Prader-Willi Syndrome and the National Disability Insurance Scheme

A report prepared by the University of Melbourne for the PWS Better Living Foundation Ltd in collaboration with the Prader-Willi Syndrome Association of Australia Inc

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EXECUTIVE SUMMARY

This report provides recommendations to guide the allocation of services and funds to people with Prader-Willi Syndrome (PWS) and their families under the National Disability Insurance Scheme (NDIS). PWS is a rare and highly complex genetic condition that affects appetite, growth, intellectual capacity and psychological well-being including aggression and violence.

This report concludes that the five separate factual matters a person has to meet for NDIS access (listed in s.24 of the NDIS Act), have been illustrated for all people with PWS.

People with PWS have multiple impairments, and:

- have a disability that is attributable to one or more intellectual, cognitive, neurological or physical impairments, and
- the impairments are permanent, and
- the impairments result in substantially reduced functional capacity to undertake, or psychosocial functioning in undertaking many activities, and
- the impairments affect the person's capacity for social and economic participation, and
- the person is likely to require support under the National Disability Insurance Scheme (NDIS) for their lifetime.

Recommendations for support were based on the results from an online survey, completed by 106 primary carers for people with PWS, telephone interviews with 21 of these carers and 6 health professionals and support providers.

Recommendations cover

- The toll of psychological and social problems on the individual with PWS including maladaptive and autistic behaviours and violence
- Social and emotional impact on the family
- Financial burden

- Dietary management
- Activities of daily living
- Community participation
- Living out of the family home support
- Education and
- Communication.

Recommendations are based on the NDIS core, capacity and capital model. Core supports are those required by all people with the disability at a given life stage. Capacity supports are those required by some individuals, depending on their goals. Capital describes funding for equipment, home modifications and other one-off costs.

The central recommendations based on the NDIS model are identified across the key areas of core, capacity and capital supports (Table 1) and further summarised to identify the needs across the lifespan (Table 2).

It is hoped that these recommendations will clarify understanding of the needs of individuals with PWS and assist with their interactions with the National Disability Insurance Agency (NDIA).

Table 1: Summary of recommendations for core, capacity and capital support services required for individuals with PWS

Core supports	Capacity supports	Capital supports
Social and activity groups	Pre-emptive psychologist advice for families with preschool aged children with PWS	Exercise equipment, gym memberships and personal trainers
Psychology services, beginning in the primary school period	Counselling and/or psychological therapy for family members	Specialised equipment and home modifications
Allied health practitioners with with an understanding of the PWS condition – Physiotherapy – Dietician, beginning during the preschool period – Dentistry – Occupational therapy – Speech therapy	Day programs which include physical activity and skills training for individuals not attending full time schooling or employment	Equipment and/or home modifications to ensure food can be locked away within the home environment
Appropriate exercise programs and carers to facilitate participation	Outside school hours care	Electric toothbrushes
Assistance to gain employment for people with PWS over 15 years of age	Full time supervision within the community for individuals who display food seeking behaviours	Orthotics and other mobility aids
Personal care assistance for individuals living out of the family home	Carer support when travelling to and participating in employment	Security aids
Respite care	Carer support for full time supervision within the home environment	
Carer support to allow the individual to participate in community activities such as attending schooling, a day program, employment and medical appointments	Disability support workers who are trained in the care of people with PWS to provide substantive and meaningful support programs in place of babysitting services	
Appropriate accommodation supported by carers trained in managing PWS for individuals over the age of 18	Personal care assistance for individuals living in the family home	

Table 2: Summary of recommendations for core, capacity and capital support services required in each group of individuals with PWS, based on age and living situation.

Report subheadings	Preschool	Primary	Secondary	Adult living in the family home (ALH)	Adult living in supported accommodation (ALSA)
Allied Health					
Psychology for person with PWS	Capacity	Core	Core	Core	Core
Psychology for families	Capacity	Capacity	Capacity	Capacity	Capacity
Physiotherapy	Core	Core	Core	Core	Core
Expert dietician management	Core	Core	Core	Core	Core
Occupational therapy	Core	Core	Core	Core	Core
Speech therapy	Core	Core	Core	Core	Core
Podiatry	Core	Core	Core	Core	Core
Self care					
Personal care assistance in the home	Core	Core	Core	Core	Core
Electric toothbrushes	Capital	Capital	Capital	Capital	Capital
General tasks and community life					
Social groups	Core	Core	Core	Core	Core
Access to exercise programs & gym memberships	Core	Core	Core	Core	Core
Day program	Capacity	Capacity	Capacity	Core	Core

Outside school hours care		Capacity	Capacity		
Exercise equipment, gym membership, personal trainers	Capital	Capital	Capital	Capital	Capital
Mobility aids and orthotics	Capital	Capital	Capital	Capital	Capital
Security aids	Capital	Capital	Capital	Capital	Capital
Supervision for travel to school		Capacity	Capacity		
Accommodation					
Full time supervision in the home environment	Capacity	Capacity	Capacity	Capacity	Core
Supported accommodation specific to PWS	Capacity	Capacity	Capacity	Core	Core
Specialised equipment & home modifications	Capital	Capital	Capital	Capital	Capital
Home modifications to secure food	Capital	Capital	Capital	Capital	Capital
Employment					
Employment assistance			Capacity	Capacity	Capacity
Carer support to attend employment			Capacity	Capacity	Capacity
Family supports					
Counselling/psychology for the family	Capacity	Capacity	Capacity	Capacity	Capacity
Respite care	Core	Core	Core	Core	

BACKGROUND

Prader-Willi Syndrome (PWS) is a multi-system disorder associated with the absence of a paternally-inherited gene usually expressed on chromosome 15 (Angulo, Butler & Cataletto, 2015). It has been estimated to affect 1 in 10,000 - 30,000 people (Angulo, Butler & Cataletto, 2015), or approximately 400,000 people worldwide and 800 Australians (The PWS Better Living Foundation, 2013a). However, The Royal Australian College of General Practitioners (2012) argues that the incidence in Australia may be closer to 580 individuals, based on a birth prevalence of 1 in 17,000 and average life expectancy of 33.2 years.

Clinical features

Clinical manifestations in infants with PWS include hypotonia, feeding difficulties, failure to thrive and lethargy (Angulo, Butler & Cataletto, 2015). In childhood, clinical features including short stature, delayed growth velocity, developmental delay, cognitive disability and maladaptive and autistic behaviours can emerge (Angulo, Butler & Cataletto, 2015; Butler, Lee & Whitman, 2006). Older children with PWS typically begin to display hyperphagia and food seeking behaviours which, compounded by a typically low muscle to fat ratio and low energy requirements, can result in rapid weight gain (Allen, 2011). Obesity and its complications are major causes of morbidity and mortality for people with PWS (Angulo, Butler & Cataletto, 2015). Some people with PWS experience improvements in appetite and temperament in adulthood, however many continue to suffer from an insatiable appetite and aggression related to food seeking (Angulo, Butler & Cataletto, 2015; Miller et al., 2011). Impaired speech and communication are also common across the lifespan in individuals with PWS.

PWS is most commonly associated with a genetic deletion abnormality or uniparental disomy (genetic duplication) abnormality. It is thought that the genetic cause underlying the condition influences the behaviours exhibited by the person. Uniparental disomy has been demonstrated to be associated with significantly greater presentation of Autistic symptoms and motor control deficits when compared to cases of deletion (Veltman et al., 2004). It is also suggested that those with Uniparental Disomy may have greater impairments in visuospatial processing and therefore have greater care needs for assistance with daily activities (Veltman et al., 2004).

Diagnosis

The diagnosis of PWS is currently based on genetic testing for children, while previously most individuals were diagnosed from clinical symptoms (Holm, 1993; Lionti et al., 2014). As a result of the early DNA testing, individuals with PWS are being diagnosed typically within 8.6 weeks of birth (Lionti et al., 2014). Early diagnosis allows families time to organise management strategies for diet and growth hormone replacement therapy, which is crucial to the health and development of the child. It is suggested that all infants that display symptoms including severe hypotonia, genital hypoplasia and failure to thrive should be genetically tested for PWS (Goldstone et al., 2008). On the other hand, adolescents should be tested if they suffer from a severe cognitive impairment, excessive eating and maladaptive behaviours (Cassidy et al., 2012).

Management

Infants who display poor sucking and severe hypotonia are often managed with tube feeding, although within this population there is no evidence on the optimal feeding regime (Goldstone et al., 2008). The treatment of infantile hypotonia is often managed by physiotherapists. Exercise programs designed throughout the development period are the most effective management plan to prevent poor muscle mass. The most important treatment plan early on in the life of a PWS child is education to the parents, including support from the allied health team.

Commonly at approximately 6 years of age, children with PWS transition from an inability to thrive to the onset of hyperphagia that causes obesity. Management of obesity is the priority for individuals with PWS as they increase in age. The management plan involves low calorie, well balanced diet, regular exercise, strict supervision around food, restricted access to food and psychological and behavioural counselling (Goldstone et al., 2008). Allen (2011) identified the key behavioural interventions for managing PWS as restricting access to food, keeping the person occupied and the use of a routine. Parents and carers commonly manage hyperphagia by locking food cabinets and fridges, supervising children with PWS around food and having low-fat snacks available (Goldberg, 2002).

Growth hormone replacement therapy is used in the management of PWS and has been shown to increase muscle mass and decrease fat mass. Infants treated with hormone replacement therapy have been shown to have greater head circumference and height, lower body mass index, improved body composition, and better acquisition of motor skills and language (Cassidy et al., 2012). However, older children and adolescents who are treated with hormone replacement therapy do not show the

same improvements.

As the medical and behavioural management of PWS improves, the population of Australians affected by the condition continues to age (Thomson, Glasson & Bittles, 2006). This is expected to lead to greater demand for supported accommodation for these older individuals. In addition, there is an increased burden on disability support services and healthcare services due to a greater prevalence of age-associated disorders such as diabetes mellitus (Thomson, Glasson & Bittles, 2006).

Support associations

Although the PWS population within Australia is small, there are several support groups available for both families and those with PWS. The Prader-Willi Syndrome Association of Australia (PWSA) is an

Australia-wide support organisation that is available for all members of the community. Victoria, New South Wales and Queensland have active State Associations, while other states and territories have informal support groups. The PWS Better Living Foundation is an Australian not-for-profit organisation which advocates for equal opportunities for people with PWS to live independent of their family in a supported residential setting and to enjoy an active role in their communities (The PWS Better Living Foundation, 2013b). The PWSA and PWS Better Living Foundation are partners in their goals to provide support and solutions to families affected by PWS to access housing and other living supports (The PWS Better Living Foundation, 2013b).

National Disability Insurance Scheme

In 2011, the Australian Government Productivity Commission released a report into disability care and support which determined that disability services were unfairly funded and influenced by factors such as location and cause of disability (Australian Government Productivity Commission, 2011). The report recommended the implementation of a National Disability Insurance Scheme (NDIS) to provide disability support funding based on individualised assessment of need, rather than a standard allocation (Australian Government Productivity Commission, 2011; Mathews, 2012). The NDIS was launched by the Australian Federal Government on the 1st of July 2013 to be progressively rolled out over 5 years (NDIS: Transforming Lives, 2014b). It is a Federal Government initiative that aims to provide individualised and coordinated supports and services for all Australian citizens with a significant and permanent disability (NDIS: Transforming Lives, 2014a). The NDIS is currently in the early transition phase for selected age groups eligible in selected regions of each state and territory with the roll out of the full scheme to commence progressively from July 2016 (NDIS: Transforming

Lives, 2014b).

The NDIS aims to enable individuals to control how, when and where supports are provided to maximise their independence (Australian Government, 2015). The National Disability Insurance Agency (NDIA) was established to administer the NDIS and oversee the operation of the scheme. Supports are divided into core, capacity and capital for each disability category (NDIS, 2015a; see Table 3) Core support items are those required by all people with a certain disability at a certain life stage, regardless of severity of condition, to allow individuals to participate in daily activities and enable them to work towards goals and aspirations. Capacity building support items allow some individuals to build independence and skills to achieve realistic goals. Capital supports describe funding for equipment, home modifications and other temporary funding requirements (NDIS, 2015a). Funding support is organised into the domains of education, employment and housing and independent living (NDIS, 2014; see Table 4). It is hoped that the NDIS will allow more Australians with a disability to enter higher education and the workforce due to a greater focus on early intervention services and support for schooling (NDIS: Transforming Lives, 2014c)

Table 3: Support categories as determined by the National Disability Insurance Scheme (2015a).

Support category	Description
Core	Required by all people with the disability at a given life stage
Capacity	Required by some individuals, depending on their goals in life
Capital	Funding for equipment, home modifications and temporary equipment requirements

For individuals to qualify for access to the NDIS they must fulfil the five criteria detailed in the NDIS Act (2013). These criteria include:

1. The person has a disability that is attributable to one or more intellectual, cognitive, neurological, sensory or physical impairments or to one or more impairments attributable to a psychiatric condition and
2. The impairment(s) are likely to be permanent, and
3. The impairment(s) result in substantially reduced functional capacity to undertake, or psychosocial functioning in undertaking, one or more of the following activities:

a. Communication

b. Social interaction

c. Learning

d. Mobility

e. Self-care

f. Self-management, and

4. The impairment(s) affect the person's capacity for social and economic participation, and
5. The person is likely to require support under the National Disability Insurance Scheme (NDIS) for the person's lifetime.

A major challenge of the NDIS is building the capacity of individuals and their families and carers to make informed choices about how they use their support package. Information packages regarding the NDIS have thus been developed by advocacy groups such as Epilepsy Foundation Victoria (Disability Care Australia, 2013) to assist people in navigating the NDIS and obtaining the supports and services that are most appropriate. Currently, no such advice exists to assist the relatively small population of Australians living with PWS.

This report is designed to directly address the NDIS Operational Guidelines for Access (NDIS, 2014) including how the needs of people with PWS might align with the purpose of the funded supports and the NDIA Outcomes Framework (NDIS, 2015b). It will be used by carers of people with PWS, Australia's National and State PWS Associations, the PWS Better Living Foundation and the NDIA to inform the application for and allocation of disability support services under the NDIS in Australia.

Table 4: List of funding support proposed to be provided under the NDIS scheme (adapted from NDIS website, 2014)

Support category	NDIS funding support
Education	**Preschool** ▫ Assistance with self-care at school (e.g. eating, mobility) ▫ Early intervention supports to prevent deterioration and improve functional capacity (e.g. speech therapy, physiotherapy) ▫ Behavioural support ▫ In special cases where the early childhood system cannot intervene, personalised supports to assist the child to attend an early learning service **Primary and High school** ▫ Assistance with self-care at school (e.g. eating, mobility) ▫ Special transport to school ▫ Transportable equipment e.g. wheelchair ▫ Support to transition between schools **Adult** ▫ Support with education, training and occupation options following completion of schooling
Employment	**Adult** ▫ Assistance to find and retain employment ▫ Supported employment ▫ Assistance with capacity building for those not eligible for Disability Employment Services and Job Services Australia ▫ Assistance with personal care ▫ Assistance with transport
Housing and independent living	**Adult** ▫ Capacity building for independent living (e.g. living skills training, money and home management, social and communication skills) ▫ Home modifications (own or rental) ▫ Support with personal care ▫ Domestic assistance ▫ Funding for supported accommodation, where the cost is greater than normal rent ▫ Other costs are covered under the scheme only when incurred as a result of disability

AIMS

This project aims to illustrate the drastic impact and permanent impairments associated with PWS on the individual and family and to demonstrate the need for PWS to be considered within the NDIS List A conditions as stipulated within the Operation Guidelines for access and disability requirements (NDIS 2014. Operational Guidelines). It also aims to identify the services frequently required for a life of dignity and independence for individuals with PWS and to map the support needs of individuals affected by PWS through the different life stages. Ultimately the findings and recommendations will be used to guide further discussion with the NDIA regarding appropriate funding requirements to meet the detailed needs and supports of individuals with PWS based on the core, capacity and capital model.

Methods

A systematic search of the current literature regarding PWS and the Australian NDIS was performed. The Medline, Cinahl, Cochrane and Embase databases were searched on 13th August 2015 using the search terms; ("Prader-Willi Syndrome" OR "PWS") AND ("NDIS" OR "national disability insurance scheme" OR "disability support" OR "disability services"). Results were limited to literature published after 2005. Each of the three authors independently reviewed the results of the searches then discussed how a quantitative and qualitative research report could add to the current literature available in this area.

To rigorously address gaps in the current literature including support requirements, carer expectations of the NDIS and access to NDIS schemes data was collected from three different sources i) online survey ii) follow-up telephone interviews and iii) written feedback.

i. Online survey

An online survey using SurveyGizmo was developed (see Appendix 1) for primary carers of people with PWS to explore their experiences and perceived support requirements for the affected individual. The survey was emailed to affiliates of the PWS Better Living Foundation including the PWSA branches in each state and territory with a request for primary carers of children and adults with PWS to respond. Respondents were asked to provide their name and contact details if they consented to provide further information in the form of a telephone interview. A total of 106 responses were collected using the online survey. Identifying data including name and contact details were collected

and stored separately to the results of the survey to de-identify the information. Some questions asked participants to rate the extent to which a certain behaviour was a problem or the extent to which the person required assistance for a certain task using an 11-point numeric rating scale. Where respondents indicated there was no problem or the person required no help, they were asked whether this was because the question was not relevant due to the person's age or capabilities. If the respondent indicated the question was not relevant, the response to the particular question was not included in the analysis. For the purposes of this report, responses were divided into five groups according to the age and living situation of the person with PWS; preschool, primary school, secondary school, adult living within the family home (ALH) and adults living in supported accommodation outside the family home (ALSA).

All online survey data were reviewed and analysed using Microsoft Excel. Means and standard deviations were reported and relevant data are graphed below.

ii. Telephone interviews

A total of 21 carers participated in semi-structured telephone interviews (see Appendix 2) which were of approximately 30-60 minutes in duration. The participants were chosen using a convenience sample of those first to respond to the online survey and consent to the interview and aiming for a spread across the lifespan. Carers were interviewed by one of three researchers who had no previous relationship with the participants. The telephone interviews further explored participant experiences with PWS including physical activity, uncontrolled eating, behavioural issues and use of disability support services. Carers of people with PWS over the age of 18 were also asked about experiences regarding the person with PWS living out of the family home or considering doing so, as well as interactions with the police, legal or paralegal services and hospital admissions. Telephone interviews were recorded using a secure hotline to allow the researchers to identify themes and transcribe important quotations. The recordings were later deleted.

