impact of caring for an individual with PWS across groups

Participants were also asked if they or their partner ever had to reduce their working hours in order to care for the individual with PWS. Over 40% of respondents in the 0-4, 5-12, 13-17 and adults living at home group reported having to give up work in order to care for the individual with PWS. In addition, over 30% of respondents in the 0-4, 5-12 and 13-17 group reported having to reduce their working hours. Across all age groups, respondents reported that they or their partner had given up work to become carers for their relative with PWS. Taken together this ranged from 30-70% of individuals across all age groups. It was lowest in the adults in supported accommodation group, likely reflecting that this group were on average significantly older and for this generation of parents it may be more likely that mothers were not working outside the family home.

Only 13% of carers of those in the 5-12 group, 6% of the adults living at home group and 22% of the adults living in supported accommodation group reported that caring for an individual with PWS had no impact on their working hours (see figure 36).

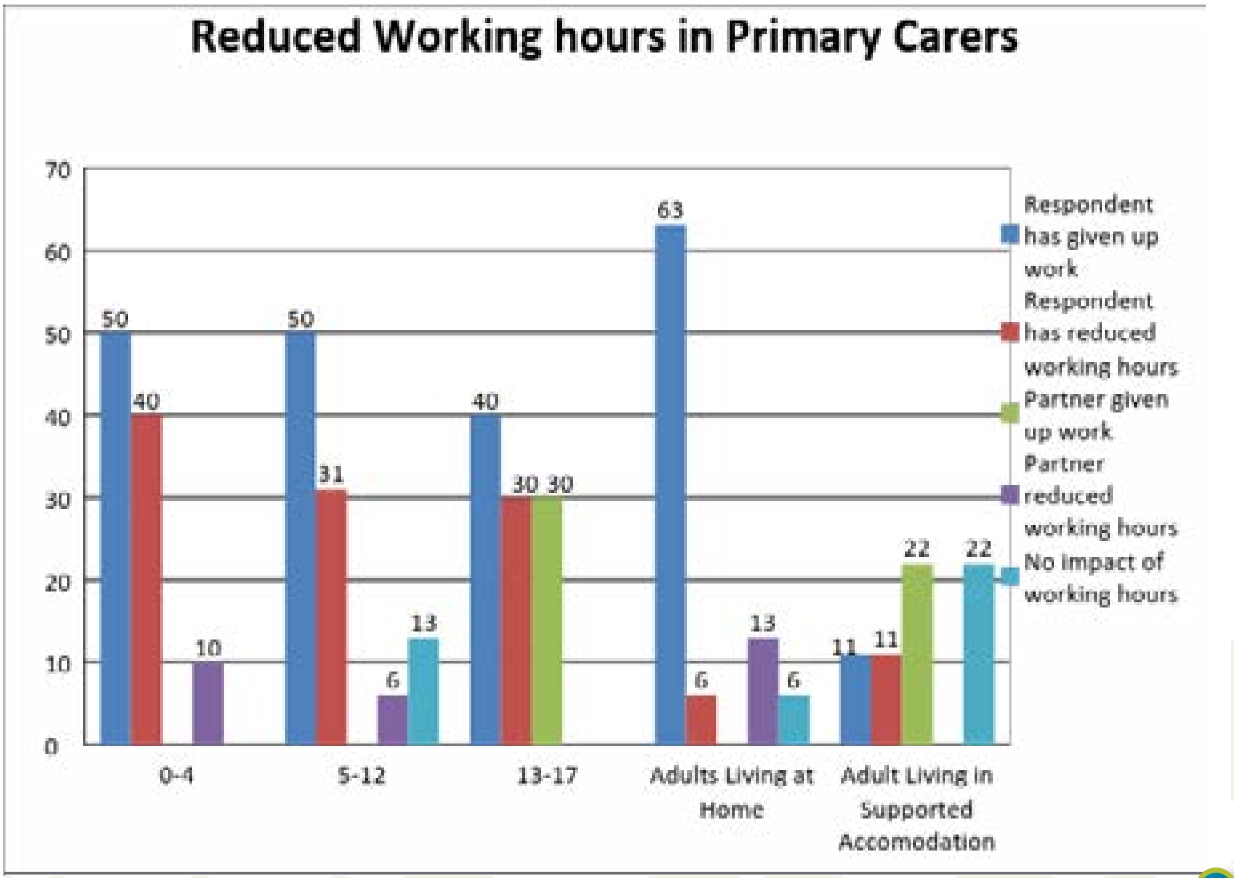


Figure 36: Graph illustrating the impact of caring for an individual with PWS on carer's employment

Participants were also asked about access to financial supports. Despite the chronicity and high medical need associated with PWS a significant proportion of individuals had no access to a medical card (50% of 5-12 year olds, 50% of 13-17 year olds and 12% of adults living at home). In order to determine a better understanding of the level of difficulty in obtaining a medical card, participants were asked if they had ever been refused a medical card. Sixty-nine per cent of children aged 5-12, 44% of children aged 13-17, 27% of adults living at home and 13% of adults living in supported accommodation had been previously refused access to medical cards (see table 5).

Table 5: Summary of financial supports currently being received by individuals with PWS

	5-12	13-17	Adults Living at Home	Adults Living in Supported Accommodation
Domiciliary Care Allowance	94%	70%	n/a	n/a
Received Medical Card	50%	50%	88%	100%
Refused Medical Card before	69%	44%	27%	13%
Long-term Illness Scheme due to	39%	30%	44%	67%
Mental Illness	13%	0%	0%	0%
Intellectual Disability	13%	20%	38%	67%
Other	13%	10%	6%	0%
Carers Allowance	25%	50%	63%	0%
Carers Benefit	0%	3%	0%	0%

Section 12

Impact on Family

Managing symptoms of PWS often requires modifications in the entire family's daily routine, often including round-the-clock supervision, locking of food cabinets and refrigerators, monitoring and regulating food intake, accommodating cognitive delays and behavioural problems, and appropriately responding to, managing and coping with emotional outbursts. Given that PWS is associated with very high care needs we asked respondents to share their experiences of the impact of PWS on a number of areas of family life: emotional toll of caring; physical toll of caring; impact on family relationships and impact on siblings.

Respondents reported that there was a significant emotional toll on the family related to caring for an individual with PWS. Taking responses that included 'significant' or 'extreme' negative impact together, 60-70% of respondents endorsed these responses. The emotional toll was lower in the group of individuals living in supported accommodation, with around 40% reporting a 'significant' or 'extreme' negative impact (see figure 37).

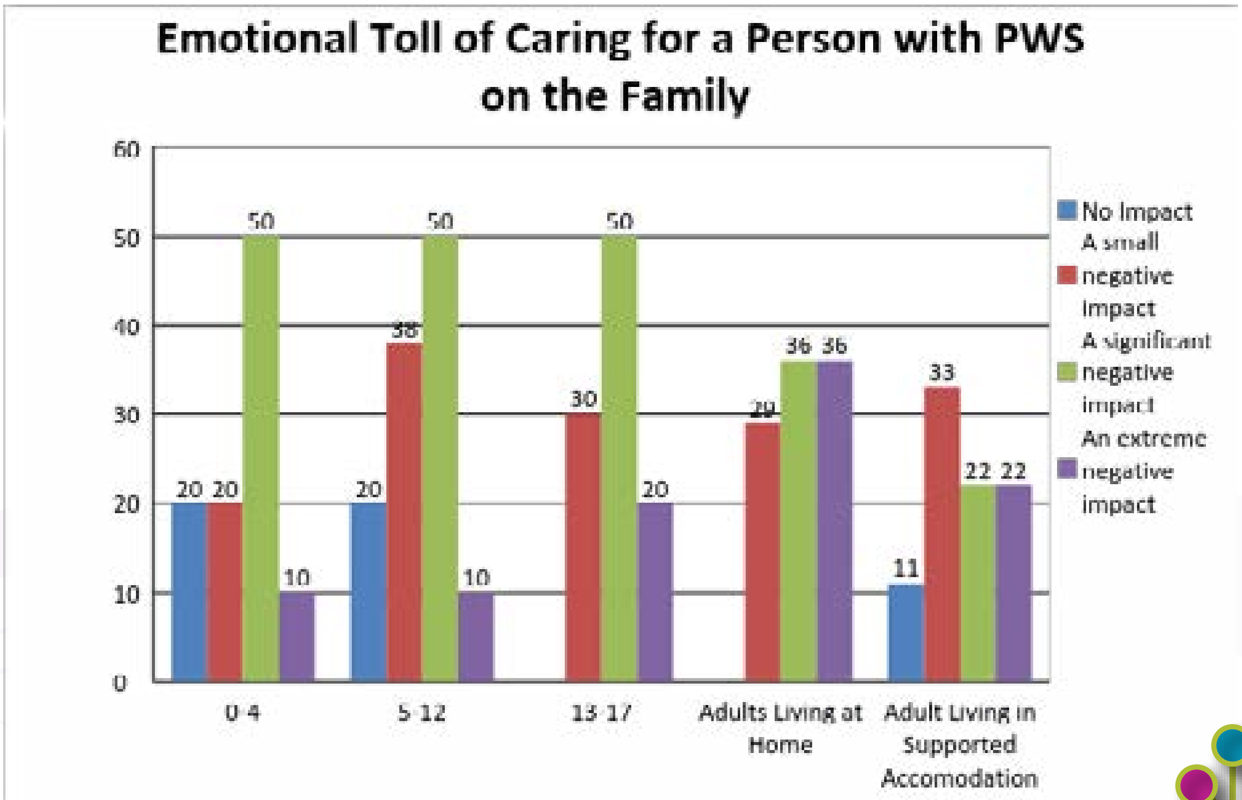


Figure 37: Emotional toll of caring for a person with PWS on the family

Physical demands relating to PWS may be associated with assistance with personal care or aggression towards family. This was most marked in the teen years where 60% of respondents indicated a 'significant' negative impact of caring for an individual with PWS (see figure 38).

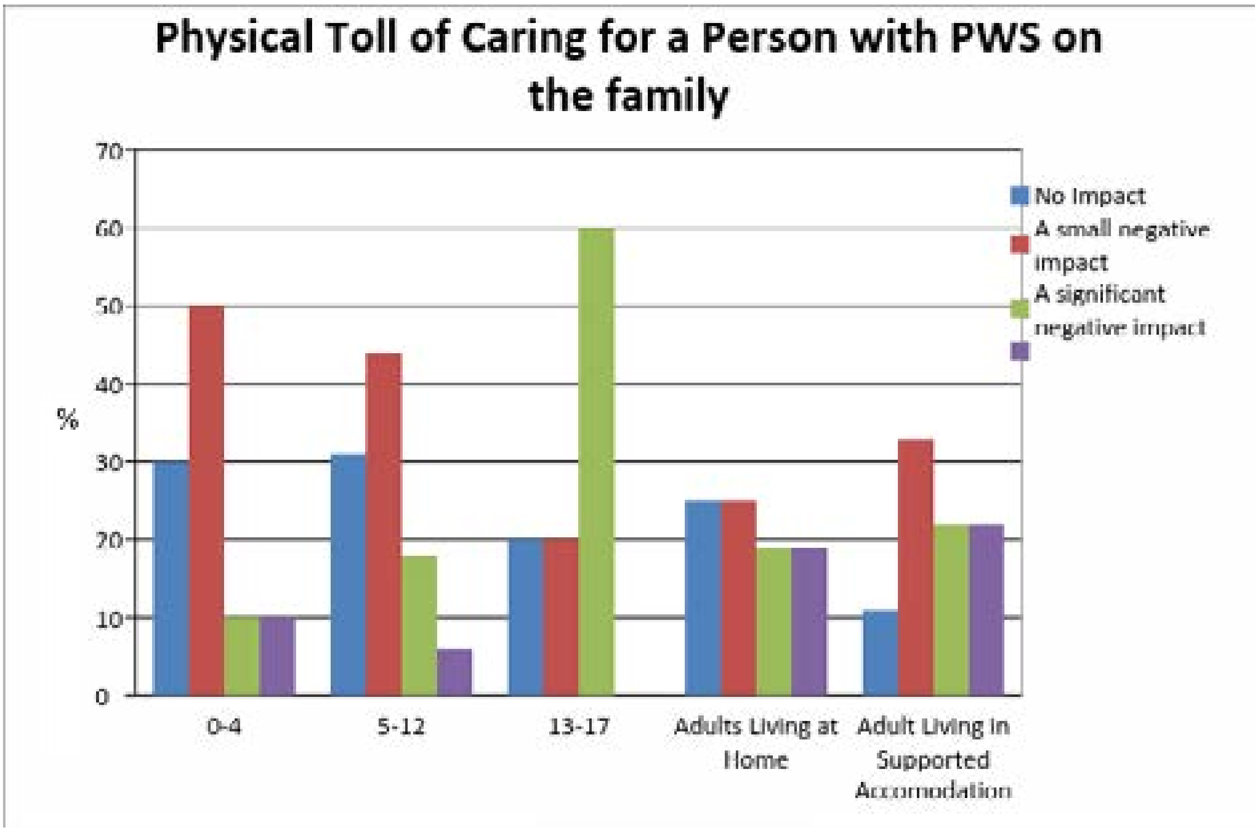


Figure 38*:* Physical toll of caring for a person with PWS on the family

All groups reported that caring for an individual with PWS had an "extreme" negative impact on family relationships. Once again, the 13-17 groups was the most severely affected with over 60% reporting either a "significant" or "extreme" negative impact. Over 40% of the 8-12 group and adults living at home group reported similar results. The lowest levels of impact on family relationships was reported in the adults living in supported accommodation group suggesting that when the individual with PWS moves out of the family home, the burden of care is reduced and family relationships improve (see figure 39).

