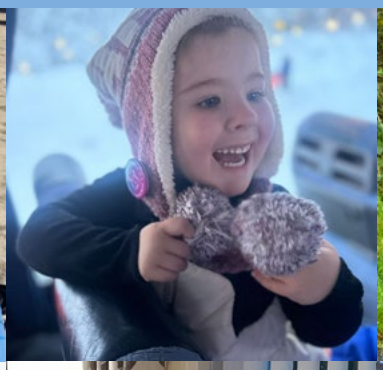
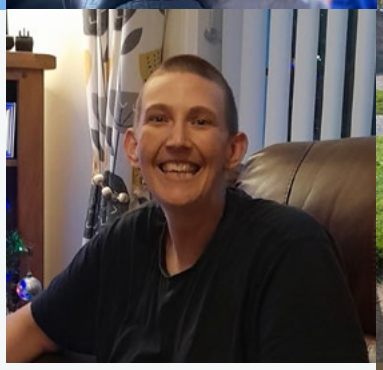
HSE National Disability Operations Community Services & Prader-Willi Syndrome Association of Ireland

A Report on the Joint National Pilot of the Health Service Executive and the Prader Willi Syndrome Association of Ireland

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PRADER WILLI SYNDROME ASSOCIATION IRELAND

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Remembering Dr. Michael Byrne.

This report is dedicated to the memory of Dr Michael Byrne. From the outset, Michael was able to hear the voices of people and families living with Prader-Willi syndrome. PWSAI are grateful for Michael's empathetic guidance and miss his wisdom.

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Introduction from Rory Tierney, Chairperson, PWSAI.

Since established as a charity, Prader Willi Syndrome Association Ireland (PWSAI) has benefited from the dedicated efforts of numerous committees over the years. Their advocacy and work have consistently aimed at enhancing services across all facets of life to meet the needs of individuals with Prader-Willi Syndrome (PWS) and their families.

The genesis of this report is a testament to the unwavering commitment and determination of those who have devoted their time to PWSAI, setting a robust foundation for the charity with a singular, clear vision: to create a world where individuals with PWS, their families, and caregivers are supported in care, education, and well-being.

This report represents the next phase in our development, embracing a holistic life approach to support people with PWS and their families across all stages of life.

Following the publication of «A Population-Based Profile of Prader-Willi Syndrome in Ireland» in 2017, it became evident that further work was needed to advocate for services and enhance the state›s understanding of PWS to adequately support individuals and their families.

In late 2017, Simon Harris called for the establishment of a joint working group to examine all aspects of Prader-Willi Syndrome. From this collaboration emerged the Report on the Joint National Pilot of the Health Service Executive and the Prader-Willi Syndrome Association of Ireland (PWSAI).

This report amalgamates evidence-based literature, family and caregiver experiences, and the voices of individuals with PWS, spanning all aspects of the syndrome from birth to adulthood.

The recommendations outlined here are crucial elements in the care and support of individuals with PWS, promising to enhance the lives of those presently affected and establish robust foundations for future generations.

This report serves as a powerful advocate for individuals with PWS and their families, empowering them to comprehend their needs and advocate for the highest standards of care and well-being from service providers, while also highlighting areas for improvement in the future.

PWSAI extends heartfelt gratitude to all contributors to this report, especially the individuals and families whose knowledge and experiences were instrumental in its creation. Additionally, we express our thanks to the HSE for their support and efforts in bringing this report to fruition.

Without a doubt, this report will make a significant positive impact on the lives of those with PWS, both today and into the future.

Introduction from Bernard O'Regan, Head of Operations for Disability Services, Health Service Executive.

I would like to welcome the publication of this comprehensive Report on the Joint National Pilot of the Health Service Executive and the Prader Willi Syndrome (PWS) Association of Ireland. This is the first time in Ireland such a focused approach has been taken to the needs and experiences of people with this complex genetic disorder, and will greatly assist the HSE and other service providers to understand the specific needs of people with PWS and their families. This document aims to provide an exploration of the current policy and service framework, the evidence based literature, the experiences of existing services from family carers, and give voice to the perspectives of service users, as well as highlighting potential areas for improvement in service provision. I believe the approach taken by the Joint Working Group, and all those who took part in this endeavour, contributes valuable insights that clearly articulate how to enhance the quality of support for individuals and their families affected by PWS.

This report centres on the voices and perspectives of those directly impacted by PWS to offer an authentic understanding of the lived experience of those affected and sets out realistic ways that services can improve those experiences.

The HSE extends particular gratitude to all involved but especially the individuals and families who generously shared their insights, contributing to a detailed exploration of the current support systems surrounding Prader-Willi Syndrome, and to a person centred exploration of the best ways to improve these systems.

National Working Group

The National Working Group on Prader-Willi syndrome (NWG) was established in 2018 by Minister for Health Simon Harris to review the effectiveness of HSE services accessed by people with Prader-Willi syndrome. The NWG is a partnership between the HSE and the Prader-Willi Syndrome Association Ireland (PWSAI). The group was tasked with developing and testing positive models of social care provision for people with PWS and their families.

Context of the Report

This report sets out evidence-based recommendations for the development of high quality, safe and effective health and social care services that meet the needs of those with PWS and their families. High levels of carer burden are associated with PWS, and particular attention was paid to developing models of social care provision that, not only meet the unique and complex needs of individuals with the syndrome, but also those of their parents and siblings.

What is Prader-Willi Syndrome?

Prader-Willi Syndrome (PWS) is a complex multi-system disorder caused by the lack of expression on paternally active genes in the PWS critical region on chromosome 15. 4 – 5 new babies are diagnosed in Ireland annually. Disruption in the hypothalamic region of the brain results in persistent and insatiable appetite, hyperphagia and hunger related eating behaviours. Hyperphagia and subsequent obesity are cardinal features of this rare genetic disorder: excessive overeating, stomach necrosis and rupture, cardiovascular disease, respiratory failure, sleep apnoea and diabetes mellitus are related life-threatening comorbidities. PWS is associated with intellectual disability, and a wide variety of physical and mental health difficulties.

PWSAI

Prader-Willi Syndrome Association Ireland (PWSAI) is a registered charity. Its mission is to raise awareness and understanding of PWS and to improve the choice and quality of care, education and support for persons with PWS. PWSAI's National Development Manager funded by the HSE will drive the implementation of the recommendations of this report. In 2022 PWSAI hosted the 11th international PWS conference, in partnership with the International Prader-Willi Syndrome Organisation, (IPWSO).

Executive Summary

Chapter 1. Introduction

- The National Working Group on Prader-Willi syndrome was established to review the effectiveness of HSE services accessed by people with PWS, particularly social care provision.
- The NWG was tasked with developing and testing positive models of social care provision that effectively meet the needs of individuals with PWS and their families.
- The National Clinical Programme for People with Disabilities (NCPPD) task group worked to implement the key recommendations of this report.
- The needs associated with the syndrome are unique and complex, and require specialist, syndrome-specific responses.
- A universal feature of PWS is a lack of satiety which leads to hyperphagia (excessive eating).
- The hunger felt by the individual is of genetic origin and outside the control of the sufferer: there is no cure or treatment at present.
- Individuals with PWS become morbidly obese and die in early adulthood (or sooner) unless their access to food is strictly controlled.
- Food security is achieved by securing food access across all environments, supervising food access across all environments, posting mealtimes and menus, and training all team members.
- In addition to hyperphagia, other behaviours, such as severe temper outbursts, may further complicate and increase the need for support

Chapter 2. Literature review

- Early diagnosis and parental knowledge about PWS help families understand and manage the early 'failure to thrive' stage in the syndrome. It also prevents over-feeding and obesity once the uncontrollable appetite or urge to eat, known as hyperphagia, develops in early to middle childhood.
- The lack of ability to regulate energy intake and the lack of satiety are crucial to understanding the needs of people with PWS
- Dietary, behavioural and environmental interventions, for example, locked kitchens and provision of 'psychological food security', or one to one supervision must be put in place to support the individual with PWS, and to prevent early mortality and obesity related comorbidities.
- The core discipline in the management of PWS is endocrinology with support from key specialities such as dietetics, orthopaedics, respiratory, ENT, dentistry, psychiatry, psychology, GI.
- PWS is associated with high rates of mental health diagnoses such as anxiety and mood disorders. Psychotic illness is very common among those with the mUPD genetic subtype.
- Challenging behaviours associated with PWS include food seeking, emotional outbursts, skin picking, repetitive questioning, difficulty with transitions and non-compliance.
- High pain threshold, lack of vomiting, and poor temperature regulation can make the recognition of developing physical illness difficult.
- The impact on carers and families is enormous, affecting carer health, carer career, family relationships and sibling mental health.
- Specialist respite is recognised, both nationally and internationally, as a necessary support for families of people with PWS
- Individuals with PWS continue to need intensive support throughout adulthood and require full time specialist residential services when they leave the family home. Food security is achieved by securing food access across all environments, posting mealtimes and menus, and training all team members, (Tauber et al, 2014).

Chapter 3. Workstream 1

- PWS is a complex, multi-systems condition: management requires involvement from all arms of the HSE -Acute, Mental Health, Primary/ Community, Disability / Social Care.
- Workstream 1 examined PWS family carer experience of all HSE services accessed or needed by their family member with PWS, particularly social care provision. (Educational provision was also included.)
- A qualitative research project was undertaken which consisted of two phases – an online survey and a peer-facilitated workshop.
- Three overarching themes emerged from the research that were consistent and present across all types of service provision and applied to all age groups: (1) Dissatisfaction with services, (2) Behavioural Issues and (3) Impact on Family.
- The subthemes that emerged from the key themes were
  - » The subthemes for 'Dissatisfaction with Services' were: 'Lack of Resources/Inequity of Provision', 'Lack of Knowledge', 'Poor Communication/Coordination', and 'Delays in Access'.
  - » The subthemes for 'Behavioural Issues' were: 'Challenging Behaviours' and 'Destructive Behaviours'.
  - » The subthemes for 'Impact on family' were: 'Pressure on Caregivers',' 'Relationships within the Family' and 'Anxiety about the Future'.

Chapter 4. Workstream 2

- Workstream 2 set out to examine the optimal, evidence-based model of Social Care provision for people with PWS and their families with respect to Respite Supports, Residential Supports, Home Support and Day Services/New Directions.
- Methodology included facilitated workshops that looked at the totality of needs of individuals with PWS across the lifespan, a review of the literature, attendance at the 5th International PWS Professional Caregivers Conference, and examination of models of social care provision abroad, including site visits.
- Workstream 2 found that specialist approaches specific to PWS are necessary in order to ensure the physical safety and psychological wellbeing of the person with PWS.
- Food security is achieved by securing food access across all environments, supervising food access across all environments, posting mealtimes and menus, and training all team members.
- PWS is a complex, multi-systems condition: management of PWS requires a full-systems model approach as the individual needs to have access to acute and community services at any given time.
- The level of support needed by the individual must be rigorously assessed in advance of participating in education, work or volunteering: evidence suggests that one to one supervision is almost always necessary.
- Psychological and Physical Food Security is the cornerstone of PWS care as it frees the individual to live his/her life without constant anxiety related to food.

Chapter 5. Voices

- This chapter looks at what people with PWS in Ireland and beyond have to say about living with the condition.
- The chapter references findings from the Stepping Up Adults Symposium (SUAS) stream at the 11th International Prader Willi Syndrome conference in Ireland in July 2022.
- The participants in SUAS group were unanimous in their wish that everyone involved in their lives should have a greater awareness of PWS. They spoke about times when professionals supporting them did not understand them and how this had led to conflict. A strong desire for increased independence was expressed but participants spoke about how difficult that is without appropriate support and supervision.
- This chapter also looks at research by Prof Elizabeth Dykens, Vanderbilt University. Twenty-one young people with PWS were interviewed regarding their input on clinical drug trials as part of the Prof Elizabeth Dykens study that assessed how they perceive their syndrome and clinical trials. Five themes emerged:
    1. Struggles with chronic hunger and food-seeking that impede goals and relationships;
    2. Struggles with anxiety and outbursts, schedule changes and school;
    3. Distancing from PWS;
    4. Needs for clinical trials that cure PWS, reduce hunger or anxiety, and lead to improved outcomes;
    5. Needs for advocacy and awareness of PWS
- This chapter includes the voices of a number of children and adults with PWS who are members of PWSAI.

Recommendations

The implementation of the recommendations of this report would substantially change the life of those with PWS, leading to better physical and mental health outcomes, reducing morbidity and mortality, and enhancing quality of life by reducing suffering and providing supports to safely access what others take for granted such as further education and community participation . Other positive outcomes would be improved mental health and quality of life for siblings and family carers, and the protection of family units and relationships within families. The implementation of this report would also contribute to the promotion of equality in our society as it has been established in the literature that most care for individuals with PWS in Ireland is provided by women in the home (Gallagher et al., 2017).

Recommendation 1 - Quality and Delivery of Health and Social Care Services

1.1. Provision of a training for all stakeholders involved in the care and support of individuals with PWS.

1.2. Provision of professional training programmes to ensure that new graduates are competent to work with individuals with PWS.

1.3. Establish a voluntary register of individuals with PWS to inform immediate and future service planning and development

Recommendation 2 - Respite Services

2.1. Dedicated, PWS-appropriate, centre-based, residential respite is recommended as it allows for the development of infrastructure and practices that are oriented specifically* towards the needs of people with PWS.

2.2. It is essential that respite is regular and planned ahead of time.

Recommendation 3 - Residential Services

3.1. Residential services for persons with PWS should be PWS-specific* and meet the standards of IPWSO's Best Practice Guidelines for Standard Care in PWS

3.2. There should be at least one PWS specific residential service in each HSE Health area and the option of providing cluster type services in areas that require more than one service.

Recommendation 4 - Day Services / New Directions

4.1. PWS-specific supports should be provided for persons with PWS attending Day Services.

4.2. People with PWS will require one-to-one, PWS-specific support and an appropriate assessment of additional needs.

Recommendation 5 - Acute Services

5.1. Establishment of a National Centre of Expertise for Prader-Willi Syndrome, in line with National Rare Disease policy.

5.2. Provide additional support to existing multi-disciplinary, endocrinology led, PWS adult and paediatric clinics.

5.3. Development of Transitional Care for adolescents moving to adult services.

5.4. Timely delivery of time critical services.

Recommendation 6 - Mental Health Services

6.1. Consultation mental health services should be provided as part of multidisciplinary team provision at National Centres of Expertise for PWS.

6.2. Access to Mental Health Intellectual Disability (MHID) adult services based on a diagnosis of PWS.

6.3. Clear functioning pathways to adult MHID services and Child and Adolescent Mental Health Services (CAMHS and CAMHS-ID) in the community.

6.4. Early access to specialist behaviour support services should form part of the mental health services provision for children and adults with PWS.

6.5. Comprehensive planning for transition from paediatric to adult services.

Recommendation 7 - Educational and Lifelong Learning Supports for Individuals with PWS

7.1. Development of professional learning materials for teachers of students with PWS.

7.2. Consultation on the development of teacher education in PWS between PWSAI, the NCSE and other relevant stakeholders.

7.3. Educating individuals with PWS requires a 'whole school' or 'whole college approach'*: the pupil or student with PWS needs advocacy within the organisation

7.4. The level of support needed by the individual must be rigorously assessed in advance of participating in education. Assessment to include consideration of the supports needed around food security, food certainty, mobility and transport, evidence suggests that one to one supervision is almost always necessary.

7.5. Effective inclusion of adult learners with PWS in Further and Higher Education and Lifelong Learning services will require enhanced collaboration between the relevant stakeholders.