A thematic analysis was completed independently by all three researchers to evaluate the data collected in the telephone interviews. The themes were then collated and agreed upon based on consensus among all three researchers. Quotations from telephone interview participants are used throughout the discussion of the results to further support the findings.

iii. Written feedback

Health professionals with knowledge of PWS and its management were also invited via email to provide written feedback on PWS supports they perceived to be critical to the wellbeing of the individuals with PWS and their families across the lifespan. Six health professionals submitted written comments. This data was included to enrich the responses provided by the families and carers of PWS individuals and to provide a health professional viewpoint.

iv. NDIS domains

Recommendations were made for each NDIS domain according to previous research regarding PWS together with the results of the online survey and telephone interviews. Supports were considered to be a core need if greater than 90% of survey participants indicated that the person with PWS displayed issues with regard to this behaviour or task. Where less than 90% of participants indicated that the particular behaviour or task was an issue for the person, the support was deemed to be either a capacity requirement if required by certain groups of individuals or largely unnecessary.

v. Ethics approval

Ethics approval for this project, inclusive of the electronic survey and telephone interviews, was granted by the University of Melbourne Human Research Ethics Committee on 3rd August 2015. All survey respondents and telephone interview participants were asked to confirm that they had read the Plain Language Statement (see Appendix 3) and indicate their consent to participating in the research project.

RESULTS – ONLINE SURVEY

The survey results have been reported to directly address the disability requirements as stipulated by the NDIS (listed in s.24 of the NDIS Act 2013)

Personal characteristics of person with PWS

A total of 106 primary carers for a person with PWS across Australia participated in the online survey. The majority (83%) of primary carers were the mother of the person with PWS.

Personal characteristics of the person with PWS according to age group and living situation are described in Table 5. There was a greater percentage of males represented in the sample (63%) due to chance as PWS is not known to affect either sex preferentially (Angulo, Butler & Cataletto, 2015; Foundation for Prader-Willi Research, 2014).

According to The Australian Government Department of Health (2015) and World Health Organisation (2000) body mass index (BMI) categories, on average, the secondary school and adult living out of the family home with support (ALSA) groups were moderately obese and the adults living in the family home (ALH) were severely obese. Of the secondary and adult age groups, 43% of respondents reported a BMI of severely obese and higher. Two respondents within the preschool age group (13%) also reported a BMI within the severely obese category. However, the large standard deviation (SD) values indicate a wide variation of BMI scores within all age groups.

Fifty three percent of respondents cared for a person with deletion PWS, 36% cared for a person with uniparental disomy and 11% of cases were of unknown genetic cause. This is similar to reports that 70% of cases of PWS are the result of deletion and 30% are due to uniparental disomy (Copet et al., 2010; Foundation for Prader-Willi Research, 2014).

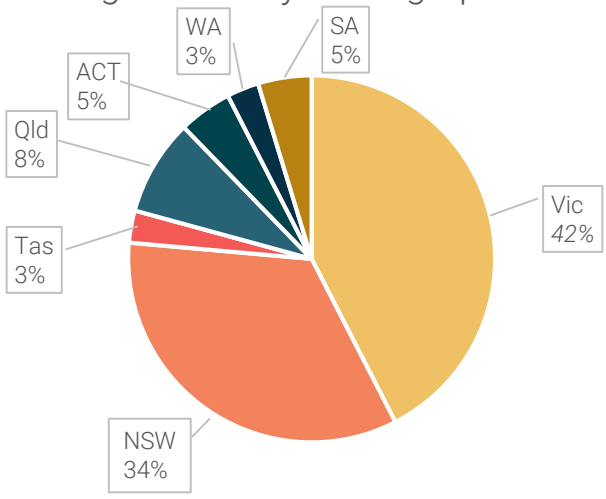
Table 5: Characteristics of the person with PWS for whom participants in the online survey cared for. Values are given as mean (standard deviation) except where percentages are indicated.

Group	Preschool	Primary	Secondary	Adult living in the family home (ALH)	Adult living out of the family home with support (ALSA)	Overall
Number of participants	15	22	13	38	18	106
Age (years)	2.4 (±1.7)	8.4 (± 2.1)	15.2 (± 2.1)	26.0 (± 7.9)	31.1 (± 7.9)	18.5 (± 11.9)
Male gender (%)	73%	59%	31%	50%	89%	63%
Body Mass Index (BMI)	20.7 (± 10)	20.4 (±8.1)	32.4 (±14.3)	44.0 (± 31.9)	34.0 (± 7.9)	33.2 (± 22.4)
Type of PWS (%):						
Deletion	20%	45%	77%	68%	39%	53%
Uniparental Disomy	67%	50%	15%	18%	44%	36%
Unknown	13%	5%	8%	13%	17%	11%

Demographics of person with PWS

Across the age groups, 42% of respondents were living in Victoria, 34% in New South Wales, 8% in Queensland, 5% each in South Australia and the Australian Capital Territory and 3% each in Tasmania and Western Australia. There were no respondents living in the Northern Territory (*Fig. 1*).

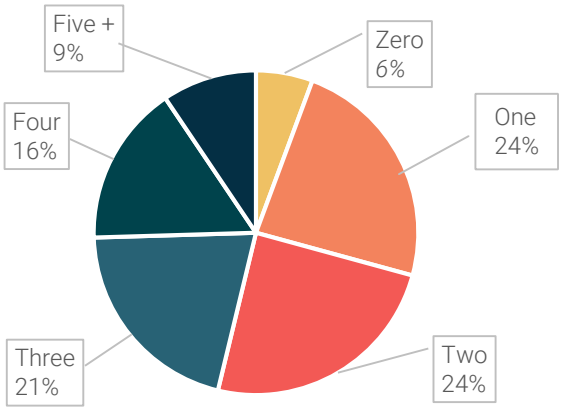
Fig. 1 - Survey Demographics



Medical practitioner usage

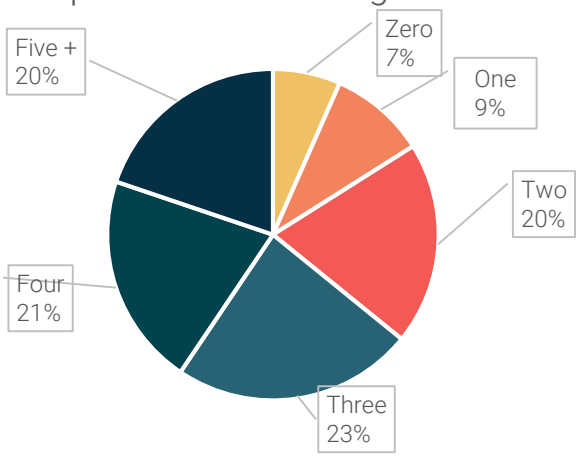
Most individuals with PWS were currently seeing at least one medical specialist. Medical specialists included Endocrinologists, Psychiatrists, Orthopaedic surgeons and Ophthalmologists. Preschool and primary groups were most likely to be seeing three specialists, the secondary group most likely to be seeing two specialists and the adult groups most likely to be seeing one medical specialist. Only 6% of respondents were not seeing a medical specialist while 24% were seeing one specialist, 26% seeing two specialists, 21% seeing three specialists, 16% seeing four specialists and 9% indicated the person was currently being managed by five or more medical specialists. (*Fig. 2*)

Fig. 2 - Number of medical specialists being seen



A high utilisation of allied health professionals was also reported including Physiotherapists, Dieticians, Dentists, Occupational Therapists, Audiologists, Podiatrists, Exercise Physiologists and Psychologists. The preschool and primary groups were most likely to be under the care of five or more health professionals followed by secondary school; ALSA and ALH groups seeing two to four, four to three and three allied health professionals respectively. Only 7% of respondents indicated the person was seeing no allied health professionals with 20% of individuals with PWS currently seeing five or more health professionals. (*Fig. 3*)

Fig. 3 - Number of allied health professionals being seen



Medications

Eighty-seven percent of individuals with PWS were taking at least one medication including growth hormones, sex hormones and psychotropic. Growth hormones were reported to be taken by 44% of all PWS individuals, with the rates highest in preschool, primary and secondary, 73%, 95% and 62% respectively. Sex hormones and psychotropic medications were also widely used in the secondary and adult groups. (*Fig. 4*)

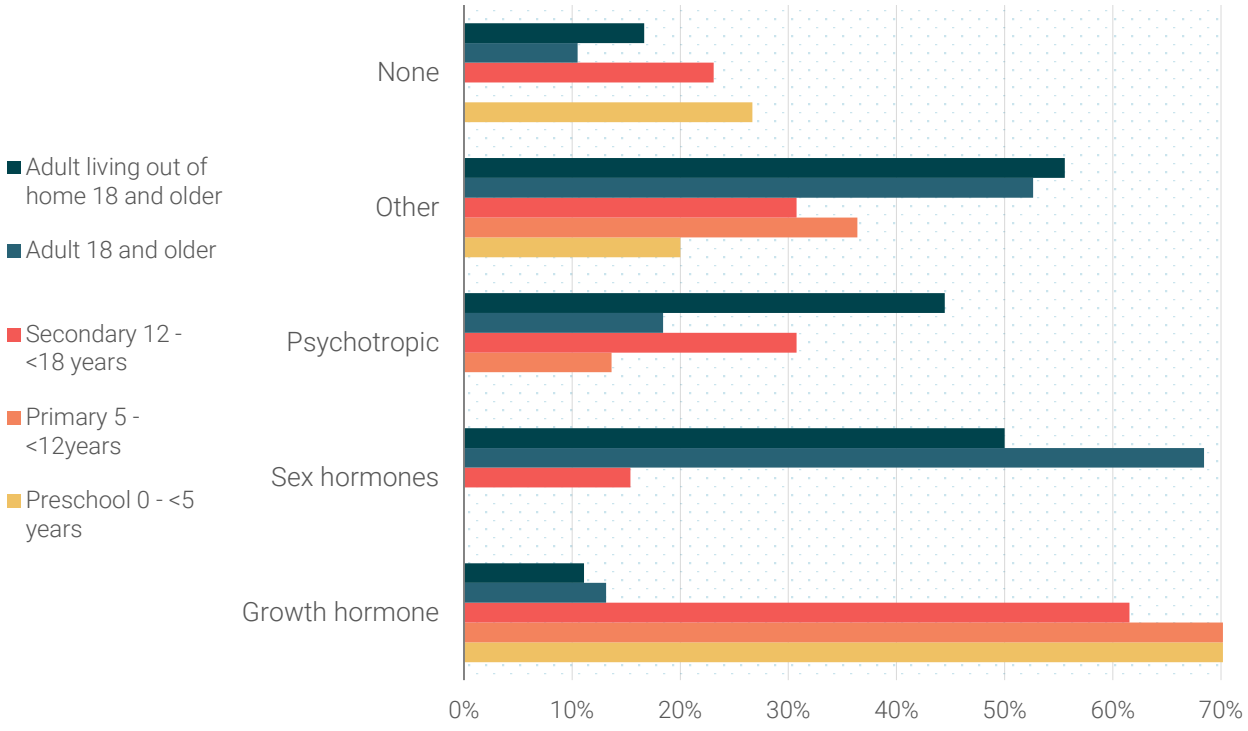


Fig. 4 - Medications

Intellectual, cognitive, sensory and physical impairments associated with PWS

vi. Associated medical conditions

Within the sample several medical conditions were identified to be associated with PWS. Scoliosis was common, particularly in the older age groups, reported by 57% of adult participants (ALH and ALSA combined) and 46% of all participants. Visual impairment was also common across all age groups affecting 35% of participants. Diabetes was increasingly common in older age groups affecting 8% in the secondary school group and 27% in the combined adult groups (*Fig. 5*). This is unsurprising given the higher BMI scores of these groups.

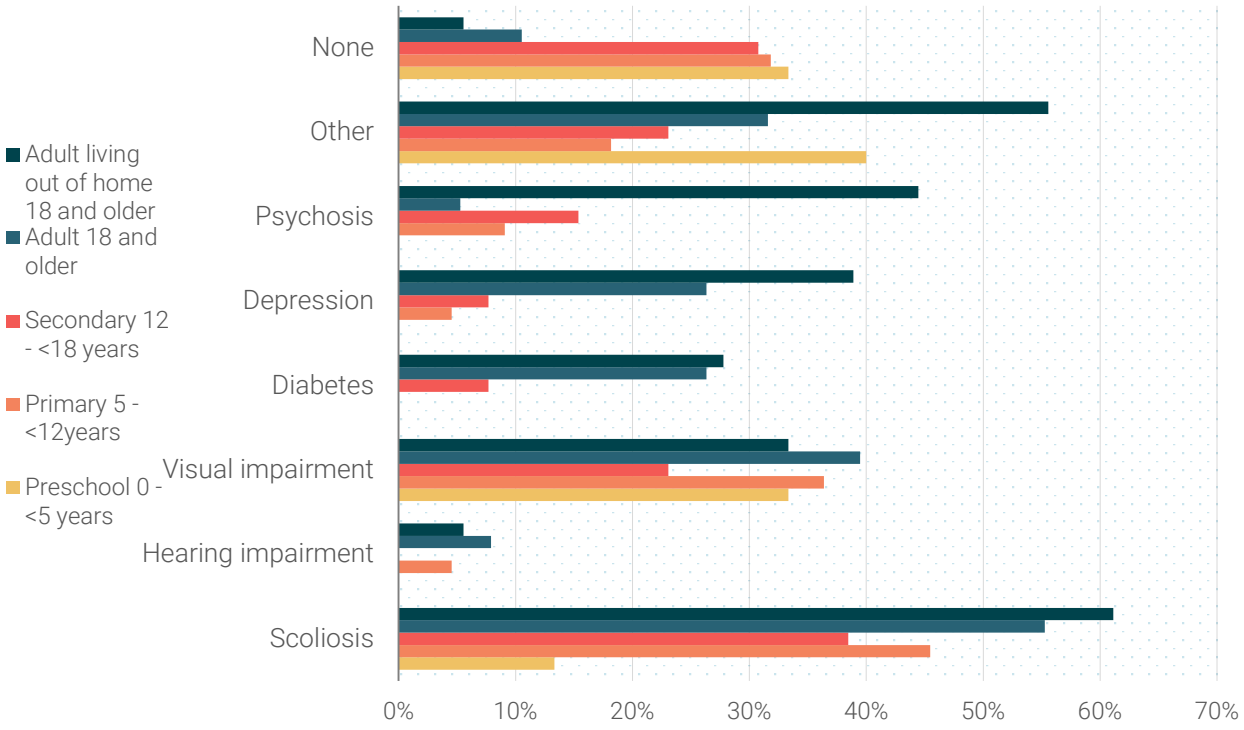


Fig. 5 - Associated medical conditions

vii. Mental health

Mental health issues were identified in all age groups with the exception of preschoolers. Depression became more common in older age groups, affecting 30% of all adults, 5% in the primary school group and 8% in the secondary school group. Psychosis was also more common in older people with PWS, affecting 9% of primary school, 15% of secondary school and 44% of the ALSA group. Only 5% of the ALH group were reported to be affected by psychosis (*Fig. 6i*). Twenty seven individuals with PWS (25%) were affected by one or both of these mental health conditions.

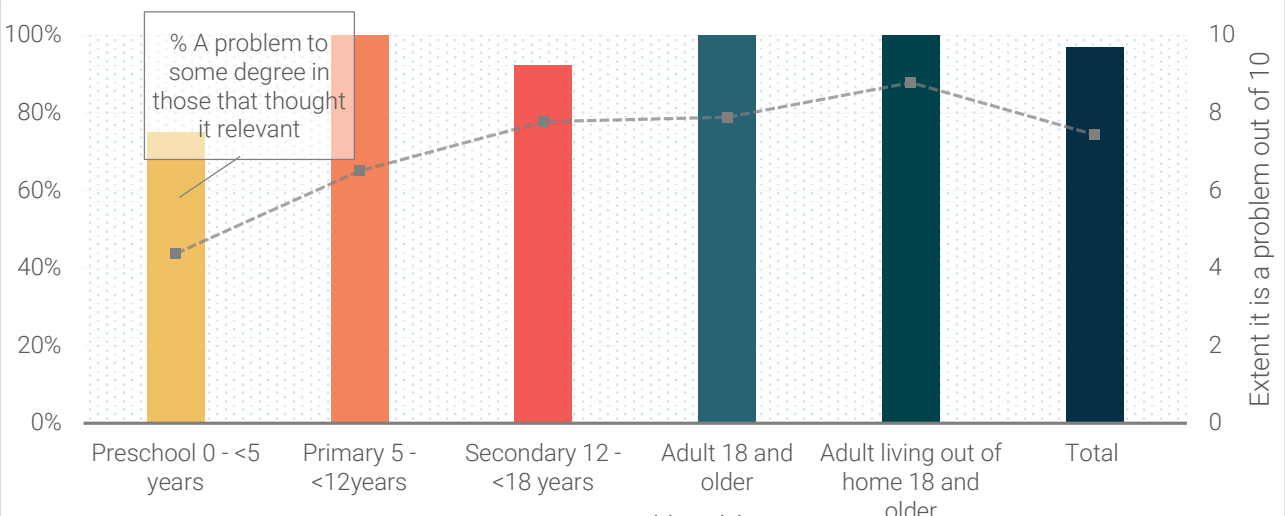


Fig. 6i - Mental health - Anxiety

When looking at low mood and anxiety 98% of individuals of an age where this could be a problem reported this behaviour to some degree. The spread of responses was relatively wide amongst the primary, secondary and adult age groups for both anxiety and low mood. The average rating for anxiety increased incrementally between age groups, suggesting that it becomes a greater problem as the individual ages. The average severity across all age groups of low mood was rated 5 out of 10 (SD 3). Anxiety was greater in severity, with an average rating of 7 out of 10 (SD 3) (Fig. 6ii).