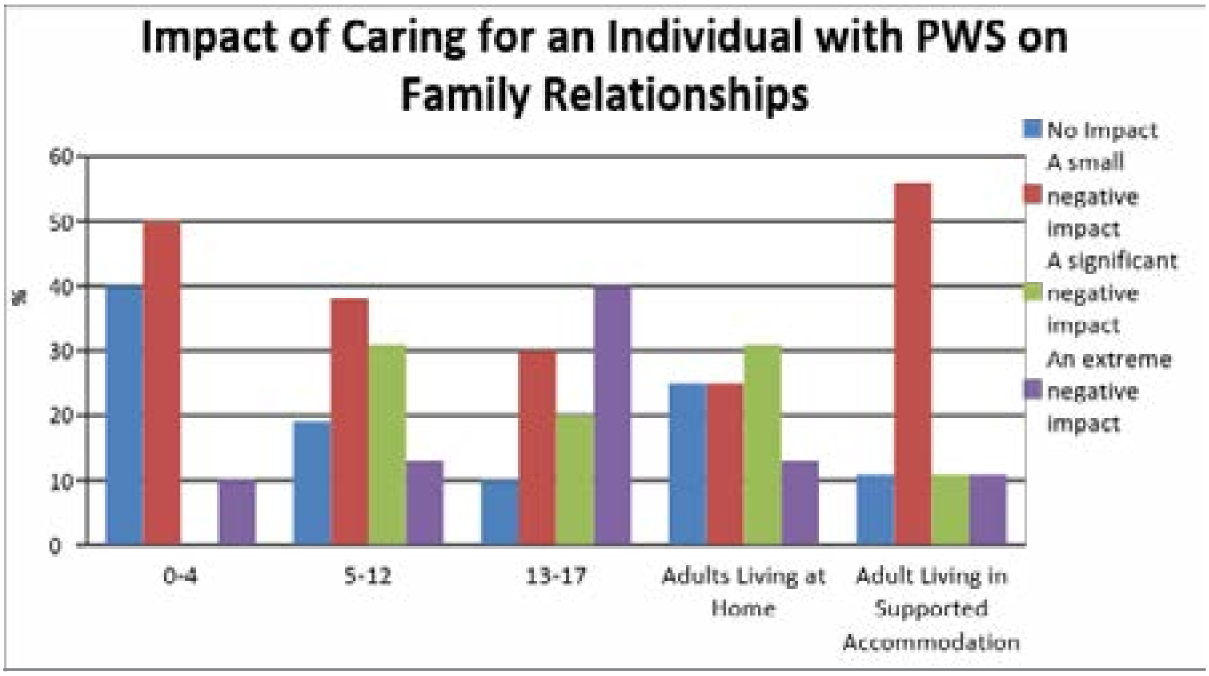


Figure 39*:* Impact of caring for a person with PWS on family relationships

Impact on Siblings

The multi-dimensional manifestations of PWS can significantly and adversely impact on unaffected siblings of an individual with PWS. Healthy children with a chronically ill sibling are at an increased risk of developing emotional and behavioural problems. In order to ascertain the impact on siblings we asked parents how they felt having a sibling with PWS affected their other children's mental health and social life. Having a sibling with PWS under the age of four did not appear to have any significant impact on sibling's mental health. There was an increase in the 5-12 group which reported that 15% of parents felt that having a sibling with PWS had a "significant" negative impact on their child's mental health. For the 13—17 group, 25% of parents reported either a "significant" or "extreme" negative impact on their unaffected child's mental health. Similarly for the adults living at home group, 25% of parents reported that having a sibling with PWS had an "extreme" negative impact on their other children's mental health (see figure 40).

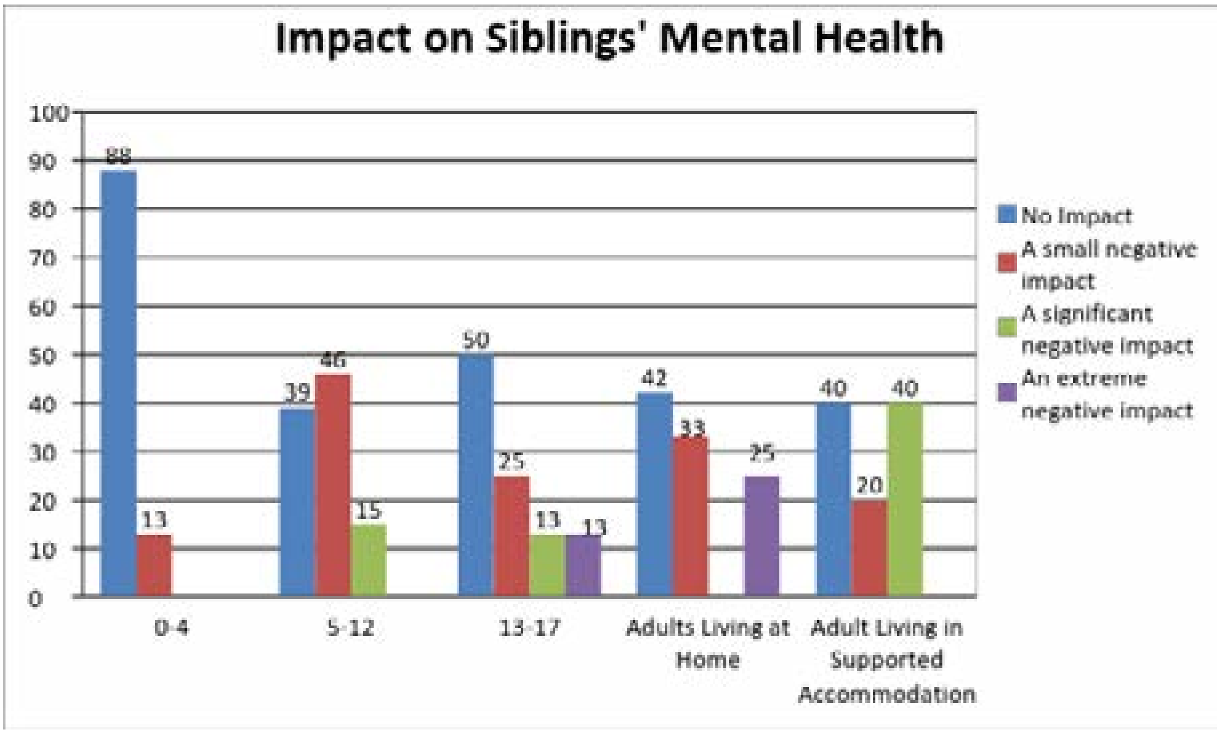


Figure 40: Impact of caring for a person with PWS on siblings' mental health

In terms of impact on siblings' social life, the 5-12 group appeared to be the most affected with only 8% of parents reporting that having a sibling with PWS had "no impact" on their child's social life. Over 50% of the 13-17 group reported that having a sibling with PWS had a "significant" or "extreme" negative impact on their other children's social life (see figure 41).

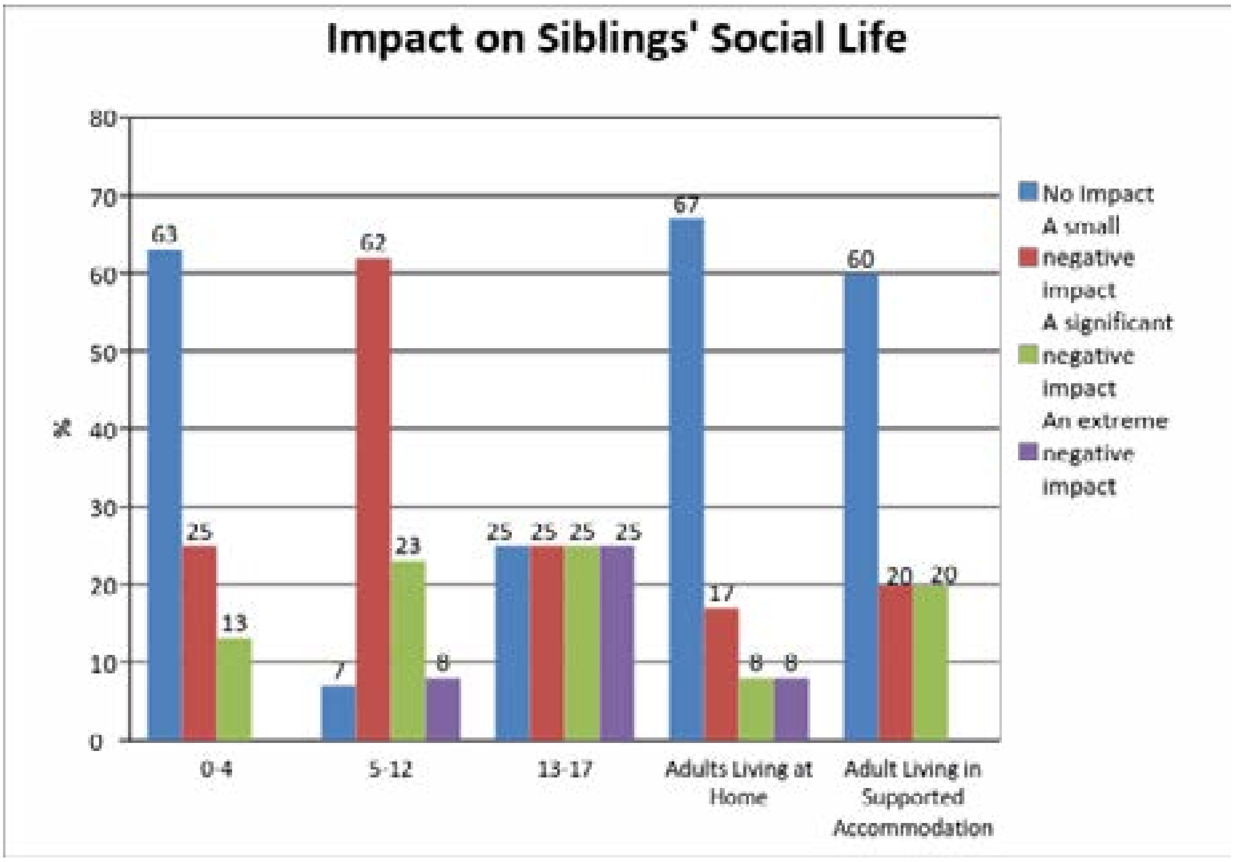
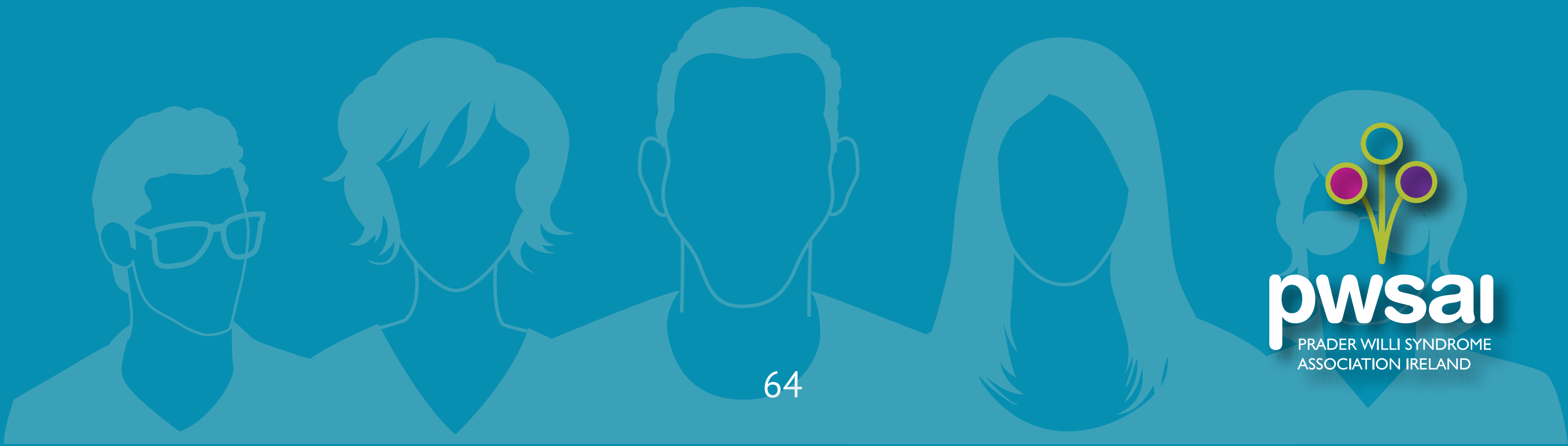


Figure 41: Impact of caring for a person with PWS on siblings' social life



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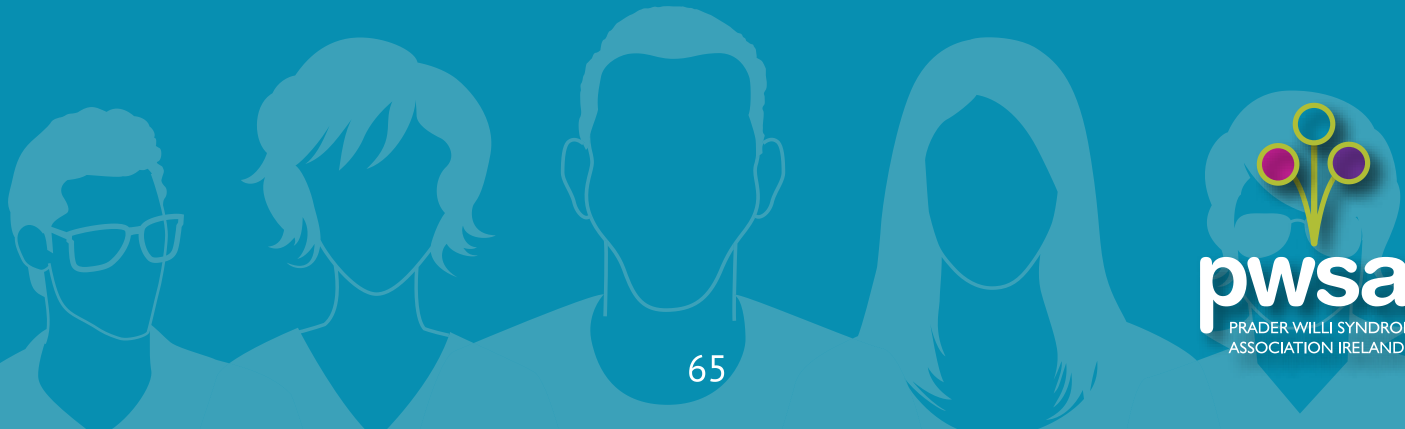
ASSOCIATION IRELAND

Discussion and Key Findings

This report is the product of an extensive survey of the needs of people with PWS living in Ireland today. PWS is a rare syndrome affecting approximately 100 individuals in Ireland. Sixty-one respondents completed the survey on behalf of their relative with PWS representing over 60% of the total population of individuals with PWS in Ireland. These data are highly representative therefore of the community of people in Ireland impacted by PWS, both individuals and families, and may be used to inform the development of improved service provisions to aid in the management of this highly complex condition.

The research conducted here was questionnaire based and completed on behalf of individuals with PWS by their caregivers. The majority of respondents were mothers of people with PWS (82%). The questionnaires were distributed through the PWSAI and the PWS specialist endocrinology clinic at Tallaght hospital. All questionnaire data that was returned was included in this report. We don't have information on those individuals who did not respond to the initial mailshot; therefore it is not possible to comment on the needs of non-responders, i.e. whether they represent a group of individuals who are more or less impacted by the condition.