* A whole school approach is cohesive, collective and collaborative action in and by a school community that has been strategically constructed to improve student learning, behaviour and wellbeing, and the conditions that support these.

Recommendation 8 - Psychological Supports for Families

8.1. Access to therapy services for family members living with or caring for a family member with PWS.

CHAPTER ONE
Introduction

Prader – Willi Syndrome

Prader-Willi Syndrome (PWS) is a complex multi-system disorder caused by the lack of expression on paternally active genes in the PWS critical region on chromosome 15 (15 Q 11.2 - Q13). The birth incidence is approximately 1 in 10,000–25,000 (Smith et al., 2003). Taking data from Growing up in Ireland, 2011 and reports from the National Centre for Medical Genetics, Gallagher estimates a minimum birth frequency in Ireland of 1 in 11,000 annually (Gallagher et al., 2017), with 5-6 new cases being diagnosed each year. This is in keeping with international studies such as Smith et al., 2003.

Individuals with PWS are characterised by short stature, several endocrine conditions such as hypogonadism, growth hormone deficiency, hypothyroidism, central adrenal insufficiency, dysmorphic features, scoliosis osteoporosis, intellectual disability and behavioural and psychiatric issues.

One of the main characteristics of PWS is severe obesity, whose prevalence varies according to age, although in all cases it reaches a significant percentage. A prevalence of overweight and obesity of 40% has been reported in children and adolescents. When considering adulthood, the sum of obese and overweight individuals with PWS reaches a percentage ranging from 82% to 98%.

Although subjects with PWS have poor feeding and appetite in infancy, they develop uncontrolled appetite leading to weight gain after around three years. It is important to note that obesity in people with PWS has different characteristics than simple obesity. Lean body mass is lower while fat mass is higher in people with PWS compared with subjects with simple obesity having body mass index.

The development of obesity in PWS is mainly related to a dysfunction in the hypothalamic satiety centre and its hormonal regulatory circuitry, affecting food intake and energy expenditure. Disruption in the hypothalamic pathways of satiety control results in persistent and insatiable appetite, hyperphagia and hunger related eating behaviours. Aggressive and obsessive food seeking and storage, eating of inedibles, stealing of food or money to buy food are common among individuals with PWS. Controlling and manipulative behaviour, compulsivity, and difficulty with changes in routine, also contribute to abnormal eating. Alterations in several brain areas the hypothalamus, amygdala, hippocampus, orbital frontal and media prefrontal cortex play a crucial role in the abnormal food intake regulation in PWS .

PWS is associated with a high risk of specific physical illnesses that may go undetected and therefore untreated (Pellikaan et al 2020). Such physical morbidity is also associated with increased mortality rates across the lifespan (Bellis et al 2021). This increase in morbidity and mortality is largely as a consequence of hyperphagia and the development of severe obesity or the occurrence of choking and/or aspiration and resultant respiratory complications. Type 2 diabetes mellitus and obstructive sleep apnoea (central sleep apnoea can also occur) are common and related to levels of obesity

Hyperphagia

"Hyperphagia is excessive eating due to a lack of satiety. Individuals think about food all the time and live in a constant state of food-related anxiety. There is no cure or treatment for this brain disorder which prevents the person from feeling full. It is a lifelong condition and individuals do not have the capacity to keep themselves safe, regardless of levels of intelligence or adaptive functioning".

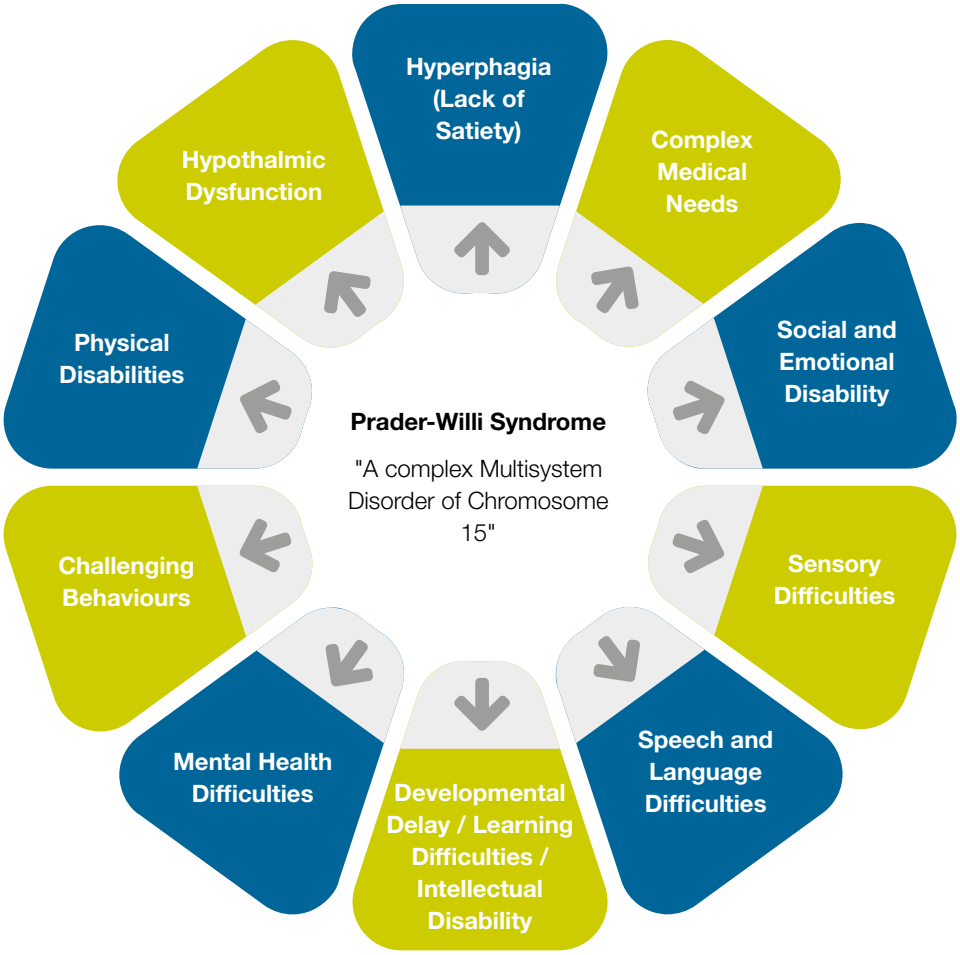
Hyperphagia is excessive eating due to a lack of satiety. People with PWS always feel hungry and never feel full: they cannot control the impulse to seek and eat food - even food normally considered inedible such as rotten food and pet food. Individuals think about food all the time and live in a constant state of food-related anxiety. At present there is no cure or treatment for this brain disorder which can present in individuals as young as three years of age and prevents the person from feeling full. It is a lifelong condition and the great majority of people with PWS do not have the capacity to keep themselves safe, regardless of levels of intelligence or adaptive functioning. Because hyperphagia and subsequent obesity are cardinal features of this rare genetic disorder, excessive overeating, stomach necrosis and rupture, cardiovascular disease, respiratory failure, sleep apnoea, diabetes mellitus, and related comorbidities can be life-threatening.

Eating of excessive amounts of food can be associated with the paralysis of the stomach so that it cannot empty (gastric paresis) and also in rupture of the stomach (gastric rupture) (Stephenson et al 2007). Severe constipation may occur as a result of gut muscle hypotonia, over-eating, and a failure to appreciate the need to defecate. All of these are acute and life-threatening conditions, requiring immediate medical attention. Whilst such problems

may not always be prevented, the level of risk will be influenced by the knowledge of those providing support and the extent to which preventative strategies are in place.

Unique risk factors apply to people with PWS that must be acknowledged, understood and expertly managed by implementing specific supports in order for the person with PWS to live safely and happily within their communities. 1 to 1 support is crucial as it frees the individual to live his/her life without constant anxiety related to food.

This report sets out evidence-based recommendations for the development of high quality, safe and effective health and social care services that meet the needs of those with PWS and their families. High levels of carer burden are associated with PWS, and particular attention was paid to developing models of social care provision that, not only meet the unique and complex needs of individuals with the syndrome, but also those of their parents and siblings.



Context for Current Report

At the request of the Minister for Health, Simon Harris, the Health Service Executive (HSE), in partnership with Prader-Willi Syndrome Association Ireland (PWSAI), established a National Working Group to develop, implement and test positive models of intervention and practice in supporting people living with Prader-Willi Syndrome and their families. The Working Group brought together parents in collaboration with professionals, both national and international, in order to lead and drive an agreed work programme as set out in the Terms of Reference (See Appendix?).

Membership of the steering group:

- Co-Chair- Dr Cathal Morgan, Head of Disability Operations, HSE
- Co-Chair –Mr Anthony Carr, Chair, PWSAI
- Member-Ms Helen McDaid, National Disability Advisor, HSE
- Member-Dr Michael Byrne, National Families and Children's Team, Social Care Division, HSE
- External Expert Advisor-Tony Holland, Professor Emeritus of the Psychiatry of Intellectual Disabilities, University of Cambridge; President of International Prader-Willi Syndrome Organisation (IPWSO)

- Member-Mr Don Tallon, PWSAI
- Member-Ms Jane Cox, PWSAI
- Member- Mr Gary Brennan, National Development Manager, PWSAI

Methodology

Two workstreams were established (see membership in Appendix?) Workstream 1 (WK1) examined PWS carer experience of HSE service provision. A summary of its work and findings form the content of chapter 3. Workstream 2 (WK2) detailed the optimal model of service provision for PWS in respect of New Directions (Day Services), Residential Supports, Respite Supports and Home Support: the model sets out what works and for who, based on best available evidence. The work and findings of WK2 are detailed in chapter 4.

The international literature on the management of PWS was reviewed, including the findings and recommendations of *A Population-Based Profile of Prader-Willi Syndrome in Ireland, (Gallagher et al, 2017) and the comprehensive standards set out in Best Practice Guidelines for Standard of Care in Prader-Willi Syndrome, (IPWSO, 2010)*. In addition, members of the steering group and Workstream 2 attended the 5th International PWS Caregivers Conference in Munich, 28-30 August 2018.

The National Working Group also built on the research and experience of PWSAI over the last 30 years. PWSAI's membership of the International Prader-Willi Syndrome Organisation (IPWSO) enables access to current evidential research and service delivery models.

Understanding the policy context

This section outlines a number of relevant key documents and policies which set the stage for future health service development in Ireland: these policies provide the platform on which the recommendations of this report will stand The philosophy underpinning these policies is the full participation in society for a person with a disability with the right supports at the right time in the right place.

Primary Care Strategy (2001)

A key objective of this strategy is to ensure that the majority of patients and clients who require urgent or planned care are managed within primary and community settings.

The Disability Act (2005)

The Disability Act 2005 places a statutory obligation on public service providers to support access to services and facilities for people with disabilities. Under the Act, people with disabilities are entitled to have their health and educational needs assessed and have individual service statements drawn up.

The UN Convention on the Rights of Persons with Disabilities (2005)

Ireland signed this UN Convention in 2007 and ratified it in 2018. The purpose of the Convention is to promote, protect and ensure the full and equal enjoyment of all human rights and fundamental freedoms by all persons with disabilities, and to promote respect for their inherent dignity.

A Vision for Change (2006)

This document details a comprehensive model of mental health service provision for Ireland. It describes a framework for building and fostering positive mental health across the entire community and for providing accessible, community-based, specialist services for people with mental illness.

Time to move on from Congregated Setting: a strategy for Community Inclusion (2011).

This strategy delivers a model of support where people with disabilities are supported to move from large institutions (congregated settings) to their own homes in the community with the supports they need. It is about supporting people to live 'ordinary lives in ordinary places'.

Value for Money and Policy Review of Disability Services (2012)

This Review proposes a fundamental change in approach to the governance, funding and focus of the Disability Services Programme from an approach predominantly centred on group-based service delivery towards a model of person-centred and individually chosen supports.

New Directions proposes that day services should take the form of individualised outcome-focused supports to allow people with disabilities live a life in accordance with their own wishes, needs and aspirations.

National Standards for Residential Services for Children and Adults with Disabilities (2013)

These standards were published by The Health Information and Quality Authority (HIQA) and outline to providers what they must do to ensure safe and effective care is provided to people living in, or using, residential and residential respite services.

The National Rare Diseases Plan for Ireland (2014)

This plan, published by the Department of Health, includes such initiatives as the *National Clinical Programmes for Rare Diseases and the National Rare Diseases Office.*

Sláintecare (2017)

Built on political consensus, Slaintecare is a ten-year programme to transform Ireland's health and social care services. It aims to provide the majority of care at or closer to home and to create a system where care is provided on the basis of need not ability to pay. It also emphasises the importance of person-centred services for people with disabilities living in their own communities while acknowledging that some people with disabilities will remain in residential care.

Mental Health Services for Adults with Intellectual Disabilities National Model of Service (2021)

This model proposes that solutions for people with intellectual disability and mental health needs lie in establishing effective partnerships between healthcare providers, service users and their carers, in a community-wide context. Furthermore, it looks at the comprehensive range of interventions provided, including the use of inpatient settings where necessary.

Sharing the Vision, A mental health policy for everyone (2022)

Sharing the Vision is a broad-based, whole system mental health policy that will address the mental health needs of the population through a focus on the requirements of the individual.
The policy is underpinned by the values of respect, compassion, equity and hope. It takes a person-centred approach with a focus on enabling and supporting the recovery journey of each individual in the context of their personal lived experience.

Assisted Decision Making (Capacity) Act (2015)

The Act supports decision-making and maximises a person's capacity to make decisions. It applies to everyone and is relevant to all health and social care services. This Act will assist in complying with human-rights obligations contained in the Constitution of Ireland, the European Convention on Human Rights, and the United Nations Convention on the Rights of Persons with Disabilities.

Current Service Provision for Individuals with PWS and their Families

Current Service Provision

"There are unique risk factors which apply to people with PWS that must be acknowledged, understood and managed in order for the person with PWS to live safely and successfully within their communities. Complex ethical issues exist around rights, autonomy, mental capacity, and restrictive practices around food, money and medications".

Introduction

This section details HSE and HSE funded services accessed by individuals with PWS, and also any PWS-specific provision that currently exists. Current research and best practice models indicate the need for specific models of intervention for people with PWS and their families across all areas of healthcare, including acute, mental health and social care services among others.

- As noted above, the general principles, policies and standards underpinning Health and Social Care provision in Ireland apply to people with PWS. However, there are unique risk factors which apply to people with PWS that must be acknowledged, understood and managed in order for the person with PWS to live safely and successfully within their communities. Complex ethical issues exist around rights, autonomy, mental capacity, and restrictive practices around food, money and medications. Food security is achieved by securing food access or with one to one supervision across all environments, posting mealtimes and menus, and training all team members.

In addition, mental health and behavioural crises can occur which necessitate the involvement of the emergency services, and *Garda Síochána** or the courts.

* (Ireland›s National Police Service)

Endocrinology is the core discipline in the management of PWS.

Irish children and adolescents with PWS currently attend the paediatric endocrine departments at Children's Hospital Ireland (CHI) at Tallaght, CHI at Crumlin, CHI at Temple Street and Cork University Hospital (CUH). The largest cohort attend the specialised, age-banded, Multi-Disciplinary PWS outpatient clinics provided at TUH which also provide support, education and training for parents. The latter service at TUH is currently reliant on charitable funding.

Despite advances in international standards of care, including the use of growth hormone therapy in adulthood, some adults with PWS in Ireland are linked to acute services with no expertise in PWS. A dedicated PWS Multi-Disciplinary PWS outpatient clinic began working with adults with PWS in February 2023 at Tallaght University Hospital (TUH). The clinic have adopted recommendations from a Dutch study by Pelikaans et al 2020 on screening and treatment for adults with PWS to avoid missed diagnosis and health problems in people with PWS.