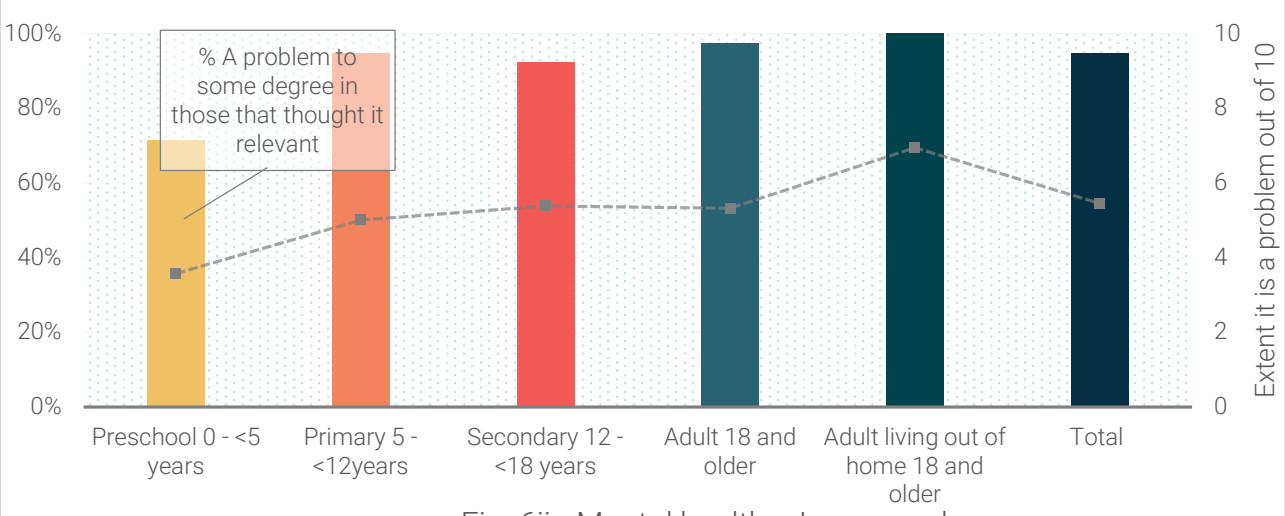


Fig. 6ii - Mental health - Low mood

viii. Food seeking behaviours

Food seeking and associated behaviours for the person with PWS were reported in 98% of all individuals that were of an age to exhibit the behaviour with 100% of secondary school and adult groups exhibiting this to some extent. The adult groups reported high severity of food seeking with an average of 8 out of 10, with 10 being extreme severity. The SD for adult scores was 2, indicating a

relatively narrow spread of answers to this question. Behaviours related to stealing food and money to buy food were reported as a less severe problem than food seeking across all groups. Although less severe a problem, other than stealing money in the preschool group, each of these behaviours was still rated to be a problem to some extent. Participants rated stealing food from plates, lunch boxes, shops and bins an average severity of between 3 and 5 out of 10 depending on the group. Stealing money was most problematic in the ALH group, for which participants rated it an average severity of 6 out of 10.

ix. Skin Picking

Skin picking was reported to affect 89% of individuals that were of an age to exhibit the behaviour, with severity increasing steadily across the age groups and being particularly high in both adult groups averaging 7-8 out of 10.

Communication (understanding and being understood by others)

Ninety seven percent of respondents reported that communication and language was a problem for the individual with PWS if they were of an age to exhibit this as an issue; of this 100% of pre-school, primary and secondary school children exhibited difficulties to some extent with an average severity of 5/10. When specifically looking at the average amount of help required for spoken understanding, written understanding and being understood the spread was high for all groups in each *(Fig. 7)*. In spite of this, it is clear that difficulties with communication exist across all ages, even the adult groups. In total an average rating of 5 out of 10 (SD= 3) was given for spoken understanding, while 7 (SD= 3) was given for written understanding. The extent of help required for being understood was rated an average of 6 (SD= 3) by all.

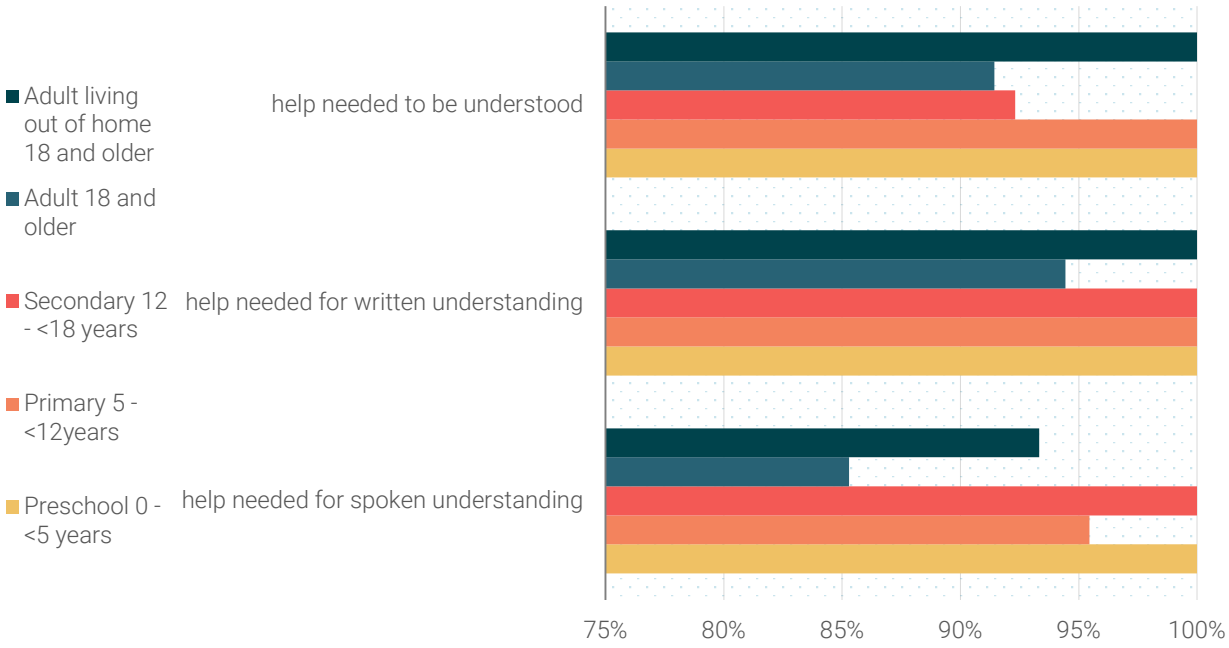


Fig. 7 - Communication

Social interaction (making and keeping friends and relationships, behaving within limits accepted by others, and/or coping with feelings and emotions)

Social behaviour in regard to tantrums and repetitive questioning was identified as a major issue in all age group categories with 98% of respondents reporting these maladaptive and autistic behaviours to be a problem to varying extents.

Repetitive questioning was a major behavioural problem across all age groups with 98% of individuals that were of an age to exhibit the behaviour reporting an issue. All individuals in the primary school, secondary school and ALSA groups exhibited this behaviour. As well as this behaviour being present in nearly all respondents the average severity rating was high at 7 out of 10.

Temper tantrums were exhibited by 100% of all PWS individuals in the primary, secondary and both adult groups at a severity of 6-8 out of 10. Eighty two per cent of pre-schoolers exhibited this behaviour at a relatively low severity of 3 out of 10. *(Fig. 8ii)*

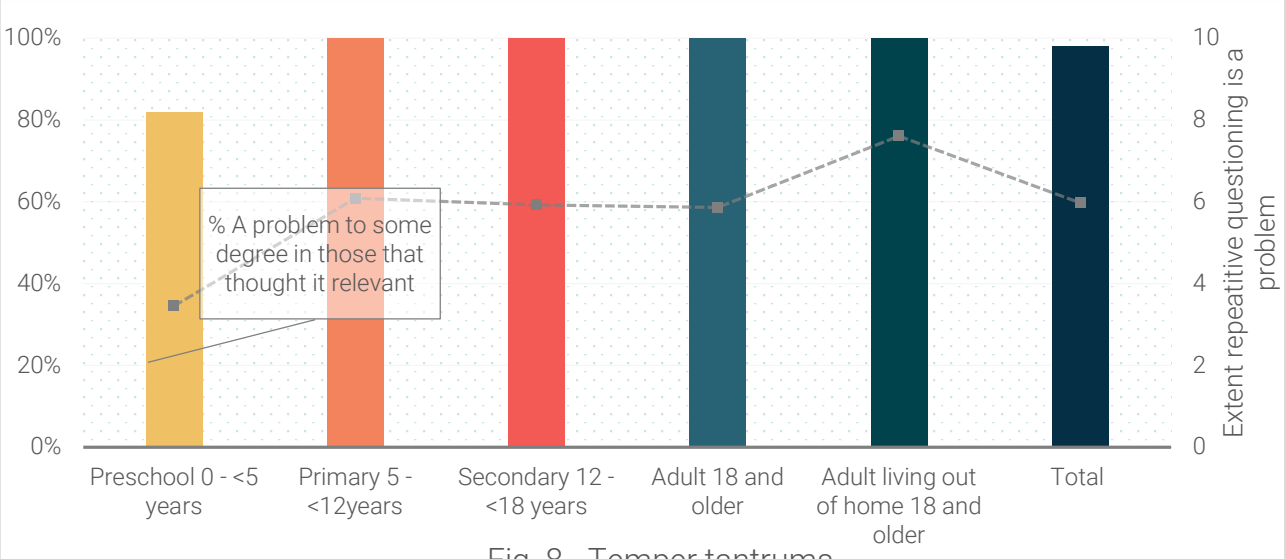


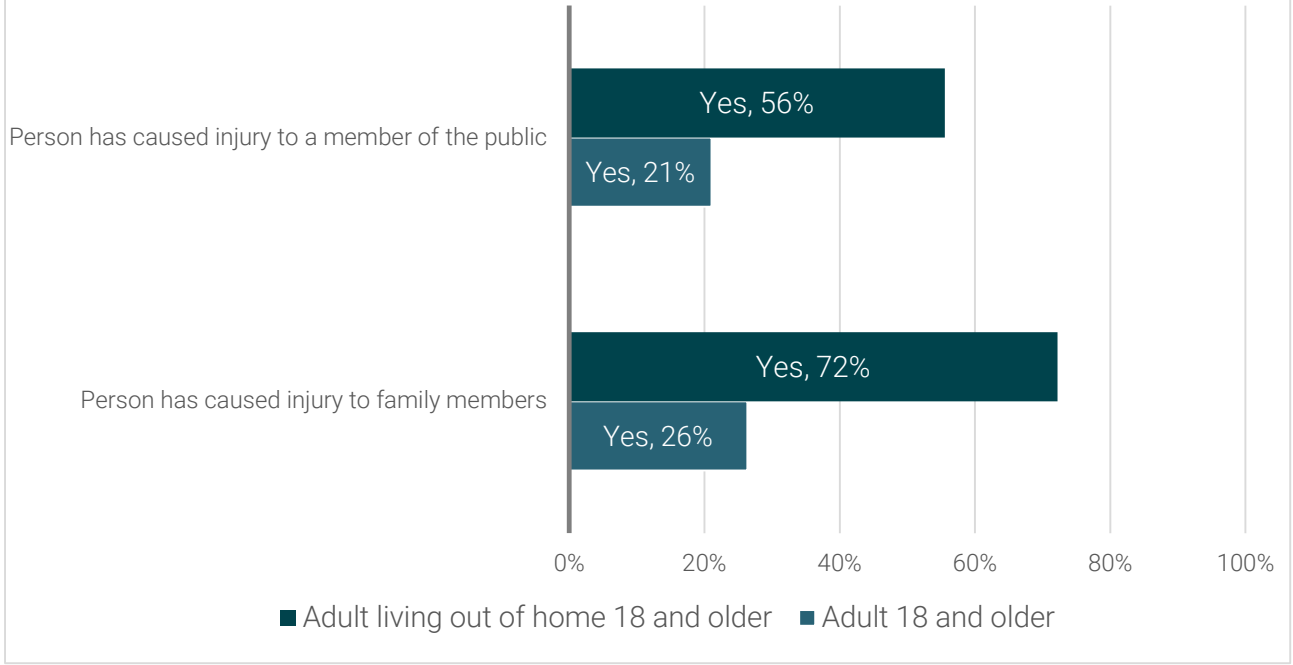
Fig. 8 - Temper tantrums

In developing friendships it was indicated that an individual with PWS required a large amount of help with 100% of primary, secondary and ALSA individuals requiring some degree of assistance. When asked to rate the help required to foster friendships out of 10 with 10 being maximal help, primary school, secondary and ALSA groups required a level of 8 and ALH 7.

In regard to coping with emotional and social problems a staggeringly high burden was identified for the primary, secondary and adult groups where 100% of individuals who were of an age to exhibit this as an issue were having severe difficulties. On a scale of 0 to 10 across all age group categories the average rating for the amount of help required for coping with feelings was severely high across all groups with 8 for primary, secondary and ALH categories and 9 out of 10 for the ALSA group.

Violent behaviour towards others was also identified as a common maladaptive behaviour. There was a dramatic rise in the incidence of violent behaviour from the ALH group to the ALSA group, increasing from 26% to 72% for violence against family members and 21% to 56% for violence against members of the public. *(Fig. 9)*

Fig. 9 - Comparison adults living in and out of home



There was a greater incidence of aggression against family members, compared to against members of the public, across all age groups. In total, 29% reported violence against family members across all age groups, and 22% against members of the public.

Learning (understanding and remembering information, learning new things, practicing and using new skills and ideas)

Ninety-nine percent of participants reported difficulty with remembering or learning tasks, with only one individual reporting no issue. When asked the degree of help an individual needed out of 10 with 10 being maximal help, learning new tasks was rated an average of 7 out of 10 (SD= 2) and remembering 5 (SD= 3) *(Fig. 10)*.

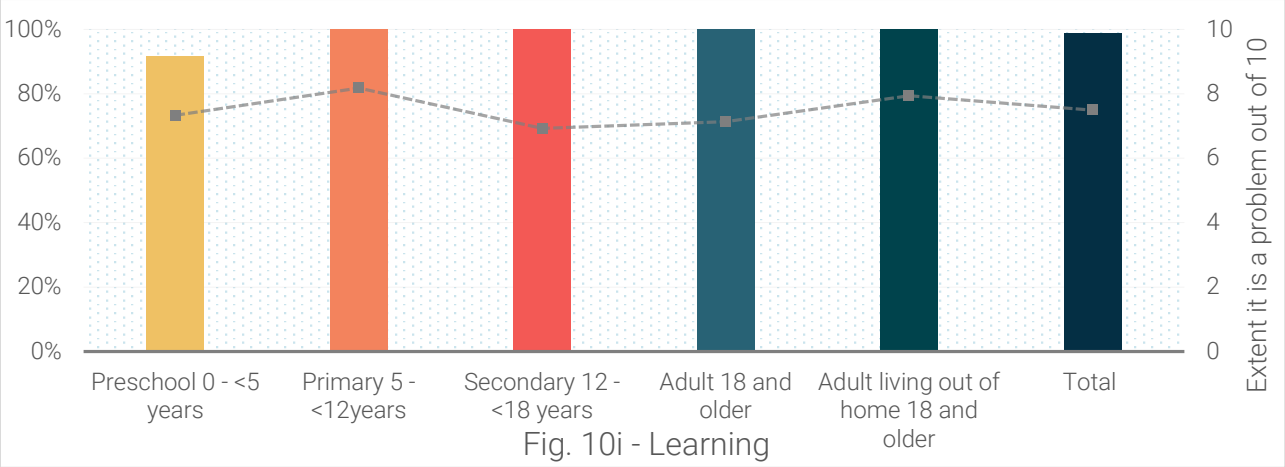


Fig. 10i - Learning

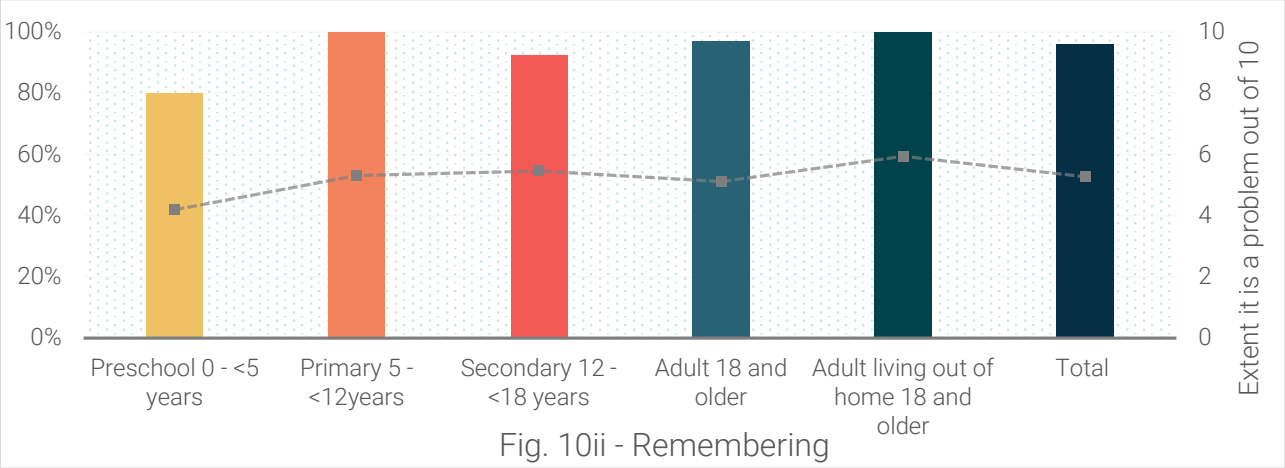


Fig. 10ii - Remembering

Mobility (moving around their home and community and/or performing other tasks involving movement, e.g. using hands and arms)

Mobility was identified as a major problem for all age groups with 100% of individuals needing some degree of assistance. When looking at mobility in the home more assistance for mobility was required within the preschool group with an average of 6 out of 10 with a SD of 2. After the preschool age, mobility assistance was required to the same extent for all groups. Individuals with PWS require significant help getting around the community at all age groups with scores averaging greater than 6 out of 10. *(Fig. 11)*

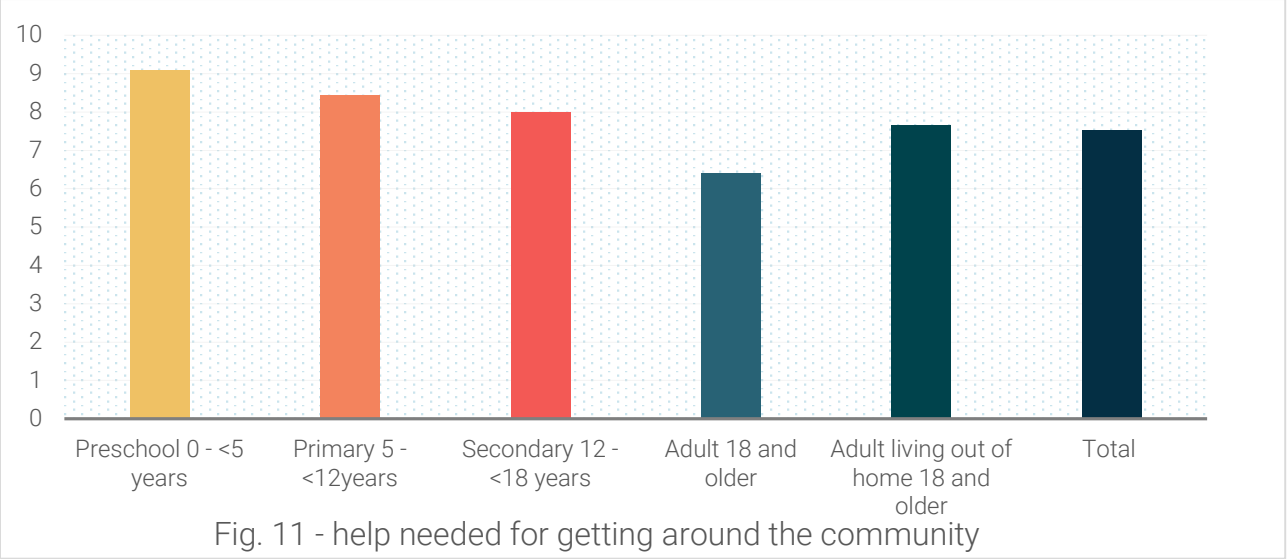


Fig. 11 - help needed for getting around the community

In regard to community engagement the person with PWS required a significant amount of help getting around the community and with road safety with 98% and 100% of individuals requiring assistance, respectively. In all groups, the average degree of help was rated high at 8 out of 10 for both tasks.