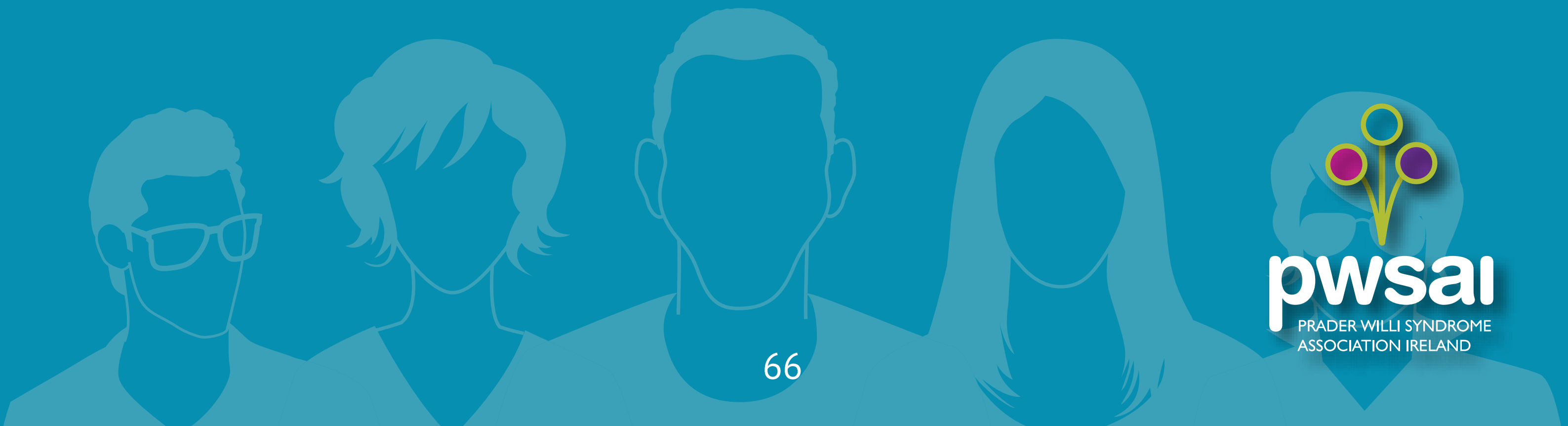
Individuals with PWS across a broad age range were included here (6 months-52 years) and both males (43%) and females (57%) were well represented. The genetic anomalies underlying PWS, i.e. paternal deletions (42.6%), uniparental disomy (26.2%) and imprinting defects (3.3%) were all represented. This is important as the anomalies have been associated with differing medical, behavioural and psychiatric needs. The participants were



widely geographically distributed across all four provinces; the majority as might be expected resided in areas of high population density, i.e. Leinster. The survey focused on the most important areas of need for people with PWS and for their family members.

These included:
- Early life and development.
- Physical health.
- Mental health and behaviour.
- Education and employment.
- Residential and respite support needs.
- Impact on families.

A summary of the findings in each of these areas is presented below together with recommendations for each of these areas. Recommendations are framed around the areas of need that can be immediately addressed through service provision and those that require further research.



Early Life and Development

The signs and symptoms of PWS are very apparent when infants are born. Infants frequently have an obvious physical appearance, low tone and feeding difficulties. As a consequence intensive medical care is often required. This was reflected in the responses to this survey; participants spent on average 26 days in the neonatal intensive care unit and a high proportion needed nasogastric feeding (57%). The introduction of clinical diagnostic criteria (Holm et al, 1993) and genetic testing has led to reductions in the length of time required to establish a diagnosis. The average length of time for a diagnosis to be established reported here was between 0-4 weeks, whereas adults born at the start of the millennium waited on average up to 19 weeks for a diagnosis. This improvement is far more favourable and supports timely initiation of medical therapies with the potential to reduce morbidity over the lifetime of the individual.

PWS is typically associated with developmental delay and intellectual disability. A large number of the individuals included in this research had a mild intellectual disability (65%) and a smaller number had moderate intellectual disability (26%). Around 9% of individuals had borderline intellectual disability or were in the average range. These developmental and cognitive needs are associated with increased care, educational, occupational and support needs across the life-course and suggest that planning for these is essential for the individual with PWS. Previous research has also shown that IQ is not a good indicator of adaptive functioning in PWS, i.e. that the individuals ability to manage themselves and the demands of daily living is lower than expected. Significantly increased needs in all areas of self-care were required across all age groups. In adult life for example, over 40% of individuals needed help with toileting, showering and tooth brushing and over 50% needed help with dressing. These rates were even higher in the teen years (>60%). A significant proportion of adults, almost 90%, required help in managing their own health and finances. These needs highlight that achieving independence as an adult is severely restricted for people with PWS and lifelong supports are required.

However the gap between measured ability and adaptive function also raises questions regarding how best to deliver services for people with PWS. The structure of service provision for people with disabilities in Ireland may pose some challenges in this regard. Typically intellectual disability services are delivered to people with moderate, severe and profound intellectual disabilities and would thus preclude a high number of people with PWS who may have need for such services since adaptive function appears significantly lower than expected. Progressing Disabilities Services (PDS) is the National Program for disability services in Ireland which was established to address inequities within disability service provision nationally. However currently PDS is not being implemented for adults and there is as yet no clarity regarding the pathway of care for people with PWS as they move into adult life. This is an issue that needs to be urgently addressed. The course of PWS is well-established and the need for services provision can be anticipated and planned based on the known needs of individuals with PWS and their demographic profile.

Main findings:

- The majority of people with PWS have significantly delayed adaptive functioning relative to their measured ability.

Recommendations:

- Service provisions need to be planned for people with PWS across their life-course. Key transitions should be anticipated and planned, e.g. at preschool, commencing primary school, transitioning to secondary school, transitioning to adult services. The adaptive needs of people with PWS need to be considered in addition to measured IQ when determining appropriate service provision.

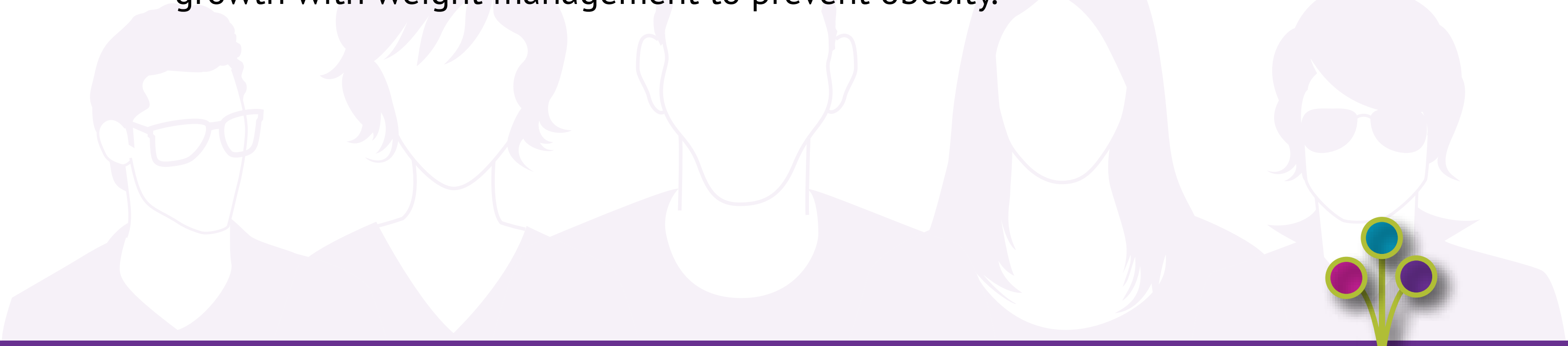
Physical Health

PWS is a multi-systemic condition associated with highly complex medical needs in addition to the developmental and cognitive needs that have been discussed above. These needs are apparent from early in life and include proactive management of feeding and swallowing, nutrition management, hypotonia and intensive early intervention to build core strength, muscle tone and mobility.

Our data illustrated the high need for intensive supports such as high rates of admission to the neonatal intensive care unit (87%). A wide range of medical specialities were required across the life-course for people with PWS. Infants required access to paediatric endocrinology (90%), paediatrics (70%), paediatric dentist (70%), paediatric respiratory specialist (60%) and ophthalmologist (40%). In addition, orthopaedics (44%), psychiatry (31%) and gastroenterology (25%) were also required by school age children. The rates requiring psychiatry (40%) and orthopaedics (80%) increased further in adolescents and the need for psychiatry significantly increased in adults. Increasingly, through active management we are preventing obesity and through intensive assessment and monitoring are trying to prevent issues. The most commonly reported medical problems in addition to obesity were sleep difficulties, hypotonia, scoliosis and flat feet, visual impairment, sensory difficulties, delays in pubertal development and mental health difficulties.

The use of growth hormone in PWS medical care together with extensive multidisciplinary intervention and commitment from caregivers has vastly improved outcomes for a number of medical conditions. It can remediate problems with short stature and osteoporosis, improve body composition and reduce the tendency towards obesity. The decision regarding initiation of growth hormone treatment (GHT) is based on the individual's needs and other medical conditions. GHT in PWS was licensed for use in Ireland in 2001; consequently our data illustrated a trend of increased use of growth hormone over time. Lower percentages of adults had received GHT (25%) compared with 93.8% of individuals in the 5-12 year age group. In young children, 86% reported that GHT was not initiated as quickly as expected. Certain treatments, such as GHT, require multi professional input and with restricted access to professional input treatment can be delayed. From our data, and in discussion with members of PWSAI, it appears that difficulty accessing sleep studies for investigation of obstructive sleep apnoea may be a particular barrier to commencing GHT in a timely way as is access to ENT services.

Obesity remains a significant challenge in the management of people with PWS and is closely related to hyperphagia, the uncontrollable drive towards food that leads to excessive eating. There was a progressive increase in body mass index (BMI) over each age category in our data. (There is a natural tendency to increase BMI with age but also a treatment effect). We observed that almost all of the adults with PWS were reported to be obese. Younger patients do not tend to have the same body habitus as ten years ago with earlier interventions however across all age categories, including the very young children, there were a number of individuals who were significantly obese. Amongst adults, there were some adults still residing at home with extreme obesity. This highlights the enormous difficulties with managing dietary intake in PWS to support individuals to maintain a healthy weight that is likely to be more of a challenge in the home as opposed to a residential setting. People with PWS typically require caloric intake around 50-80% of that recommended for their age. Careful management of diet is therefore required to ensure that it is nutritionally balanced, especially in childhood where it is necessary to balance nutritional requirements for growth with weight management to prevent obesity.



A broad multidisciplinary approach is required for the medical management of PWS. Specialist medical centres are provided in Tallaght Hospital and Cork University Hospital. There are many competing demands on existing clinical services and timely access for patients with PWS can be challenging. Adequate resourcing of these services will help to reduce extensive waiting lists and provide services in Ireland to obviate the need to access services outside the State. Early intervention services are critical to optimize development and support families of those with PWS. These services are invaluable but are often affected by core staff deficiencies, with many teams lacking Speech and Language therapy or Occupational therapy. The provision of these interventions are time critical and comprehensive input at the appropriate developmental stage optimizes outcome. Dietary management is critical in this condition at all life stages, as highlighted previously. Early in life particularly, working intensively with families to optimize muscle tone and mobility is vital and has far reaching effects in terms of muscle strength and body composition. Given that PWS is a rare disorder a skilled multidisciplinary team with experience in PWS that includes dieticians, speech and language therapists, occupational therapists, physiotherapists, psychologists, art and play therapists and medical social workers should be available in the main centre(s) to provide support in a hub and spoke model to the local and regional centres and support families. The aim is to provide as much supported care as close to home as possible but in a co-ordinated seamless way.

Main findings:

- People with PWS have highly complex medical needs and access a large number of medical specialists throughout all stages of their lives.
  The core discipline in the management of PWS is endocrinology both in paediatric and adult endocrinology with support from other key specialties such as respiratory, ENT, and orthopaedics amongst others.
- Each life stage in PWS presents new medical challenges that require access to expertise.
- There are high levels of obesity in adults with PWS, some of which may be ameliorated in future generations by the use of GHT.

- Apparent delays in commencing GHT may be attributable to poor access to medical investigations such as sleep studies.

There is a progressive need for access to psychiatry supports across the lifespan.

Recommendations:

- Adequate resourcing of key medical services to reduce waiting times and increase access.
- The creation of skilled multi-disciplinary team to support the specialist medical teams. Tallaght Hospital is recognised as a national centre for PWS that would ultimately be based at the National paediatric Hospital NPH. This may link with a regional service in Cork and liaise with the European rare diseases network.
- This would serve also as a resource for the development of education and training for community based teams supporting people with PWS and their families within their own locality.
- The provision of specialist dietetic services as part of key medical services is essential for weight management and to promote adequate and balanced nutrition.

Mental Health and Behaviour

Mental health and behavioural difficulties are highly prevalent in PWS. PWS is associated with a characteristic behavioural phenotype. A behavioural phenotype refers to an observed set of behaviours that appear to be consistently part of the clinical presentation for an individual with a genetic syndrome. In PWS common behaviours that are observed include temper outbursts, self-harm (skin-picking), mood swings, repetitive questioning, compulsive behaviour and hoarding. The most persistent characteristic is hyperphagia or over-eating, which leads to significant obesity and can be life-threatening. If unrestricted an individual with PWS may consume over three times as many calories as are required.

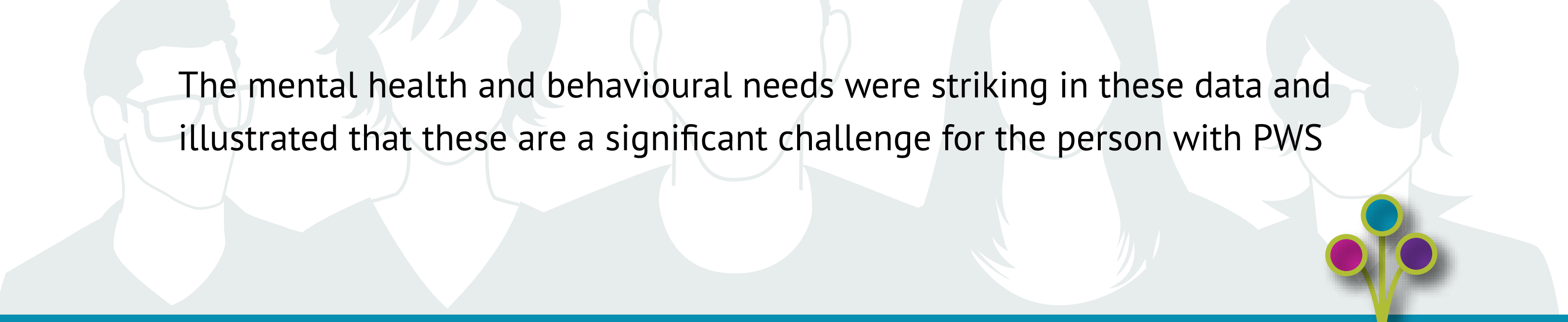
Rates of mental health disorders are significantly elevated compared with the general population with intellectual disability (Whittington and Holland, 2004). In particular people with PWS present with significantly elevated rates of mood disorders and anxiety, obsessive compulsive disorder and major mental health disorders such as bipolar illness and psychosis. Mental health disorders and PWS related behaviours are significantly impairing for the individual and represent some of the most challenging aspects of the condition for the caregivers of people with PWS.

In this study we identified a persistently increased prevalence of mental health disorders across each age group from childhood to adulthood. Sixty-eight per cent of all participants were diagnosed with a mental disorder in their lifetime. This was most marked in the adult group, where 60% had a mental disorder. Anxiety disorders were the most common condition, reported in 76% of adults with PWS, 19% of adolescents and 30% of children aged 5-12 years with a comorbid mental disorder. A significant number of adults with comorbid mental disorders also had depression, psychosis and bipolar illness. These conditions, particularly bipolar illness, may place them at increased risk of overeating. Additionally medications may be associated with increased appetite and weight gain. Therefore timely, appropriate and well supervised management of mental health disorders is required in the context of overall care.