Mental Health Services

There are no clear pathways to appropriate mental health services for individuals with PWS and no specialist service currently exists.

Some adults access generic community mental health services: others have access to community Mental Health Intellectual Disability (MHID) teams but currently there are areas of the country with no MHID teams: many of the teams are poorly resourced. Some children and adults access specialist ID mental health services for those with moderate to severe ID provided by section 38 and 39 Voluntary Agencies[1] as part of their disability services.

The HSE National Model of Service for Adults with ID, published in 2021, states that *"People with intellectual disability should access support from mental health services in the same way as the general population, within a framework which is multi-disciplinary and catchment area-based. Team members should have appropriate training and expertise, and teams should be suitably resourced. Recommendations for the mental health of intellectual disability (MHID) model of service follow similar international models. It is imperative that MHID services capture the work they do, and use evidence based practice", (HSE, 2021)*

Generally, children and adolescents have access to community Child and Adolescent Mental Health Services (CAMHS) but again provision is patchy across the country. The establishment of community Child and Adolescent Mental Health Services (CAMHS-ID) teams is also currently being progressed.

Respite Provision

The purpose of respite care provision is to provide respite for caregivers.

Currently there are no dedicated, PWS-specific residential respite services for children or adults. In 2021 PWSAI, in collaboration with the NCPPD, submitted a proposal for funding for a PWS-appropriate residential respite care. This proposal was approved, and funding provided for respite service provision. This service will provide a number of nights respite for children and adults with PWS and their families and is due to commence operating in 2024.

Residential Provision

In December 2003, RehabCare opened a dedicated PWS-specific community-based residential service for adults with PWS; Graifin House continues to support four individuals full-time and is located in South County Dublin. A number of individuals with PWS had previously been funded by the Irish Health Service at a PWS specialist residential service in the UK.

Resilience Ireland established a dedicated, PWS-specific, community-based residential service in Enniscorthy, County Wexford in October 2019; six young adults with PWS currently live in Lemongrove House. Resilience opened a second PWS specific service in Monasterevein in 2023 supporting four adults, with a plan to open a third service in Cork in 2024.

In the 2017 report, (Gallagher et al 2017) found a need for 16 new full-time residential placements for adults with PWS by 2021 among those who participated in *A Population-Based Profile of Prader-Willi Syndrome in Ireland, 2017.* Gallagher believed that this is a conservative estimate of need as anecdotal evidence suggests additional need outside the cohort studied. Due to the necessity for specific support for all individuals with PWS, it is envisioned that there will be an ongoing need for full-time residential placements.

Day Services/New Directions

Many adults with PWS access HSE funded day services or supports. Anecdotal evidence suggests a mix of experiences, on a continuum from successful placements to long suspensions from services due to food stealing or other challenging behaviours. Carers report that where services and supports work, there is adherence to best practice recommendations in terms of food security, and consistent planning and scheduling.

Early Development and Education

Due to the complex nature of the syndrome, Disability Services are delivered to children with PWS through the Children's Disability Network Teams

1 Related to those agencies provided with funding under Section 38 and section 39 of the Health Act, 2004.

(Early Intervention Teams and School Aged Teams) in their local Community Healthcare Organisation. Younger children generally attend mainstream preschool and primary educational settings with supports: most switch to special educational settings at second level.

PWSAI

Prader-Willi Syndrome Association Ireland (PWSAI) was founded by parents in the late 1980s as a support group for individuals with PWS and their families in Ireland. It became a registered charity in 2002 and currently supports over 100 families. Its mission is to raise awareness and understanding of PWS and to improve the choice and quality of care, education and support for persons with PWS.

PWSAI

"Its mission is to raise awareness and understanding of PWS and to improve the choice and quality of care, education and support for persons with PWS".

The appointment of a Development Manager for PWSAI was approved and fast tracked by the chairperson of the National Working Group and is funded by the HSE. The Development Manager works to establish a sustainable foundation from which future PWS education, awareness, training programs, and services provision can develop and will drive the implementation of the recommendations of this report.

In 2022 PWSAI hosted the 11th international PWS conference, in partnership with the International Prader-Willi Syndrome Organisation, (IPWSO). The conference took place from 6 to 10 July at the University of Limerick with 519 delegates attending from 38 different countries.

National Clinical Programme for People with Disabilities (NCPPD)

In 2021 Minister Anne Rabitte and Minister Mary Butler met with members of PWSAI. Following that meeting, a task force on PWS was established under the auspicious of the NCPPD, chaired by Prof. Mac McLachlan. This task force worked to progress key recommendations of this report.

Summary

- The National Working Group on Prader-Willi syndrome was established to review the effectiveness of HSE services accessed by people with PWS, particularly social care provision.
- The NWG was tasked with developing and testing positive models of social care provision that effectively meet the needs of individuals with PWS and their families.
- The NCCPD task group worked to implement the key recommendations of this report.
- The needs associated with the syndrome are unique and complex, and require specialist, syndrome-specific responses.
- A universal feature of PWS is a lack of satiety which leads to hyperphagia (excessive eating).
- The hunger felt by the individual is of genetic origin and outside the control of the sufferer: there is no cure or treatment at present.
- Individuals with PWS become morbidly obese and die in early adulthood (or sooner) unless their access to food is strictly controlled.
- Food security is achieved by securing food access across all environments, one to one supervision of food access across all environments, posting mealtimes and menus, and training all team members.
- In addition to hyperphagia, other behaviours, such as severe temper outbursts, may further complicate and increase the need for support

CHAPTER TWO
Literature Review

Prevalence

Taking data from Growing up in Ireland, 2011 and reports from the National Centre for Medical Genetics, Gallagher estimates a minimum birth frequency in Ireland of 1 in 11,000 annually (Gallagher et al., 2017), with 5-6 new cases being diagnosed each year. This is in keeping with international studies such as Smith et al., 2003.

Hyperphagia

Hyperphagia (overeating or over appetite) emerges in early childhood (Angulo et al., 2015). This leads to severe obesity (Crino, 2018). Because hyperphagia and subsequent obesity are cardinal features of this rare genetic disorder, excessive overeating, stomach necrosis and rupture, cardiovascular disease, respiratory failure, sleep apnoea, diabetes mellitus, and related comorbidities can be life-threatening. Complications of obesity remain the major cause of death in adults with PWS (Tauber et al, 2014).

> *'At no time can a person with PWS be expected to voluntarily or independently control his/her own food consumption' (Gourash et al. in Butler, p.414).*

The drive for food remains a lifelong source of stress for individuals with PWS: people with PWS display a permanent obsession with food and excessive food searching, food storage, foraging and hoarding, (Bellis et al, 2020). Tauber et al, 2014 asserts that obsession with food, particularly the timing of the meals and the hedonic part of food, is overexpressed. Holland argues that PWS is a 'starvation' syndrome rather than an over-eating one in which the brain keeps telling the stomach, "you're starving, you need food", and the drive to find food overrides everything else (Holland et al, 2003). Food security, would in the majority of cases, include one to one supervision across all environments where access to food is uncontrolled (Tauber et al, 2014).

Medical Issues

Individuals with PWS present with low muscle tone, short stature, incomplete sexual development, cognitive disabilities, and behavioural issues (Butler et al., 2022). Endocrine issues include growth hormone deficiency necessitating daily Growth Hormone Treatment (GHT), the need for sex Hormone Replacement Therapy (HRT) and increased risk for thyroid dysfunction and type 2 diabetes (Emerick and Vogt, 2013). Other comorbid health problems include orthopaedic issues such as scoliosis, kyphosis and hip dysplasia, sleep apnoea, excessive daytime sleepiness and sleep disturbances, issues around bone health including osteoporosis, ophthalmic issues, dental issues associated with enamel quality and saliva production, gastrointestinal issues and hypothalamic dysfunction (Jin, 2012; Shim et al., 2010: Van Bosse, 2010; Sedky, Bennett & Pumariega, 2014). Hypothalamic dysfunction complicates the care of the individual across all settings: effects include altered pain threshold, issues around attention, atypical reactions to medication including anaesthesia, temperature instability and mood lability (Butler et al, 2022; Emerick and Vogt, 2013). People with PWS have a high pain threshold and rarely vomit thus serious illness (e.g. appendicitis) may not present with the classical symptoms until it is too late. In addition, lack of satiety can lead to binge eating and, where excess amounts are eaten quickly, to stomach paralysis (paresis) which in turn leads to stomach rupture which is fatal (McAllister, Whittington & Holland, 2011). Death by choking is another hazard (Crino, Fintini, Bocchini and Grugni, 2018).

Obesity is a major cause of premature morbidity and mortality in PWS: people with PWS die prematurely from complications conventionally related to obesity, including diabetes mellitus, metabolic syndrome, sleep apnoea, respiratory insufficiency, and cardiovascular disease, (Crino et al., 2018).

The core discipline, particularly in childhood, in the management of PWS is endocrinology with support from key specialities such as dietetics, orthopaedics, respiratory, ENT, dentistry, psychology, GI (Butler et al., 2022). Centres of Excellence in PWS, in the form of multi-disciplinary clinics led by an endocrinologist with clinical expertise and research interest in PWS, exist in other countries e.g. Sweden, Denmark, France, Brazil, Australia and USA. This model of provision is in line with the recommendations of the Rare Diseases Plan for Ireland 2014 and the National Clinical Programme for Rare Diseases, in particular 'ultra-rare' diseases such as PWS. IPWSO's Clinical and Scientific Advisory Board has produced comprehensive medical evaluation, guidance and overview documents relating to infants, children, adolescents and adults with PWS. www.ipwso.org/medical-professionals

Structured transitional care between paediatric and adult specialist PWS services is essential for good health outcomes (Paepegaery et al., 2018). The latter is in line with Ireland's Model of Care for Transition from Paediatric to Adult Healthcare Providers in Rare Diseases (HSE and RCPI, 2018).

Mental Health

Prader-Willi Syndrome is associated with a range of mental health diagnoses and challenging and destructive behaviours. As discussed above, hyperphagia (due to lack of satiety) leads to challenging food related behaviours and food related anxiety. PWS is also associated with a marked propensity to other problem behaviours such as temper outburst, repetitive and ritualistic behaviours, repetitive questioning, skin picking and other compulsive and self-harming behaviours (Whittington and Holland, 2004: Didden, Korzilius & Curfs, 2007; Woodcock and Oliver, 2009; Novel-Alsino et al., 2019). In a recent Irish study, Feighan found the most common challenging behaviours were skin-picking (92% of adolescents), repetitive questioning, difficulty transitioning, and non-compliance: she also reported high levels of physical and verbal aggression directed at family caregivers (Feighan, Hughes, Maunder, Roche and Gallagher et al., 2019). A number of studies describe severe temper or emotional outbursts that are distressing and disabling for the individual with PWS and endure into adulthood (Rice et al., 2015 & 2018; Bianco-Hinojo et al., 2019).

Gallagher found high levels of major mental health difficulties among Irish persons with PWS and very significant and frequent behavioural challenges (Gallagher et al., 2017).

Anxiety disorders are highly prevalent, (Butler et al., 2022; Woodcock et al., 2009; Wigen etal., 2005; Tsai et al., 2019; Feighan et al., 2021). Soni found very high rates of mental illness, both affective and psychotic in individuals with PWS (Soni et al., 2007 & 2008). Those with the mUPD genetic subtype are at heightened risk for psychotic illness, often with a depressive or affective component (Boer et al., 2002; Sinnema et al., 2011).

Gallagher found high levels of major mental health difficulties among Irish persons with PWS and very significant and frequent behavioural challenges (Gallagher et al., 2017). An earlier study conducted in Ireland found significant mental health problems among children with PWS (Skokauskas et al., 2011). Ireland's National Clinical Programme for Rare Diseases 'Model for Care in Transition' references PWS and recommends that 'preparation and anticipation by genetic subtype, and for declining mental health in early adulthood should begin during childhood'. Feighan found individuals with PWS in Ireland had to wait on average 22 months for an appointment with a psychologist (Feighan et al., 2019).

Early Development, Education and Employment

The infant with PWS presents with severe hypotonia and delayed milestones. Intensive, multi-disciplinary early intervention is required: intervention should include physiotherapy, speech and language therapy, occupational therapy among other disciplines (Butler et al., 2022).

The neurodevelopmental profile of PWS includes a distinctive cluster of behavioural features along with intellectual disability or learning difficulties (Whitman and Thompson, 2006). Difficulties with task-switching, insistence on sameness in routines and resistance to change are prevalent (Haig and Woodcock, 2017: Dykens et al, 2017). Students with PWS experience challenges with social cognition including theory of mind, difficulty perceiving the intentions of others, recognising emotions etc (Dimetropolis and Ho, 2013; Dykens and Roof, 2019; Debladis et al., 2019; Schwartz et al., 2021). Cognition and social development are further influenced by genetic subtype and research in this area can inform educational strategies and supports (Whittington and Holland, 2016).

Whittington reports academic underachievement among students with PWS (Whittington et al., 2004). Effective teaching and learning is further complicated by the impact of hyperphagia, attention difficulties, poor adaptive functioning, hypothalamic dysfunction and the pupil's individual presentation of medical and mental health difficulties; the child with PWS needs to be understood within the context of the challenges placed on him by the syndrome (Waters, 1999). Materials for teachers have been developed by IPWSO and by PWS associations in the UK, Canada, USA and Australia, see for example www.pwsausa.org/wseat-webinar-series/

Gallagher reported poor learning outcomes for Irish pupils with PWS: most adults left education with no qualification and only one Irish adult with PWS was in paid part-time employment. Most Irish pupils with PWS switch from mainstream to special settings in late childhood or early adolescence (Gallagher et al., 2017).

Impact on Family

'Constant and obsessive food seeking behaviour can make life very difficult for both the family and caretakers' (Crino et al., 2018). Recent research conducted in Ireland found caring for a person with PWS was associated with a significant negative impact on family relationships, carer health, sibling mental health and sibling social life (Gallagher et al., 2017). When compared to families of children with a variety of causes of intellectual disability or complex health conditions, parents of children with PWS report higher levels of stress and mood disruption,

and more difficulties coping with their child's symptoms (Lanfranchi & Vianello, 2012, Skokauskas, Sweeny, Meehan & Gallagher, 2012)). Mazaheri found difficulties in family functioning, reduced quality of life and significant behavioural distress symptoms among mothers and siblings of those with PWS: 92% of siblings indicated moderate-to-severe symptoms of PTSD (Mazaheri et al, 2013). Studies have shown a strikingly high caregiver burden associated with caring for a person with PWS that peaks in adolescence and early adulthood (e.g. Kayadjanian et al., 2018). Having a relative with PWS is associated with higher prevalence of experienced traumatic events, and of PTSD, affecting the wider family system, (Bos-Roubos et al, 2022). Psychosocial supports are recommended for caregivers and siblings to support coping and prevent deterioration in mental health (Gallagher et al., 2017; Mazaheri et al., 2013).

> *'Constant and obsessive food seeking behaviour can make life very difficult for both the family and caretakers' (Crino et al., 2018).*

Raising awareness in health care professionals of the typical presentation of trauma symptoms in PWS relatives may contribute to effective treatment of their psychosocial stress, (Bos-Roubos et al, 2022)

Social Care Supports

The Professional Provider Caregiver Board of IPWSO has published, and regularly updates, comprehensive and detailed guidelines for those providing social care supports to adults with PWS (Best Practice Guidelines for Standard of Care in PWS). The basic premise of intervention acknowledges that "at no time can a person with PWS be expected to voluntarily or independently control his/her own food consumption" (Gourash et al in Butler, p.414).