Self-care (e.g. daily showering, bathing, dressing, eating, toileting and grooming; and/or special health care needs attended to by self, family members or carers)

Within all groups personal care was a significant issue with 99% of all respondents requiring help with activities of daily living. Only one individual in the ALH group was reported as independent, although still requiring verbal reminding. The preschool group overall required the most assistance with all personal activities of daily living, which due to the age of the individuals within this group, is not surprising. The care needs trended down after primary school, although the needs increased again within the ALSA group. Tasks requiring the most assistance overall by carers were showering/bathing and cleaning teeth both averaging 6 out of 10 with a SD of 3.

Of individuals who are either living in a group home or living with supports 100% of individuals required help with dressing and cleaning teeth, while 93% required assistance with showering and bathing. Furthermore 81% of individuals required assistance with toileting. *(Fig. 12)*

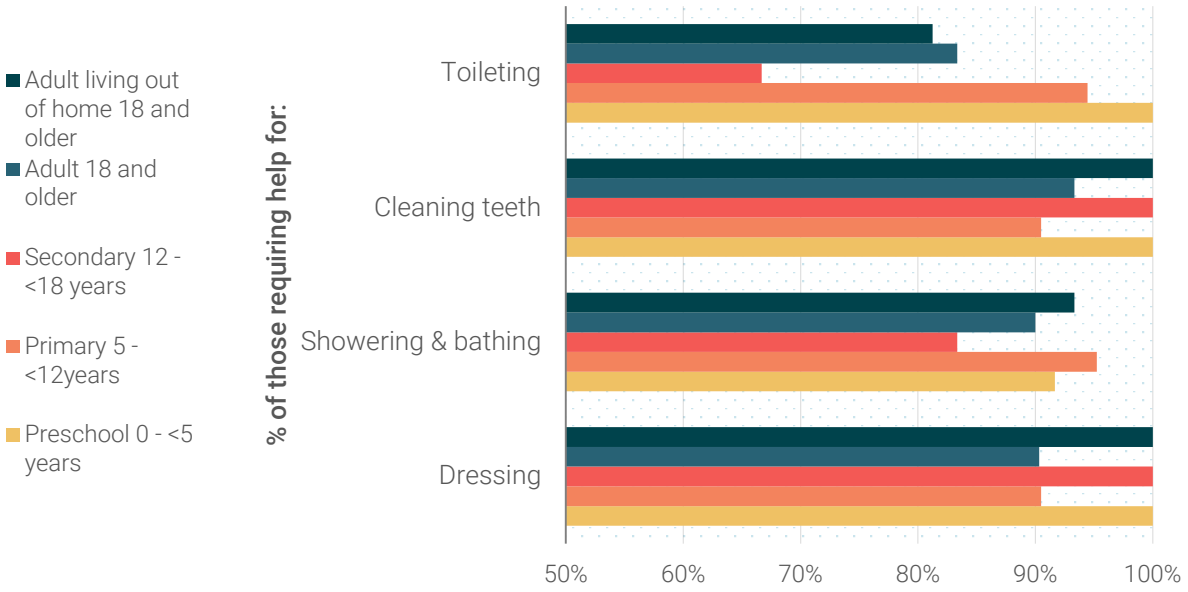


Fig. 12 - Activities of daily living

Self-management (planning and organising daily life and managing household personal finances)

Assistance was required to some extent across all areas of self-management for all individuals with PWS (100%).

When looking at the domains of self-management including managing health, managing money, performing daily tasks around the house, problem solving and making decisions a high degree of assistance was required over all age groups ranging from 7-9 out of 10, with only a small amount of variance in responses. *(Fig. 13)*

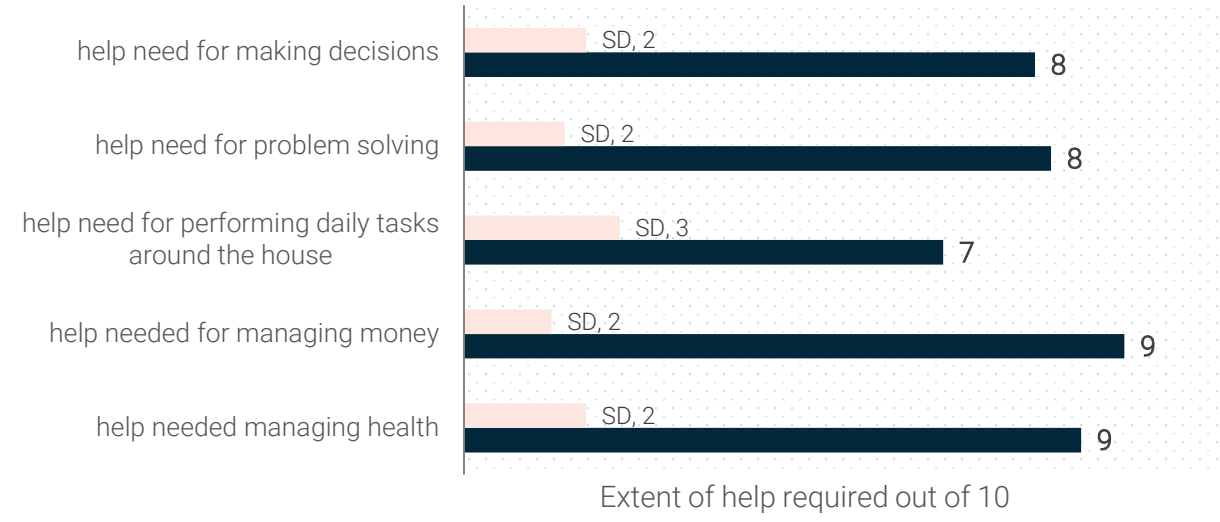


Fig. 13 - Help with self management tasks

An individual's social and economic participation (including their ability to work)

i. Housing

Participants who indicated that the person with PWS was living in the family home were asked about the time frame in which housing for the person is an issue. 82% of all participants of the survey indicated that housing would be an issue for the person with PWS at some point in the future. In the preschool and primary groups, the most common selection was "in the next 20 years". In the secondary group, 77% of responses were that housing would be an issue in the next 5 years. For the ALH group, 13% of carers reported that housing was an issue now (urgently), 11% indicated it would be an issue in the next 6 months, 24% in the next 2 years and 42% in the next 5 years.

Participants who indicated that the person with PWS was living in the family home also identified a need to have access to short term supported accommodation to support the person to live out of the family home *(Fig. 14)*.

In the adult groups, there remained a significant reliance on parents and siblings as carers for the person with PWS.

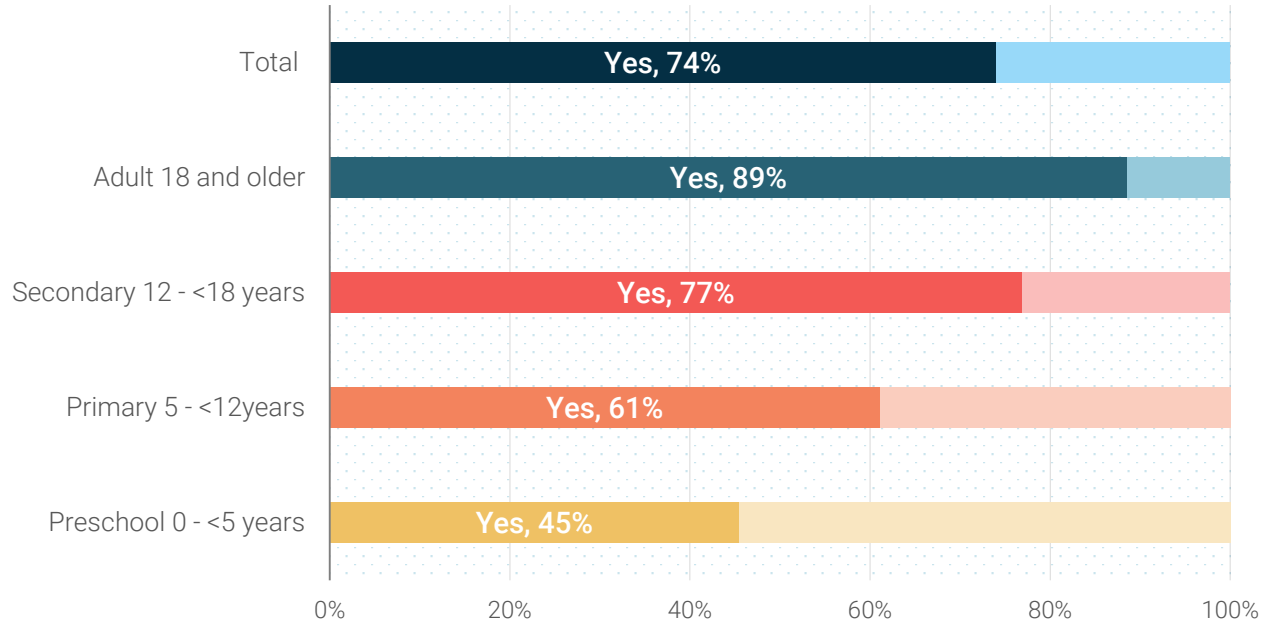


Fig. 14 - Desire for access to short term accomodation

ii. Education

Participants were also asked about the education the PWS individual was currently undertaking. Of the primary school age individuals 82% were receiving primary school education including attending mainstream school without an aid (11%), mainstream school with an aid (44%), special school (39%) and part primary-part special school (6%). Within the secondary school age group, 85% were attending school. This included those attending mainstream school with an aid (45%) and special school (45%). One child within the group secondary group was still attending primary school with an aid. These results demonstrated relatively good school attendance rates in both the primary and secondary school age categories. Only 4 children of primary school age and 2 of secondary school age were not attending any formal education.

iii. Day program utilisation

Day program attendance was reported in the adult groups with 50% of ALH and 70% of ALSA individuals attending a day program.

iv. Employment

Respondents caring for an adult with PWS indicated whether the person was engaged in employment. Of all adults with PWS, 7% were employed full-time and 21% were employed part-time. Those living at home were more likely to be employed (32%) than those living out of the family home (22%).

Family impact

As well as the profound impact on the individual with PWS the data revealed a significant toll on the family unit regarding emotional, physical and financial stressors as a result of caring for a person with PWS *(Fig. A15)*.Each domain rated highly across all groups, indicating a significant burden on the family in multiple respects. In total the average emotional toll was rated 8 out of 10 (SD 2) while the physical and financial tolls were both rated 7 (SD 3).

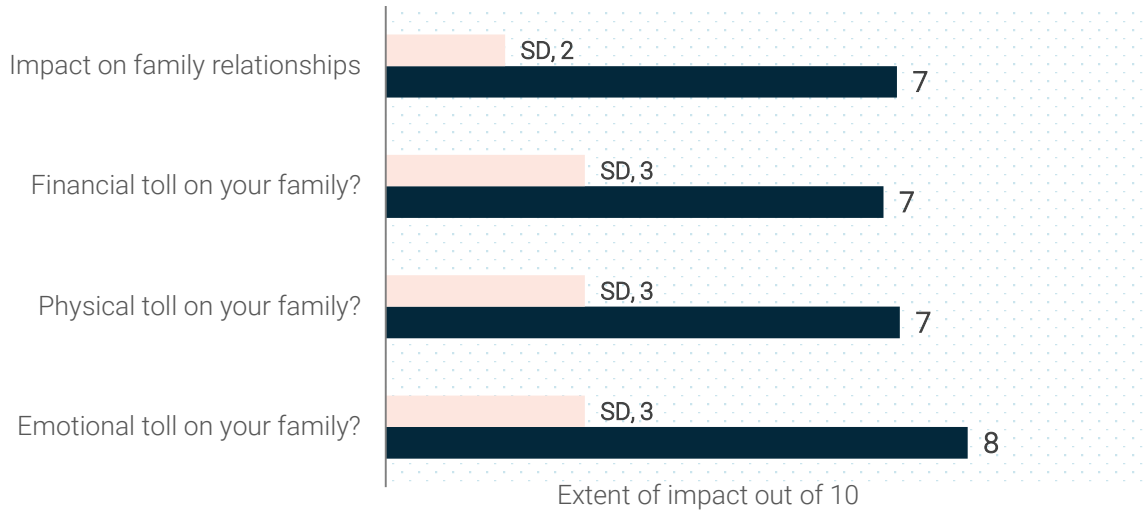
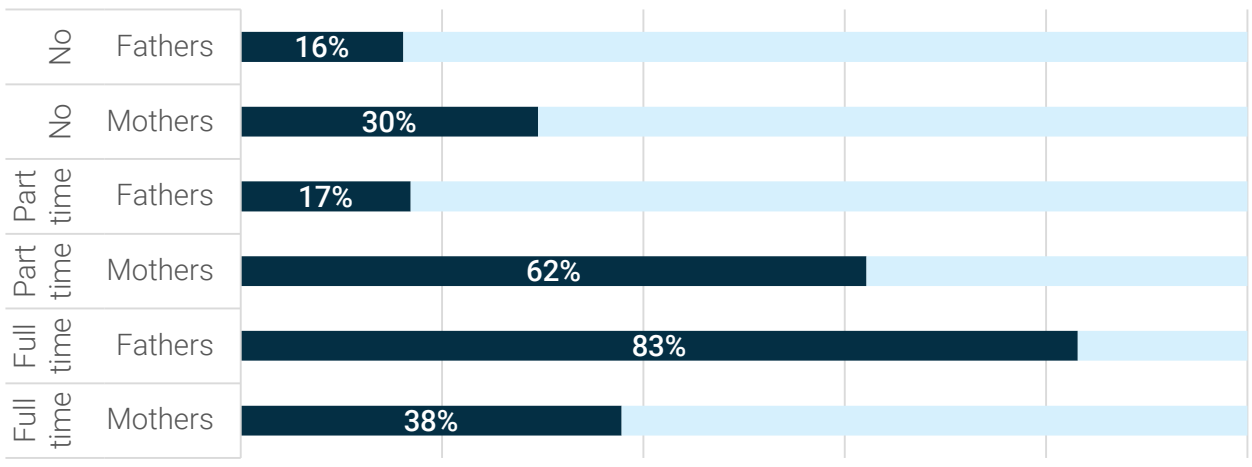


Fig. 15 - Family Impact

Within the survey participants who indicated that the person with PWS was living in the family home, 82% of carers for an adult and 54% of all respondents reported that they would like access to respite services. Unfortunately the survey did not identify those participants who already had access to respite services and wanted to continue accessing these services. For respondents caring for an adult with PWS, 88% reported relying on parents for care and 51% reported relying on siblings. Respite services were utilised by 77% of participants in the secondary school group, 63% of the adult groups and 48% of all survey participants.

Another area of family impact identified was the reduced ability of mothers and fathers of people with PWS to sustain full time employment. 84% of fathers and 71% of mothers were employed in some capacity and 16% of fathers and 30% of mothers were not currently employed. Of those employed only 83% of fathers and 38% of mothers were employed full time while 17% of fathers and 62% of mothers were employed part time. *(Fig. 16)*

Fig. 16 - Parents employment status



RESULTS – TELEPHONE INTERVIEWS

A total of 21 carers for people with PWS participated in a telephone interview regarding effects of the condition on the family socially, emotionally, physically and financially as well as aspects of dietary management, daily activities and education for the person. Table 6 lists the personal characteristics of telephone interview participants for each group. The average age, gender and BMI of participants were similar to those of participants of the online survey, indicating a representative sample of the larger group of survey participants.

Table 6: Characteristics of people with PWS cared for by participants of the telephone interviews. Values are given as mean (standard deviation) except where percentages are indicated.

Group	Preschool	Primary	Secondary	Adult living in the family home (ALH)	Adult living out of the family home with support (ALSA)	Overall
Number of participants	5	4	4	5	4	21
Age (years)	2.9 (±1.7)	8.0 (± 2.2)	13.8 (± 6.1)	22.6 (± 1.1)	30.5 (± 8.7)	15.3 (±11)
Male gender (%)	60%	50%	25%	50%	50%	52%
Body Mass Index (BMI)	17.1 (±1.9)	19.3 (±3.4)	30.3 (±3.3)	45.8 (±8.2)	36.7 (±8.2)	29.8 (±15.1)

Fig. 17 - Demographics of telephone interviews

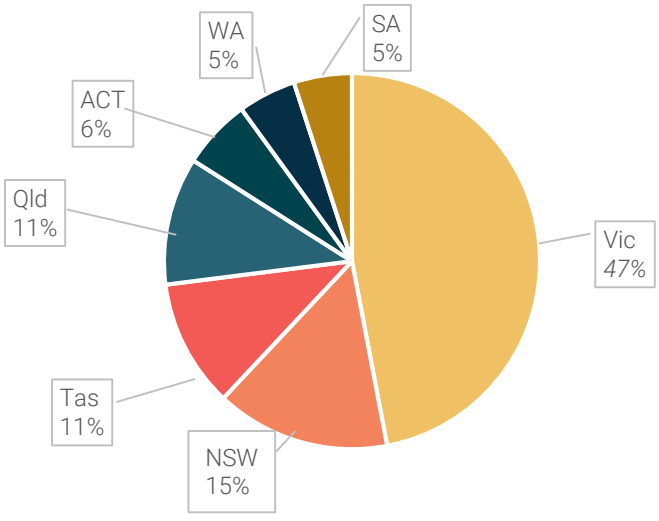


Fig. 17 The percentage of telephone interviews completed from each Australian state and territory

Figure 18 shows there was a wide spread of states represented in the data collected for the telephone interviews. Every state that was included in the online survey had at least 1 individual participate in a telephone interview. Victoria had the greatest contribution overall with 47% of the interviews conducted. New South Wales had 15% contribute to the telephone interview, while Tasmania and Queensland contributed 11% and the ACT 6% of the interviews. South Australia and Western Australia contributed individually to 5% of the additional interviews.