The commonly reported PWS related behaviours included self-injury (scratching), repetitive questioning, obsessions and compulsions, and non-compliance. Over 40% of respondents reported that they found these behaviours relatively difficult to manage. Emotion regulation refers to the processes involved in managing one's emotions and those of others (Cole et al, 1994). It may be impaired in mental health disorders and in the context of atypical neurodevelopment and is thought to contribute to presentation of volatile emotions and temper outbursts. We investigated emotion regulation in people with PWS as reported by caregivers and found that it was significantly impaired in all age groups. Around 60-70% of all individuals needed a maximal or significant amount of support with coping with feelings and emotions. This is likely to be a highly stressful component of caring for an individual with PWS and is likely to contribute significantly to the perceived emotional toll on carers (see below).

As expected hyperphagia was highly prevalent in all age ranges. It typically emerges after infancy and our data concurred with this; hyperphagia was least prevalent in the 0-4 year old age group (30%) in comparison with over 80% in the other age ranges. It appeared more prevalent in adults in residential care compared with those still living at home, suggesting that this is a critical factor to precipitate residential care arrangements (see below). As noted above hyperphagia has a significant impact on food intake and obesity and requires careful behavioural and dietetic management.

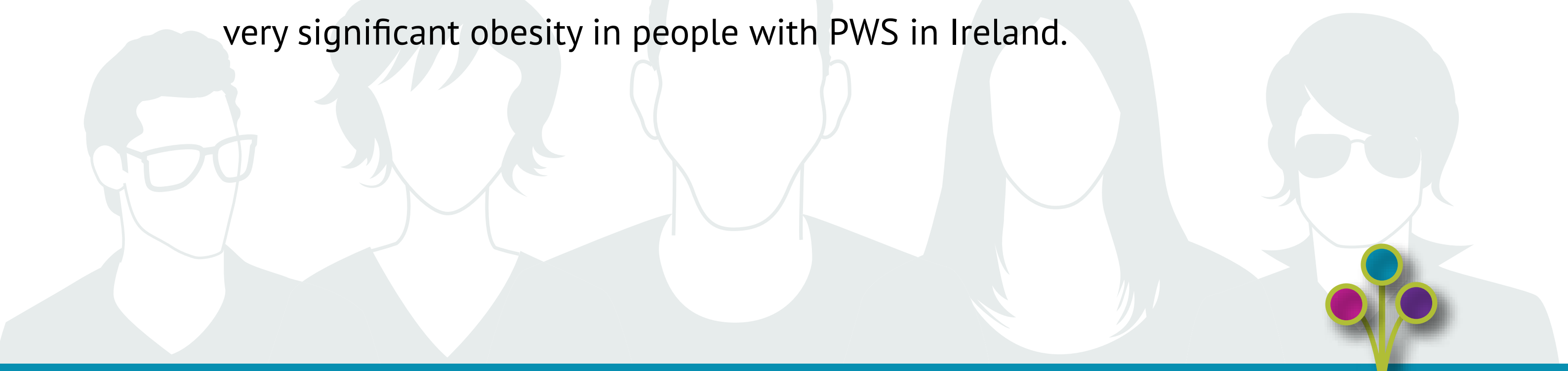
The mental health and behavioural needs were striking in these data and illustrated that these are a significant challenge for the person with PWS



and those caring for them. The complexity of mental health and behavioural needs will require skilled multi-disciplinary professionals who can provide appropriate assessment and intervention. In Ireland, mental health intellectual disability services typically provide services to individuals with moderate, severe or profound intellectual disability. Since the majority of people with PWS do not fall within this range of intellectual disability, they are frequently directed towards generic child and adolescent or adult mental health services. This is problematic as clinicians in these services are not typically trained in managing people with intellectual disabilities and the clinical care pathways are not appropriate. The provision of specialist teams with expertise in treating a number of rare syndromes would help to provide adequate and effective services for people with PWS and comorbid mental health disorders. Four regional adult mental health teams and 1-2 child and adolescent mental health teams nationally with a special interest in management of rare syndromes could be established. These could liaise closely with medical teams to ensure that the management of their psychiatry needs is integrated into overall medical care. They could also provide consultation to mental health and community teams to support the development of PWS specific programs and interventions that are accessible in the person's locality.

Main findings:

- People with PWS have high levels of major mental health difficulties that require specialist mental health treatment and services.
- People with PWS have very significant and frequent behavioural challenges that are likely to impact negatively on them and on their family members.
- Hyperphagia is highly prevalent, difficult to manage and associated with very significant obesity in people with PWS in Ireland.



Recommendations:

- Regional teams with a special interest in rare syndromes with behavioural phenotypes should be established for the provision of specialist mental health support for adults with PWS.
- National teams with a special interest in rare syndromes with behavioural phenotypes should be established for the provision of specialist mental health support for children and adolescents with PWS.
- A national resource for rare syndromes associated with behavioural and mental health presentations should be developed to provide education, training and consultation to professionals regarding the management of conditions, including PWS.
- Early access to specialist behaviour support services is vital to manage the behavioural challenges, especially hyperphagia to prevent the development of obesity in those with PWS. When obesity becomes established it is more difficult to address and the focus must therefore be on prevention.

Education and Employment

In Ireland, the Department of Education and Skills provides for the education of children with special educational needs that have been set out as part of the Education for Persons with Special Educational Needs (EPSEN) Act, 2004. This emphasises inclusive education within a mainstream setting where possible unless the individual's assessed needs indicate that alternative school placements are required. Typically people with special educational needs may be supported in a mainstream classroom, attend a special class within a mainstream school or attend a special school. There are no arrangements for specialist preschool provision for younger children with PWS. This contrasts with the provision of the home tuition grant for preschool age children with Autism Spectrum Disorders (ASD) which in practice is frequently used to access specialist preschool provision.

The preschool age children included here were very young on average and consequently only two were in preschool. All the school-aged participants with PWS in this survey were attending school. In primary school aged children (5-12 years), 63% were in mainstream primary school and the remainder were in special educational settings. In adolescence the majority (67%) were attending a special educational setting. Young people in primary and secondary school generally had access to the support of a Special Needs Assistant, either on a full or part time basis. Differences between primary and secondary school age children in the type of setting may be related to a number of factors. Assessed ability of mild intellectual disability may lead to a recommendation for mainstreaming in the primary school aged children. However given the observations above regarding adaptive functioning, it seems likely that the developmental gap between individuals with PWS and their peers is larger by the time they reach adolescence and therefore mainstream education may no longer be appropriate to their social and personal care needs. Additionally, the behavioural needs of people with PWS increase significantly in adolescence, as we observed (see below). Consequently mainstream schools may not have the appropriate behavioural resources to meet these needs. Additionally behavioural difficulties may be exacerbated by stress associated by mainstream education, due to excessive demands on the individual in an environment that is not fully appropriate to their needs.

Success in education in Ireland is frequently measured by the completion of the two national state examinations, the Junior Certificate Examination and the Leaving Certificate Examination. Some schools offer part of the Junior Certificate program for people with mild intellectual disability. The Leaving Certificate Applied is offered to students where their needs are not adequately catered for by the Leaving Certificate program. In our survey, there were 23 responses for adults regarding the highest level of education achieved. 13 (56%) had no education qualification, 2 (13%) completed Leaving Certificate Applied (LCA) and 3 (9%) had completed some part of the Junior Certificate program and 5 (22%) were still in education. These data may reflect the ability of the participants and that these exams are not suited to their educational needs.

Level of employment might also be viewed as a further measure of success in education. A stark observation was that only 1 adult with PWS was in paid employment, 3 (12%) were in voluntary unpaid employment and 17 (68%) were in a 'day programme'. It is not clear from our data what the characteristics of the day programmes that individuals with PWS were attending were.

The stated primary purpose of the LCA is to prepare participants for adult and working life and the development of literacy and numeracy skills (LCA program statement, DES). Since these are important areas of development, further research should investigate what are the factors influencing the completion of the LCA by people with PWS. This is particularly relevant in consideration of the proportion of individuals accessing employment.

These data did not also extensively explore the challenges facing young people in education or the perspectives and experiences of professionals involved in the education of people with PWS. Further we have no data here regarding the challenges for adults with PWS in accessing paid employment. Work is central to the lives of people and provides purpose and social contact and status (Shepherd, 1989). It is important to promote mental health and wellbeing (Rowland and Perkins, 1988). Ireland has particularly low rates of employment of people with disabilities. The Comprehensive Employment Strategy for People with Disabilities, (Dept. of Justice and Equality, 2015), sets out key priorities that include building skills, capacity and independence, providing supports into work, making work pay, supporting job retention, providing support and engaging with employers. Further research is required to explore how people with PWS may successfully access paid employment.

Main findings:

- The majority of people with PWS accessed mainstream school placements in primary school.
- The majority of people with PWS accessed special school placements in secondary school.
- A high proportion of adults in the survey left education without any educational qualification.
- Only one adult with PWS who completed education had paid employment.

Recommendations:

- More research is required to determine the factors that contribute to successful school placement and to explore the training and education needs for professionals regarding PWS
- Research is required to determine how to support people with PWS into paid employment, in line with the key priorities of the Comprehensive Employment Strategy for People with Disabilities
- Training for education professionals teaching individuals with PWS is required. Excellent guidelines for teachers have been developed by the International Prader Willi Syndrome Organisation (IPWSO, 2010) which can be used to inform this training and could be adapted by the Special Education Support Service.
- Consideration should be given to the provision of home tuition to preschool children with PWS to support access to specialist preschool education.

Residential and Respite Needs

Residential care refers to the provision of a home environment outside the family home. This may be funded by the health service but is frequently provided by a private provider. Respite care provision refers to alternative accommodation that may be provided to a person with a disability on an occasional or regular basis to provide respite for caregivers. Residential and respite care frequently form part of the management plan for people with PWS.

36% of the adults with PWS who were included in the survey were already living in residential care. We asked caregivers of people with PWS who were residing at home for their views on the expected requirement for residential care for their relative with PWS. Two individuals were reported to likely require long-term residential provision in the next two years and a further 14 individuals within the next five years. It should be noted that there were a large number of adults still residing at home with parents, many of whom are likely to be older parents. Therefore this is likely to be a conservative estimate of the need for residential care. The majority, 60% of respondents, reported that the preference was for PWS specific residential care. Those with PWS have very specific requirements that need to be addressed in residential or respite accommodation.

There were large differences in the numbers of individuals accessing respite care across the different age ranges. The largest proportion was in the adolescent group (60%). Only 25% of adults who were still residing in the family home accessed respite care and 6% of children under 12 years. Across all age-groups over 50% of individuals indicated that they had a requirement for respite care, therefore there is a significant gap between the proportion who are receiving this service and those who identify as requiring it. To our knowledge there are no PWS specific respite care provisions and currently respite care is likely to be accessed in a mixed environment. Anecdotal reports indicate that unsuitable respite can be disruptive and problematic.

It has continually been highlighted throughout this report that people with PWS have significant challenges in their cognitive and adaptive functioning and in relation to their behaviour. All of these are likely to impact and reduce the ability for the individual to live independently and for caregivers to provide adequate levels of care. The most significant issue is the management of hyperphagia and obesity, which is critical to reduce morbidity and mortality. Residential care becomes critical in the context of adult life when the person with PWS will naturally wish to have greater independence and self-management. Best practice recommendations indicate that people with PWS require an appropriate environment that has consistent supports for the management of diet and behaviour. At a minimum this necessitates a plan to secure food and a structured daily schedule. There is a tacit acceptance among experts that providing the appropriate environmental supports for the person with PWS is most effectively delivered in the context of a PWS specific program that can take account of the very specific needs of PWS. Furthermore in a mixed environment, adherence to the strict food controls required for people with PWS could be considered an infringement of the rights of those who don't have the condition. In Ireland there is one Prader-Willi specific residence. It is operated by RehabCare, located in Dublin and currently accommodates 5 people with PWS. The remaining provision is within settings that are not specific to PWS. This report highlights a critical requirement for residential and respite care that can meet the needs of people with PWS. Best practice indicates that PWS specific environments are likely to be the most successful in providing this type of care.

Main findings:

- There is a high need for residential care for people with PWS, particularly among adults who are residing at home with ageing parents.

Main findings .....continued:

- Respite provision is not available to the majority of adults with PWS and children under 12 years, and the current provision is not PWS specific.
- The majority of residential care provisions and none of the respite care provisions are PWS specific.

Recommendations:

- There is an urgent need to scale up the provision of PWS specific residential care places for adults with PWS. The provision of a structured predictable environment that controls access to food and reduces the anxiety of the individual is critical to the long term management of PWS. This type of provision has been shown to prolong life and reduce morbidity associated with extreme obesity and provides consistency in treatment and a sense of fairness for the individual.
- Similarly appropriate respite care provision is urgently required to support parents and relatives in their roles as caregivers. As demonstrated in this survey, PWS is associated with a significant impact on the affected person and on their families. Respite is an important part of management to help support the person to reside at home for as long as they can and to prevent carer burnout. Similarly the provision should be PWS specific for consistency in the management.

Impact on Families

This report also aimed to investigate the reported impact of PWS on caregivers and family members. For a complex condition such as PWS, frequently the focus may be on the medical and behavioural needs of the individual. However it is important to look at the broad effects of the condition on the caregiver and the family to identify how else families can be better supported. To this end we explored the reported financial, physical and emotional impacts and how the lives of siblings of the individual with PWS may be impacted.

It is difficult to estimate the financial impact fully. In terms of added costs, a small but significant number of respondents reported an extreme negative financial impact that appeared to increase with age. In addition it was noted that there had been a negative impact on the employment status of caregivers. This was noticeable across all age-groups, where more than 50% reported that either they or their partner had given up work and a high number in each age group had also reduced working hours. Caregivers of the adults residing in supported accommodation were least likely to report they had given up work; however these are on average an older group of adults and possibly represent more families where only one partner routinely worked outside the home. Change in employment status underscores the loss of income to families and the loss of participation in the workforce that are likely to be directly attributable to PWS. Moreover, as the majority of the respondents were mothers, the corollary is that it is largely women who are moving out of the workforce and therefore being disadvantaged financially and with respect to future employment.

Respondents also provided information regarding entitlements and benefits received in respect of their relative with PWS. Many were in receipt of benefits such as Domiciliary Care Allowance and a smaller number had a Medical Card. It was concerning however to note that quite a number reported that they had been refused a medical card which may imply that they have to personally fund aspects of their relative's medical care.