Respite Provision

Respite is recognised, both nationally and internationally, as a necessary support for families of people with PWS (Akindola et al., 2015; National Clinical Programme for Rare Diseases, 2013; Gallagher et al., 2017).

Gallagher et al. highlighted the 'critical need for appropriate, PWS-specific respite services in Ireland' and stated, 'provision should be PWS-specific for consistency in management'. The Department of Health's Rare Diseases Plan for Ireland, (2014) 'acknowledges that, for some rare diseases, specialist respite may be needed', and recommends 'residential respite for children with rare diseases' and 'assessment of carers' needs'. In terms of the benefits of adequate respite care, *Towards best practice in the provision of respite services for people with intellectual disabilities and autism* (2007) informs us that "the benefits for the carer were clustered around health and well-being, family functioning and concrete supports. For service users, social development and independence were the main benefits" (HSE, 2007)

Residential Provision

PWS associations in other countries have highlighted the need for appropriate residential services to be provided for people from the age of 18 onwards (Akindola et al., 2015). Such provision is available, for example, in Denmark, Sweden, Germany, Austria, UK, USA and Australia. IPWSO's (2010) *Best Practice Guidelines for Standard of Care in PWS* state 'the preferred form of living arrangement is in a PWS-specific environment'. PWSA UK's (2016) publication 'Residential Care and Supported Living' describes how semi-independent living or living in non PWS-specific residential settings can lead to "severe obesity, failing health and early death". Food security may include one to one supervision, securing food access across all environments, , posting mealtimes and menus, and training all team members, (Tauber et al, 2014).

The urgent need to scale up the provision of PWS-specific residential care places for adults with PWS in Ireland has been highlighted (Gallagher et al., 2017).

Day Services

The adaptive needs of people with PWS need to be considered in addition to measured IQ when determining appropriate day services and supports (Gallagher et al., 2017). The HSE's document *Interim Standards for New Directions, Services and Supports for Adults with Disabilities,* (2015), acknowledges that 'disability-specific conditions are a key consideration when individualised personal plans are being developed', and that 'there are specific disability conditions that present challenges with regards to the level of community inclusion that may be appropriate for an individual' with 'some people requiring purpose built facilities to support their needs; however 'these factors should not compromise the principles of Community Inclusion or Active Citizenship but rather reinforce the approach to individualised planning and solutions'.

Summary

- Early diagnosis and parental knowledge about PWS help families understand and manage the early 'failure to thrive' stage in the syndrome. It also prevents over-feeding and obesity once the uncontrollable appetite or urge to eat, known as hyperphagia, develops in early to middle childhood.
- Due to the complex nature of the syndrome, Disability Services should be delivered to children with PWS through the Children's Disability Network Teams (Early Intervention Teams and School Aged Teams) in their local Community Healthcare Organisation.
- The lack of ability to regulate energy intake and the lack of satiety are crucial to understanding the needs of people with PWS
- Dietary, behavioural and environmental interventions, for example, locked kitchens, one to one supervision and provision of 'psychological food security', must be put in place to support the individual with PWS, and to prevent early mortality and obesity related comorbidities.
- The core discipline in the management of PWS is endocrinology with support from key specialities such as dietetics, orthopaedics, respiratory, ENT, dentistry, psychiatry, psychology, GI.
- PWS is associated with high rates of mental health diagnoses such as anxiety and mood disorders. Psychotic illness is very common among those the mUPD genetic subtype.
- Challenging behaviours associated with PWS include food seeking, emotional outbursts, skin picking, repetitive questioning, difficulty with transitions and non-compliance.
- High pain threshold, lack of vomiting, and poor temperature regulation can make the recognition of developing physical illness difficult.
- The impact on carers and families is enormous, affecting carer health, carer career, family relationships and sibling mental health.
- Specialist respite is recognised, both nationally and internationally, as a necessary support for families of people with PWS

Individuals with PWS continue to need intensive support throughout adulthood and require fulltime specialist residential services when they leave the family home. Food security usually includes one to one supervision and securing food access across all environments.

CHAPTER THREE

Workstream 1: PWS Carer Experiences of Health Service Provision in Ireland

Introduction

In line with its Terms of Reference, Workstream 1 sets out to examine PWS carer experience of health service provision in Ireland, particularly social care provision.

PWS is a complex, multi-systems condition: management requires involvement from all arms of the HSE- Acute, Mental Health, Primary/Community, Disability /Social Care. In addition, the educational progress of the individual with PWS is in part dependent on services provided by the HSE.

Methodology

Workstream 1 undertook a research project using a qualitative design. The project consisted of two phases – an online survey and a peer-facilitated workshop.

Purposeful sampling was used, specifically total population sampling. Participants were recruited through PWSAI and the Paediatric Endocrinology Clinic in Tallaght University Hospital. 57 carers took part in the online survey and 25 in the follow up workshop. Participants were from 14 counties across Ireland. While almost half of the participants in the online survey were caring for an individual under the age of twelve years, 80% of those who attended the workshop were caring for a teenager or adult with PWS, perhaps reflecting the greater need of the latter cohort for improved services.

Online Survey

The purpose of the online survey was to examine carer experience of service provision. The questions used were open-ended and qualitative, covering the range of services used and needed by individuals with PWS and their families, and were designed to assess the experience of caring for an individual with PWS in the following areas:

1	Living with your child/family member with PWS
2	Community care supports
3	General & PWS-specific Respite provision
4	Medical Services
5	Education Provision
6	Residential Provision

Topics addressed in online survey conducted by Workstream 1 'Care Experience of HSE Services'

Questions and prompts on these topics were tailored to evaluate the experience of the relevant services used. As the needs of individuals with PWS change across the lifespan, three versions of the survey were designed: each version addressed specific services relevant to the age of the individual with PWS being cared for (0 – 12 years, 13 – 17 years, and 18+ years).

Peer-facilitated Workshop

The purpose of the workshop was to expand on the data gathered from the online survey. The workshop acted as a sanity check to the data collected from the survey and the themes developed from it and allowed the themes to be examined in greater detail.

Participants joined one of four groups depending on the age of the person with PWS cared for (0 – 12 years, 13 – 17 years, 18 – 25 years and 25+ years). Each group was peer-facilitated by a carer of an individual with PWS in the same age range as the rest of the group. Each group had a peer note taker and the research team also alternated between groups. The workshop followed a semi-structured format, with the peer facilitators following a series of questions and prompts. The same areas were examined in the workshop as for the online survey, see Table? above.

Following the group stage of the workshop, the peer facilitators presented the data from their group to the rest of the workshop. This was followed by a lively open forum where participants shared their experience of PWS service provision.

Qualitative Analysis

All qualitative data was analysed manually following the Braun & Clarke (2006) model of thematic analysis. The researchers familiarised themselves with the data, actively searching for patterns and meaning, and took note of potential codes. Initial codes were generated, and the data were then collated together based on these. Themes and sub-themes were developed which contained the essence of the data as a whole and accurately depicted the story of the data

Findings

The findings of both the online survey and the peer-facilitated workshop are presented together. Three overarching themes were developed that were consistent and present across all types of service provision and applied to all age groups: Dissatisfaction with Services, Behavioural Issues and Impact on Family. (See Fig. 2)

Only one of the main themes which emerged refers directly to services but by examining the impact of PWS on behaviour and family life, the needs of individuals with PWS and their carers can be better understood and by extension the services required.

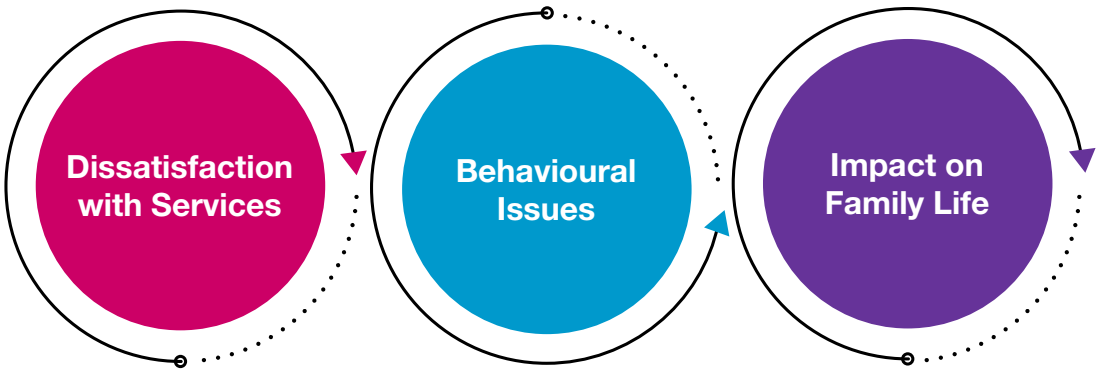


Fig.2

Theme 1. Dissatisfaction with Services

The theme of Dissatisfaction with Services was overwhelmingly prevalent in carers' responses and was consistent across all age ranges and most types of service provision (with perhaps the Positive feedback exception of Early Intervention Services and the paediatric endocrine clinic in TUH). The core tenet of the theme is that services are not fit for purpose and the sub-themes relate to various aspects of this. The sub-themes which emerged are presented in Fig 3.

Fig.3

Lack of Resources/Inequity of Provision

Overall carers believed that services do not have the resources to provide the level of care that is required in PWS. Important aspects of care are not provided and services that are on offer are not provided at an adequate level.

The majority of respondents said there were no suitable respite services: those who had experienced general respite services said they were poor; support staff hadn't understood their child's level of anxiety and their behaviours, nor did they know how to manage hyperphagia. Carers stated no PWS-specific respite provision exists. Many parents recorded details of failed respite provision:

- One family told how, despite huge planning, they had to collect their 14 year old son one day into a planned three day respite break following a massive temper tantrum over an apple (this was one of only two experiences of respite this family had ever had):
- Another parent detailed how her adult daughter with PWS was offered big fried suppers late at night. However, some families in the north east of the country had experienced respite which met the complex needs of their children.

Mental Health services were a particular area of concern. Many with adult children lived in areas with no Mental Health Intellectual Disability (MHID) provision. Some had accessed unsuitable general psychiatric services at times of crises that had brought them to A and E, while others had been denied access to MHID teams as they did not meet the IQ criteria. Some families with children and adolescents reported a dearth of CAMHS/CAMHS-ID services in their area. An inability to access behavioural/psychological supports was an issue across all age groups.

For parents of younger children, services depended on the resources and composition of their local Community Disability Network Team. Regarding SLT, physio and OT, some families had good access to one or two of these disciplines but none at all to others. There was huge concern about the drop off in necessary therapy services when children moved to School Aged Teams. For carers of adults with PWS, access to appropriate endocrine services was a major issue but of greatest concern to this cohort was the lack of dedicated, PWS-specific residential provision.

Lack of Knowledge

Across educational, community care, medical, day service and respite settings, carers reported a lack of knowledge of the nature and extent of the needs of the person with PWS. Lack of knowledge about the syndrome among local HSE disability officers and managers was also noted. As a result, parents considered it unsurprising that comprehensive supports did not exist. Parents believed professionals need knowledge about how PWS presents across the lifespan in order to be able to adequately support their child in the present. Parents detailed incidents with health professionals which illustrated a lack of knowledge of PWS:

- Parents had been asked by doctors at endocrine clinics if the individual with PWS had a 'good appetite'.
- Dieticians had included sugary foods on their child's diet plan.
- Lack of knowledge around managing hyperphagia had led to the expulsion of one young adult from a further education course targeted for people with disabilities, precipitating a mental health crisis.
- Professionals had discussed issues in front of individuals with PWS that were way beyond the young persons' capacity to understand or cope with, triggering mental distress and challenging behaviours.

Poor Communication/Coordination

Carers stated they had experienced a lack of communication and coordination of care between services and across disciplines. Families often had to make multiple trips to Dublin, rather than having coordinated appointments, increasing pressure on carers especially as the associated disruption of routine caused huge anxiety and resultant challenging behaviours in their family member with PWS. Family carers were very unhappy with the lack of communication between acute and mental health services, between the education and health sectors, and between hospital doctors from various disciplines involved in their family members care. Communication was also poor between carers and services, resulting in carers having difficulty knowing how to access or navigate services. These difficulties were amplified at transitional stages and added to carer anxiety (further explored below under 'Theme 3: Impact on Family'). Some family carers expressed a desire for a HSE care coordinator, preferably assigned at the time of diagnosis so 'families don't have to be so heavily involved, micromanaging everything and getting exhausted'.

Delays in Access

Carers all had difficulty in accessing services for their children: difficulties increased as their children grew older and peaked in adulthood.

Individuals with PWS had to languish on long hospital outpatient waiting lists before they could be seen- often for time sensitive care. Delays in accessing ENT services, such as sleep studies, led to delays in starting vital Growth Hormone Treatment. The status of those with complex spinal disorders deteriorated as they waited for orthopaedic consultant appointments, MRIs and surgeries. Delays in accessing DEXA scans affected bone health. Of particular concern to parents of adults with PWS was delays in accessing endocrinology.

Parents of children attached to Early Intervention Services reported delays in accessing SLT, physio and O.T, and infrequent blocks of therapy. Parents were generally happy with the quality of EI services, once accessed. Waiting lists for CAMHS was another issue of concern.

"Early Intervention in Mayo was very good. We had access to all the services…But they drop children like a hot coal once they go to school."

"There is no psychological support."

"By the time his splints and special shoes are ready, sometimes he's outgrown them"

"CAMHS in my area is almost non-existent."

"Respite does not work…It is not PWS specific…carers don't understand the behaviours."

"Provide respite care on a regular basis. Let me see light at the end of the tunnel."

Theme 2. Behavioural Issues

The theme, Behavioural Issues, details the carer experience of the behaviour of individuals with PWS. Sub-themes which emerged are contained in fig.

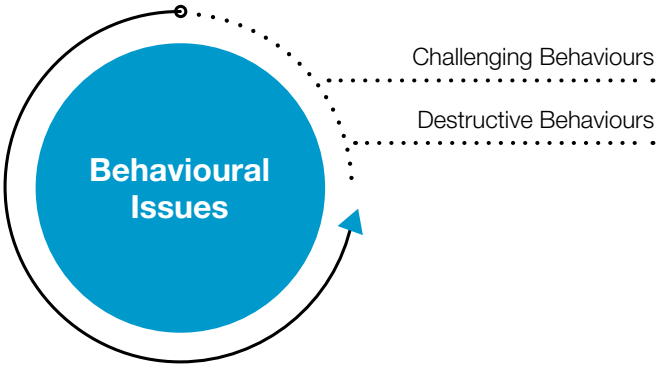


Fig.4

Challenging Behaviours were a concern at all ages, with Destructive Behaviours emerging as a greater concern in the teen years and early twenties. Parents report anxiety (both generalised and food-related) as a root cause. Behaviours existed on a continuum, ranging from incessant questioning to violent outbursts, according to the degree of anxiety experienced by the individual with PWS.

Challenging Behaviours (or behaviours that challenge)

Challenging behaviours (Behaviours of Concern) occurred across the lifespan. Behaviours reported include repetitive and obsessive-compulsive behaviours (both food and non-food related), meltdowns, arguing, pleading and sobbing, taking of food in the home, shoplifting food, skin picking, rectal picking, hoarding and impulsive buying.

Repetitive questioning and fixations over particular people, places and objects were particularly 'wearing and distracting' for carers: some parents reported near misses while driving as a result. Obsessions could go on for days, weeks or even months. Skin-picking was very prevalent throughout the data: wound care after surgeries, poor personal hygiene and rectal picking made keeping sores free from infection very difficult for carers. Any minor cut could develop into a sore that remained open for months.