Table 7 outlines the themes to emerge from the telephone interviews listed below according to age group and commonly discussed areas of impact; social/emotional, physical, financial, dietary management and education

Table 7: Themes to emerge from telephone interviews within each age group and area of impact identified

	Psychological and social problems (including maladaptive and autistic behaviours)	**Physical**	**Financial**	**Dietary management**	**School/community participation**
Pre-school	Tantrums, repetitive behaviours, fixation and stress Behavioural issues manageable, however psychology input would be beneficial in providing guidance on how to manage a child's behaviours Importance of language to manage behaviour	Importance of physiotherapy Difficulty with engaging in organised activity/sport without an aid/assistance Mostly play based activity Early intervention services effective	Costs associated with physical activity programs, healthy diet and equipment Cost of parental time spent caring for the child with PWS	Pre-emptive changes to the environment and family diet Other family members having difficulty adapting to eating healthily in front of the child with PWS Need to restrict access to food in the home	Kinder aids helpful

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Primary	Unpredictable, disrupted sleep routine Lack of access to psychiatrists trained to treat PWS Social exclusion of the child and the parents/family Psychological issues – stress & anxiety for family members	Inability to participate with other children of the same age, availability of aids, fatigue, cost Physiotherapy from birth Early intervention programs very effective	Cost associated with private carers Lost opportunity cost due to under employment of parents that are involved with care/support provision Costs associated with installing alarms on doors and locks on cupboards and fridges Costs associated with health foods	Strategies included using a food routine, restricting access to food, keeping food out of sight and the use of non-food rewards Families members buying unhealthy food outside the home for person with PWS Importance of full time supervision to limit food seeking behaviours Access to a dietician who understands PWS would be valuable	

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Secondary	Running away from home Concrete thinking Rewards system doesn't work for everyone Psychologist input helpful for person with PWS Psychologist services difficult to access in rural areas & limited	Barriers: Larger groups, gyms not designed for children without support staff, financial expense for gym, horse riding etc., age restrictions for gyms, rural locations Making activity fun and social - need to do activity with somebody else for motivation.	Cost associated with healthy foods Costs associated with time away from work to collect the child from school when behavioural issues arise Lost opportunities cost (see above)	Phases of food seeking vary from moderate to strong Restricting spending money Planning prior to attending events with food Difficulties finding casual carers who understand the importance of diet	Important for food to be controlled and the person to have access to something that keeps them busy and active in the community Respite and independent living important for person's social development and autonomy

	choice of health professionals Positive reinforcement helpful, reward charts Lack of access to respite in rural areas	Supportive school group sport activities can be really difficult - usually evening but often tired. Organised sports and activities at a routine time/place work better for motivation levels Use of achievement charts Need for carer support to attend programs		Hyper-awareness of food	Carer support needs to be tailored to PWS

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Adult living in family home (ALH)	Stubbornness, tantrums in reaction to change. Lack of control of behaviour leading to depression/anxiety due to the individual recognising their disability Mental health issues for siblings, parents Effect on the ability of the family to socialise Exclusion risk due to temper tantrums less socially acceptable in older age groups Impact of stress on relationships, intimacy and privacy	Helps if the person has control over choosing what they want to do Unable to take family holidays due to mobility limitations Try to give individual ownership of their personal decisions to help minimize resistance to exercise Support workers helpful – barrier if not available Fatigue issues Difficulty motivating Older parents – difficult to encourage person	Inability to work due to high care needs Lost opportunity cost (see above)	Greater access to food in adulthood can unleash extreme food seeking behaviours Food security methods helpful to ensure there is no false hope about food availability Use of a food roster Restricting spending money to avoid overeating – this means that person needs to be taken to work/day program	Full time supervision in public spaces is essential The individual with PWS not wanting to move out of the family home Food stealing leading to trouble with the law if unsupervised in public

	Importance of routine and stability for the person with PWS Verbal aggression	and be active with them Day program useful for encouraging physical activity			

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Adult living in supported accommodation (ALSA)	Competitive, obsessive, tantrums, violence, aggression, running away Behavioural issues improved with personal growth over time and improved management & support Depression, anxiety, OCD Consistency of support key for behavioural management Dominating behaviours, perseverative, persisting, narrow thinking Psychologist services difficult to access in rural areas and limited choice of health professionals Issues with skin picking and general personal hygiene	Supervised activity walking program, one to one sessions work better at engaging individual Assistance/supervision of another person required for physical activity Barriers to physical activity: motivation, understanding, lack of carer availability for individual support e.g. to take them through a gym program More cooperative than they were as teenagers Better understanding of the condition and benefits of physical activity Physical activity enforced previously, now an accepted part of routine	Expense described in time spent caring for the family member and inability to work Planning for eating at events prior to attending helpful	Importance of food routine x2 for reducing stress levels and allowing person to think about other aspects of life Planning for eating at events prior to attending helpful	Behavioural issues often prevent the move out of home Need for greater availability of accommodation with staff who are trained to work with people with PWS Importance of building relationships in readiness for moving out of home with doctors, dieticians etc. to promote safe transition to supported living Barriers to moving out included poor availability of appropriate housing, financial costs and the person with PWS not wanting to move out

RESULTS – HEALTH PROFESSIONAL COMMENTS

Six health professionals who are active in treating individuals with PWS provided their feedback in regard to the supports required for individuals with PWS and their families to reach their goals and live a life of dignity. Their expertise included a Paediatric Endocrinologist, Occupational Therapist, Psychiatrist, Physiotherapist and Disability Service Managers

Table 8: Themes to emerge from health professional comments in regard to appropriate support requirements within each age group and their relative impact identified

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Pre-school	Allied health assessments and ongoing therapy including - - speech pathology - occupational therapy - psychology Parent counselling at the time of diagnosis.	Allied health assessments and ongoing therapy including - - physiotherapy - speech pathology - dietician, - exercise physiology - occupational therapy	Costs associated with allied health management	Allied health assessments and ongoing therapy - dietician	Learning ability assessment before primary school. (Not to be confused with the less effective but widely used IQ test)

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Primary	Allied health therapies ongoing. Psychologist reviews ongoing	Allied health therapies ongoing	Allied health therapies ongoing	Allied health therapies ongoing	Teacher's aid appropriately trained in PWS.

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Secondary	Psychologist support ongoing for individual and family Behavioural intervention plan Assessment and provisions for a secure and safe environment – to prevent absconding and injury.	Allied health support for day-to-day living Participation and appropriate supervision for exercise – incidental and programmed.	Regular PWS-appropriate respite care Home modifications	Coordination between parents and the school vital - particularly around excursions and cooking lessons or when money is being used. Food security: inaccessible bins (e.g. cone topped bins), no access to other children's lunch boxes or staff room (e.g. lockers). Sometimes a "buddy" is useful (when going to the bathroom or between classes to maintain a distraction and prevent food seeking.)	School counselling Special needs support class within a mainstream school or special needs school (SSP) or one on one teacher's aid. IO or IM (can be difficult to access when the child has an IQ of greater than 70 – but doable with letters of support) PWS education for teachers from a "clinical" professional to cover all aspects of PWS and emphasise 'duty of care' for the student with PWS. A scribe for exams (if in mainstream school). Supervision at all times and given structured timetables for weekly lessons Supported/supervised transport including to and from school (to prevent foraging). Supported work experience (to prevent foraging and job failure).

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Adult living in family home (ALH)	Psychologist support ongoing for individual and family Behaviour Intervention Support Plan Support and supervision required at all times	Established activity based programs including exercise equipment at home or regular gym attendance (may require personal trainer)	Monetary control with the possibility of guardianship. Home modifications	Supported work environment including inaccessible food sources.	Post school programs with high level of supervision for further learning and preferably, transition to work Supported/supervised transport High level of support including 1 on 1 supervision in the community (shopping areas, food courts etc). Regular PWS-appropriate respite if still living in family home. Provision of appropriate daytime activities. Depending on the level of support required this could be a supported employment or community participation program. Transitional arrangements need to be put in place.

	Psychological and social problems (including maladaptive and autistic behaviours)	Physical	Financial	Dietary management	School/community participation
Adult living in supported accommodation (ALSA)	Ongoing psychologist/counselling support Regular health monitoring - health signs, skin checks Behavioural support including behaviour intervention plan Provision of additional supervision when things go wrong (e.g. when behaviours are out of hand and more than one staff member is required or when medical requirements are high and additional staffing is needed for personal care).	Regular access to a gym with appropriate supervision required.	Supervision/support for monetary management. Home based exercise equipment (preferable) Guardianship orders set in place due to section 32 Monitoring system put in place Home modifications		PWS specific/appropriate residential care – including high level of caregiver support, restrictive practices environment Training of staff is a must - the right staff for people with PWS is vital. Coordinated care - All staff must work as a team and follow service requirements. Travel support/supervision. Suitable PWS daytime activity with supervision

DISCUSSION

The toll of psychological and social problems on the individual with PWS including maladaptive behaviours and violence

Individuals of all ages with PWS are greatly affected by psychological, behavioural and social difficulties including violent outburst. Characteristic of the condition, at varying severities, are behaviours such as skin-picking, compulsions and temper tantrums (Cassidy, 2012). This is reported in the findings of this report with all respondents reporting repetitive questioning, temper tantrums and poor emotional/social coping skills to be a moderate to highly severe issue.

Consistent with previous findings (Cassidy, 2000) the study identified a lower impact (affecting 82% and severity 3/10) of temper tantrums among pre-schoolers. As identified in the findings of this report the social impacts of fixation, temper tantrums and compulsive behaviours appear to become more evident and problematic when the child begins school, this being supported in the literature (Ho,A, 2010). Issues appear to progress further in the secondary school and adult groups with interviewees identifying continuing difficulties in managing the behavioural and psychological concerns including that the individual no longer responds to rewards systems, while others function poorly at school or attempt to run away from home.

These maladaptive behaviours were also reported to lead to other problems including social exclusion of the child and family.

> *"It's hard for other kids to understand that kind of behaviour… she's (becomes) socially excluded"*

While the maladaptive and autistic behaviours of individuals with PWS may be a social deterrent previous studies have suggested additional social inadequacies. **Individuals typically exhibit poor relationships with their peers, often lack friends and prefer solitary activities** (Dimitropoulous, 2013). Traits of Autism Spectrum disorder are also identified amongst PWS individuals (Dimitropoulous, 2007). Similarly the findings identify a high reliance on assistance by the individual with PWS to develop friendships. Unfortunately, this does not appear to improve with age (Rosner, 2004) with ALSA and ALH groups continuing to require on average significant assistance. This identifies that a **high level of social isolation exists within the PWS community**. As a result social groups and activities should be made accessible and tailored to meet the needs of individuals with

PWS. While those with PWS may not thrive socially within their environment, this may be improved by being able to play and interact with people who display similar behavioural traits.

Mental health problems were also a major concern amongst all participants, with the exception of preschoolers. These included depression, anxiety, low mood and psychosis. These findings are mirrored in a recent study that identified individuals with PWS suffer more severely from withdrawal, depression and thought problems than their age and IQ matched counterparts (Skokauskas et al., 2012).

Professor Stewart Einfeld, Chair of Mental Health and Senior Scientist Brain and Mind Institute, University of Sydney indicated that in his 35 years of care for people with PWS, he had not met any individual who had managed to function independently, without outside supervision and support. In his opinion most people with PWS require a high level of support.

Einfeld also commented that intellectual ability is not a good predictor of level of support required, particularly in PWS. His research has shown that there is a major discrepancy between academic scores as measured by IQ assessments and emotional development and adaptive functioning. The latter are much more severely impaired in persons with PWS.

The Australian Child to Adult Development Study (ACAD) is a longitudinal cohort study that followed 967 families of children with developmental disabilities over a 20-year period. Through ACAD, Einfeld compared family well-being (parental mental health and family function) across a broad range of developmental disabilities. The findings were that the families of people with PWS had the most impaired family well-being. Even more so than the families of children with autism. Reduced family well-being was directly associated with the severity of the individual's behavioural problems, particularly the rage attacks and hyperphagia. He also found that family well-being greatly improves with provision of supports to families.

Psychosis was also demonstrated in the data as a concern, being echoed by Dykens et al., (2003) who reports high rates of psychosis in PWS, when compared with other individuals with intellectual impairment. One parent mirrored the view of several;

> *"Our biggest issue has been the psychosis"*

Focusing specifically on aggression, the results revealed violence to be more problematic as the individual ages. Generally PWS children can be somewhat easy-going and affectionate, with personality problems developing between 3 and 5 years of age, progressing later to violent outbursts (Butler, 2011). Interestingly, in the findings a substantial increase in violence was identified between adults living at home and those living out of the family home in supported accommodation, 26% to 72% respectively for violence against family members and 21% to 56% respectively for violence against members of the public. This highlights how vital specialised respite care is but also highlights the necessity of specialised and appropriate living conditions for PWS individuals living in the community. This is echoed in the health professionals' and parents' feedback;

> *"It is important that when going into supported accommodation the person with PWS is well matched with others that live there, it is also important that the restrictions that are placed on PWS are upheld in the house with everyone. Daily routines of when to eat, exercise and do daily living chores also must be adhered to by everyone"*

> *"Having people living in a service that doesn't have PWS has not worked for us; it has caused more issues and anxiety than is needed"*

The importance of consistent and specialised support was highlighted by family and carers identifying improvements in maladaptive behaviours as a result of having access to appropriate supports. Unfortunately, access to such services can be problematic due to living in remote locations and where suitable supervision for transport is not available, thus limiting the choice of health care professionals that can be accessed. Additionally, parents cited poor access to psychological support by those specially trained in the management PWS and felt that the individual with PWS would benefit from psychological review.

> *"I would really like a psychologist's point of view"*

Currently, emotional and behavioural concerns in PWS individuals can be managed with a combination of medication and behavioural interventions (Ho, 2010). Psychologists can provide management strategies and support the individual through their behavioural, emotional and social

difficulties. Therefore, **access to psychological treatment should be a core funding for individuals with PWS.** Health professionals also identify the need for psychological assistance commencing at primary school age to manage maladaptive behaviours as well as psychological supports focused on specific PWS training to assist families and carers manage behavioural issues is also necessary.

Skin picking, which is reported as another compulsive maladaptive behaviour in PWS (Dykens, 1992) was reported in 89% of participants at a high severity in adults groups in particular. This is similar to other findings where skin picking is reported to be a significant problem in both youth and adults, 69–95% and 81% respectively (Morgan, 2010). Skin-picking has both physical and psychological health consequences including bleeding, sores, scarring, infections, permanent disfigurement, hospital admissions and even death (Morgan, 2010).The impact of the emotional toll on the individual should not be ignored with social embarrassment and social isolation being linked to skin picking. **Psychological interventions have shown promise in reducing chronic skin picking** (Flessner, 2008).

When maladaptive behaviour is managed appropriately, it is possible that social functioning will be enhanced.

Recommendations	
Core	• Assessment and ongoing management by a psychologist for individuals with PWS, beginning in the primary school period. • Access to appropriate social and activity groups for individuals with PWS including supervision by trained staff able to manage maladaptive behaviours.
Capacity	• Access to pre-emptive psychologist advice and counselling for families of preschool aged children to assist with management of maladaptive behaviours.

Social and emotional impact on the family

Despite the obvious effect of emotional and behavioural issues on the individual, the toll on family members can be devastating. It is documented that parents of children with an intellectual disability experience a significant amount of psychological stress and as a result have an increased likelihood of developing mental health issues themselves (Blatcher & McIntyre, 2006). It is vital to note that it is not just the individuals themselves that need support, but also the families. One mother of a 6 year old provides her honest reflection on this impact;

"What have I lost in the last 6 years? My sanity, my sense of humour, my husband, unable to commit to my work, time with my other children"

Parents and carers endure a great deal of social and emotional trauma as a result of PWS. One study found that the parents of individuals with PWS suffer a greater degree of anxiety, obsessive compulsion and other psychological conditions, when compared to the parents of age and IQ-matched children without PWS (Skokauskas et al., 2012). The study also found that the severity of the psychological state of the child is linked with the severity of the psychological state of the parent (Skokauskas et al., 2012**). All except one respondent reported a severe emotional toll on the family due to PWS.** Considerable levels of stress associated with anticipating the maladaptive and autistic behaviours of the individual with PWS was frequently discussed in the interviews.

As well as the emotional toll, a family's ability to be a part of society including the workforce was reduced due to the burden of caring for an individual with PWS.

> *"You've got to be on your toes 24/7"*

There was also a significant impact on family relationships with family breakdowns and an inability to socialise due to the maladaptive behaviours of the individual with PWS.

> *"It's just hard to go anywhere or do anything because we don't know what she's going to do"*

> *"Once they're an older teenager and they're moving into adulthood… you don't socialise as much because it's just too much stress"*

> *"It puts significant pressure on a marriage"*

The physical and financial tolls were also rated highly with almost all respondents reporting this to be an issue. The physical toll also took the form of aggression against family members. As stated earlier, injury caused to family members is not uncommon.

> *"There certainly has been a lot of verbal abuse, emotional abuse and even physical abuse"*

In order to minimise the stress placed on siblings, parents and carers of those with PWS as well as to protect their social and emotional wellbeing, families should be given access to respite care. This would ameliorate the psychological effects of the condition on family members. To the same effect, **families benefit from counselling or access to a psychologist** to assist with the management of their psychological health. This should perhaps be provided as a capacity support for family members who indicate suffering emotional distress.

"I think one of the things that would be great for all parents to have is somebody to talk to, like a counsellor, psychologist… someone who knows something about Prader-Willi Syndrome and can understand what parents are going through"

Recommendations	
Core	• Access to respite care for all families with a child with PWS to minimise the impact of psychological stress and allow family to engage socially.
Capacity	• Access to counselling and psychological services should be offered to family members of those with PWS

Physical

The findings of this report clearly demonstrate that within all age groups physical activity and **access to physical activity is important for weight management, social interaction, emotional wellbeing and muscle strength.** Butler et al., (2006) identified that individuals with PWS have decreased energy expenditure during physical activity due to their body composition and metabolic rate when compared to obese individuals. Furthermore this reduction in energy expenditure leads to an increase in energy stores being converted to fat (Butler et al., 2006). Studies also show a reduction in the spontaneous physical activity for all PWS individuals which leads to a reduction in muscle mass and increased adipose tissue (Schlumpf et al., 2006). Due to this, individuals with PWS are required to be physically active to avoid obesity and obesity-related conditions. Eilholzer et al., (2003) showed that **with motivation, support and encouragement, individuals with PWS are able to adhere to a regular physical activity program**. As a result, these individuals experience improved body composition and increased spontaneous activity levels. Both studies indicate the importance of physical activity as an addition to the lives of all PWS individuals. This is well supported by the carers interviewed in the current study.

All telephone interview participants in the preschool group reported that their child was involved in the early intervention program, which includes physiotherapy. Many preschool children only received their exercise and exercise programs through a physiotherapist as parents reported difficulty with their children being able to participate in organised group activities. This difficulty in participation is related to the need to have an aid present or participate in a one-on-one exercise program to keep the child engaged and compliant with exercise.

> *"He's probably doing more than he would do (exercise) if he didn't have an aid"*
> *"....needs one to one just to help her get up steps at the playground"*

Within the primary school group, many individuals had developed regular and organised exercise routines, with all of the parents interviewed reporting that their child was engaging in at least 60 minutes of physical activity daily. These activity levels reach the recommended physical activity guidelines for children and young people, which are stated as 60 minutes of moderate to vigorous activity daily (Department of Health, 2014). As with the preschool group, many individuals involved in the early intervention program found it extremely beneficial. Most of the primary school children were receiving physiotherapy and completing physiotherapy-based programs weekly. Finding appropriate team and individual sports in the community was reported to become increasingly difficult in the primary school years. Sporting teams and other exercise-based co-curricular activities often were

unable to provide the support or aids required for the PWS child to participate. Aids are often used to help children access these programs but this can increase the financial stress on the families. There are appropriate group activities available for PWS children although they are limited and families struggle to travel the distance to these locations, particularly in rural and remote areas. These activities can often be associated with significant financial costs.