In relation to family life, respondents frequently reported a significant or extreme emotional toll on them related to caring for their relative with PWS. This is not surprising, based on what we learned about the demands of managing the PWS related medical, psychiatric and behavioural needs. We previously reported elevated rates of anxiety symptoms in parents of children with PWS compared with parents in an IQ matched control group (Skokauskas et al, 2012). Others have also reported high levels of parental stress (Tvrdik et al, 2015) and reduced quality of life (Mazaheri et al, 2013). Moreover, stress amongst families with PWS was shown to be higher than in relation to some other developmental disabilities (Lanfranchi and Vianello, 2012). Physical demands were also reported to be high for caregivers, particularly for adolescents and adults with PWS. This is likely to be related to the high physical needs and supports required in relation to activities of daily living. In addition respondents all reported some negative impact of caring on family relationships across all age groups; this was less marked amongst the respondents of adults who were living in supported accommodation. Caregivers also reported an overall negative impact on the mental health of siblings without PWS and on their social life, particularly in the latter case where the individual with PWS was an adolescent. Mental health symptoms were also observed by Mazaheri et al, 2013, who recommended psychosocial supports for carers and siblings due to the multiple challenges of living with PWS.

Main findings:

- Respondents reported a negative impact on their financial and employment status related to caring for an individual with PWS.
- Some individuals were unable to access social welfare supports, such as the medical card or carer's allowance.
- A significant emotional and physical toll was associated with caring.

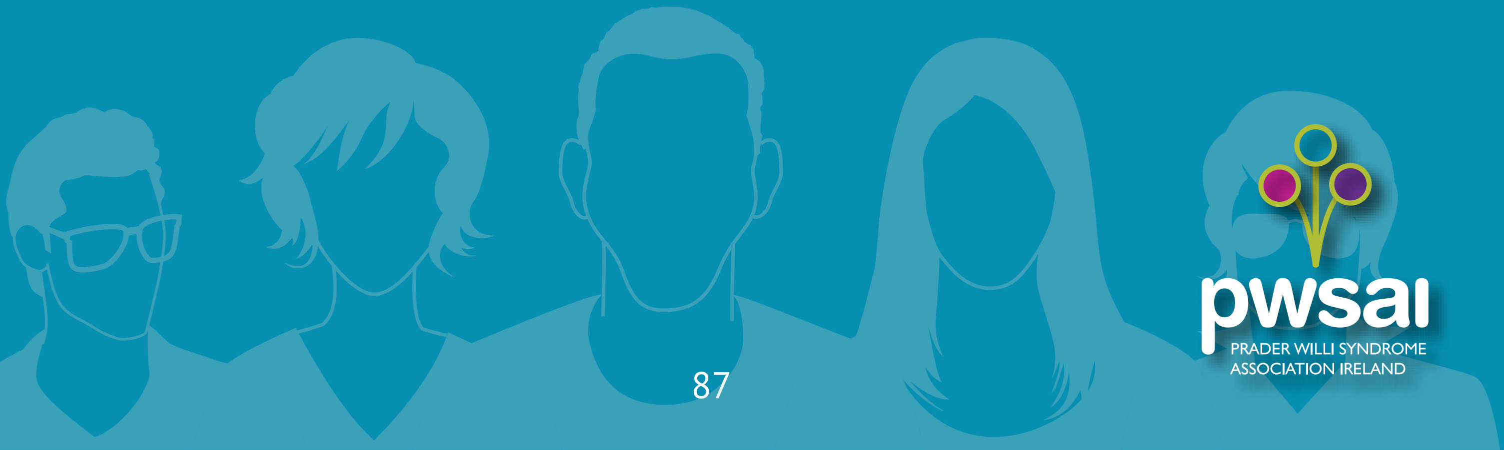
- Caring for an individual with PWS was associated with a negative impact on family relationships.
- A negative impact was also reported on the mental health and social life of siblings.

Recommendations:

- More research is required to investigate the factors associated with decreased employment in caregivers.
- A review of the access to social welfare supports would ensure that there is equitable access to entitlements. The recent announcement in Budget 2016 that all individuals with the Domiciliary Care Allowance will access the Medical Card is welcome.
- Improved service provision, particularly supporting a structured care plan and environment for the individual with PWS may help alleviate carer burden and reduce the pressure on family relationships.
- Psychosocial supports for caregivers and siblings are also required to support coping and prevent deterioration in mental health.

Conclusion

This is the report of a comprehensive and representative survey of caregivers of people with PWS in Ireland today. There have been significant advances in recent decades in the clinical care of individuals with PWS, particularly in relation to medical care. As a consequence of earlier detection, comprehensive targeted interventions and specific therapies such as growth hormone therapy, more people with PWS will live longer and experience less morbidity. This report highlights that there are still considerable challenges facing people with PWS and their families. Obesity rates remain high, educational attainments are not reflected in paid employment, mental health and behavioural concerns continue to progress across the life-course and as we have seen are associated with significant carer burden and negative impact on family functioning. There are significant gaps in service provisions that if addressed through our key recommendations could serve to reduce negative outcomes and improve the lives and health of people with PWS and their families.



Key Recommendations

1. The provision of specialist multidisciplinary supports with expertise in dietary management and behaviour support will help to prevent obesity and manage behaviour.

2. Specialist multidisciplinary medical teams that are well resourced will ensure that people with PWS can get timely access to medical care and prevent avoidable secondary disability.

3. Establishment of a number of mental health teams for adults and children/ adolescents with expertise in rare syndromes, including PWS, will support the provision of expert and timely access to essential psychiatric care.

4. Specialist medical, psychiatry and multidisciplinary supports can provide support to community teams locally to ensure that aspects of service provision, e.g. behaviour management can be delivered locally.

5. PWS specific residential care and respite care that can provide the appropriate environmental controls in a family like environment are critical particularly in respect to the prevention of life-threatening hyperphagia and obesity.

6. Most importantly, people with PWS are individuals with their own set of needs, interests, likes and dislikes. Therefore, it is essential that services are person-centred and take account of the perspectives of people with PWS. This calls in the future for greater facilitation of the involvement of people with PWS in service planning.

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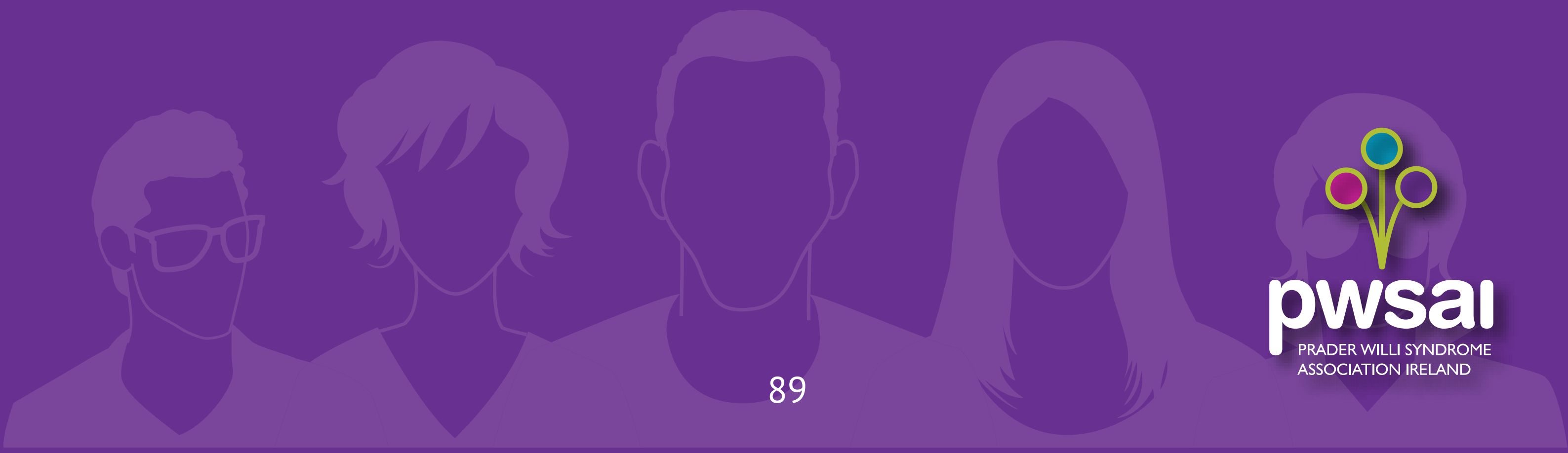

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PRADER WILLI SYNDROME
ASSOCIATION IRELAND

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Appendices

Appendix 1: Diagnostic Criteria for PWS

Diagnostic Criteria for Prader-Willi Syndrome

Major Criteria

1. Neonatal and infantile central hypotonia with poor suck, gradually improving with age.
2. Feeding problems in infancy with need for special feeding techniques and poor weight gain/failure to thrive.
3. Excessive or rapid weight gain on weight-for-length chart (excessive is defined as crossing two centile channels) after 12 months but before 6 years of age: central obesity in the absence of intervention.
4. Characteristic facial features with dolichocephaly in infancy, narrow face or bifrontal diameter, almond-shaped eyes, small-appearing mouth with thin upper lip, down-turned corners of the mouth (3 or more requires).
5. Hypogonadism – with any of the following, depending on age
    a. Genital hypoplasia (male: scrotal hypoplasia, cryptorchidism, small penis and/or testes for age [<5th percentile]; female: absence or sever hypoplasia of labia minora and/or clitoris.
    b. Delayed on incomplete gonadal maturation with delayed pubertal signs in the absence of intervention after 16 years of age (male: small gonads, decreased facial and body hair, lack of voice change; female: amenorrhea/olgiomenorrhea after age 16).
6. Global developmental delay in a child younger than 6 years of age; mild to moderate mental retardation or leraning problems in older children
7. Hyperphagia/food foraging/obsession with food.
8. Deletion 5q11-13 on high resolution (>650 bands) or other cytogenetic/ molecular anbornmality of the Prader-Willi chromosome region; including maternal disomy.

Diagnostic Criteria for Prader-Willi Syndrome

Minor Criteria

1. Decreased fetal movement or infantile lethargy or weak cry in infancy, improving with age.

2. Characteristic behaviour problems - temper tantrums, violent outbursts and obsessive/compulsive behaviour, tendency to be argumentative, oppostional, rigid manipulative, possessive and stubborn, perseverating, stealing and lying (5 or more of these symptoms required).

3. Sleep disturbances or sleep apnoea.

4. Short stature for genetic background by age 15 (in the absence of growth hormone intervention).

5. Hypopigmentation – fair skin and hair compared to family.

6. Small hands (<25th percentile) and/or feet (<10th percentile) for height age.

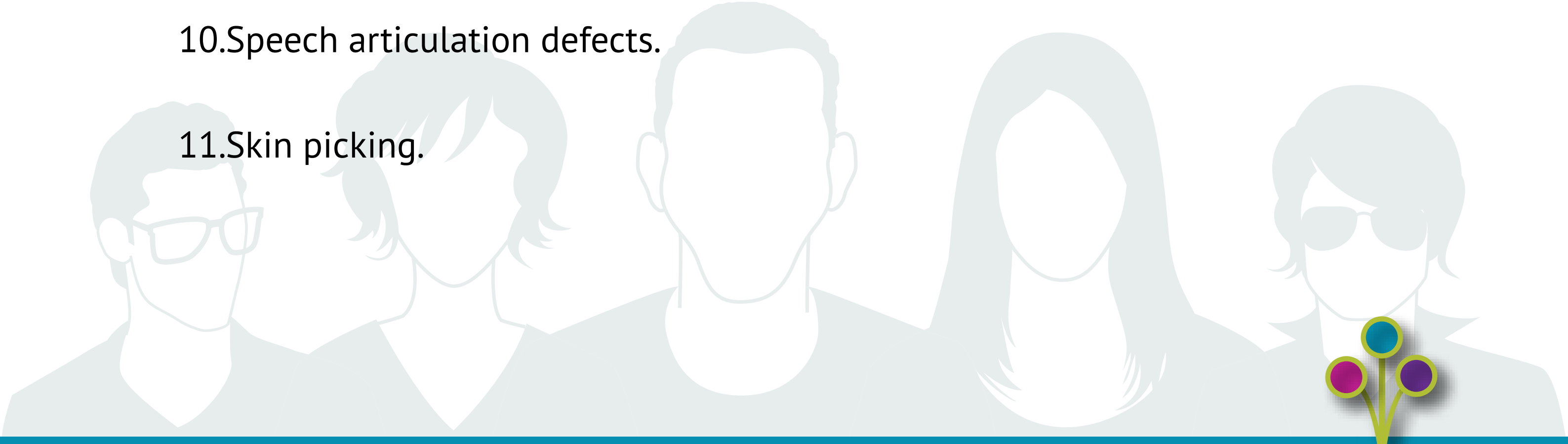
7. Narrow hands with straight ulnar border.

8. Eye abnormalities (esotropia, myopia).

9. Thick viscous saliva with crusting at corners of the mouth.

10. Speech articulation defects.

11. Skin picking.



Diagnostic Criteria for Prader-Willi Syndrome

Supportive Findings

1. High pain threshold.
2. Decreased vomiting.
3. Temperature instability in infancy or altered temperature sensitivity in older children and adults.
4. Scoliosis and/or kyphosis.
5. Early adrenarche.
6. Unusual skill with jigsaw puzzles.
7. Normal neuromuscular studies

Scoring

Major criteria are weighted at one point each.

Minor criteria are weighted at one half point.

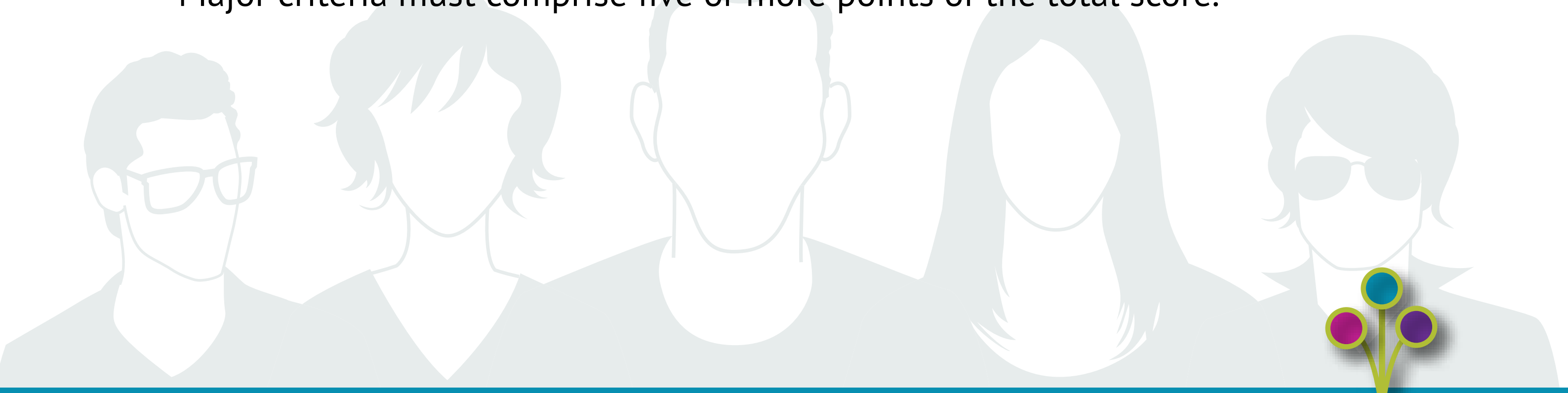
Children 3 years or younger:

Five points are required for diagnosis, four of which should come from the major group.