Changes or disturbances to routine caused severe emotional distress in individuals with PWS and would quite often induce meltdowns that were extremely difficult to control. Even where the changes were to events and people that the individual preferred, upset would still occur. Some parents suggested that such behaviour was often not intentional, reporting that their children experienced extreme guilt and self-loathing after a tantrum or upset.

Parents reported:

- Lack of danger awareness when out and about, for example, around traffic or animals,
- Inappropriate social behaviour e.g. invading personal space, interrupting, asking strangers very personal questions, skipping queues.

Manipulative behaviour and oppositional defiance were also commonly reported: family members with PWS were often able to gain access to food or money from strangers or carers not familiar with PWS. Quite often parents reported:

- A sense of ruthlessness in their child when they became goal-oriented.
- When in possession of money, individuals had very little control in shops, compulsively purchasing toiletries, stationary etc.
- Others with more developed skills, bought compulsively online or ordered over the phone.

"He hoards things. He has to buy two of everything. My garage is full to the ceiling of unopened boxes of electrical items."

I have to lock all the food away and have it up high… I have to climb up a ladder to get my own cereal every morning."

"When we deviate from the routine, she cannot accept it and will get emotionally upset."

"She has stolen food from local shops and from her fellow students on her course."

Destructive Behaviours

The Destructive Behaviours mentioned in this theme generally emerged during the teens and early twenties. Destructive behaviour was directed both at others and towards the individual himself. Among the behaviours reported by parents were:

- Hitting, swearing, pulling, pushing, choking, throwing and breaking objects (including windows) and absconding.
- Also reported were aggressive skin picking (gouging) and other forms of self-harm including suicide attempts and verbal abuse towards self.

Destructive behaviours often followed on from challenging behaviours that had escalated. Carers considered many destructive behaviours to be impulsive by nature. The main caregiver was often the target due to their control over the individual's diet and food, however destructive behaviours were experienced by the whole family:

- One teenage sibling had to move out of the family home and other families had considered similar action.
- Carers reported tension and behaviours at dinnertime as the individual with PWS would become upset and aggravated when comparing their portion to that of other family members.

As a result, some families no longer ate together.

"Very bad skin picking at times and can keep cuts open for a month a year and longer."

"Repetitive questions are an ongoing battle. Crying for no reason and not stopping. Sobbing if not getting her own way"

"When he decides he wants food other than what he's allowed, it ends up in a massive argument, shouting, name calling, breaking stuff in the house & hitting me."

"It's wild in our house, windows get smashed and everything is broken."

"He verbally abuses and belittles himself in front of the mirror."

"She has tried to end her life four times."

"Can be very violent when her mental health is bad and also phones the guards and the social worker…Violence will be hitting, choking, cursing and breaking things."

Theme 3. Impact on Family Life

The theme Impact on Family Life was prevalent in responses across all age ranges. This theme relates to the strain and stress caregivers and other family members experience, and how that relates to Service Provision especially Social Care supports. Sub-themes which emerged were: (1) Pressure on Caregivers, (2) Relationships within the Family and (3) Anxiety about the Future See Fig.4.

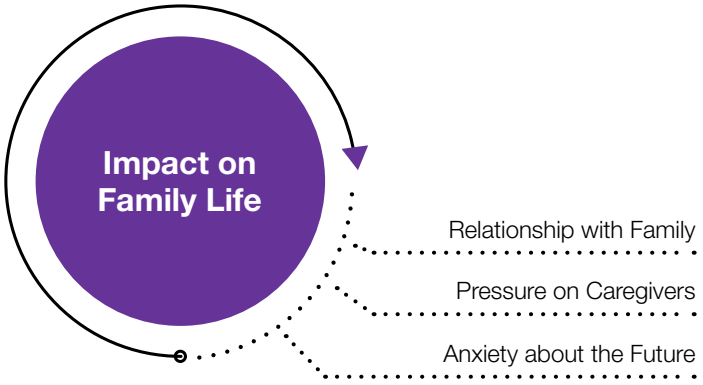


Fig.5

Pressure on Caregivers

Caregivers were required to be constantly vigilant as food security had to be maintained around the clock: meals had to be on schedule, low calorie and tailored to dietary needs. Huge energy was expended on distracting, protecting and bargaining with the person with PWS over food. Carers had to continually manage their child's anxiety and inability to cope with change. Much time and emotional energy was spent chasing up and fighting for services, attending appointments, and communicating their child's needs to carers at school and in disability services.

Families were under financial pressure; in most families there was the loss of one income due to the needs of the individual with PWS. Many families were paying for private services to fill holes in provision: others had rebuilt or undertaken renovations to their kitchen to improve food security.

Carers were concerned about their own mental health and expressed an urgent need for PWS-specific respite. They expressed regret that they had not been offered counselling and support at the time of diagnosis, and that no one was checking to see if they were coping. They wished social care workers and disability managers understood PWS needs as they were exhausted from having to micromanage everything themselves.

Relationships within the Family

Caregivers reported that living with an individual with PWS can put a major strain on family relationships. Family life revolved around the member with PWS and their needs. For many families with a teenager or adult member with PWS, even simple things like family meals, traveling together in the family car or going on holiday together were not feasible. One teenage sibling had to move out of the family home for her own safety, and other families had considered similar action.

Siblings were often reported to be helpful and empathetic but some harbour resentment at having to take on a carer role, at not being able to bring friends home due to challenging behaviours or at the dietary restrictions imposed on the whole family due to the needs of the sibling with PWS. Some siblings experience mental health issues due to the strain of living with an individual with PWS; others were reported to have no relationship with the family member with PWS.

Marital tension was reported, with in many cases the loss of career of the primary caregiver being cited as a significant factor. Many families lived in a highly charged atmosphere which greatly affected the development of healthy relationships and emotional functioning leading to a negative impact on the mental health of all members. Parents were often absent due to appointments and hospital stays: they worried that the pressure of caring had also reduced their emotional availability to their other children. In some cases, parents have described their other children as having to 'grow up too fast' and act as

secondary carers to their child with PWS. Because of their child's complex needs, families often could not avail of natural support structures such as friends and extended family to allow them time together away from the family member with PWS.

Anxiety about the Future

For carers, anxiety about the future was present across the lifespan but especially so at times of onset or increase of PWS symptoms, and at times of transition between services. Fears were expressed about future inability to cope if respite did not become available. Parents reported extreme anxiety about the lack of PWS-specific residential provision particularly when caring for the individual at home became unsustainable for reasons of sibling mental health, damage to the integrity of the family unit or the needs of the person with PWS himself. Parents also worried about the capacity of schools and disability day services to meet the needs of their child. Parents also worried about their other children's physical safety, the fact that they had been exposed to acts of self-harm in the home, and whether they were at risk of eating disorders because of the family's peculiar (but necessary) relationship with food. Many carers reported being on constant alert, worrying at every moment and in every environment, about possible triggers for behavioural meltdowns or unexpected food in the environment.

"His brother has to lock himself in the bathroom if he wants to eat a treat."

"Everything revolves around food. It is continuous questioning, distraction and security… Our child needs constant monitoring."

"I have high blood pressure and am constantly prescribed anxiety medication – it's non-stop."

Her siblings are very stressed and we had to bring them to a private psychiatrist."

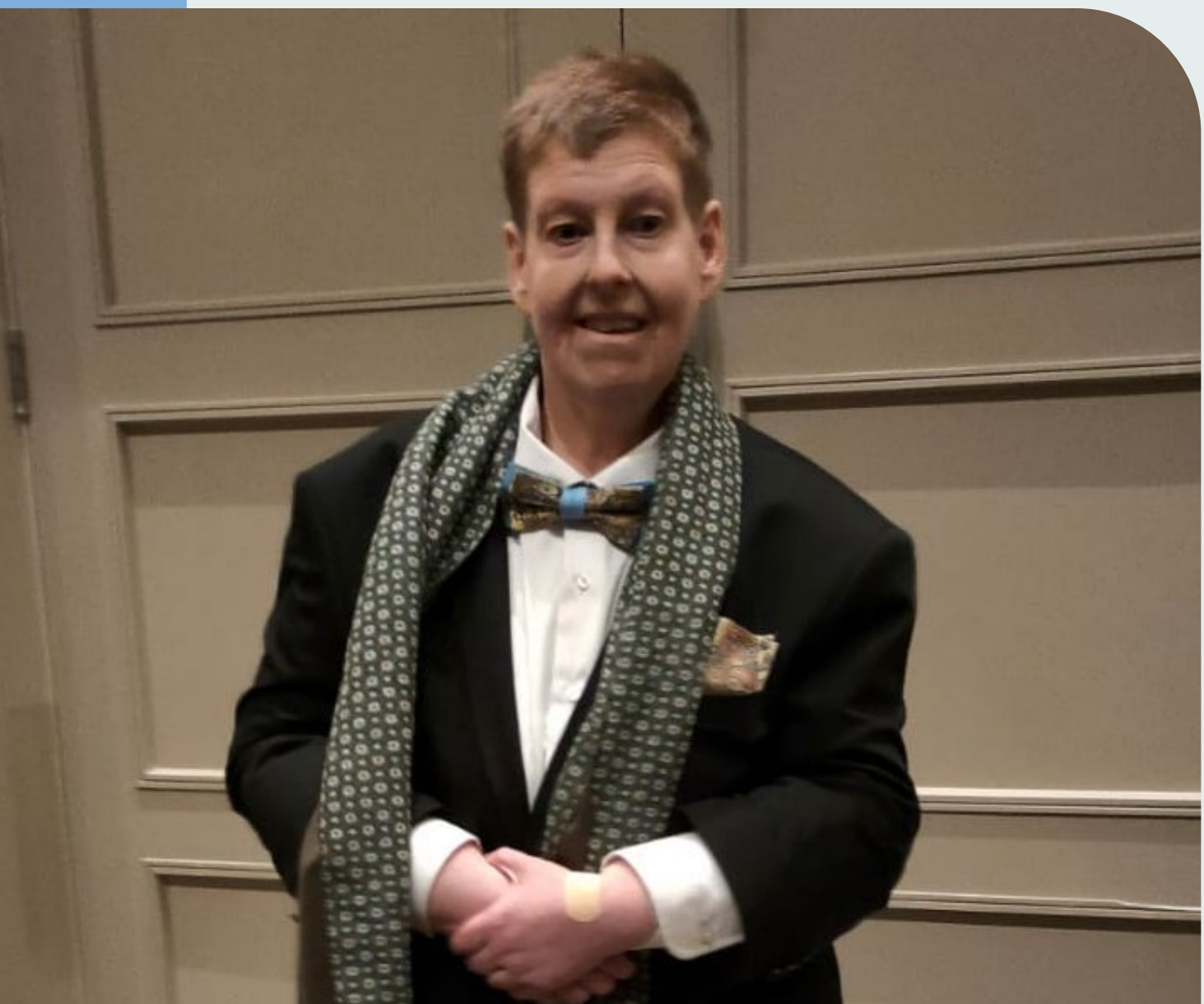
"We constantly worry about siblings, a real risk of physical injury – feelings of guilt."

Summary

- PWS is a complex, multi-systems condition: management requires involvement from all arms of the HSE- Acute, Mental Health, Primary/ Community, Disability /Social Care.
- Workstream 1 examined PWS family carer experience of all HSE services accessed or needed by their family member with PWS, particularly social care provision. (Educational provision was also included.)
- A qualitative research project was undertaken which consisted of two phases – an online survey and a peer-facilitated workshop.
- Three overarching themes emerged from the research that were consistent and present across all types of service provision and applied to all age groups: (1) Dissatisfaction with services, (2) Behavioural Issues and (3) Impact on Family.
- Subthemes were developed from each overarching themes.
- The subthemes for 'Dissatisfaction with Services' were: 'Lack of Resources/Inequity of Provision', 'Lack of Knowledge', 'Poor Communication/ Coordination', and 'Delays in Access'.
- The subthemes for 'Behavioural Issues' were: 'Challenging Behaviours' and 'Destructive Behaviours'.
- The subthemes for 'Impact on family' were: 'Pressure on Caregivers',' Relationships within the Family' and 'Anxiety about the Future'.

CHAPTER FOUR

Voices of people with PWS



Introduction

This chapter looks at the voice of people with PWS. There is a growing awareness of the need to listen and action the voice of people with disabilities.

The chapter references findings from the SUAS (Stepping Up Adults Symposium) stream at the 11th International Prader Willi Syndrome conference in Ireland in July 2022, research from Prof. Eizabeth Dykens and the voices of adults and children from Ireland.

SUAS

The SUAS programme provided a forum for adults with PWS from across the world to talk about their lived experience of the syndrome.

One of the primary goals of the SUAS programme was to encourage adults with PWS to think about their future in terms of self-advocacy, education, employment and living as independently as possible. The two main themes to come out of this discussion were:

1. PWS awareness across all areas of their lives

The group were unanimous in their wish that everyone involved in their lives should have a greater awareness of PWS. They spoke about times when professionals supporting them did not understand them and how this could lead to conflict. They spoke about how they were excluded from social events because they struggled when food was available. They felt that some friends and family didn't understand how it impacted them. Some members of the group suggested that people with PWS should be supported to educate others about PWS.

2. The wish for greater independence with supports around food and diet

The group expressed a strong desire for increased independence but spoke about how difficult that is without support. Many felt that it was unfair on their parents and families and wanted their own place to live, as many of their siblings had. They also stated that they would struggle to do so with unsupported access to food and a managed diet.

At one of the sessions, Living with Hyperphagia, one of the delegates from Ireland discussed the impact hyperphagia has had on their quality of life and mental health.

"Other people with a craving can choose not to do that if they want to, the difference is that we can't."

"Prader Willi syndrome has turned me into someone that I'm not, then trying to explain to people no matter how much you explain, people still don't understand."

"I started taking food from shops which totally goes against me as a person and my values, so I started feeling really guilty about that. I felt really bad about myself, people really judged me for that . I had to leave both the training centres because of those issues and very little support."

Dykens (2021), *Food is a poisonous drug.*

The findings from the SUAS programme are echoed in recent research by Prof. Elizabeth Dykens, Vanderbilt University, entitled 'Food is a poisonous drug'.

Twenty-one young people with PWS were interviewed as part of this study that assessed how they perceive their syndrome and clinical trials. Five themes emerged:

1. struggles with chronic hunger and food-seeking that impede goals and relationships;
2. struggles with anxiety and outbursts, schedule changes and school;
3. distancing from PWS;
4. needs for clinical trials that cure PWS, reduce hunger or anxiety, and lead to improved outcomes;
5. and needs for advocacy and awareness of PWS.

"PWS affects the jobs that you get and how things go in the community, you can't go where they have food around."

"Like the food around me, it's like a drug that kills me. The food is a poisonous drug that kills me."

3. Voices from Ireland on the lived experience of PWS

Adults and children from around Ireland were asked to comment on life with PWS.

"I feel anxious when food is around, I need people to help me. Sometimes I feel not happy in my head and I feel sad."

"If they don't know about my food, they don't know about me."

"I feel hungry all the time, sometimes it stops me from doing things that I love to do."

"If there is a change, it makes me worried and sometimes other people don't understand PWS and it makes me feel worried."

"I can get very upset when the staff at my centre offer me sweets even though they say they understand my PWS."

"People at my day service need to make sure that they understand my PWS. PWS is not me, I hate it."

"It's hard for me to make friends, everything we do seems to be based around food and it makes me sad."

"I need constant reassurance that my plans for the day are going to go ahead. My plans are what keep me from losing my mind."

"I feel constantly tired, I don't sleep well and my weak muscles makes it harder for me to do things that others in my centre can do."