> *"We tried AusKick for a while but that didn't work. He is a very determined kid and wants to do things his own way, when he realises that others are more capable he struggles to be motivated"*

> *"Location and cost are the biggest barriers"*

Within the secondary school population, **physical activity is extremely important for social engagement, behaviour management and weight control.** As individuals become older, a reluctance to participate in physical activity can occur. It is particularly important for exercise in this age group to be considered a social activity. Interviewees commonly reported that the individual was not motivated to exercise alone, so exercise with groups or aids had proved more effective. The use of achievement charts was also reported to help encourage individuals to participate in regular exercise. Unfortunately, both aids and specialised exercise programs have a financial and time cost. Those in rural locations are further disadvantaged due to a lack of access to appropriate aids and appropriate exercise groups.

> *"Physical activity through the use of others helps, he is better in a supervised activity program"*

> *"We have tried charts for goals for him to reach, this works sometimes but sometimes it doesn't"*

> *"If it comes from us [the individual's parents] he will not do it"*

Carers for adults with PWS described similar issues. They identified that access to appropriate activity and the acquisition of appropriate aids and carers can be costly and inaccessible. One interviewee also discussed their family's inability to take a family holiday, due to mobility limitations

and unrealistic costs. It should be noted that **in the adult category many of the parents of those with PWS are themselves increasing in age** and as a result have their own health problems to manage, which can limit their ability to be active. **These problems lead to a further financial burden on the family as aids and carers are always required to enable the person to participate in physical activity.**

> *"I've got chronic back pain and my husband's on oxygen so it makes it hard for us to get out and do it with her"*

The day program was reported to be particularly important for facilitating physical activity in adults with PWS. Many day programs offer group exercise, with the appropriate amount of aids/carers. This is incredibly important especially for those who live out of home, as regular exercise is achieved through the program. The program was currently being attended by 33% of people who completed the survey. The day program was more popular in the older age groups, as it is designed for those who have left school or are not currently working. A total of 50% of the ALH and 70% of the ALSA group were attending day program, compared to 0% of primary and 8% of preschool and secondary. These results suggest that the day program should be a core support, offered to all individuals with PWS who are not currently engaged in full time schooling or employment.

The phone interview and survey data reveal the need for supports and programs to enable PWS suffers to engage in regular physical activity. **Physical activity is vitally important for all PWS sufferers to prevent poor mobility, obesity and reduced quality of life** (Eiholzer et al., 2003). Therefore physical activity should be actively encouraged for all individuals with PWS and should be supported by the NDIS. Access to appropriate physical activity and aids/carers to enable participation is a core need for all individuals with PWS and should be appropriately funded.

With a lack of physical activity, there is often a decrease in mobility for individuals affected by PWS. A theme that appeared during the interviews **was the effect that mobility and lack of mobility has on the care needs of the individual. Eighty-two per cent of PWS individuals required help for mobility around the house. Furthermore 98% of respondents reported that the individual required significant assistance for mobility either within the home or the community.** These results indicate that physiotherapy is a core support that needs to be offered to all children and adults with PWS through the NDIS.

Recommendations	
Core	• Physiotherapy for all children and adults with PWS. • Appropriate exercise programs including suitably trained carers to facilitate participation should be offered to all people affected by PWS. • Day programs which include physical activity and skills training should be offered to all people with PWS who are not attending full time schooling or employment.
Capital	• Funding is required for individuals to purchase exercise equipment, gym memberships and utilise personal trainers to facilitate and encourage physical activity for the person with PWS. • Funding for orthotics and mobility aids should be provided for all individuals with PWS.

Financial

The economic benefits of increasing employment of people with disability are well recognised and one of the aims of the NDIS (National Disability Services Policy Research Unit, 2011). However, the results of this study demonstrate that the financial burden associated with PWS encompasses much more than low employment rates for individuals with the condition.

The capacity of parents of PWS individuals to participate in the work force was identified in our findings to be negatively impacted with rates of unemployment being much higher than the national average for Australian adults of 6.2% in September 2015 (ABS, 2015a). High rates of part time employment were also reported. Of those who were employed, 17% of fathers and 62% of mothers were working part time hours. This is significantly higher than the reported statistics regarding employed Australian adults that 13.5% of males and 43.2% of females were employed part time in 2011-2012 (Australian Bureau of Statistics, 2013). This identifies that parents of PWS who are employed are less likely to work full time compared to the general Australian population. Such findings are supported by the ABS (2015b) which demonstrate that primary carers for people with a disability are significantly less likely to participate in the Australian workforce. As a result **there is a need for improved supports to allow parents to better engage in employment.**

The costs associated with PWS were reported frequently in the telephone interviews, including time away from work for the parents of the person with PWS. Many parents discussed situations in which they took leave from work to take the person with PWS to medical appointments or to bring them

home from school or day program when behavioural issues arose. Another strong theme from the preschool and primary school groups was **the inability to use after-school care and babysitting services due to the availability of food and the inability of those services to adapt to the needs of the child with PWS.** Many participants reported paying for private carers or reducing their working hours in response to this.

> *"The big cost for me is the inability to go back to work"*

The ability for PWS individuals to sustain their own employment is a challenge. One health professional expressed:

> *"Very few people with PWS can work in open employment without having major health implications"*

This sentiment was reflected in the responses regarding adults from the online survey where employment was a low 29%. This is substantially lower than the labour participation rate of 52.8% for Australians aged 15-64 with a disability as reported in 2012 (ABS, 2015c). ALH were more likely to be employed than those living in supported accommodation, however, it is not clear why this is the case. People with PWS living in the family home may be less severely affected and therefore able to participate in employment more readily, however, this is only speculative.

It is clear that support is required for people with PWS to gain appropriate employment within a well-supported environment. Health professionals **identified the necessity of support and supervision when attending employment** where issues surrounding food, theft and behaviour may occur.

> *"…workplaces must be aware of the issues around food and ensure that food is not accessible to the people with PWS"*

Home modifications and equipment are also described as vital by both health professionals and carers of individuals with PWS. Carers of preschool aged children who were interviewed identified the cost of equipment such as specialised prams, orthotics and play equipment that would not normally be required for a child of the same age. Also described was the cost associated with replacing cupboards and fridges to ensure that they could be securely locked to assist in the management of food seeking behaviours. One mother of a primary school aged child with PWS described installing an alarm system in response to the child repeatedly leaving home during the night. In a few cases,

household items needed to be replaced after being damaged during temper tantrums by the person with PWS. In the school aged groups, interviewees described the added cost of a strict healthy diet and fees associated with attending exercise programs and gymnasiums. As described earlier, physical activity is an important strategy for the management of the physical, social and emotional components of the PWS condition (Goldstone et al., 2008). A strong theme from the secondary and adult group interviews was the importance of a supported environment for allowing the person with PWS to be physically active. This was described to include exercise programs in which one-on-one care was available and other participants were of similar age and physical function. Hence, it is clear that **structured exercise programs are an important intervention for the ongoing management of the PWS condition** and should be funded under the NDIS as a core support.

Stealing money is one of many behaviours which is associated with food behaviours (International Prader-Willi Syndrome Organisation, 2010) which carers who participated in the online survey and phone interviews reported as a major problem for secondary and adult groups. This was particularly the case for adults living at home where **financial costs associated with theft of money and goods from family members, friends and shops is a major issue**. Carers described **the need for very strict supervision in all environments and at all times** to limit this. This indicates the importance of constant supervision in the home and community for the person with PWS.

Recommendations	
Core	• Exercise programs with appropriate supervision including gymnasium memberships should be offered to all people affected by PWS.
Capacity	• Assistance to gain employment offered for all people with PWS over 15 years of age to support employment participation in the most appropriate capacity for the individual. • Carer support when attending employment is required by some people with PWS, depending on the employment environment and the individual's capacity.
Capital	• Funding will be required by some families, particularly with younger children affected by PWS, to address unreasonable costs for items including specialised equipment and home modifications.

Dietary management

Food seeking behaviours are a major issue in PWS as demonstrated in the literature (Allen, 2011; Goldstone et al., 2008) and supported by the findings of this report with **food seeking behaviours is a very severe problem across the age groups**. Of the respondents to the online survey in the secondary and adult age groups, **43% of respondents reported a BMI within the severe or very severe** obesity categories, indicating the importance of early intervention to avoid weight gain and the many potential and costly complications of severe obesity.

It is clear that families require assistance to manage food seeking behaviours and the unique dietary requirements of individuals with PWS. It has been reported that the management of individuals with PWS from two to three years of age by an experienced dietician is required to minimise problems associated with excessive weight gain and osteoporosis (Butler, 2006).

Close supervision from a dietician is required to monitor the effects of the recommended restricted caloric intake in combination with vitamin and calcium supplementation (Butler, 2006). **Early dietary intervention to assist families in the implementation of a well-balance restricted-energy diet from two years of age has been demonstrated to avoid excessive weight gain at ten years of age** in multiple studies (Miller, Lynn, Shuster & Driscoll, 2013; Schmidt et al., 2008). Many interviewees reported that intervention from a dietician was helpful, however, some respondents living in rural areas discussed an inability to access the services of a dietician who understood the clinical picture of this rare genetic condition. These findings indicate that **assessment and management by a dietician experienced in the management of PWS should be a core service offered to all children and adults with PWS under the NDIS.**

> *"We've seen a dietician but the dietician had no idea what Prader-Willi even was"*

As discussed above within the financial domain the need to purchase equipment and pay for home modifications to limit food seeking and foraging behaviours was common. Another expense described was the cost of fresh and healthy food that was low in fat to suit the optimal diet for people with PWS.

> *"Being disciplined in the approach to food for the whole family, not just her, is important"*

> *It's expensive to buy good quality food that's low in fat"*

Further to this is the impact of the restrictive diet on siblings and the rest of the family. Sibling resentment was also widely highlighted in the interview responses as a result of dietary restrictions, home modifications and change to home life activities which were necessary due to the individual with PWS's food and other maladaptive behaviours. Alarmingly Mazaheri et al., (2013) in their study investigating the impact of PWS on family's quality of life, identified 92% of the siblings in the study to have moderate-to-severe symptoms of post-traumatic stress disorder. This, once again, highlights the **necessity of psychological support to be offered to all family members** of individuals with PWS.

A tendency for family members to indulge in unhealthy food at every opportunity when outside the family home or when the child with PWS was not around was also reported.

> *"Even though we're eating healthily it's actually not a healthy lifestyle for us"*

Key interventions for managing food seeking behaviours associated with PWS have been reported to include restricting access to food, distraction, use of routines, supervision of children around food and availability of low-fat snacks (Allen, 2011; Goldberg, 2002). Similarly, interview participants reported that the use of a food routine or roster, keeping food out of sight, restricting spending money and the use of non-food rewards were helpful strategies for managing diet and food seeking behaviours. All interviewees reported physically restricting access to food for the person with PWS using locks and argued it was part of their duty of care for the individual to do so. Hawkins, Redley & Holland (2011) described the conflict between the duty of care to manage the person's diet and the promotion of independence and autonomy experienced by support workers caring for people with PWS in a United Kingdom group home. The authors concluded that recognising the autonomy of the person with PWS entails risk and is, in certain situations, "setting the person up for failure" (Hawkins, Redley & Holland, 2011).

A central theme from the telephone interviews in the secondary and adult groups was **the importance of food restriction and certainty about food allowing the person with PWS to direct their focus away from food**. It was argued that the use of a food routine created a sense of security for the person with PWS and enabled them to focus their attention on other aspects of their life more easily. This theme reflects the ideas presented by Gourash & Forster (2005) who argue that **food security can reduce stress and behavioural problems exhibited by the person with PWS whilst managing**

their weight. The authors argue that this can be achieved by removing any doubt about the food the person will be served or hope that they can obtain food by other means (Gourash & Forster, 2005). Participants of the telephone interviews indicated the importance of full time supervision around food in order to provide this security. Participants in the preschool, primary and secondary groups highlighted difficulties accessing carers for after-school hours and occasional care with the capacity to provide care according to the needs of the individual with PWS, particularly regarding food restriction. These findings indicate that all people with PWS should have access to full time supervision within the home environment, whether this is provided by a family member or paid carer. Support workers trained in the care of individuals with PWS should be available to provide occasional care in place of babysitting and after school care services.

"The key for a young adult or an adult is that there is adequate support to enable that person to feel good about themselves, to get on with their life and for food - and the potential access to food – not to be the thing that kind of dominates their thinking"

In the adult groups, a central theme was the difficulties associated with greater access to food once the person became more active in the community. Results from this report demonstrated an **increased severity of problems associated with food seeking in the adult age groups.** Food seeking behaviours within the community were also indicated as a problem by the majority of respondents, including stealing food from plates, lunch boxes, shops and bins. **Seventy-five per cent of respondents indicated that stealing food from shops was an issue to some extent for the person with PWS, while 76% indicated that stealing food from other people's plates was an issue, 77% indicated that stealing food from bins was an issue and 85% indicated that stealing food from lunch boxes was an issue.** It is not uncommon for people with PWS to also steal money as discussed in the financial section of this discussion in order to purchase food.

As 91% of respondents indicated that food seeking behaviours within the community was an issue

to some extent, full time supervision within the community is recommended as a core support offered to all people affected by PWS.

Recommendations	
Core	• Assessment and management by a dietician with an understanding of the PWS condition for all people with PWS, beginning during the preschool period.
Capacity	• Full time supervision within the home environment is required for all people with PWS. Carer support should be offered to assist families with this. • Disability support workers who are trained in the care of people with PWS should be available to provide occasional care in place of babysitting services. • Full time supervision within the community is required for people with PWS.
Capital	• Funding will be required to ensure food can be locked away within the home, school and work environment.

Activities of daily living (ADLs)

Individuals with PWS often require assistance with personal care tasks in the home environment. This further places a significant care burden on the family. During the interviews conducted, a common theme was a lack of concern for personal hygiene. Many individuals had problems with skin picking as mentioned above, especially those in the older age groups (Hustyi et al., 2013). This issue is compounded by poor personal hygiene, leading to delayed healing and a risk of infection. Issues with personal hygiene are often related to a reluctance to shower and bathe. Some interviewees indicated that the individual with PWS would not spontaneously shower and bathe further reflected with **93% reporting the need to provide the person with PWS help for showering and bathing.**

> *"Personal hygiene is not his strongest point"*

PWS individuals regularly experience problems with dental conditions including oral thrush, cavities and oral infections (Scardina, Fuca & Messina, 2007). Research has demonstrated that **individuals affected by PWS regularly display poor oral hygiene** and recommended that dentistry management is a necessity to prevent oral complications in PWS individuals (Scardina, Fuca & Messina, 2007). Seventy-two of the 106 individuals who completed the survey reported that their child regularly saw a dentist. While these numbers are encouraging, the evidence suggests it would be beneficial for 100% of individuals with PWS to visit a dentist regularly (Scardina, Fuca & Messina, 2007). The issues related to oral hygiene may be linked to the need for help with cleaning teeth. The survey results showed that **96% of individuals reported the person needed moderate help for cleaning teeth.** This suggests that significant support is required for individuals to maintain a regular and efficient brushing routine, likely contributing to the dental problems associated with PWS. Research suggests that the use of an electric toothbrush is more effective than a manual toothbrush, especially for those who require assistance from another individual to brush (Bernal, 2005) indicating that provision of an electric toothbrush to improve oral hygiene should be considered.

Additional personal activities of daily living including dressing and toileting further increase individuals' care needs. **Ninety-five per cent of individuals reported that the person with PWS required at least moderate help with dressing and 90% of individuals reported the person with PWS needed moderate assistance for toileting.**

To reduce the personal care burden on families, occupational therapists are able to provide modifications with the home environment. Installing rails, shower chairs and elevated toilet seats may all assist individuals to improve independence with these tasks. Additionally, these modifications may reduce the physical demand on carers (McCandleness et al., 2011). Occupational therapy can also increase the independence of individuals with PWS through practicing tasks such as dressing, toileting and showering. Training to perform these personal activities of daily living can both improve the quality of life in the individuals and reduce carer burden. Access to occupational therapy should be available to all individuals regardless of age to assist with the development of independence and reduce carer reliance. Furthermore, occupational therapy services should be available when individuals move home or when an individual's function has changed, leading to a reduced ability to perform personal activities of daily living.

Overall, **the online survey found that 99% of individuals required assistance for personal activities of daily living.** This places a significant burden on the families of individuals with PWS. Personal care assistance and occupational therapy represents a core need for all individuals with PWS. Furthermore, the significant personal care needs reported indicate that 24-hour care is required for an individual to live out of the family home.

Recommendations	
Core	• Occupational therapy for preschool and primary groups to assist with home modifications and practicing personal care activities. • Regular dental care for all individuals with PWS.
Capacity	• Occupational therapy for secondary and adult groups moving house and those whose level of function has changed. • An electric toothbrush should be provided to assist individuals with oral hygiene. • Home modifications to the bathroom for those who require assistance with personal care.

Community participation

Community engagement is known to promote self-esteem and resilience for the individual participating (Sweet, 2013). Increased participation in community activities such as employment, sport and leisure has been directly associated with improved mental and social wellbeing (Kawachi & Berkman, 2001; Strawbridge et al., 2001; VicHealth, 2008; United Nations, 2006). The importance of a level of community independence and choice was expressed by one health professional;

> *"Giving people with PWS choices is important - they can't have choices around food but they can choose what they do and how they want to live their life (socially)"*

Sadly, for the secondary and adult groups, 12 participants (17%) reported that the person with PWS was not attending school, employment or a day program. This indicated a low level of community engagement for these individuals and the need for greater support to enable participation in structured activities for physical, social and mental wellbeing.

For individuals with PWS, support to enable community participation is exceedingly important once schooling ceases to provide a means for social engagement. One health professional comments;

> *"When I consider working with somebody with PWS what jumps to my mind is how they think and how best to get them into life - food and the impact is secondary to this. If you solve the first part, the second part comes; if you only do the second part, the person is alive but doesn't live"*

Adults with PWS are more likely to attend a day program than children as reflected in the data. Day programs are a valuable opportunity to participate in physical activities and learn skills such as gardening and horse riding as expressed by many interviewees. Participants also reported that attending a day program provided an important focus for the person with PWS aside from food.

Telephone interview participants discussed the importance of carer availability to enable the person to safely attend employment and assist with certain employment types such as newspaper delivery. As demonstrated in the survey data employment rates were low which indicates that further carer supports in this area could potentially increase workforce training and subsequent workplace participation.

A central theme from telephone interviews in the primary, secondary and adult groups was the importance of full time supervision for the person with PWS within the community to ensure the person's safety. Further to this **100% of the online survey respondents indicated that the person required a significant amount of help for getting around in the community and for road safety.** Violence in the community was also highlighted as a concern with **22% of survey respondents reporting that the person with PWS has caused injury to a member of the public,** particularly in the adult groups. These results indicate that full-time supervision in community spaces is necessary for every person with PWS to ensure safe community engagement.