Children 3 years of age to adults:

Total score of eight is necessary for the diagnosis.

Major criteria must comprise five or more points of the total score.



Appendix 2: Survey Questions: A Population-Based Profile of Prader-Willi Syndrome in Ireland

Contents

Please note an abbreviated version of this questionnaire was completed by parents/ guardians of children under 5.

Section A: Profile

In this section, you will be asked questions about the profile of both you and the person with PWS that you care for.

1. Study ID		
2. Your Relationship to the Person with PWS	☐ Biological Mother ☐ Biological Father ☐ Step-Mother ☐ Step-Father ☐ Adoptive Mother ☐ Adoptive Father ☐ Partner of child's parent ☐ Sibling	☐ Foster Mother ☐ Foster Father ☐ Aunt ☐ Uncle ☐ Other relative/in law ☐ Unrelated guardian ☐ Yourself
3. If known, please tick the highest level of education completed by the mother and father of the person with PWS.	**MOTHER** ☐ Level 0: Early Childhood Education ☐ Level 1: Primary Education ☐ Level 2: Junior Certificate/ Intercertificate ☐ Level 3: Leaving Certificate ☐ Level 4: Post-secondary non –tertiary education ☐ Level 5: Short-cycle tertiary education ☐ Level 6: Bachelor or equivalent ☐ Level 7: Master or equivalent ☐ Level 8: Doctoral or equivalent ☐ Unknown	**FATHER** ☐ Level 0: Early Childhood Education ☐ Level 1: Primary Education ☐ Level 2: Junior Certificate/Intercertificate ☐ Level 3: Leaving Certificate ☐ Level 4: Post-secondary non –tertiary education ☐ Level 5: Short-cycle tertiary education ☐ Level 6: Bachelor or equivalent ☐ Level 7: Master or equivalent ☐ Level 8: Doctoral or equivalent ☐ Unknown
4. Date of Birth of Person with PWS	Day: _______ Month: ________ Year:___________	
5. Gender of person with PWS	☐ Male ☐ Female	
6. Person with PWS's Ethnic/Cultural Background	White: ☐ Irish ☐ Irish Traveller ☐ Any other White background Black or Black Irish: ☐ African ☐ Any other Black background Asian or Asian Irish: ☐ Chinese ☐ Any other Asian background ☐ Other (Please Specify) ___________________________	

Section B: Birth History

In this section, you will be asked questions about the birth history of the person with PWS. We will also ask questions about their diet after they were born.

1. What was the length of the pregnancy with the person with PWS? (Please provide exact weeks on the line if known).	☐ Very premature or very early (fewer than 34 weeks) ____________________ ☐ Premature or early (34 weeks through 36 weeks) ____________________ ☐ On or near due date (37 weeks through 41 weeks) ____________________ ☐ More than 2 weeks late (42 weeks or more) ____________________ ☐ Don't know
2. How much did the person with PWS weigh at birth? (Answer in kgs or pounds)	☐ Kgs_______ Pounds ___________ ☐ Unsure
3. Do you know the age of the biological father at the time of the person with PWS's birth?	☐ Yes (Please Specify) _______________ ☐ No
4. Do you know the age of the biological mother at the time of the person with PWS's birth?	☐ Yes (Please Specify) ________________ ☐ No
5 (a) Was the person with PWS placed in the neonatal or perinatal (NICU/Special care) ICU for any reason?	☐ Yes **(if "yes" selected, please answer 5b-c)** ☐ No **(If "no" selected, please skip to Q6)** ☐ Unknown **(If "Unknown" selected, please skip to Q6)**
5 (b) How long did the person with PWS spend in the neonatal or perinatal ICU after birth?	Days:_________ Weeks:________ Months:_________
5 (c) Please outline the reason(s) why the person with PWS was placed in the neonatal or perinatal ICU? *E.g. Ventilation (breathing support), for feeding monitoring, infection etc.,*	____ ____ ____ ____ ____
6 (a) After birth, how was the person with PWS fed? **<u>(i.e. received the majority of their nutrition)</u>**	☐ Mouth (Bottle fed) ☐ Mouth (Breast fed) ☐ Nasogastric tube ☐ Intravenously ☐ Unsure ☐ Other (Please Specify) ____________________
6 (b) How long was the person with PWS fed a liquid diet only?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not applicable
6 (c) At what age were high energy liquid foods stopped?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not applicable
7 (a) At what age were solid foods introduced to the person with	Weeks:______ Months:_______ Years __________ ☐ Unsure

PWS?	☐ Not relevant
7 (b) How long was the person with PWS on one meal a day?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not relevant
7 (c) At what age was a second meal introduced?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not relevant
7 (d) At what age was a third meal introduced?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not relevant
7 (e) At what age was the person with PWS fully weaned to a solid diet providing the majority of the nutrition?	Weeks:______ Months:_______ Years __________ ☐ Unsure ☐ Not relevant
8. Were there any difficulties encountered when weaning from a liquid diet to a solid diet?	☐ Yes (Please Specify) ___________________________________ ___________________________________ ___________________________________ ___________________________________ ☐ No ☐ Unsure
9. Were any food aversions encountered? *Food aversions refer to refusal to try or to eat certain foods or textures*	☐ Yes (Please specify) ___________________________________ ___________________________________ ___________________________________ ☐ No ☐ Unsure

Is there any additional information or comments you would like to add about the person with PWS's birth history?

Section C: History of PWS Diagnosis

In this section, we would like to ask you a few questions about when the person with PWS received their diagnosis. The answers you provide in this section will help us to get an overall picture of the average age of diagnosis for people with PWS and any issues with obtaining a diagnosis that need to be addressed to ensure better access to diagnosis for people with PWS in the future.

1. At what age was there a diagnosis of PWS?	Weeks: ________ Months: ________ Years: ________
2. Was the diagnosis of PWS confirmed by genetic testing?	☐ Yes ☐ No ☐ Unsure
3. What type of PWS was the person diagnosed with?	☐ Deletion ☐ Uniparental Disomy ☐ Imprinting defect ☐ Not known ☐ Other (Please Specify) __________________
4. At which service/hospital was the diagnosis received?	__________________
5. Before PWS was diagnosed, had the person with PWS received a different diagnosis?	☐ Yes (If yes, ***please specify the other diagnosis here)*** ________________________________ ☐ No
6 (a) Has the person with PWS ever had an IQ assessment?	☐ Yes ***(Is "yes" selected, please answer Q6b)*** ☐ No ***(Please skip to Section D, Q1)*** ☐ Don't know ***(Please skip to Section D, Q1)***
6 (b) Please tick which of the following best describes the result of the person's most recent IQ assessment.	☐ No learning disability ☐ Borderline learning disability ☐ Mild learning disability ☐ Moderate learning disability ☐ Severe/Profound learning disability ☐ Unsure

Is there any additional information or comments you would like to add about the person with PWS's diagnosis?

Section D: Developmental History

This section will ask about the person with PWS's early development. We will ask you to remember at what age the person with PWS reached specific milestones. It can be very difficult to remember the exact age in years and months but please approximate to the best of your ability.

1. Please give the exact age (in years and months) that the person with PWS first walked.	☐ Years:_____ Months:_____ ☐ Has not yet walked ☐ Unsure
2. Please give the exact age (in years and months) that the person with PWS said his/her first words.	☐ Years:_____ Months:_____ ☐ Has not yet said his/her first words ☐ Unsure
3. How old was the person with PWS when he/she was able to use two to three words together for meaningful speech?	☐ Years:_____ Month:______ ☐ Does not yet have meaningful speech ☐ Unsure
4. How old was the person with PWS when he/she was toilet trained during the day?	☐ Years:_____ Months:______ ☐ Not yet toilet trained during the day ☐ Unsure
5. How old was the person with PWS when he/she was toilet training during the night?	☐ Years:_____ Months:______ ☐ Not yet toilet trained during the night ☐ Unsure

Is there any additional information or comments you would like to add about the person with PWS's developmental history?

Section E: Educational history This section has been split into two parts, (1) participants who are currently in education and (2) participants who are not currently in education. Please read the instruction carefully to help navigate your way through this section	
1 Is the person with PWS currently in education?	☐ Yes **(Please answer Question 2)** ☐ No (**Please skip to Question 3)**
QUESTION 2: TO BE ANSWERED ONLY IF PERSON WITH PWS IS CURRENTLY ATTENDING AN EDUCATIONAL SETTING	
2 (a) Which of the following descriptions best suits the person with PWS's ***current*** educational setting?	☐ Mainstream Preschool ☐ Special Needs Preschool ☐ Special Needs Primary School ☐ Mainstream Primary School ☐ Special class in a Mainstream Primary School ☐ Home-schooled ☐ Special Needs Secondary School ☐ Mainstream Secondary School ☐ Special class in a Mainstream Secondary School ☐ Higher Certificate Course ☐ Third Level Institution ☐ Other (Please Specify) ______________________
2 (b) On average, how many days per week does person with PWS attend this educational setting?	☐ 1 ☐ 4 ☐ 2 ☐ 5 ☐ 3
2 (c) What educational supports does the person with PWS currently receive?	☐ Special Needs Assistant full-time ☐ Special Needs Assistant part-time ☐ Resource teaching (Please indicate how many hours per week __________) ☐ July Provision ☐ Other (Please specify) ___________________________________ ☐ None of the above
2 (d) How is the person managing with the level of support they are receiving?	☐ Not managing at all ☐ Managing a little ☐ Somewhat managing ☐ Managing very well ☐ Managing extremely well
2 (e) How satisfied are you with the level of educational support the person with PWS is receiving?	☐ Not satisfied at all ☐ A little satisfied ☐ Somewhat satisfied ☐ Very satisfied ☐ Extremely satisfied
You have now completed the education section. Please skip to Section F: Occupational History and Daily Living Skills	
QUESTION 3: PLEASE ANSWER IF PERSON WITH PWS IS CURRENTLY NOT ATTENDING AN EDUCATIONAL SETTING	

3 (a) Why is the person with PWS currently not attending an educational setting?	☐ Has not begun their education ***(If the person with PWS has not begun their education, please skip to Section F)*** ☐ Has completed education ☐ Has not completed education but is currently not attending an education setting **(Please specify why)** ___________________________________ ___________________________________ ___________________________________ ☐ Home-schooled
3 (b) Before the person with PWS stopped attending an educational setting, what educational settings did they attend? (Tick all that apply)	☐ Mainstream Preschool ☐ Special Needs Preschool ☐ Special Needs Primary School ☐ Mainstream Primary School ☐ Special class in a Mainstream Primary School ☐ Home-schooled ☐ Special Needs Secondary School ☐ Mainstream Secondary School ☐ Special class in a Mainstream Secondary School ☐ Third Level Institution ☐ Other (Please Specify) ____________________ ☐ None of the above
3 (c) What educational qualifications did they receive? (Tick all that apply)	☐ None ☐ Intermediate Certificate/Junior Certificate ☐ Leaving Cert Applied ☐ Leaving Cert ☐ Advanced Certificate ☐ Higher Certificate ☐ Bachelor's Degree ☐ Master's Degree ☐ Doctoral Degree Other (Please Specify) _________________________ ☐ Not Applicable
Is there any additional information or comments you would like to add about the person with PWS's educational history?	

Section F: Occupational History and Daily Activities	
1 (a) Is the person with PWS in paid employment?	☐ Yes ***(If "yes" selected, please answer Q1b)*** ☐ No ***(If " no" selected, please skip to Q2)***
1 (b) How often (on average) does the person with PWS undertake this paid employment?	☐ 5 or more days per week ☐ 4 days per week ☐ 3 days per week ☐ 2 days per week ☐ 1 day per week ☐ Other (specify) ☐ ________________________________
2 (a) Is the person with PWS in voluntary employment?	☐ Yes ***(if "yes" selected, please answer Q2b)*** ☐ No ***(If " no" selected, please skip to Q3)***
2 (b) How often (on average) does the person with PWS undertake this voluntary employment?	☐ 5 or more days per week ☐ 4 days per week ☐ 3 days per week ☐ 2 days per week ☐ 1 day per week ☐ Other (specify) ☐ ________________________________
3 (a) Is the person with PWS attending a day program or group activities?	☐ Yes ***(if "yes" selected, please answer Q3b)*** ☐ No ***(If "no" selected, please skip to Section G)***
3 (b) How often does the person attend a day program?	☐ 5 or more days per week ☐ 4 days per week ☐ 3 days per week ☐ 2 days per week ☐ 1 day per week ☐ Other (specify) ________________________________
4. Is there any additional information or comments you would like to add about the person with PWS's occupational history and daily activities?	

Section G: Independence and Daily Living Skills	
1. Does the person with PWS require the use of the following aids for mobility?	☐ Walking Rollator ☐ Walking frame ☐ Wheelchair ☐ Other (Please specify) _________________ ☐ No aids required
2 Please rate on a scale of 1-4, how much help does the person with PWS need for mobility around the house?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help)
3 (a) Have any modifications been made to your home to facilitate independence for the person with PWS (e.g. for self-care activities, mobility, food security etc.)	☐ Yes **(please answer Q3b and then skip to Q4)** ☐ No **(please skip to 3c)**
3 (b) For what purpose were these modifications to the home made?	☐ For self-care activities (e.g. showering, toileting, etc.) ☐ To improve mobility around the home ☐ To ensure food security ☐ Other (Please specify) _________________________________ _________________________________ **(Please skip to Q4)**
3 (c) If modifications have ***not*** been made to your home, please tick why?	☐ Do not need modifications in the home ☐ Too costly ☐ Other (Please specify) __________________________________ __________________________________
4. Do you plan to make modifications to your home in the future to facilitate independence for the person with PWS?	☐ Yes **(Please Specify when and the type**) ○ For self-care activities (E.g. showering, bathing toileting, etc.) ○ For mobility ○ To ensure food security ○ Other ___________________ **When:** Month ______ Year ______ ☐ No
5. How much help does the person with PWS need for getting around the community?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
6. How much help does the person with PWS need for spoken understanding	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
7. How much help does the person with PWS need with written understanding?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help)

	☐ 4 (Maximal help) ☐ Not applicable due to age
8. How much help does the person with PWS need for being understood?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
9. How much help does the person with PWS need for making and keeping friends?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
10. How much help does the person with PWS need for coping with feelings and emotions?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
11. How much help does the person with PWS need for dressing?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
12. How much help does the person with PWS need for showering and bathing?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
13. How much help does the person with PWS need for cleaning teeth?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
14. How much help does the person with PWS need for toileting?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
15. How much help does the person with PWS require for managing their health e.g. managing their medication, exercise and diet?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
16. How much help does the person with PWS need for learning new tasks?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
17. How much help does the person with PWS need for remembering?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
18. How much help does the person with PWS need for	☐ 1 (No help) ☐ 2 (A little help)

managing money?	☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
19. How much help does the person with PWS need for performing daily tasks around the house?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
20. How much help does the person with PWS need for problem solving?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A Not applicable due to age significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
21. How much help does the person with PWS need for making decisions?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
22. How much help does the person with PWS need for road safety?	☐ 1 (No help) ☐ 2 (A little help) ☐ 3 (A significant amount of help) ☐ 4 (Maximal help) ☐ Not applicable due to age
Is there any additional information or comments you would like to add about the person with PWS's independence or daily living skills?	