"I don't have friends in the centre, they don't understand me. I spend most of my time on my own. It's very boring."

"Then in the morning, food is like waking me up, that's the problem with me."

"At home, if food is left out, it's just calling to me. It feels like I can't think of anything else until I eat."

CHAPTER FIVE

Work Stream 2: Model of Support

Introduction

In line with its Terms of Reference, Workstream 2 set out to examine the optimal, evidence-based model of Social Care provision for people with PWS and their families with respect to Respite Supports, Residential Supports, Home Support and Day Services/New Directions.

PWS is a complex, multi-systems condition: management of PWS requires a full-systems model approach as the individual needs to have access to acute and community services at any given time. For this reason, it was necessary for Workstream 2 to map out the presenting needs of the individual with PWS and general service provision requirements across the lifespan, before looking in detail at Social Care needs and provision.

The general principles, policies and standards underpinning Social Care provision in Ireland apply to people with PWS. However, there are unique risk factors which apply to people with PWS that must be acknowledged, understood and managed in order for the person with PWS to live safely and successfully within their communities. Complex ethical issues exist around rights, autonomy, mental capacity, and restrictive practices around food, money and medications. Current research and best practice models indicate and support the need for specific models of intervention for people with PWS and their families.

Methodology

Workshops

The members of Workstream 2 used a facilitated workshop approach: each workshop focused on the needs of particular age cohorts (see Table 1) and expanded on the following themes:

- Current service provision and care supports
- Challenges identified to current service provision and care support
- Additional requirements needed to enhance current service provision and care support
- Response to additional requirements
- Evidence based material to support models of intervention

Workshop 1	0-5 years, 6-12 years
Workshop 2	13-17 years, Transitional care 16-25 years
Workshop 3	Transitional care 16-25 years, 18+ years,

Table.1

Best Practice International Models of Care

Reference to current international models and the extant literature formed part of the process, including the findings and recommendations of *A Population-Based Profile of Prader-Willi Syndrome in Ireland,* Gallagher et al, 2017.

Of further influence and significance to the outcomes of Workstream 2 was the attendance by members of Workstream 2 at the *5th International Caregivers' Conference for PWS* hosted by IPWSO (Munich, 28-30 August 2018). (Workstream members attending included members representing the HSE, PWSAI and a professional care organisation.) Ireland is an active participant and leader within this international organisation which enables access to current evidential research and service delivery models. IPWSO's publication *Best Practice Guidelines for Standards of Care in PWS* is updated following each conference.

In addition, immediately prior to the setting up of the National Working Group, PWSAI and service provider representatives visited centres providing best practice social care supports to individuals with PWS in Denmark, Germany and the UK, and had consulted with representatives of services from Australia and the USA.

Findings and Discussion

The findings of Workstream 2 are discussed below under the following headings;

Finding 1: General Service Needs

Finding 2: Need for Transition planning

Finding 3: PWS-Specific Care: The Optimal Model of Social Care Provision

Finding 4: Service-Specific Considerations:

4.1 Respite 4.2 Home Sharing

4.3 Home Support

4.4 Outreach

4.5 Residential

4.6 Residential Shared Care

4.7 Day Services/New Directions

Finding 1: General Service Needs

Workstream 2 first mapped out the presenting needs of the individual with PWS across the lifespan and general services required to meet those needs. The complex and changing interplay of multiple needs pointed to a requirement for a holistic, person-centred approach to the individual with PWS in line with the HSE's Social Care Model for service provision for persons with disabilities. The findings concerning general service requirements are presented below in table formc:

Services Required	Service Components
Acute Hospital Services	Endocrine, Respiratory, ENT, Orthopaedic, GI (list not exhaustive)
Mental Health Services	Psychiatry, Psychology, Behavioural Support Team
Primary Care	GP, Physio, SLT, OT, Dental, Ophthalmic, Audiology, Podiatry.
Dietetics	PWS-specific (acute and community)
Social Care Services	Home Support, Respite, Outreach, Residential, New Directions/Day Services
Progressing Disability Services	Early Intervention Team, School Based Team
Information/Training	PWS-specific training/CPD for all professionals involved
Planning	Person Centred Planning, Transition Planning, Care Co-Ordinator, Register
Educational Supports	SNA Access, Resource Teaching, Supports to access Further Education
Family Supports	Information/Training, Psychological Supports, Genetic Counselling

Essential Services. Table.2

Finding 2: Need for Transition Planning

Workstream 2 found there is a huge need for coordination, collaboration and structured protocols at times of transition in order to pass on essential information, mitigate family stress, prevent service breakdown, and protect the mental health and behavioural status of the individual with PWS. Difficulties are encountered across all services and all stages of transition but particularly during the transition between paediatric and adult services. Members examined in detail difficulties and solutions across acute, mental health, educational and disability/social care services for those aged 16-25 years. Discussions around the needs of this age cohort inform many of the recommendations made in the final chapter of this report and are in line with The National Clinical Programme for Rare Diseases document *Model of Care for Transition.*

Finding 3: PWS-Specific Care: The Optimal Model of Social Care Provision

Workstream 2 found that specialist approaches specific to PWS are necessary in order to ensure the physical safety and psychological wellbeing of the person with PWS. The individual with PWS has a genetically driven, life-threatening, excessive drive for food which has to be skilfully managed and the physical environment must be adapted to make it 'food safe'. Anxiety, and challenging and manipulative behaviours around food must be managed. The person with PWS cannot actively and safely participate in the wider community without an appropriate level of support. The adult with PWS is vulnerable to exploitation in return for food and may also fall foul of the law, with shoplifting of food items common. Food related premature death is also a risk (e.g. gastric rupture, choking, accidental deaths e.g. by impulsively crossing the road to obtain food).

IPWSO's Consensus Statement on PWS Care states 'The preferred form of living arrangement is in a PWS-specific environment, This allows for consistency in treatment and a sense of fairness to the individual.' Gallagher highlights as critical that Residential and Respite Services 'are PWS-specific and provide the appropriate environmental controls in a family like environment, providing a structured predictable environment that controls access to food and reduces the anxiety of the individual.' She points out that this type of provision has been 'shown to prolong life and reduce morbidity associated with extreme obesity'. Food secure environments, which are essential for people with PWS, can rarely be assured outside of PWS-specific residential settings run by staff trained in the management of PWS. PWSA UK's publication 'Residential Care' describes how semi-independent living or living in non-PWS specific residential settings can lead to "severe obesity, failing health and early death". German services providing dedicated, PWS-specific residential care have reported reduced use of psychotropic medication and increased self-regulation skills among adults with PWS in this kind of environment.

Those supporting the individual with PWS, both at frontline and organisational level, need training and ongoing continuous professional development (CPD) in PWS. Staff must be trained to deal with PWS related medical issues and emergencies, and skilled in implementing Food Security (both physical and psychological*), Anxiety Management, Positive Behaviour Support, Crisis Management and at managing PWS related challenging behaviours such as Skin-picking, temper outbursts, impulse buying etc. Food security usually includes one to one supervision, securing food access across all environments. The hyperphagia experienced by individuals with PWS necessitates that robust Restrictive Practices Policies, and Safeguarding measures are in place. (See section *Nutritional Phases and Hyperphagia* on page 15 of this report.)

All necessary environmental supports, (including any necessary adaptations to the building), must be put in place prior to the person with PWS entering the service. Non-negotiable supports include daily visual schedules, personal development plans and caregivers trained in PWS care, access to psychological and behavioural supports, and a continuum of care throughout the day, (including 'waking staff' at night for residential provision). Structure, Predictability, and Communication protocols and tools are essential, so too individual behaviour plans, meticulous recording of behavioural data and health status, and protocols for reviewing same. The physical environment must be 'food safe'. Also essential are generous personal space to allow the person to disengage socially whenever necessary.

The findings of Workstream 2, in relation to the model of care needed to properly support the individual with PWS, are presented below in tabulated form (Table?). A fuller description of the optimal model of PWS social care provision can be found in IPWSO's comprehensive document *Best Practice Guidelines for Standard of Care in PWS.* See also IPWSO's *Consensus Statement on Environmental Structure for PWS Living* in Appendix.

Guiding Principles	• Collaboration and Trust. • Training, Education and Information. • Clearly defined Statements of Purpose. • Person Centred/Social Care Model. • Holistic Assessment of Need, regardless of IQ. • Community Based
Food related Supports	• Food Security (both Psychological* and Physical) • Distance from food shops/outlets • Weight maintenance/weight loss where indicated • Dietician led, balanced, healthy, attractive meal planning • Safeguards around access to money and medications • Robust Restrictive Practices Processes • Robust Risk Assessment/Management • Robust Safeguarding Practices
Access to Services	• Access to Medical Services in the Community/Primary Care • Access to Multi-Disciplinary Team • Access to Mental Health Services • Access to Dietician with expertise in PWS

Staffing Requirements	• Service provider knowledge of PWS at organisational level • Support team trained in Social Care/Disability training and PWS • PWS specific training to the standard of IPWSOs 'Best Practice Guidelines for Standard of Care in PWS'/ ongoing CPD • RNID (Intellectual Disability Registered Nurse) on staff. • Staff/Client Ratio based on assessment of need • Substantive 24-hour supervision/support to include waking night duties. • Staff trained in medication management/specialised medical equipment
Behavioural Supports	• Psychological Food Security* • Consistency (of Staff/ Support/Approach/Training) • Anxiety Management/Reassurance/Understanding • Structure/ Communication protocols and tools e.g. daily visual schedules, routines, display boards • Positive Behaviour Support/led by experienced Behaviour Support Team • Behaviour Support Plans/Functional assessments • Knowing triggers/Knowledge of 'attention switching' deficits
Community/Social Supports	• Supported to access Work, Education, Recreation in the community • Supported to maintain relationships with Family • Access to Transport
Environmental Design Requirements	• Food Security e.g. separate locked kitchen for food preparation, distance from food outlets, specialised bins, movement monitors. • Sensory needs taken into account • Accessible bathrooms/Custom made furniture (obesity related) • In house gym (community-based gym sufficient for Day Services) • Need for adequate private, communal and outdoor space Weighing centre in private location (fulltime residential only) • Appropriate place for daytime rest/nap (Day Services only)

Table No. 4 Model of Support for Social Care Provision for Persons with PWS: Respite, Residential, New Directions / Day Services

Finding 4: Service-Specific Considerations

4.1 Respite

Respite care is the provision of alternative care for a person with a disability in order to enable the family carer to take a short break, holiday or rest.

For individuals with PWS, respite that is residential in nature should be available, as endorsed by Ireland's Rare Diseases Plan.

As with all social care provision for individuals with PWS, respite supports must be PWS-specific. The PWS-specific model of support is outlined above in the section *PWS-Specific Care: Optimal Model of Social Care Provision* and in tabulated form in Table? Essential elements of support include the provision of a food safe environment, 'psychological food security'*, a structured environment which includes the provision of visual supports such as daily visual schedules, and staff trained in the complexities of PWS.

Dedicated, PWS-appropriate, centre-based, respite is recommended as it allows for the development of infrastructure and practices that are oriented specifically towards the needs of people with PWS. Food security may include one to one supervision, securing food access across all environments, , posting mealtimes and menus, and training all team members. This model enables people with PWS

to develop positive relationships with others with the syndrome and help meet their social needs. It contributes to a sense of wellbeing and fairness in the person with PWS as everyone is subject to the same necessary restrictions around food, money and medications.

It essential that respite is regular and planned ahead of time: without certainty for the person with PWS, it will be of no benefit to families - on the contrary, without certainty around all aspects of the respite plan, it will cause severe anxiety and possible behavioural breakdown in the person with PWS, thereby further compounding the family's difficulties and level of stress.

While taking into account HSE disability policy with respect to community-based services and decongregated settings, it may be useful to place respite, and residential services in the same general locality. Such general proximity of services may allow providers to develop their expertise in relation to PWS and may facilitate greater efficiency and cost-effectiveness through the sharing of some support services. In addition, when more people with PWS live in or visit a particular area, it increases the likelihood that PWS will become understood locally, both in the wider community and among relevant community-based services such as GP and Mental Health Services. The latter has been the experience of PWS services internationally, for example, in the towns of Regens Wagner in Bavaria, Oconomowoc in Wisconsin and Kettering in England.

Respite that works best for both the family and the individual with PWS is:

- Residential/Overnight
- Centre-based
- Regular/Planned
- PWS-specific care
- Dedicated

4.2 Home Sharing

Home Sharing is defined as the provision of care to people with a disability in the Home Sharing family's home.

Home Sharing, used as a respite model for persons with PWS, presents considerable challenges.

Lengthy and detailed PWS-specific training is required for caregivers, including how to provide 'psychological food security'*. Physical modifications to ensure food security may be necessary. Food security may include one to one supervision, securing food access across all environments, posting mealtimes and menus, and training all team members.

Family homes, by their very nature, cannot guarantee the level of predictable environment needed for the person with PWS. Home Sharing models of respite for persons with PWS are at greater risk of placement breakdown, leading to great anxiety for the person with PWS and a further crisis of confidence in services for carers.

4.3 Home Support

Home Support is a model of care that provides supports to a person with a disability in their home environment in order to enable the person to remain in his/her own home for as long as possible- its primary purpose is not the provision of respite care for the primary carer or family.

As a model of support for persons with PWS, Home Support presents considerable and, at times, insurmountable challenges; providers must have the specialist knowledge and skills needed in order to deliver appropriate care in line with current, evidence-based, best practice in PWS care. In addition, any necessary environmental adaptations, including those specific to the maintenance of 'food security' (both physical and psychological[1]) must be put in place.

In respect of the family caring for a member with PWS, the Home Support model can present considerable challenges. When the person with PWS is present in the family home, the many structures needed to support that person (e.g. locked kitchens, carefully scheduled meals and rigid timetables) must also remain in place for the rest of the family. Siblings will not have the opportunity to experience 'a normal food environment', even briefly. Food security (both physical and psychological) is a redline issue in professional PWS care: if an individual home environment is not food safe, then the professional carer will have difficulty providing best practice care.

In addition to issues around hyperphagia, siblings and carers may continue to be exposed to challenging and destructive behaviours and be

1 Psychological Food Security is state of mind and allows the person with PWS to engage in and enjoy a range of activities and pursuits without the continual distraction of food-related thoughts.

Food security is the essential ingredient for managing the food related behaviours associated with PWS. Food security has been defined as the ready availability of nutritionally adequate and safe foods with an assured ability to acquire these foods in socially acceptable ways. In PWS, food security provides no doubt when meals will occur and what will be served; no hope of getting anything different from what has been planned, and no disappointment related to false expectations. Food security is achieved by securing food access across all environments, super-vising food access across all environments, posting mealtimes and menus, and training all team members. When the individual with PWS is experiencing food security, that is, no doubt, no hope and no disappointment related to food, a generalized behavioural improvement typically occurs. For this reason, food security is the mainstay of PWS management before considering the implementation of behavioural interventions and pharmacotherapy (adapted from "Pittsburgh Partnership Psychiatrists' Primer for Prader-Willi Syndrome" by Janice Forster, MD and Linda Gourash, MD

unable to socialise within the home environment with extended family and friends- or indeed among themselves. Carers report that family members with PWS suffer great anxiety about who is 'in charge' when both the primary caregiver and a paid caregiver are in the house, and report behavioural breakdown as a consequence; rigid thought processes, a need for clear structures and hierarchies, and chronic anxiety are fundamental characteristics of the PWS personality.

4.4 Outreach

An Outreach support program provides individualised services and supports to children with disabilities and their families, within their own homes and communities. Supports are tailored around the needs identified during assessment and formulated into a personalised support plan. Outreach supports should be creatively designed and provided in the location suited to the child and family's needs.