Respite care reduces parental stress significantly, at least in the short term, for the majority of parents of children with a developmental disability (Chan & Sigafoos, 2001). The results of the online survey support the importance of respite care for the families of people affected by PWS. **Eighty-two per cent of carers for an ALH and 54% of all respondents reported that they would like access to respite services.** Telephone interview participants also discussed the importance of respite care for relief from the high carer burden associated with PWS.

The results of this report also identified high usage of community health care associated with attending medical appointments, particularly in the younger age groups where preschool and primary groups were currently seeing on average 3 medical specialists. Furthermore telephone interview

participants indicated attending general practitioner (GP) visits for the person with PWS as often as monthly. Not only does this create a financial burden for the family but also difficulties associated with appropriate supervision and travel to and from appointments. **Carer support to attend medical appointments is a must.**

Finding appropriate care particularly for preschool and primary school children was a problem indicated by survey respondents who use siblings for care 48% of the time and extended family and friends 86% of the time. Providing this care places a significant burden on the family members and their friends as a result of managing the special dietary requirements, food seeking and behavioural issues associated with PWS. Hence, respite care is not only important for parents but also to reduce carer stress for the extended family. Another benefit of respite care identified is that **respite care provides social interaction and an opportunity for greater autonomy for the person with PWS.** Difficulties accessing appropriate respite care in rural areas were also discussed. These findings highlight the significant positive effects of respite care for the person with PWS and their family and support the recommendation that respite care should be a core support for all families affected by PWS.

Recommendations	
Core	• Carer support for community activities such as attending a day program, employment and medical appointments should be offered to all people with PWS. • PWS appropriate respite care should be offered to all families affected by PWS, including those living in rural and remote areas. • Day programs which include physical activity and skills training should be offered to all people with PWS who are not attending full time schooling or employment.
Capacity	• Assistance to gain employment for all people with PWS over 15 years of age to support employment participation in the most appropriate capacity for the individual. • Carer support when attending employment is required by some people with PWS, depending on the employment environment and the individual's capacity.

Living out of the family home with support

Supported accommodation has demonstrated positive effects for the autonomy and quality of life for people with a range of disabilities (Social Policy Research Centre, University of New South Wales, 2014). For adults with intellectual disability, supported accommodation has been shown to increase personal choice and self-determination, social behaviours and participation in community-based activities (Walsh et al., 2010). Similarly, many interview participants of the secondary and adult groups discussed the importance of supported accommodation outside the family home for the person's autonomy and quality of life. **Interviewees expressed that living out of the family home in a supported environment was the safest means for adults with PWS to gain independence from family members. Many reported that this was important to the person with PWS as they were observing their peers mature and become more independent. Moving out of the family home also improved family relationships as verbal and physical aggression was often directed towards support workers, rather than family members.** Older interviewees indicated the importance of availability of supported accommodation to support the person with PWS once parents become too elderly to provide the necessary care.

> *"My relationship as a mother is really good with [the person with PWS] now because I'm not having to control what he's doing with his food and I don't have to tell him what he can and can't do, I'm free to be his mother"*

> *The aggression is directed "... to those that are providing the care and you recognise that there's a resentment towards that care for a young person who's wanting to be independent"*

The results of the online survey identified that there were concerns that housing would become an issue at some point in the future of the individual with PWS and indicated the importance of availability of supported accommodation in the future. These findings indicated **that a significant proportion of families affected by PWS will be seeking supported accommodation under the NDIS and the majority of people with PWS in the secondary and adult groups will require supported accommodation within the next 5 years.**

> *"As a parent, feeling that he's going into a safe environment is a big thing"*
> *"It's going to be the best thing for him to have in life"*

Telephone interview participants in the adult groups identified a range of barriers to the person with PWS moving out of the family home as did health professionals.

> *"This is a very stressful time for not only the person with PWS but also the family"*

A strong theme from the secondary and adult group interviews was the **importance of accommodation specific to the needs of people with PWS including restricted access to food and assistance for food preparation.** An individualised approach has been described as an important strategy for successful supported living (Social Policy Research Centre, University of New South Wales, 2014). **Interviewees described unsuccessful attempts at moving out of the family home into supported accommodation without strict dietary management and full time supervision.** This was also echoed by health professionals.

> *"At no time should a person with PWS be left unattended either in the service or in the community"*

It is important to note that the average BMI was lower for the ALSA group. This appears to indicate that **appropriately supported living situations can provide the necessary dietary management and intervention.**

> *"I think it is something that really is necessary but it needs to be a specific Prader-Willi home, not just any home. She's had respite care where they've gone out and bought her fish and chips"*

> *"The thought of her moving into a home where food wasn't controlled or where she could manipulate staff, I find quite scary"*

Furthermore**, individuals with PWS who moves out of the family home will require assistance for personal activities of daily living.** Within the online survey participants, all individuals required help with both dressing and cleaning teeth and 93% and 81% required help with showering and toileting respectively.

> *"He has his own support worker in the house who assists with personal care"*

> *"It's important that the level of support he has is maintained, he needs 24-hour care"*

Many interviewees noted a lack of availability of PWS-specific supported accommodation and an inability to self-fund carer support for the person with PWS to live in individual accommodation. A final barrier described by some carers was resistance from a person with PWS who did not want to live out of the family home. Interviewees identified that **whilst it was difficult for them to convince the person with PWS of the need for them to live outside the family home, it was inevitable with ageing parents that this occurs.**

> *"I'm not going to be around forever and I have to make sure that she gets looked after"*

> *"There's only one specific PWS house in Melbourne and we're extremely lucky that we have access to it"*

Participants of the telephone interviews indicated that **it would be impossible for the person with PWS to live independently outside the family home without full-time support.** One interviewee described the experience of her daughter who is affected by PWS moving out of the family home to live independently for a prolonged period. She discussed issues including the person inviting her

friends to visit with food, weight gain and avoidable police interactions. The participant reported that police officers eventually recommended that the person return home or move to a fully supported living situation. A further **strategy to increase the success of supported accommodation for people with disability is the facilitation of linkages in the community.** One telephone interview participant from the living out of the family home group argued that it was important to build relationships with carers, doctors and allied health workers to enable a situation of "safe independence" for the person with PWS.

> *"She was calling the police every other day when she was on her own"*

The average age of those living out of the family home is 5 years older when compared to the average of the ALH group. This suggests that many families delay the move out of the family home until the individual's long-term future requires serious consideration. Furthermore, the increased age is likely related to the waiting lists associated with specific PWS housing. Some states and territories provide no access to PWS specific housing, while those that do only have small numbers of places available. **Many families are unwilling to allow their child to live in generic disability supported accommodation as they believe that these services are not able to provide for the complex needs of individuals with PWS.** A common concern cited by telephone interview participants was open availability of food in these houses. As a result, many carers reported waiting until a position became available in a specific PWS house or paying high costs to access disability support workers to provide living assistance privately. There is a need for increased access to specific PWS supported accommodation or access to funding to cover the costs of full time care. In light of this**, individuals with PWS would highly benefit from access to the NDIS funding stream 'user cost of capital', where financial support is provided to subsidise the costs of living in specialist disability housing** (Schedule J Supports for specialist disability housing). This is particularly important for individuals with PWS with the impact of low employment rates and the implication of social and maladaptive behaviours, making it very unlikely that the individual will earn an income high enough to independently fund their own housing

An interesting statistic revealed by the online survey data was that only 5% of ALH were reported to be affected by psychosis, compared to 44% of those living out of the family home. This finding may support the speculation that adults with PWS are more likely to live in the family home if they present a lesser carer burden. ALH were also less likely to have caused injury to a family member or member of the public. However, under the **NDIS all persons with PWS should be offered the opportunity to**

access supported accommodation outside the family home to allow for greater autonomy and independence, regardless of the carer burden placed on the family.

Research has revealed that considerable savings in support funding may be possible by implementing detailed living skills programs for individuals that are living out of the family home in supported accommodation. Such a program has been implemented by the Arc of Alachua in Gainesville, Florida. Of the 80 persons with PWS living in their supported accommodation homes, 12 have 'graduated' to apartment style living with a lover level of support. Savings in the residential support component of the program for these individuals is running at around 65%. Further research into this program and funding to encourage similar programs in Australia could lead to substantial future savings for NDIS.

Recommendations	
Core	• Accommodation supported by carers trained in managing PWS in an environment with food restriction should be offered to all people with PWS over the age of 18.
Capital	• Funding for house modifications required for all individuals who are moving out of home. • Funding for electronic and physical security aids for all individuals who are moving out of home.

Education

PWS is characterised by mild to moderate mental retardation (Gross-Tsur, 2001). Although individuals with PWS have varying degrees of cognitive ability, it is widely accepted that their relative cognitive strengths lie in visuospatial perception, organization and solving jigsaw puzzles (Whittington, 2004; Chen, 2007). Nevertheless, they typically have low cognitive function, language deficits, poor short-term memory, and difficulty with executive processing (Ho, 2010; Chen, 2007). This was reflected in the survey results with all but one individual requiring on average a significant amount of assistance. High levels of school attendance was noted in both primary and secondary age children, however, this was typically either a special school, or if a mainstream school, using the assistance of an aid. Despite the high attendance rate, there remained 6 children in the primary and

secondary school groups combined that were not attending any formal education. Although there may be a range of explanations for this, the IQ-based entry system for special schools may contribute, impeding access and funding for special school attendance in PWS individuals with above 70 IQ**. The IQ benchmark of 70 and below for acceptance into special schools, is based on the narrow definition for intellectual functioning. This does not take into account the maladaptive behaviours and high supervision requirements, that occur irrespective of IQ level of an individual with PWS, which often make mainstream school unacceptable and inappropriate.** One mother reported in a telephone interview that her child had been rejected from a special school because his IQ is just above the minimum required for entry. However, he is also unable to attend a mainstream school.

> *"He has an IQ that is too high to attend a special school so he has to attend a mainstream school. But his care needs are too high for him to be able to attend a mainstream school. So there is no place for him"*

Although average IQ is between 60-70 in the PWS population (Cassidy, 2012), it has been previously shown that this does not correlate with cognitive ability (Gross-Tsur, 2001). Therefore, despite having relatively high IQs, individuals with PWS are unable to cognitively function at this level and typically have poor academic achievement (Whittington, 2004; Gross-Tsur, 2001). In concert with this, the survey result found that all bar one respondent required on average, a significantly high level of assistance for problem solving. Furthermore, moderate issues with remembering were also present in 96% of the respondents. While it would have been ideal to obtain further data to explore the relationship between IQ and other factors such as care needs, only 33 respondents reported the IQ of the individual with PWS in the survey responses. Therefore, it is not possible to conduct statistical analyses that would yield significant findings.

> *"Some individuals show higher IQ, doesn't often work like that, cognitive and social skills do not relate to the IQ"*

While it would appear that individuals with PWS that are attending school are well supported by the education system - whether in a special school or a mainstream school (through the provision of personal aids) - the NDIA could provide further support and ease carer stress. Respondents claimed

that on average, the individual with PWS required 8 out of 10 level of assistance for road safety. When combined with the need for constant supervision in the community to manage food seeking behaviours, it is quite evident that individuals with PWS are unable to safely travel to school independently. This means that parents and carers are tasked with transporting the individual to school. While this may not be a problem for some families, for others it could further limit their ability to work. Given the financial burden of the condition which has been discussed earlier, any measure that would enhance the ability of carers to undertake employment would be beneficial. Hence, the provision of supervision (in the form of carers or aids) to assist individuals with PWS to arrive safely at school should be a capacity provision that will be especially relevant to the high school group.

Many interviewees described schools which provide support that is sensitive to the needs of the individual with PWS. For example, one school provided a locker space in the library in order to eliminate the temptation of seeing other students' lunches in the hallway. Many other schools stock special, healthy treats for the individual as rewards and to eat during class celebrations such as birthdays, where cake would otherwise be provided to them. However, some interviewees recalled situations where the effects of the condition were not considered and led to episodes of anxiety or tantrums for the individual with PWS. In particular, one mother described having to collect the individual with PWS from school due to the open display of food which resulted in an anxiety attack. Therefore given the unique needs of the PWS population**, it is important that those responsible for the continuum of care of the individual with PWS are knowledgeable about the condition and are well trained** to minimise and manage the incidence of maladaptive behaviour. To ensure this, teachers and carers of children with PWS should receive adequate PWS-specific training.

Recommendations	
Core	• Teachers and carers responsible for individuals with PWS should be adequately trained to provide supervision and care that is sensitive to the needs of the individual with PWS.
Capacity	• Supervision should be provided for individuals with PWS to travel to school in order to relieve carer burden whilst also ensuring safety and minimising food seeking behaviours on the way to school. • A scribe could be offered for school examinations if the individual with PWS is attending a mainstream school.

Communication

Speech and language development is delayed in individuals with PWS to variable degrees (Lewis, 2002). Typically PWS sufferers do not communicate with vocabulary until 18 months of age, which is 6 months later than their non-disabled, same age counterparts (Ho, 2010). In more severe cases, verbal language is not displayed until 6 years of age (Ho, 2010). They have poor speech-sound development, reduced oral motor skills and language deficits which can proceed into adulthood (Ho, 2010). **Almost all individuals with PWS from the online survey have difficulties with language, communication, spoken communication, written communication and being understood.** Speech pathologists are often involved from infancy and throughout the preschool age as part of the early childhood intervention services (Chen, 2007). They play an important role in improving oral motor skills for feeding, and later, receptive and expressive communication skills (Chen, 2007). However, given the extent of communication impairment in this population the need for speech therapy extends beyond this scope.

While previous studies (Lewis, 2006) and the current survey results show that the extent of language difficulties decreases with age (likely the result of early intervention), it still remains a problem to some extent in the older age groups. **All respondents in the preschool aged group were undertaking speech therapy, reducing in primary school and further in secondary school** (86% and 23% respectively). Sadly no adults from the online survey were having current speech therapy, despite the average rating for language and communication being moderately high for almost all participants. It would appear, then, that while speech and language problems extend into adulthood, individuals are not utilizing services which could improve their ability to communicate, and thus consequently influence their social and emotional health. This may be due to the associated costs of sourcing a speech therapist privately.

The provision of augmentative and alternative communication (AAC) tools such as Makaton sign language can also be beneficial. Makaton is a recognised communication system for people with learning difficulties and it is used extensively by both children and adults. Toddlers and preschool children with expressive language delays in particular have been shown to benefit from this form of communication (Foreman 1998).

Speech therapy should be a core support for individuals at school age through the NDIS to enhance communication and ability to socialise at school. It should also be a core support for adults to facilitate the transition into new roles and different environments such as work or a group home.

Recommendations	
Core	• Speech therapy should be offered to individuals across all ages in order to improve their ability to communicate.
Capacity	• Training and education in Makon sign language could be offered to facilitate communication and learning.

JUSTIFICATION FOR INCLUSION OF PWS ON 'NDIS LIST A - PERMANENT IMPAIRMENT/FUNCTIONAL CAPACITY – NO FURTHER ASSESSMENT REQUIRED'

It can be seen from the information in this report that PWS causes a permanent impairment with substantially reduced functional capacity in all diagnosed individuals. Therefore, 'no further assessment required' should be the position of the NDIA/S, and PWS should appear on List A for access assessment.

The evidence suggests that 100% of people with PWS in Australia, who meet the residency and age criteria, will qualify for support funding under the NDIS. This statement is based on the following analysis of the 5 qualifying criteria detailed in the NDIS Act (2013):

1 The person has a disability that is attributable to one or more intellectual, cognitive, neurological, sensory or physical impairments or to one or more impairments attributable to a psychiatric condition:

- All people with PWS suffer from a multiplicity of these impairments and conditions. In fact, the disease is so complex is its presentation, you could argue that the individuals suffer from not just one (as they would need to qualify) but all 7 of these impairments and conditions.
- Clinical features including short stature, delayed growth velocity, developmental delay, cognitive disability and maladaptive behaviours can emerge (Angulo, Butler & Cataletto, 2015; Butler, Lee & Whitman, 2006).

2 The impairment(s) are likely to be permanent:

- The disease can be found in the body at the chromosomal level. While there are many interventions designed to minimize the impact on the sufferer, there is no known cure at this stage.
- PWS is a multi-system disorder associated with the absence of a paternally-inherited gene usually expressed on chromosome 15 (Angulo, Butler & Cataletto, 2015)

3 The impairment(s) result in substantially reduced functional capacity to undertake, or psychosocial functioning in undertaking, one or more of the following activities:

- Communication – 100% of the preschool, primary and secondary school groups reported communication to be a problem.
- Social interaction – 100% of the primary, secondary school and ALSA groups reported repetitive questioning as a major behavioral problem. 100% of the respondents over the age of 7 reported tantrum behaviours with a severity of 6-8 out of 10. 100% of the primary, secondary and ALSA required assistance in developing and fostering friendships. 100% of primary, secondary and adult respondents reported problems coping with social and emotional issues. Individuals with PWS suffer more severely from withdrawal, depression and thought problems than their age and IQ matched counterparts.
- Learning – all but one participant indicated problems with learning tasks with an average degree of difficulty of 7 out of 10.
- Mobility – 100% of participants require mobility assistance. 91% of respondents indicated that food seeking behaviours within the community was an issue to some extent, full time supervision within the community is recommended as a core support offered to all people affected by PWS.
- Self-care – 105 of the 106 respondents to our survey indicated that assistance was required in this area. The one acceptation indicated that, while their 23yo son was able to perform these tasks independently, he still required verbal prompting from a carer to carry out the process.
- Self-management – 100% required assistance with self-management with the severity of the problem ranging between 7 to 9 out of 10. 100% of people with PWS in the secondary school and adult categories require assistance with food related activities. 100% of all respondents require assistance with money management.

4 The impairment(s) affect the person's capacity for social and economic participation:

- As is evident from the above statistics, having PWS dramatically impairs the capacity of individuals to participate socially and emotionally.
- Low mood and anxiety were reported as a problem by 98% of individuals where this was relative to their age.

- Mobility was identified as a major problem for all age groups with 100% of individuals needing some degree of assistance
- People with PWS working are substantially lower than the labour participation rate of 52.8% for Australians aged 15-64 with a disability as reported in 2012 (ABS, 2015c).
- While the maladaptive behaviours of individuals with PWS may be social deterrents, previous studies have suggested additional social inadequacies. Individuals typically exhibit poor relationships with their peers, often lack friends and prefer solitary activities (Dimitropoulous, 2013).

5 The person is likely to require support under the National Disability Insurance Scheme (NDIS) for the person's lifetime:

- PWS is currently incurable, resulting in the individuals requiring support from the NDIS for their lifetime.
- The average rating for anxiety increased incrementally between age groups, suggesting that it becomes a greater problem as the individual ages.
- The adult groups reported high severity of food seeking with an average of 8 out of 10, with 10 being extreme severity
- Skin picking severity increasing steadily across the age groups
- Difficulties with communication exist across all ages, even the adult groups.