Section H: Medical History and Medications	
1(a) Last date person with PWS was weighed	Day: ______ Month: _______ Year:__________
1(b) Location where person with PWS was last weighed	☐ Clinic ☐ Home ☐ Other ______________________
1(c) Weight of person with PWS on last date weighed (answer in kgs or stone)	Kgs: _______ Stone: __________
2(a) Last date person with PWS was measured in height?	Day: ______ Month: _______ Year:__________
2(b) Location where person with PWS was last measured	☐ Clinic ☐ Home ☐ Other ______________________
2(c) Height of person with PWS on last date measured (answer in cms or feet)	Cms: _______ Feet: ____________
3. In what hospital does the person with PWS receive most of his/her PWS-related medical care?	____________________________________
4(a) Has the person with PWS ever been admitted (overnight) to a hospital due to PWS?	☐ Yes (If "yes" selected please answer 4b-4e) ☐ No
4(b) How many times in total has the person with PWS been admitted (overnight) to a hospital due to PWS but **not counting sleep studies?**	☐ < 5 times ☐ 5-10 times ☐ 11-15 times ☐ 16-20 times ☐ >20 times
4(c) How many times in total has the person with PWS been admitted (overnight) to a hospital due to PWS in the last 12 months but not counting sleep studies?	☐ < 5 times ☐ 5-10 times ☐ 11-15 times ☐ 16-20 times ☐ >20 times
4 (d) Has the person with PWS ever been admitted overnight in hospital for sleep studies?	☐ Yes **(Please answer Q4e)** ☐ No **(Please skip to Q5)**
4(e) How many times in total has the person with PWS been admitted (overnight) to a hospital for Sleep studies?	☐ < 5 times ☐ 5-10 times ☐ 11-15 times ☐ 16-20 times ☐ >20 times
5 Which of these medical	☐ Paediatrician

specialists has the person with PWS seen in the past? (Tick all that apply)	☐ Paediatric Endocrinologist ☐ Endocrinologist ☐ Respiratory doctor ☐ Orthopaedic surgeon ☐ Psychiatrist ☐ Dentist ☐ Gastroenterologist ☐ Ophthalmologist ☐ None ☐ Other (Please specify) ___________________________________	
6(a) Has the person with PWS ever been diagnosed with any of these orthopaedic problems? (Tick all that apply)	☐ Scoliosis/Kyphosis/Lordosis ☐ Osteopenia/Osteoporosis ☐ Hypotonia ☐ Pronated or flat feet ☐ Hip dysplasia ☐ Leg deformities (e.g. knock knees, bow legs) ☐ Other orthopaedic problems (Please Specify) _____________________ ☐ None of the above	
6(b) Has the person with PWS ever been treated for orthopaedic problems in any of these ways? (Tick all that apply)	☐ Back brace ☐ Back cast ☐ Back surgery ☐ Orthotics ☐ Other (Please specify) ______________________ ☐ None of the above	
7(a) Has the person with PWS ever had any bone fractures?	☐ Yes **(If yes, please complete table below)** ☐ No **(If "no" selected, please skip to Q8)**	
7 (b) Please list the part of the body fractured as well as the cause of the each fracture.	***Part of Body Fractured***	***Cause of Fracture***
	1. ___________________	☐ Accident/Trauma ☐ Repetitive Force (e.g. running) ☐ Fall from more than 3 feet ☐ Spontaneous /Unexplained ☐ Other (Please specify) ____________________
	2. ___________________	☐ Accident/Trauma ☐ Fall from more than 3 feet ☐ Spontaneous /Unexplained ☐ Other (Please specify) ____________________
	3. ___________________	☐ Accident/Trauma ☐ Fall from more than 3 feet ☐ Spontaneous /Unexplained ☐ Other (Please specify)

			______________________
	4. ____________________		☐ Accident/Trauma ☐ Fall from more than 3 feet ☐ Spontaneous /Unexplained ☐ Other (Please specify) ______________________
	5. ____________________		☐ Accident/Trauma ☐ Fall from more than 3 feet ☐ Spontaneous /Unexplained ☐ Other (Please specify) ______________________
8. Has the person with PWS ever been diagnosed with or displayed any of these sleep problems? (Tick all that apply)	☐ Sleep apnoea ☐ Excessive daytime sleepiness ☐ Frequent or early waking (not appropriate for age) ☐ Other ***(Please specify)*** ________________________________ ☐ None of the above		
9. Has the person with PWS ever been used any of the following aids for use while sleeping?	☐ CPAP ☐ BiPAP ☐ Oxygen ☐ None of the above ☐ Other ***(Please specify)*** __________________________		
10. Has the person with PWS been diagnosed with any of the following? (Tick all that apply)	**Diagnosis**	**Age at time of Diagnosis**	**Additional Information**
	☐ Hearing impairment		Does the person require a hearing aid? ☐ Yes ☐ No
	☐ Visual Impairment		Does the person require visual aids other than glasses? ☐ Yes (Please Specify) _____________ ☐ No Was ophthalmic surgery required? ☐ Yes ☐ No
	☐ Thyroid Disease		☐ Overactive ☐ Underactive
	☐ Diabetes		☐ Type 1 ☐ Type 2
	☐ Delayed Puberty		

	☐ Sleep apnoea		
	☐ Adrenal disease		Did the person with PWS require steroid replacement? ☐ Yes ☐ No
11 (a) Has the person with PWS ever experienced sensory difficulties?	☐ Yes **(If "yes" selected, please answer Q11b)** ☐ No **(If "no" selected, please skip to Q12)** ☐ Unsure **(If "no" selected, please skip to Q12)**		
11(b)	☐ Sounds ☐ Sights ☐ Smells ☐ Textures (e.g. dislike the feel of certain clothes) ☐ Tastes ☐ No reaction to certain stimuli (e.g. cold, heat, pain) ☐ Other (Please specify) ____________________		
12. Has the person with PWS ever received Growth Hormone Treatment?	☐ Yes **(If "yes" selected, please answer Q13a-d)** ☐ No **(If "no" selected, please skip to Q13e)** ☐ Unsure		
13 (a) What age was the person with PWS when they began receiving Growth Hormone Treatment?	__________Years _________Months		
13 (b) Was there a delay in starting Growth Hormone treatment for any reason?	☐ Yes (Please specify why) ____ ____ ____ ☐ No		
13 (c) For what duration did the person with PWS receive growth hormone treatment?	☐ Currently still on treatment ☐ _________ Years _________ Months		
13 (d) At any point was the treatment stopped early?	☐ Yes (Please specify why) _______________________________ ________________________________ ________________________________ ☐ No ***Please skip to Q13f***		
13 (e) If the person with PWS was not treated with Growth Hormone Treatment, why was this?	☐ Not available at the time ☐ Treatment was not offered to person ☐ Person was not suitable for treatment due to other illnesses ☐ Person with PWS/their family declined treatment ☐ Too young ☐ Not applicable ☐ Other (Please Specify) ______________________________		

13 (f) **PLEASE ANSWER IF PERSON WITH PWS IS FEMALE** What age was the person with PWS when she had her first period?	☐ Years_______ Months _______ ☐ Has not yet had first period
14 (a) Is the person with PWS currently taking or has in the past taken any medications to support pubertal development or sex hormones?	☐ Yes (Please answer Q14b-d) ☐ No (Please skip to Q14e)
14 (b) If yes, what medication did the person with PWS take?	________________________ ________________________ ________________________
14 (c) What age did they begin taking this medication?	☐ Years_______ Months _______
14 (d) For what duration did the person with PWS take this medication?	☐ Years _________ Months________ ☐ Currently still taking medication ***Please now skip to Q15***
14 (e) If no, why has the person with PWS not taken medications to support pubertal development or sex hormone?	☐ Not relevant due to age/stage ☐ Not required puberty as was achieved spontaneously ☐ Do not wish to take as concerned due to possible effect on mood ☐ No due to other reasons

15. Please list any other medications the person with PWS is currently taking or has taken in the past. ***PLEASE DO NOT INCLUDE PSYCHOTROPIC (MEDICATIONS IN THIS SECTION.***	**Medication**	**Age**	**Duration**

Is there any additional information or comments you would like to add about the person with PWS's medical history or medications?

Section I: Mental Health and Behaviour

1 (a) Has the person with PWS ever been diagnosed with or received treatment for any mental health difficulty?	☐ Yes **(If “yes” selected, please answer 1b-c)** ☐ No **(If “no” selected, please skip to Q2)**	
1 (b) Please tick any mental health difficulty the person with PWS has ever been diagnosed with or received treatment for. Please also list the year they received this diagnosis or treatment.	**Anxiety Disorder:** ☐ Generalised Anxiety Disorder ☐ Separation Anxiety Disorder ☐ Panic Disorder ☐ Agoraphobia ☐ Social Anxiety Disorder ☐ Simple Phobia ☐ Obsessive Compulsive Disorder **Depressive Disorders** ☐ Major Depressive Disorder ☐ Dysthymic Disorder ☐ Depression **Other Disorders** ☐ Bipolar Disorder ☐ Psychosis ☐ Autism Spectrum Disorder ☐ Attention-Deficit-Hyperactivity-Disorder ☐ Oppositional Defiant Disorder ☐ Conduct Disorder ☐ Schizophrenia ☐ Enuresis ☐ Encopresis ☐ Tourette Syndrome ☐ Chronic Tic Disorder **Other(s) (Please Specify)** ____________________________ ____________________________	**Year** ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______ ______

1 (c) Where has the person with PWS attended mental health services? Please list all the services (*e.g. Disability Service, CAMHS, Primary Care, other (Please specify*)

☐ Not Applicable

Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________
Place:_________________________________ Year _____ Intervention ________________________________

2 (a) Please rate on a scale of 0-4	**Self Injurious Behaviours**	

by circling the number, **<u>HOW OFTEN</u>** the following behaviours have occurred in the past 6 months. **0. Never/No Problem** **1. Monthly** **2. Weekly** **3. Daily** **4. Hourly**	Self biting	0 1 2 3 4
	Head hitting	0 1 2 3 4
	Body hitting	0 1 2 3 4
	Self-scratching (incl. skin picking)	0 1 2 3 4
	Pica (ingesting non-food items)	0 1 2 3 4
	Inserting objects (nose, ears, anus, including rectal picking)	0 1 2 3 4
	Hair pulling	0 1 2 3 4
	Teeth grinding	0 1 2 3 4
	Aggressive / Destructive Behaviours	
	Hitting others	0 1 2 3 4
	Kicking others	0 1 2 3 4
	Pushing others	0 1 2 3 4
	Biting others	0 1 2 3 4
	Grabbing and pulling others	0 1 2 3 4
	Hair Pulling	0 1 2 3 4
	Scratching others	0 1 2 3 4
	Pinching others	0 1 2 3 4
	Verbally abusive with others	0 1 2 3 4
	Destroying things (e.g. rips clothes, throws chairs, smashes tables etc)	0 1 2 3 4
	Bullying – being mean or cruel (e.g. grabbing toys or food from others)	0 1 2 3 4
	Other	
	Stealing food	0 1 2 3 4
	Stealing money	0 1 2 3 4
	Lying	0 1 2 3 4
	Repetitive questions	0 1 2 3 4
	Disruptive e.g. in class	0 1 2 3 4
	Non-compliance	0 1 2 3 4
	Difficulty transitioning	0 1 2 3 4
	Obsessions / compulsions (Please specify the type of obsession/compulsion)	0 1 2 3 4 ____________________ ____________________ ____________________ ____________________ ____________________
	Other(s) (Please Specify) ________________________ ________________________	0 1 2 3 4
2 (b) Please rate on a scale of 0-3 by circling the number, how **<u>SERIOUS</u>** the behaviour problem is. **0. No problem** **1. Mild** **2. Moderate** **3. Severe**	**Self Injurious Behaviours**	
	Self biting	0 1 2 3
	Head hitting	0 1 2 3
	Body hitting	0 1 2 3
	Self-scratching (incl. skin picking)	0 1 2 3
	Pica (ingesting non-food items)	0 1 2 3
	Inserting objects (nose, ears, anus, including rectal picking)	0 1 2 3
	Hair pulling	0 1 2 3
	Teeth grinding	0 1 2 3
	Aggressive / Destructive Behaviours	

	Hitting others	0	1	2	3
	Kicking others	0	1	2	3
	Pushing others	0	1	2	3
	Biting others	0	1	2	3
	Grabbing and pulling others	0	1	2	3
	Hair Pulling	0	1	2	3
	Scratching others	0	1	2	3
	Pinching others	0	1	2	3
	Verbally abusive with others	0	1	2	3
	Destroying things (e.g. rips clothes, throws chairs, smashes tables etc)	0	1	2	3
	Bullying – being mean or cruel (e.g. grabbing toys or food from others)	0	1	2	3
	Other				
	Stealing food	0	1	2	3
	Stealing money	0	1	2	3
	Lying	0	1	2	3
	Repetitive questions	0	1	2	3
	Disruptive e.g. in class	0	1	2	3
	Non-compliance	0	1	2	3
	Difficulty transitioning	0	1	2	3
	Obsessions / compulsions	0	1	2	3
	Other(s) (Please Specify) _________________________ _________________________	0	1	2	3

2 (c) Below are several questions that ask about your responses to challenging behaviours displayed by the person with PWS you care for. Please read each question, and tick the number on the scale that reflects your own view

How confident are you in dealing with the challenging behaviours of the person with PWS you care for?	☐ 1 Not at all confident ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 Very confident
How difficult do you personally find it to deal with the challenging behaviours of the person with PWS you care for?	☐ 1 Very difficult ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 Not very difficult at all
To what extent do you feel that the way you deal with the challenging behaviours of the person with PWS you care for has a positive effect?	☐ 1 Has no positive effect at all ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 Has a very positive effect
How satisfied are you with the way in which you deal with the challenging behaviours of	☐ 1 Not satisfied at all ☐ 2 ☐ 3

the person with PWS you care for?	☐ 4 ☐ 5 ☐ 6 ☐ 7 Very satisfied		
To what extent do you feel in control of the challenging behaviours of the person with PWS you care for?	☐ 1 Not in control at all ☐ 2 ☐ 3 ☐ 4 ☐ 5 ☐ 6 ☐ 7 Very much in control		
3 (a) Is the person with PWS **currently** taking any psychotropic medications?	☐ Yes (**Please answer Q3b)** ☐ No **(Please skip to Section H)**		
3 (b) Please select the medications the person is **currently taking or has taken in the past.** Please also list the start date and duration of the medication.	**Medication**	**Start Date (Month + Year)**	**Duration (In months)**
	☐ Aripiprazole (Abilify) ☐ Risperidone (Risperdal) ☐ Fluoxetine (Prozac) ☐ Methylphenidate (Ritalin, Concerta, Equsaym, Meikinet) ☐ Atomoxetine (Strattera) ☐ Lisdexamfetamine (Tyvense) ☐ Quetiapine (Seroquel) ☐ Clonidine ☐ Sertraline (Lustral) ☐ Phenergan ☐ Olanzapine (Zyprexa) ☐ Unsure ☐ Other (Please Specify) ________________________ ________________________		
Is there any additional information or comments you would like to add about the person with PWS's mental health or behaviour?			