Examples of the sort of supports that might be provided for families with a member with PWS could include: provision of after school support in a location outside the family home but tailored to the child's needs, provision of 1 to 1 supports in the community to allow an individual engage safely with a community activity such as Special Olympics, day time Saturday respite away from the family home, proactive supports and interventions such as functional assessments and behaviour plans.

As with all social care provision for individuals with PWS, Outreach supports must be PWS-specific and include essential elements of care such as a food safe environment, 'psychological food security'* , environmental supports including the provision of visual supports, and staff trained in the complexities of PWS.

4.5 Residential

As with all social care provision for individuals with PWS, full-time residential supports must be PWS-specific: this model of support is outlined in the section *PWS-Specific Care: The Optimal Model of Social Care Provision* above and summarised in Table? This is in line with HSE policy of individualised and person-centred supports as, when addressing the needs of any individual, account must be taken of needs that arise from a specific disability or condition. (Essential elements of PWS- specific support include the provision of a food safe environment, 'psychological food security'*, a structured environment which includes visual supports such as daily visual schedules, 1 to 1 supervision in the community and staff trained in the complexities of PWS.)

In line with HSE policy *Time to move on from Congregated Settings*, residential services for adults with PWS should be community-based. Consideration should be given to locating a number of PWS-specific residential community houses in the same general locality. This can allow providers to develop their expertise in relation to PWS and can facilitate greater efficiency and cost-effectiveness through the sharing of some support services. This model of provision entails more people with the syndrome living in the same general area, increasing the likelihood that PWS will become understood in the local community, and that relevant community-based services will develop expertise in the syndrome e.g. primary care, local MHID services, educational and leisure providers such as Special Olympics, ETBs. The latter has been the experience of a number of organisations internationally, for example, Consensus UK (formerly Gretton Homes).

PWS is associated with complex medical needs and so consideration should also be given to the distance of residential community services from a tertiary hospital.

4.6 Residential Shared Care

Shared Care is where an individual with a disability resides both at home and in a residential service, (for example, 3.5 days per week in the residential service). The principle of shared care is that individuals will spend their time equally between the family home and residential service. A shared care service provides planned shared care for families, giving them time off from caring responsibilities, and provides a home from home experience which is structured and designed around the individual needs of the person.

As with all social care provision for individuals with PWS, shared care residential supports must be PWS-specific. The discussion above on full-time residential supports also applies to the residential shared care model. (see section 4.5 Residential above).

4.7 Day Services/New Directions

As with all social care provision for individuals with PWS, day service supports must provide PWS-specific care as described in the section *PWS-Specific Care: Optimal Model of Social Care Provision* above and summarised in Table? (Essential elements of support include the provision of a food safe environment, 'psychological food security'*, a structured environment which includes the provision of visual supports such as daily visual schedules, and staff trained in the complexities of PWS.) This may be challenging for the provider as day services and supports will almost inevitably be provided in mixed disability settings.

In line with HSE policy *New Directions*, person-centred, community-based Day Services should be available to all school leavers and adults with PWS. Rigorous assessment of need, which takes account of the considerable discrepancy between IQ and adaptive functioning seen in individuals with PWS, must take place in order to ensure safe and

successful outcomes, maximise potential and ensure each person has the best possible life.

For persons with PWS in fulltime residential care, New Directions/Day Services can be either centre based or provided from the place of residence depending on the needs and preferences of the person with PWS. Where a person in fulltime residential care accesses centre-based Day Services, it is essential that the residential care provider collaborates with the day service provider to ensure that a continuum of PWS-specific care is provided throughout the day.

For adults with PWS who still live in the family home, it is essential that centre-based day services are provided. Not all homes are food secure and appropriately adapted for the needs of the person with PWS: most families struggle to put the necessary PWS behavioural interventions and structures in place. People with PWS are rigid thinkers and unable to cope with conflicting modii operandi. In addition, people with PWS generally have fixed ideas and hierarchical tendencies and suffer great anxiety about who is 'in charge' when both the primary caregiver and a Social Care provider are in the house. When conflicts inevitably arise, the family is put under additional unnecessary strain.

It is essential that community-based centres providing Day Services/New Directions for adults with PWS have the space and physical infrastructure to provide the necessary environmental supports: some community hubs/drop in facilities currently being developed in Ireland are wholly inadequate to meet the needs of individuals with PWS. The HSE's *Interim Standards for New Directions* acknowledge the need for 'premises and facilities…designed to support the delivery of safe services'. Other standards particularly relevant to supporting an individual with PWS include Standard 3.2 concerning Positive Behaviour Support and emotional well-being, Standard 5.2 concerning staff competencies and reliable supports, and Standard 2.12.8 concerning clinically directed special dietary requirements.

In order to access education, employment or volunteering opportunities in the community, individuals will need PWS-specific support in order to be successful and safe and maintain good mental health and behavioural status. The level of support needed by the individual must be rigorously assessed in advance of participating in education, work or volunteering: evidence suggests that one to one support is almost always necessary.

Summary

- Workstream 2 set out to examine the optimal, evidence-based model of Social Care provision for people with PWS and their families with respect to Respite Supports, Residential Supports, Home Support and Day Services/New Directions.
- Methodology included facilitated workshops that looked at the totality of needs of individuals with PWS across the lifespan, a review of the literature, attendance at the 5th International Professional Caregivers Conference, and examination of models of social care provision abroad, including site visits.
- Workstream 2 found that specialist approaches specific to PWS are necessary in order to ensure the physical safety and psychological wellbeing of the person with PWS.
- Food security is achieved by securing food access across all environments, supervising food access across all environments, posting mealtimes and menus, and training all team members.
- PWS is a complex, multi-systems condition: management of PWS requires a full-systems model approach as the individual needs to have access to acute and community services at any given time.
- Psychological and Physical Food Security is the cornerstone of PWS care. 1 to 1 support frees the individual to live his/her life without constant anxiety related to food.
- Other core supports include visual supports, carers trained in the management of comorbid medical problems and challenging behaviours common in PWS, anxiety management etc (See Table? 'Model of Support').
- Respite and residential supports are most effective in meeting the needs of people with PWS when provided in dedicated rather than mixed disability settings.
- The level of support needed by the individual must be rigorously assessed in advance of participating in education, work or volunteering: evidence suggests that one to one supervision is almost always necessary.
- The provision of residential respite is a necessary support for families.

CHAPTER SIX
Recommendations

Introduction

This chapter sets out the recommendations of the National Working Group on PWS. It is clear there is an urgent need for substantial improvement in health and social care provision in Ireland for those living with PWS and their families. This is evidenced by the scientific literature, from the consultation feedback and research conducted for this report, and from examining models of best practice internationally. There is an urgent need to develop new, dedicated services, particularly in the area of social care supports such as respite and residential provision, and mental health. Some services will need additional resources to more effectively meet the needs of those with PWS, for example, additional multi-disciplinary supports for adult endocrine clinics catering for patients with PWS. Other services can be improved within existing resources by adopting PWS-specific approaches outlined in Chapter 4.

'There is an urgent need to develop new, dedicated services, particularly in the area of social care supports, such as respite and residential provision, and in mental health.'

The complex interplay of needs in Prader-Willi syndrome means the success of any service is dependent on the success of all individual services, for example, the success of social care services is dependent on access to appropriate high quality mental health services, and on expert knowledge and management of medical issues. If services are not informed by an understanding of PWS, individuals with PWS typically die prematurely. It is obvious that services that do not meet the unique needs of people with PWS are an inefficient use of public money.

The implementation of the recommendations of this report would substantially change the life of those with PWS, leading to better physical and mental health outcomes, reducing morbidity and mortality, and enhancing quality of life by reducing suffering and providing supports to safely access what others take for granted such as further education and community participation . Other positive outcomes would be improved mental health and quality of life for siblings and family carers, and the protection of family units and relationships within families. The implementation of this report would also contribute to the promotion of equality in our society as it has been established in the literature that most care for individuals with PWS in Ireland is provided by women in the home (Gallagher et al., 2017).

Recommendation 1	**Quality and Delivery of Health and Social Care Services.**
Recommendation 2	**Respite Services**
Recommendation 3	**Residential Services**
Recommendation 4	**Day Services (New Directions)**
Recommendation 5	**Acute Services**
Recommendation 6	**Mental Health Services**
Recommendation 7	**Educational and Lifelong Learning Supports for Individuals with PWS**
Recommendation 8	**Psychological Supports for Families**

Recommendation 1
Quality and Delivery of Health and Social Care Services

1.1 For the HSE and PWSAI to jointly develop a training programme delivered via multiple platforms (e.g., online, workshops) for health service staff, social care service providers and all other agencies involved in the care and support of individuals with PWS.

1.2 For the HSE and PWSAI to work with the relevant professional training programmes to ensure that new graduates are competent to work with individuals with PWS across the lifespan.

1.3 For the HSE to establish a voluntary register of individuals with PWS to inform immediate and future service planning and development.

Recommendation 2
Respite Services

2.1 Dedicated, PWS-appropriate, centre-based, residential respite is recommended as it allows for the development of infrastructure and practices that are oriented specifically* towards the needs of people with PWS. This model enables people with PWS to develop positive relationships with others with the syndrome and help meet their social needs. It contributes to a sense of wellbeing and fairness in the person with PWS as everyone is subject to the same necessary restrictions around food, money and medications.

2.2 It is essential that respite is regular and planned ahead of time: without certainty for the person with PWS, it will be of no benefit to families - on the contrary, without certainty around all aspects of the respite plan, it will cause severe anxiety and possible behavioural breakdown in the person with PWS, thereby further compounding the family's difficulties and level of stress.

Recommendation 3
Residential Services

3.1 Residential services for persons with PWS should be PWS-specific* and meet the standards of IPWSO's Best Practice Guidelines for Standard Care in PWS

3.2 Although the numbers of individuals with PWS in Ireland is low, there is a large geographical spread of those potentially seeking residential placements. There should be at least one PWS specific residential service in each HSE Health area and the option of providing cluster type services in areas that require more than one service.

3.3 There should be equitable access to out of area RHA funding, and across the state, for PWS specific residential community-based care.

Recommendation 4
Day Services / New Directions

4.1 PWS-specific supports should be provided for persons with PWS attending Day Services: non-negotiable supports include but are not limited to food security, food certainty, daily schedules, personal development plans, a place to rest, caregivers trained in PWS, access to psychological and behavioural supports, and a continuum of care throughout the day including when travelling to and from the service.

4.2 People with PWS who wish to access Further Education, employment or volunteering opportunities in the community will require one-to-one, PWS-specific support and an appropriate assessment of additional needs.

Recommendation 5
Acute Services*

5.1 Establishment of a National Centre of Expertise for Prader-Willi Syndrome, in line with National Rare Disease policy.

5.2 Provide additional support to existing multi-disciplinary, endocrinology led, PWS adult and paediatric clinics in line with National Rare Disease policy to enhance the care provided and enable continuation of these clinics in the face of competing demands.

This should include (but not confined to) endocrinology, dietetics, clinical nurse specialist, psychiatry, psychological supports, OT, SLT, physiotherapy, orthopaedics, respiratory, ENT, ophthalmology, GI, dentistry, podiatry.

5.3 Development of Transitional Care for adolescents moving to adult services in line with the National Clinical Programme for Rare Diseases 'Model of Care for Transition', (including exploration of the case for Care Coordinators to manage transition in a coordinated manner across medical, mental health and social care domains).

5.4 Timely delivery of time critical services e.g. rapid access to sleep studies to enable early access to Growth Hormone Therapy, MRIs and surgery to correct spinal deformity before secondary disability occurs.

*In Ireland, Acute Services refers to hospital services provided at tertiary centres.

Recommendation 6
Mental Health Services

6.4 Consultation mental health services should be provided as part of multidisciplinary team provision at National Centres of Expertise for PWS (paediatric and adult). These can provide support, training, and outreach to community mental health services around diagnosis and treatment specific to the needs of individuals with PWS. `

6.2 Access to Mental Health Intellectual Disability (MHID) adult services based on a diagnosis of PWS.

6.3 Clear functioning pathways to adult MHID services in the community.

6.4 Clear functioning pathways to Child and Adolescent Mental Health Services (CAMHS and CAMHS-ID) in the community.

6.5 Early access to specialist behaviour support services should form part of the mental health services provision for children and adults with PWS.

6.6 Comprehensive planning for transition from paediatric to adult services.

Recommendation 7
Educational and Lifelong Learning Supports for Individuals with PWS

7.1 Development of professional learning materials for teachers of students with PWS, for example, PWS-specific Teacher Education to be included on the NCSE's (National Council for Special Education) Primary and Post-Primary CPD (Continuous Professional Development) Programme, PWS-specific online summer courses for teachers, inclusion on curriculum of the DES funded post-graduation programmes for special education teachers, inclusion on curriculum for education of Montessori Teachers and Early Educators.

7.2 Consultation on the development of teacher education in PWS between PWSAI, the NCSE and other relevant stakeholders.

7.3 Educating individuals with PWS requires a 'whole school' or 'whole college approach'*: the pupil or student with PWS needs advocacy within the organisation, The level of support needed by the individual must be rigorously assessed in advance of participating in education, work or volunteering: Assessment to include consideration of the supports needed around food security, food certainty, mobility and transport, evidence suggests that one to one supervision is almost always necessary.

7.4 Effective inclusion of adult learners with PWS in Further and Higher Education and Lifelong Learning services will require enhanced collaboration between the relevant stakeholders, including: Adult Learners with PWS, PWSAI, SOLAS (the Further Education and Training Authority), ETBs (Education and Training Boards), HEA (the Higher Education Authority), HSE, NLN (the National Learning Network), AHEAD (the Association for Higher Education Access and Disability) and AONTAS (the National Adult Learning Organisation).

* A whole school approach is cohesive, collective and collaborative action in and by a school community that has been strategically constructed to improve student learning, behaviour and wellbeing, and the conditions that support these.

Recommendation 8
Psychological Supports for Families

8.1 Access to therapy services for family members living with or caring for a family member with PWS. The purpose of these services would be to provide a safe time and place to explore adaptive coping mechanisms and strategies to support their own wellbeing.

8.2 Provision of appropriate respite supports for all families caring for a family member with PWS.

*Psychological Food Security is state of mind and allows the person with PWS to engage in and enjoy a range of activities and pursuits without the continual distraction of food-related thoughts.

Food security is the essential ingredient for managing the food related behaviours associated with PWS. Food security has been defined as the ready availability of nutritionally adequate and safe foods with an assured ability to acquire these foods in socially acceptable ways. In PWS, food security provides no doubt when meals will occur and what will be served; no hope of getting anything different from what has been planned, and no disappointment related to false expectations. Food security is achieved by securing food access across all environments, supervising food access across all environments, posting mealtimes and menus, and training all team members. 1 to 1 support is crucial as it frees the individual to live his/her life without constant anxiety related to food.