The survey responses indicated that the management of this syndrome becomes progressively more difficult as individuals approach adulthood.

We have found no evidence of persons with PWS being capable of living completely independently and in that respect they all need support at all ages. The provision of appropriate support in all residential settings is vital in order to maximise individuals' capacity, enable community inclusion, and maintain good health and wellbeing.

A 20 year longitudinal study of 967 families by Professor Stewart Einfeld found that the families of people with PWS had the most impaired family well-being. Even more so than the families of children with autism. Reduced family well-being was directly associated with the severity of the individual's behavioural problems, particularly the rage attacks and hyperphagia. This evidence justifies support for families of individuals with PWS at least equal to that of families of individuals with ASD who require substantial support.

Furthermore, in September 2015, the NDIA Intellectual Disability Reference Group (set up by the NDIA Independent Advisory Council) met in Canberra. The Victorian Representative, Mr. Kevin Stone, advises that the Group was unanimous in its recommendation that Prader-Willi Syndrome should be included on List A for Access Assessment purposes; That is, NDIS List A - Permanent impairment/functional capacity – no further assessment required.

LIMITATIONS

This study reports on a small sample size (106 participants) of people with PWS. However, PWS is thought to affect 585 - 800 Australians (RACGP, 2012; The PWS Better Living Foundation, 2013a) so the online survey has captured a significant proportion of this population. Due to many of the people with PWS being under the age of 18, the researchers were unable to seek the opinions of people with PWS directly as it was not achievable to gain informed consent from parents and guardians of these children within the timeframe. As the primary carer was the survey respondent and interview participant, the researchers relied on this person to report the toll on the entire family, including the person affected by PWS. While the online survey was comprehensive, it would have been beneficial to include further questions surrounding domestic activities of daily living including the help required for cleaning around the home and washing clothes. With this information, recommendations could have been made surrounding the requirements for help around the home.

The telephone interviews were undertaken as a sample of convenience. The choice to interview those first to respond to the questionnaire may have introduced a source of bias, as these respondents may be considered more proactive and educated regarding the role of research in advocating for people with PWS, and may be more likely to be from a higher socioeconomic background with greater ease of access to technology with which to complete the survey. Furthermore, the interviews were completed by three inexperienced students, with differing styles of questioning.

The study did not separate data into individuals without access to the NDIS and those currently receiving NDIS support in trial sites across Australia. The results were also not reported according to the person's underlying genetic abnormality, despite deletion and uniparental disomy abnormalities being known to present differently (Veltman et al., 2004). The limited statistical analysis is a further limitation, which does not allow for the representation of relationships between the different variables. It was decided that these investigations were beyond the scope of the current report.

CONCLUSION

Individuals with PWS are known to have extensive care needs in the domains of dietary management (Allen, 2011, Angulo & Cataletto, 2015), behavioural (Cassidy, 2012) and emotional issues (Skokauskas et al., 2012) cognitive function (Whittington, 2004; Gross-Tsur, 2001) and personal care (Scardina, Fuca & Messina, 2007). In addition this produces social, financial, and emotional stress on those involved in the care of the individual with PWS (Skokauskas et al., 2012).

This group will present a complex challenge to the allocation of services and funds by the NDIS because of their small population and unique care needs. In particular, they will require unique supports and supervision to live out of the family home in order to successfully manage their dietary needs, personal safety and community access.

This report details the findings from three sources; an online survey completed by 106 primary carers for Australians affected by PWS, telephone interviews with 21 of these carers and additional comments by 6 health professionals. Recommendations are provided based on the care needs reported by all three sources with the aim to guide further conversation with the NDIA.

As demonstrated throughout this extensive report PWS results in a permanent disability that substantially reduces functional capacity and as such PWS should be considered for inclusion in the List A conditions as stipulated in the NDIS Operational Guidelines for – Access – Disability Requirements (2014).

As demonstrated throughout this report, and confirmed by the NDIA Intellectual Disability Reference Group, PWS results in a permanent disability that substantially reduces functional capacity and as such PWS should be considered for inclusion in the List A conditions, ''Permanent impairment/functional capacity – no further assessment required.''

Central recommendations of this report are listed below and summarised in Table 2.

Core

- Social and activity groups supervised by appropriately trained staff
- Allied health -
- Psychology services, beginning in the primary school period
- Physiotherapy

- Dietician with an understanding of the PWS condition, beginning during the preschool period
- Dentistry
- Occupational therapy
- Speech therapy
- Podiatry
- Appropriate exercise programs and trained carers to facilitate participation
- Personal care assistance for individuals living out of the family home
- Respite care
- Day programs which include physical activity and skills training for individuals not attending full time schooling or employment
- Carer support for community activities such as attending schooling, a day program, employment and medical appointments
- Accommodation supported by carers trained in managing PWS for individuals over the age of 18

Capacity

- Psychology
- Pre-emptive psychologist advice for families with preschool aged children with PWS
- Counselling or psychology for family members
- Outside school hours care
- Full time supervision within the community for individuals who display food seeking behaviours within the community
- Assistance to gain employment for people with PWS over 15 years of age
- Carer support when travelling to and participating in employment
- Carer support for full time supervision within the home environment
- Disability support workers who are trained in the care of people with PWS to provide substantive and meaningful support programs in place of babysitting services

- Personal care assistance for individuals living in the family home

Capital

- Exercise equipment, gym memberships and personal trainers
- Specialised equipment and home modifications
- Equipment or home modifications to ensure food can be locked away within the home environment
- Electric toothbrushes
- Mobility aids and orthotics
- Security aids

While these recommendations have been made for the PWS population in Australia, it is important to note that the condition presents variably within a range. All persons present with social, cognitive and functional impairment, which can fall within a range from substantial to severe.

It is our opinion that Prader-Willi Syndrome should be placed on 'NDIS List A - Permanent impairment/functional capacity – no further assessment required'

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CONFLICTS OF INTEREST STATEMENT

The project was undertaken by the University of Melbourne on behalf of the PWS Better Living Foundation, who recognised the need for further research in this area. The researchers were Doctor of Physiotherapy students from the University of Melbourne who undertook this research as a final year project with the support of a senior research physiotherapist and research assistants within the Department of Centre for Health, Exercise and Sports Medicine. Professor Kim Bennell initiated the project following discussions with James O'Brien, a Director of the PWS Better Living Foundation. Both Kim Bennell and James O'Brien have had personal experiences with PWS. There are no other conflicts of interest to report.

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APPENDICES

Appendix 1: Online survey questions

Prader-Willi Syndrome Survey

Consent Form

THE UNIVERSITY OF MELBOURNE

CENTRE FOR HEALTH, EXERCISE AND SPORTS MEDICINE
DEPARTMENT OF PHYSIOTHERAPY

CONSENT FORM
Persons participating in research projects

Title: Supporting people with Prader-Willi Syndrome: understanding needs, challenges and strategies

Investigator(s): Prof Kim Bennell, Dr Belinda Bilney, Mr James O'Brien, Mr Matthew Etty-Leal, Ms Rachel Nelligan

1. I consent to participate in this project, the details of which have been explained to me, and I have been provided with a written plain language statement to keep.

2. I consent to my data being used to answer other questions relevant to Prader Willi Syndrome.

3. I understand that after I sign and return this consent form it will be retained by the researcher.

4. I understand what my participation will involve and I agree that the researcher may use the results as described in the plain language statement.

5. I acknowledge that:

(a) the possible effects of participating in the program have been explained to my satisfaction;

(b) I am aware that participation in this study is voluntary and I have been informed that I am free to withdraw from the project at any time without explanation or prejudice and to withdraw any unprocessed data I have provided;

(c) the project is for the purpose of research;

(d) I have been informed that the confidentiality of the information I provide will be safeguarded subject to any legal requirements;

(e) I have been informed that a copy of the research findings will be forwarded to me, should I wish.
*
[ ] I consent to participating in this research project
The plain language statement for this survey is available here*
[ ] I confirm that I have read and understood the plain language statement.
Would you like a copy of the survey outcomes?*
( ) Yes
( ) No
Email address that the survey outcomes should be sent to:*
_____

Personal details about person with PWS

1) What is the person completing this form's relationship to the person with PWS? For example, if you are the mother of the person with PWS, please write "mother".*
_____

2) What is the person's date of birth?*
_____

3) What is the person's gender?*
( ) Male
( ) Female

4) What is the person's height (cm)?*
_____

5) What is the person's weight (kg)?*
_____

6) What type of PWS was the person diagnosed with?*
( ) Deletion
( ) Uniparental Disomy
( ) Unknown

7) What is the person's postcode?*
_____

Further Characteristics

8) At what age was there a diagnosis of PWS?*
_____

9) For how long was the person hospitalised after birth? (please select one)*
( ) ≥ 1 week
( ) 1-2 weeks
( ) 2-4 weeks
( ) 4-12 weeks
( ) ≥ 12 weeks

10) Please select all family members that are currently living*
[ ] Mother
[ ] Father
[ ] Siblings... if so, how many:: _____

11) What is the person's current living situation?*
( ) At home
( ) In group home
( ) Independent with support

12) If they are living at home, who lives in the home with them? (tick all that apply)*
[ ] Mother
[ ] Father
[ ] Grandparents
[ ] Siblings
[ ] Other: _____
[ ] Not applicable

13) If they are living at home, would you like to access out of family home respite (short term supported accommodation)?*
( ) Yes
( ) No
( ) Not applicable

14) PWS Better Living Foundation is particularly interested in access to suitable housing for adults with PWS. Please complete the following sentence as it best relates to your situation.

Is housing for the person an issue:*
( ) Now (urgently)
( ) In the next 6 months
( ) In the next 2 years
( ) In the next 5 years
( ) In the next 20 years
( ) Not required

Further Characteristics

15) Would you like to access full-time supported accommodation outside the family home?*
( ) Yes
( ) No

16) Is the mother of the person with PWS working?*
( ) No
( ) Yes full time
( ) Yes part time
( ) Not applicable

17) Is the father of the person with PWS working?*
( ) No
( ) Yes full time
( ) Yes part time
( ) Not applicable

18) What is the IQ level of person with PWS? (if known)
_____

19) Select the current support networks that have been used within the last 6 months for the person with PWS? (tick all that apply)
*
[ ] Parents
[ ] Siblings
[ ] Aunts/uncles
[ ] Grandparents
[ ] Friends of family
[ ] Paid nanny/carer
[ ] Supported group outings
[ ] Camps
[ ] Overnight respite
[ ] Out of family home accommodation
[ ] None of the above

Medical History

20) Other medical conditions that the person with PWS has (Tick all that apply)*
[ ] Scoliosis
[ ] Hearing impairment
[ ] Visual impairment
[ ] Diabetes
[ ] Depression
[ ] Psychosis
[ ] Other: _____
[ ] None of the above

21) Medications that the person with PWS is taking (Tick all that apply)*
[ ] Growth hormone
[ ] Sex hormones - oestrogen or testosterone
[ ] Psychotropic
[ ] Other: _____
[ ] None of the above

22) Medical specialists that the person with PWS is currently seeing (tick all that apply)*
[ ] Paediatrician
[ ] Respiratory physician
[ ] Endocrinologist
[ ] Psychiatrist
[ ] Orthopaedic surgeon
[ ] Opthamologist
[ ] Other: _____
[ ] None of the above

23) Other health professionals the person with PWS is currently seeking (tick all that apply)*
[ ] Physiotherapist
[ ] Dietitian
[ ] Dentist
[ ] Speech therapist
[ ] Occupational therapist
[ ] Audiologist
[ ] Podiatrist
[ ] Exercise Physiologist
[ ] Psychologist
[ ] None of the above

Daily activities

24) Is the person with PWS attending school?*
( ) Yes
( ) No

25) If yes, tick one:
[ ] Primary school without aid
[ ] Primary school with aid
[ ] Special school (primary)
[ ] Special development school (primary)
[ ] Part primary school / Part special school
[ ] Secondary school without aid
[ ] Secondary school with aid
[ ] Special school (secondary)
[ ] Special development school (secondary)

26) Is the person with PWS employed?*

( ) Yes
( ) No

27) How often does the person undertake this employment (on average)?
( ) 5 or more days per week
( ) 4 days per week
( ) 1 day per week
( ) 3 days per week
( ) 2 days per week
( ) Other: _____

28) Is the person with PWS attending a day program?*
( ) Yes
( ) No

29) How often does the person attend the day program?
( ) Weekly
( ) Fortnightly
( ) Monthly
( ) Other: _____

PWS behaviours/characteristics

To what extent is food seeking a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is food seeking no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is stealing food from shops a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is stealing food from shops no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is stealing food from other people's plates a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is stealing food from other people's plates no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is stealing food from bins a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is stealing food from bins no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is stealing food from friends' lunch boxes a problem for the person with PWS?*
No problem Extreme problem

( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is stealing food from friends' lunch boxes no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

PWS Behaviours/Characteristics

To what extent is stealing money a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is stealing money no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is skin picking a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is skin picking no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is anxiety a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is anxiety no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is low mood a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is low mood no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent are temper tantrums/rages a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Are temper tantrums/rages no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is repetitive questioning a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is repetitive questioning no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is daytime sleepiness a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is daytime sleepiness no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

PWS Behaviours/Characteristics

To what extent is sleep disturbance a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is sleep disturbance no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is fatigue a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10
Extreme problem

Is fatigue no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent are difficulties with communication/language a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is difficulty with communication/language no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

To what extent is obesity a problem for the person with PWS?*
No problem Extreme problem
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Is obesity no problem because:*
( ) The person could display the behaviour but doesn't
( ) The behaviour is not relevant due to age or capabilities

Degree of independence of person with PWS

How much help does the person with PWS need for mobility around the house?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for mobility around the house because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for getting around the community?*
No help Maximal help

( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for getting around the community because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for spoken understanding?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for spoken understanding because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for written understanding?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for written understanding because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for being understood?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for being understood because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

Degree of independence of person with PWS for social interaction and self-care

How much help does the person with PWS need for making and keeping friends?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for making and keeping friends because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for coping with feelings and emotions?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for coping with feelings and emotions because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for dressing?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for dressing because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for showering and bathing?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for showering and bathing because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for cleaning teeth?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for cleaning teeth because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for toileting?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10
Maximal help

Does the person require no help for toileting because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for managing health?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for managing health because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

Degree of independence of person with PWS for learning and self-management

How much help does the person with PWS need for learning new tasks?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for learning new tasks because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for remembering?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for remembering because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for managing money?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for managing money because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for performing daily tasks around the house?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for daily tasks around the house because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for problem solving?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for problem solving because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for making decisions?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for making decisions because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

How much help does the person with PWS need for road safety?*
No help Maximal help
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Does the person require no help for road safety because:*
( ) The person could require help but doesn't
( ) The activity is not relevant due to age or capabilities

Impact of caring for person with PWS on families

How would you describe the degree of the emotional toll on your family?*
No impact Extreme negative impact
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

How would you describe the degree of the physical toll on your family?*
No impact Extreme negative impact
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

How would you describe the degree of the financial toll on your family?*
No impact Extreme negative impact
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

How would you describe the impact of the condition on family relationships?*
No impact Extreme negative impact
( ) 0 ( ) 1 ( ) 2 ( ) 3 ( ) 4 ( ) 5 ( ) 6 ( ) 7 ( ) 8 ( ) 9 ( ) 10

Has the person caused injury to family members*

( ) Yes
( ) No

Has the person caused injury to a member of the public?*
( ) Yes
( ) No

Conclusion

The researchers are interested in completing a number of 30-60 minute telephone interviews with carers to obtain further details about the needs of people with PWS. Would you be willing to participate in a telephone interview? If yes, please provide your preferred telephone number and time of contact.*
( ) Yes
( ) No
Name:
_____
Contact telephone number:*
_____
Preferred time of contact:
_____

Appendix 2: Telephone interview prompts

a) It is important for the person with PWS to participate in exercise. Tell me about the physical activity routine of _____.
   - What different exercise programs are they currently doing?
   - How much time do they spend doing this?
   - What works for you? What doesn't work?
   - What things have helped with regards to accessing exercise programs?
   - What are the barriers to accessing exercise programs?
   - What supports do you think would be needed to make sure the person can participate in physical activity? How do you think this would help?

b) Uncontrolled eating is a major issue in Prader-Willi. Tell us about your experiences with this
   - Can you explain what you have had to change, if anything, in their environment because of uncontrolled eating?
   - Can you describe the costs associated with this? Did you have to cover the costs?
   - How have you managed uncontrolled eating in other places?
   - What has worked in relation to managing uncontrolled eating in your child? What has not worked?
   - What impact has this had on you and your family?
   - What services or programs could be of benefit to help with uncontrolled eating?
   - Can you describe any other unhealthy habits that _____ engages in? (e.g. smoking, eating non-food items, skin picking)

c) Behavioural issues are another major aspect of Prader-Willi. Tell us about your experiences with this?
   - What impact has this had on your child?
   - What has helped your child in this area?
   - How has this impacted on you and your family?
   - Would any additional services assist you in this area?

- Can you describe issues with excessive property or equipment damage? Can you describe the costs associated with this?

d) What are your expectations around the National Disability Insurance Scheme?
   - What do you know about it?
   - How do you think it will help your child?
   - What would you like it to do for your child?

e) Tell us about your experiences with disability support services
   - What have you found most helpful?
   - What services are missing that you would like to access?
   - What service providers or services, if any, would you like to recommend to other Prader-Willi families?

f) We are interested in the time period when the person with Prader-Willi was just born
   - In the online survey you completed, you said that _____ was diagnosed at age _____. Is that correct?
   - Would you have preferred a diagnosis in the 1st week? Why? How do you think this may have changed your situation?
   - Do you think an early diagnosis would have resulted in better health outcomes? Can you tell me more about this? Why do you think it would/wouldn't have?
   - In your opinion, can you tell me how an early diagnosis would/wouldn't have resulted in financial savings for the health system?

ADULT GROUP ONLY:

g) We're interested to hear about experiences for people with Prader-Willi regarding moving out of the family home or considering doing so.
   - Can you tell me about your thoughts on this topic with regards to _____?
   - What barriers have you experienced?
   - What supports do you think would be needed for _____ to move out of the family home if they wished to?
   - OR… what supports or services have been helpful in supporting _____ to live out of the family home?
   - What (would be/were) the reasons for _____ (moving out/not moving out) of the family home?

h) The next few questions are about the burden of the Prader-Willi condition on the adult.
   - Can you describe any police interactions? How many have occurred in the last 12 months?
   - Have you or anyone in your family experienced any legal or para legal visits regarding _____. How many interactions with VCAT and other legal services have occured in the last 12 months?
   - How often would you typically visit the GP regarding _____?
   - How often would you visit medical specialists?
   - How many ambulance trips and hospital admissions have occurred for _____?
   - How long were the hospital admissions?
   - Can you describe any issues with _____ following doctor's orders and taking medications?