Section J: Other Health Professionals and Therapies

1. Which of these services has the person with PWS received in the past? (Tick all that apply) **Please also specify the time you waited to received your first appointment with that particular service.**	**Service**	**Time spent waiting for first appointment with service**
	☐ Speech and Language Therapy	☐ ________ Months
	☐ Physiotherapy	☐ ________ Months
	☐ Occupational Therapy	☐ ________ Months
	☐ Dietician	☐ ________ Months
	☐ Psychologist (Psychological assessment, psychological intervention)	☐ ________ Months
	☐ Behaviour Analyst (Behaviour assessment and behavioural management)	☐ ________ Months
	☐ Psychiatry (Medical diagnosis and drug treatment)	☐ ________ Months
	☐ Hydrotherapy	☐ ________ Months
	☐ Early Intervention	☐ ________ Months
	☐ Audiologist	☐ ________ Months
	☐ Podiatrist	☐ ________ Months
	☐ Dentist	☐ ________ Months
	☐ None of the above	
	☐ Other (please specify)	
	______________________	☐ ________ Months
	______________________	☐ ________ Months
	______________________	☐ ________ Months
2. In the last 12 months how many sessions of these services has the person with PWS had?	Speech and Language Therapy _________ Physiotherapy __________ Occupational Therapy _________ Dietician _____________ Psychology ___________ Behaviour Analyst __________ Psychiatry _____________ Hydrotherapy _________ Early Intervention ______ Audiologist ___________ Podiatrist ___________ Dentist __________ Other (please specify) _________________________ __________	
3. In the past 12 months how many sessions of these services did you/the person with PWS have to pay for?	Speech and Language Therapy _________ Physiotherapy __________ Occupational Therapy _________ Dietician ____________ Psychology __________ Behaviour Analyst Psychiatry_____________ Hydrotherapy _________ Early Intervention ______ Audiologist ___________ Podiatrist ___________	

	Dentist __________ Other (please specify) _________________________ __________
4(a) Is the person with PWS currently on a waiting list to receive services?	☐ Yes **(If "yes" selected please answer 4b)** ☐ No **(If "no" selected, please skip to section I)**
4(b) How long has the person with PWS been waiting for the following services?	Speech and Language Therapy _________ Physiotherapy __________ Occupational Therapy _________ Dietician ______ Psychology _____ Behaviour Analyst Psychiatry ________ Hydrotherapy _________ Early Intervention ______ Audiologist ___________ Podiatrist ___________ Dentist __________ Other (please specify) _________________________ __________
Is there any additional information or comments you would like to add about the person with PWS's treatments?	

Section K: Hyperphagia

Sometimes older children with PWS can begin to display hyperphagia and food seeking behaviours which can result in rapid weight gain. The next section is a short questionnaire designed to assess hyperphagia. Please complete this questionnaire if the person with PWS you care for has shown any signs of hyperphagia

1. Does the person with PWS display hyperphagia (over-eating) or any food seeking behaviours?	☐ Yes (Please answer Q2-14) ☐ No (Please skip to Section L) ☐ Unsure (Please answer Q2-14)
2. How upset does the person with PWS generally become when denied a desired food?	☐ Not particularly upset at all ☐ A little upset ☐ Somewhat upset ☐ Very upset ☐ Extremely upset
3. How often does the person with PWS try to bargain or manipulate to get more food at meals?	☐ A few times a year ☐ A few times a month ☐ A few times a week ☐ Several times a week ☐ Several times a day
4. Once the person with PWS has food on their mind, how easy is it for you or others to re-direct the person with PWS away from food to other things?	☐ Extremely easy, takes minimal effort to do so ☐ Very easy, takes just a little effort to do so ☐ Somewhat hard, takes some effort to do so ☐ Very hard, takes a lot of work to do so ☐ Extremely hard, takes sustained and hard work to do so
5. How often does the person with PWS look in the bin for food?	☐ Never ☐ A few times a year ☐ 1–2 nights a month ☐ 1–3 nights a week ☐ 4 to 7 nights a week
6. How often does the person with PWS get up at night to food seek?	☐ Never ☐ A few nights a year ☐ 1–2 nights a month ☐ 1–3 nights a week ☐ 4 to 7 nights a week
7. How persistent is the person with PWS in asking or looking for food after being told “no” or “no more”?	☐ Lets go of food ideas quickly and easily ☐ Lets go of food ideas pretty quickly and easily ☐ Somewhat persistent with food ideas ☐ Very persistent with food ideas ☐ Extremely persistent with food ideas
8. Outside of normal meal times, how much time does the person with PWS spend talking about food or engaged in food-related behaviours?	☐ Less than 15 minutes a day ☐ 15 to 30 minutes a day ☐ 30 minutes to an hour ☐ 1 to 3 hours a day ☐ more than 3 hours a day
9. How often does the person with PWS try to steal food (that you are aware of?)	☐ A few times a year ☐ A few times a month ☐ A few times a week ☐ Several times a week ☐ Several times a day
10. When others try to stop the person with PWS from talking about food or engaging in food-related behaviours, it generally	☐ No distress or upset ☐ Mild distress or upset ☐ Moderate distress or upset ☐ Severe distress or upset

leads to:	☐ Extreme distress ☐ Behaviours can’t usually be stopped
11. How clever or fast is the person with PWS in obtaining food?	☐ Not particularly clever or fast ☐ A little clever or fast ☐ Somewhat clever or fast ☐ Very clever or fast ☐ Extremely clever of fast
12. To what extent do food-related thoughts, talk, or behaviour interfere with the person with PWS's normal daily routines, self-care, school, or work?	☐ No interference ☐ Mild interference; occasional food-related interference in completing school, work, or hygiene tasks ☐ Moderate interference; frequent food-related interference in completing school, work, or hygiene tasks ☐ Severe interference; almost daily food-related interference in completing school, work, or hygiene tasks ☐ Extreme interference, often unable to participate in hygiene tasks or to get to school or work due to food-related difficulties
13. How old was the person with PWS when they first showed an increased interest in food?	___________ Years and __________ Months
14. How variable is the person with PWS's preoccupation or interest in food?	☐ Hardly ever varies ☐ Usually stays about the same ☐ Goes up and down occasionally ☐ Goes up and down quite a lot ☐ Goes up and down all the time

Section L. Financial Supports	
1. Do you receive domiciliary care allowance?	☐ Yes ☐ No
2 (a) Does the person with PWS have a medical card currently?	☐ Yes (If “yes” selected, please answer 2b and 2c) ☐ No (If “no” selected, please skip to Q3)
2 (b) On what basis was the person with PWS awarded his/her current medical card?	☐ Income is below threshold required for medical card eligibility ☐ Discretionary basis ☐ Don't know
2 (c) Was the person ever refused a medical card?	☐ Yes ☐ No ☐ Don't know
3 (a) Does the person with PWS have access to the Long Term Illness Scheme?	☐ Yes (If “yes” selected, please answer Q3b) ☐ No (If “no” selected, please skip to Q4)
3 (b) On what grounds was the person with PWS awarded access to the Long Term Illness Scheme	☐ Intellectual Disability ☐ Mental Illness ☐ Other ________________________________
4. Do you receive carer’s allowance?	☐ Yes ☐ No
5. Do you receive carers benefit?	☐ Yes ☐ No
Is there any additional information or comments you would like to add about the person with PWS’s financial supports?	

Section M: Living Circumstances	
1 (a) What is the current living situation of the person with PWS	☐ At home **(Please skip to Q2)** ☐ In a PWS-specific group home ☐ In another group home ☐ Independent with support ☐ Other ____________________________________ **If you answered PWS-specific group home or other group home please identify the provider of the home and its location. (Please also answer Q1b-1e)**____________________________________
1 (b) What age was the person with PWS when they entered their first group home	_____________________years
1 (c) What weight was the person with PWS when they entered the group home?	__________________Kg/s or __________Stone __________Pounds
1 (d) How many different group homes has the person lived in.	____________________________
1 (e) If relevant, what ages were the parents, when the person with PWS entered the group home?	Parent 1: ___________ years Parent 2: ___________ years
2 (a) If the person with PWS is living at home, who lives with him/her (tick all that apply)	☐ Mother ☐ Father ☐ Grandmother ☐ Grandfather ☐ Siblings (How many?) ___________ ☐ Other ________________________ ☐ Not applicable
2 (b) Does the person with PWS have access to respite (supported care or accommodation)?	☐ Yes **(If "yes" selected", please answer 2b-2j)** ☐ **No (Please skip to Q3)** ☐ Not applicable as person with PWS is in a residential home/supported living arrangement (Please skip to Q3)
2 (c) Does this respite include overnight respite?	☐ Yes ☐ No
2 (d) How many hours per month does the person with PWS have access to respite **in his/her own home?**	☐ 0-5 ☐ 5-10 ☐ 10-15 ☐ 15-20 ☐ More than 20
2 (e) How is this respite funded?	☐ State funded ☐ Charity funded ☐ Paid for by you ☐ Other ______________________________________
2 (f) Name the organisation that provides this respite	______________________________________

2 (g) How many hours per month does the person with PWS have access to respite **<u>outside his/her own home?</u>**	☐ 0-5 ☐ 5-10 ☐ 10-15 ☐ 15-20 More than 20
2 (h) How is this respite funded?	☐ State funded ☐ Charity ☐ Privately funded ☐ Other ____
2 (i) In the last 12 months how many times in total has the person with PWS accessed any form of respite?	___________________
2 (j) Are you satisfied with the quality of the respite the person with PWS receives?	☐ Yes ☐ Not applicable ☐ No (Please specify why not) ____ ____ ____ ____ ____ ____ ____
3. In the last 12 months how many times did you require respite but were unable to access it?	____________________ ☐ Not applicable
4. Do you want to receive overnight respite?	☐ Yes ☐ No
5. Do you want to receive daytime respite?	☐ Yes ☐ No
Is there any additional information or comments you would like to add about the person with PWS's living circumstances?	

Section N: Impact of Caring for Person with PWS on Families

The toll on family members caring for a person with PWS can be high. This next section has been designed to capture the physical, emotional and financial impact of caring for a person with PWS. The answers you provide in this section will be important in order to highlight the stress placed on parents and carers and identify the support families need to minimise this stress.

1. Have you or your partner ever given up work or reduced your working hours to care for the person with PWS?	☐ I have given up work ☐ My partner has given up work ☐ I have reduced my working hours ☐ My partner has reduced his/her working hours ☐ No	
2. On average, how much per month do you pay for medications for the person with PWS?	€ ________________	
3. On average, how much per month do you pay for appliances/aids for the person with PWS? (For example, orthotics, back braces, CPAP, special footwear, mobility equipment)	€________________	
4. On average, how much per month do you pay for therapies for the person with PWS? (For example, Speech and Language Therapy, Physiotherapy, Occupational Therapy)	€ __________________	
5. On average, how much per month do you spend on travel costs to appointments for the person with PWS?	€ __________________	
6. Has the person caused injury to family members?	☐ Yes ☐ No	
7. Has the person caused injury to a member of the public?	☐ Yes ☐ No	
8. Please rate on a scale of 1-4 the impact that caring for a person with PWS has had on you in the following areas: 1. No Impact 2. A small negative impact 3. A significant negative impact 4. An extreme negative impact	Emotional toll	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Physical toll	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Financial toll	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Family Relationships	☐ 1 ☐ 2 ☐ 3 ☐ 4

9. On a scale of 1 to 10, please indicate where you and your family currently are **right now,** in terms of crisis by picking **one** of the following statements.	☐ 1 Everything is fine, my family and I are not in crisis at all. ☐ 2 Everything is fine, but sometimes we have our difficulties ☐ 3 Things are sometimes stressful, but we can deal with problems if they arise ☐ 4 Things are often stressful, but we are managing to deal with problems when they arise ☐ 5 Things are very stressful, but we are getting by with a lot of effort ☐ 6 We have to work extremely hard every moment of every day to avoid having a crisis ☐ 7 We won't be able to handle things soon. If one more thing goes wrong - we will be in crisis ☐ 8 We are currently in crisis, but are dealing with it ourselves ☐ 9 We are currently in crisis, and have asked for help from crisis services (A and E Department, hospital, community crisis supports) ☐ 10 We are currently in crisis, and it could not get any worse	
The next question asks about the person with PWS's siblings. Please answer the next question if you are the parent of the person with PWS and also have other children. If you are not the parent, please skip to the comment question below.		
10. Please rate on a scale of 1-4. the impact that caring for a person with PWS has had o**n your other children** in the following areas: **1. No Impact** **2. A small negative impact** **3. A significant negative impact** **4. An extreme negative impact**	Physical Health	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Mental Health	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Social Life	☐ 1 ☐ 2 ☐ 3 ☐ 4
	Academic Performance	☐ 1 ☐ 2 ☐ 3 ☐ 4
	The amount of time you spend with them	☐ 1 ☐ 2 ☐ 3 ☐ 4
Is there any additional information or comments you would like to add about caring for a person with PWS?		

Section O: Housing Needs In this section, you will be asked questions about access to housing and other living supports. The answers you provide in this section will help us advocate for improved housing services for people with PWS in Ireland	
1(a) If the person with PWS is currently living in a group home does the person want to stay in this group home?	☐ Yes ☐ No (if "no" is selected please answer 1b) ☐ Not applicable
1 (b) To what type of new residential arrangement does the person want to move?	☐ PWS group home ☐ Mixed group home ☐ Independent with support ☐ Don't know ☐ Other (Please Specify) ______________________________ ______________________________
2. If the person with PWS is not currently in a group home, when, if ever, do you think the person with PWS will require a place in a group home?	☐ Now (urgently) ☐ In the next 6 months ☐ In the next 2 years ☐ In the next 5 years ☐ In the next 20 years ☐ Not required ☐ Don't know
3. What type of longterm residential arrangement do you think would be most appropriate for the person with PWS? *If one or more of the options would be appropriate, please rank them 1, 2 and 3 in terms of preference*	☐ PWS group home ☐ Mixed group home ☐ Independent with support ☐ Don't know ☐ Other ______________________________
Is there any additional information or comments you would like to add about the person with PWS's Housing needs?	

Section P: Future Research

The purpose of this study is to assess the needs of people with PWS. Are there any areas or issues that are not covered in this survey that you think should be mentioned? (Please fill in on the lines below)

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Thank-you very much for taking the time to take part in our research study. The answers you have provided will help us advocate for improved health, education and social care aspects of service provision for people with PWS. We hope to continue our PWS research to further improve the lives of people with PWS.

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