- When the individual with PWS is experiencing food security, that is, no doubt, no hope and no disappointment related to food, a generalized behavioural improvement typically occurs. For this reason, food security is the mainstay of PWS management before considering the implementation of behavioural interventions and pharmacotherapy (adapted from "Pittsburgh Partnership Psychiatrists' Primer for Prader-Willi Syndrome" by Janice Forster, MD and Linda Gourash, MD

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The Underlying Causes for Lack of Mental Capacity in Adults with Prader-Willi Syndrome. Presentation delivered by Prof Tony Holland, University of Cambridge

Findings from attendance of Workstream 2 members at the 5th International PWS Caregivers Conference in Munich, 28-30 August 2018

Findings from visits of PWSAI, RehabCare and Resilience Ireland to Residential Services for adults with PWS at 4 locations in Denmark in ??? and April 2017

Findings from visit of PWSAI to Residential and Day Services for adults with PWS at Regens Wagner, Bavaria, October 2017

Findings from visit of Workstream 2 members to PWS-specific Residential Provision run by Consensus (formerly Gretton Homes), England, May 2018

Findings from PWSAI meeting with the Director of Operations and the Admissions Coordinator of Prader-Willi Homes of Oconomowoc, a Residential and Respite Services Provider for Adults and Children with Prader-Willi Syndrome in Wisconsin, USA, 20

Appendices

Appendix 1

Membership of Work streams

Workstream 1 : 'PWS Carer Experience of HSE Services'
Dr. Michael Byrne, HSE
Don Tallon, PWSAI
Donnchadh Clancy, PWSAI
Ann McCrann, CHI at Tallaght
Prof. Edna Roche, CHI at Tallaght
Workstream 2: 'Model of Support'
Helen Mcdaid, HSE
Laura Keane, ResilienceCare
Paul Wright, PWSAI
Don Tallon, PWSAI
Jane Cox, PWSAI

Appendix 2

Terms of Reference

17th May 2018

Terms of Reference

National PWS Working Group (including work streams)

Piloting good models of practice to support people living with Prader-Willi Syndrome (PWS)

1. Introduction:

The HSE in partnership with the Prader-Willi Syndrome Ireland (PWSAI) is establishing a National Pilot Working Group to develop, implement and test positive models of intervention/ practice in supporting people living with PWS and their families. The work of the National Working Group will build on the positive research1 and experience of the PWSAI2 and will seek to provide practical supports for Parents/ Carers of people living with PWS. The Pilot working group will seek to bring together parents in collaboration with professionals (both National and International) in order to lead and drive an agreed work programme as set out in these terms of reference.

2. Terms of Reference:

1. The overarching aim of this group will be to develop and implement a model of intervention(s) relating to care and supports to children and adults living with PWS and their families/ carers.
2. The models developed and implemented will be based on evidenced informed evaluations of “what works” and “for whom” as a key guiding principle
3. Steps to be undertaken prior to establishment of pilots;
    a. Undertake a profiling exercise of services in place in each HSE CHO to inform a baseline of current provision and how needs are being met in collaboration with PWSAI
    b. Gather, confirm and utilise national international evidence of PWS prevalence as a basis to consider models of support intervention. This work stream will also inform the HSES own estimates process in respect of future Budget allocations/ bids
4. Identify and scope two models of intervention based on the following categories;
    a. Residential respite
    b. Home Supports
5. Subject to available resources, implement on a pilot basis the categories of services as outlined above and evaluate the effectiveness of same with the aim of “mainstreaming” effective service models specifically targeting PWS

1 Ref: https://pwsai.ie/past-events/pwsai-national-survey-launch-2017/
2 Ref: https://pwsai.ie/

3. Work Streams

Two subgroups will be established to undertake the necessary groundwork on behalf of the National Working Group. The group will comprise representatives from PWSAI, experts in the area as well as HSE input. The subgroups will comprise two work streams which are identified in the following table and which will assist the National PWS Woking Group to develop a plan of action.

Work Stream Number	Area	Actions to be undertaken by the subgroup – i.e. explore, develop and detail working papers for the National Working Group	Subgroup member
	a. Profiling and Scoping exercise b. Estimate resource requirements for PWS services	6. What provision is currently in place for people with PWS? 7. What funding is in place/ allocated presently for PWS clients in each CHO? 8. What is the experience of the service user and their families of services for PWS?	**Dr Michael Byrne/ Prof Tony Holland** Don Tallon, Paul Wright, Anthony Carr, Jane Cox.
	Care Pathways – Support Model	9. Detail the optimal model of service provision for people with PWS in respect of day, residential and home support provision. 10. Ensure the model sets out what works and for whom based on best available evidence.	**Ms Helen McDaid** Don Tallon, Paul Wright, Anthony Carr, Jane Cox.

4. National Working Group Membership:

HSE National Office	Family/ National Umbrella Group	Clinical Expert (International Advisor)	Clinical Expert (Ireland)
Dr Cathal Morgan (Chair)	Ms Jane Cox	Professor Tony Holland (UK)	
Dr Michael Byrne	Mr Don Tallon		
Ms Helen Mc Daid	Anthony Carr		

5. Modus Operandi:

The working group members will seek to work on a consensus basis.

HSE National will convene and administer the required meetings.

Expenses incurred through participation in the mediation process will be met by the individual members and their respective organisations.

Appendix 3

Best Practice Guidelines for Standard of Care in Prader-Willi Syndrome, IPWSO, 2010

Consensus Statement on Environmental Structure for PWS Living

All guidelines should take into account the assessment of the individual to assure health and safety and to promote quality of life.

1. The individual's opinion should be taken into account when following these guidelines whenever possible.
2. The preferred form of living arrangement is in a PWS-specific environment. This allows for consistency in treatment and a sense of fairness to the individual. All attempts to create a family like environment including their own space regardless of group size is important for the person with PWS to have a sense of belonging as well as a place to disengage from group living whenever necessary. In addition it is also vital for the individual to have a choice of vocational opportunities, as enjoyment and fulfilment of one's own day is crucial for anyone to feel productive and an important member of their community.
3. Optimal success for the person with PWS will be based on the type of supports that are in place. Those supports require an understanding of the unique needs associated with PWS. All areas of a person with PWS' day should have proper supervision to assist with those unique needs. There needs to be a 24 hour type of supervision in place in either the form of direct caregiver support, alarms or security systems that alert caregiver or monitoring agency that the individual has either left the designated area(s) or that they are in need of assistance. A combination of the above may also be used.
4. At times additional support may be needed due behavioural issues or need for assistance from another caregiver. Each supportive environment, both home and vocational, should have a system in place that allows for quick response from another caregiver to assist in the need at hand.
5. Routine and consistency is essential to the successful living for a person with PWS. Inclusion of their ideas and preferences should be part of the planning of the structure of their regular routine. Meals should be managed and structured in a formal way to ensure that everyone knows the plan of how meals are to be planned, monitored, and served. It is necessary for every person with PWS to have a diet plan that can be followed by all. Exercise should also be a part of their daily routine including some levels of movement or fitness every day with the amount of time to be determined on individual basis. Healthy weight management is also an important part of monitoring the progress of supporting a person with PWS. Weights should be taken at a minimum of once per week based on history.
6. Structure of the home for a person with PWS should have some form of house rules for living with others. This allows for everyone to understand and remain consistent with those guidelines, expectations, and boundaries that are needed when in a group living environment. Another area is the complete security of food, money, and medication which should be locked and managed by caregivers.
7. To assist in overall self-esteem it is important for everyone to feel a part of their community. Inclusion of those activities within the community should be individually assessed to determine environmental supports that may be needed. Understanding the person's own level of independence will assist in maintaining required needs for safety. Living in a community should include good neighbour relationships, it is important to create systems explaining about PWS.
8. It is essential to have individual behaviour management plans ranging from positive motivation to crisis management. This once again allows for consistency and proper understanding of that specific individual's needs when that person may be in a crisis or to simply avoid or redirect the situation.
9. Training of caregivers is key to the success of the individual with PWS. Caregivers need a good understanding of the complexity of the needs of the syndrome as well as the compassion that is needed when supporting a person with PWS.

10. Maintaining healthy relationships once the person with PWS is outside their family's home is important for the person with PWS. Contact and involvement with family and friends may need additional support and guidance from caregivers. That support may consist of teaching relationship building and in some incidences may require additional counselling. Human sexuality may also require support and education.
11. A clear structure is required to develop and maintain a healthy and stable relationship between the parents, the individual, and the caregiver. These communications are most effective if they are team-based and have pre-determined professional boundaries established.
12. Due to the potential for life-threatening situations, it is imperative to create an environmental structure that allows access to comprehensive medical services.
13. To support self-determination as it relates to environmental structures, choices should be offered to assist the person to individualize their room while promoting personal safety.
14. In order to provide effective support services there needs to be an established form of communication and information systems in place for everyone involved in the care and treatment of the individual.

Conclusions:

Across cultural lines, all participants agreed that in order for an individual with PWS to succeed and live a healthy and productive quality of life that a basic level of support, conducive to promoting safety and personal growth, must be in place prior to providing care. It was determined that environmental supports were non-negotiable, including but not limited to;

- food security
- daily schedules
- personal growth and development plans
- trained caregivers
- continuum of care throughout their day.

It was also agreed upon, that it is imperative that as care providers, we continue to explore this cohesive collaboration of efforts through on-going education, training, and awareness.

Appendix 4

Glossary of PWS Terms

Anxiety

Anxiety refers to feelings of nervousness, worry and tension. Anxiety disorders are highly prevalent in PWS and are central to many of the behavioural difficulties that can occur in PWS. A rise in anxiety can be directly related to an escalation in perseveration, compulsivity, non-compliant and oppositional behaviour, frustration and loss of control.

Adaptive Functioning

Adaptive functioning refers to a person's ability to effectively manage daily living tasks andsocial interactions as compared to their age, cultural, and environmental expectations.

Attention Deficit Hyperactivity Disorder (ADHD)

Attention deficit hyperactivity disorder (ADHD) is a behavioural disorder that includes symptoms such as inattentiveness, hyperactivity and impulsiveness.

Challenging Behaviour

Challenging behaviour (also known as behaviours that challenge) is defined as culturally abnormal behaviour(s) of such an intensity, frequency or duration that the physical safety of the person or others is likely to be placed in serious jeopardy, or behaviour which is likely to seriously limit use of, or result in the person being denied access to, ordinary community facilities.

Chromosome

Package of genes that carries genetic information. Most cells have 46 chromosomes (23 pairs, one copy of each chromosome from each parent), which are found in the nucleus of cells.

Cognitive Development

Cognitive development is an important aspect of overall child development. Cognition refers to how we think, pay attention, remember, and learn.

Comorbid

Relating to or denoting a medical condition that co-occurs with another.

Deletion

A missing piece of chromosomal material. In PWS, the deletion is in the proximal part of the long arm of chromosome 15 inherited from the father.

Depression

Depression is a mood disorder that involves persistent feelings of sadness and loss of interest. Symptoms of depression may include being more tearful or irritable, altered sleep patterns, negative thinking, decreased energy, focus, motivation and self-esteem. It can be associated with an increase in typical PWS behaviours eg repetitive, ritualistic behaviours, or with the emergence of new behaviours. Individuals with PWS have an increased risk for depression, especially those with the deletion genetic subtype.

Developmental Delay,

Developmental delay refers to when a child's development lags behind established normal ranges for his or her age. Physical, cognitive, communication, social, emotional, and behavioural skills can all be delayed in PWS.

Executive Functioning

Executive function is a set of mental skills that include working memory, flexible thinking, and self-control. We use these skills every day to learn, work, and manage daily life. Trouble with executive function can make it hard to focus, follow directions, and handle emotions, among other things.

Food-seeking

Resulting from an impairment of satiety at brain level, individuals with PWS usually have a significant preoccupation with food, with problem food-seeking behaviours, (such as food stealing, eating crumbs from the floor) highly prevalent throughout the lifespan.

Gene

A unit of genetic information. Usually contains the genetic code for one protein.

Genotype

The entire set of genes in an organism.

Growth Hormone (GH)

A pituitary-secreted hormone that stimulates growth in childhood and adolescence.

Hyperphagia

Consumption of more than a normal quantity of food. The hyperphagia that occurs in PWS is believed to be caused by a hypothalamic abnormality that results in a lack of satiety.

Hypothalamic Dysfunction

Hypothalamic dysfunction is a problem with part of the brain called the hypothalamus (see definition below).

Hypothalamus

Part of the brain that controls appetite, body temperature, hormones, and other vital functions.

Hypotonia

Decreased muscle tone. Decreased muscle resistance to passive movement. In PWS infantile hypotonia is a nearly universal finding, causing decreased movement and lethargy with decreased spontaneous arousal, weak cry, and poor reflexes, including poor suck.

Metabolism

The body's process of absorbing nourishment from food and turning it into energy or stored fat.

Obsessive Compulsion Disorder

Obsessive compulsive disorder (OCD) is a mental health condition in which a person has obsessive thoughts and compulsive behaviours.

Obesity

A condition in which body weight is considerably (more than two standard deviations) above the range that is normal for body height.

Osteoporosis

Demineralization (thinning) of the bones.

Person-centred

Person-centred care moves away from professionals deciding what is best for a patient or service user and places the person and their well-being at the centre, rather than their condition. Being person-centred means systems and services are orientated towards supporting persons to recognise and build upon their own strengths, preferences and goals to achieve their full potential.

Phenotype

The physical and other characteristics associated with a particular genotype; often used when talking about a genetic condition.

Positive Behaviour Support (PBS)

PBS is a behaviour management system used to understand what maintains an individual's challenging behaviour. The aim of PBS is to improve the quality of the person's life by providing the right support at the right time. Successful implementation needs a whole organisational approach and ongoing commitment.

Psychosis

A serious mental disorder where a person's thoughts and perceptions are disturbed, and the individual may have difficulty understanding what is real and what is not. There is increasing evidence that PWS is associated with high rates of psychosis.

Residential Services

Residential care refers to long-term care given to persons with disabilities who stay in residential settings other than in their own home or family home. Increasingly in Ireland, residential services support people with disabilities to take up tenancies in the community, or provide accommodation and staff supports to enable no more than 3 or 4 people with a disability to live together in a residence or house in the community.

Respite Services

Respite care is the provision of alternative care for a person with a disability in order to enable the family carer to take a short break, holiday or rest. Where the individual is cared for overnight in a dedicated facility, this is described as residential respite.

Restrictive Practices

Restrictive practices in health and social care refer to the implementation of any practice or practices that restrict an individual's movement, liberty and/or freedom to act independently without coercion or consequence.

Scoliosis

Scoliosis is a sideways curvature of the spine. It is highly prevalent in people with PWS, with affected individuals commonly requiring bracing and/or surgery. It can affect people of any age, from babies to adults, but most often starts in children aged 10 to 15.

Sensory Dysfunction.

Individuals with PWS have been described as having difficulty with regulation of sensory stimuli from the environment. It is suspected that this problem may be related to other symptoms of PWS such as poor satiety recognition, decreased sensitivity to pain, tendency to self-injure and sleep issues.

Sex Hormone Deficiencies

Sexual development in PWS is affected by the diminished or disrupted production of sex hormones. This condition is called hypogonadism (failure of the gonads to function properly – testes in men and ovaries in women.) In PWS, hypogonadism is present from birth and is thought to be caused by a combination of hypothalamic and primary gonadal deficiencies.

Sleep Apnoea

Sleep Apnoea is a condition where breathing is diminished or impaired during sleep. Individuals with PWS, including young children, are at risk for both central and obstructive sleep apnoea. Affected individuals commonly require continuous positive airway pressure (CPAP) therapy while sleeping at night.

Social Development

The gradual gaining of skills, relationships and attitudes that enables a person to interact in society.

Syndrome

A term used to describe a symptom complex characterized by many medical signs. In many syndromes, not all affected individuals will have every diagnostic sign.

Task-Switching

Task switching, or set-shifting, is an executive function that involves the ability to unconsciously shift attention between one task and another.

Uniparental Disomy

Uniparental disomy refers to the situation in which 2 copies of a chromosome come from the same parent, instead of 1 copy coming from the mother and 1 copy coming from the father. In about 25% of people with PWS, both chromosome 15s are maternal in origin, and no paternal chromosome 15 is present in the PWS person.