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

A collaboration between the Prader-Willi Syndrome Association Ireland (PWSAI) and Trinity College, Dublin.

Tallaght Hospital

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PRADER WILLI SYNDROME ASSOCIATION IRELAND

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

Research carried out in 2016.
Report published in 2017.
Full report available at: **www.pwsai.ie**

What is the PWSAI?

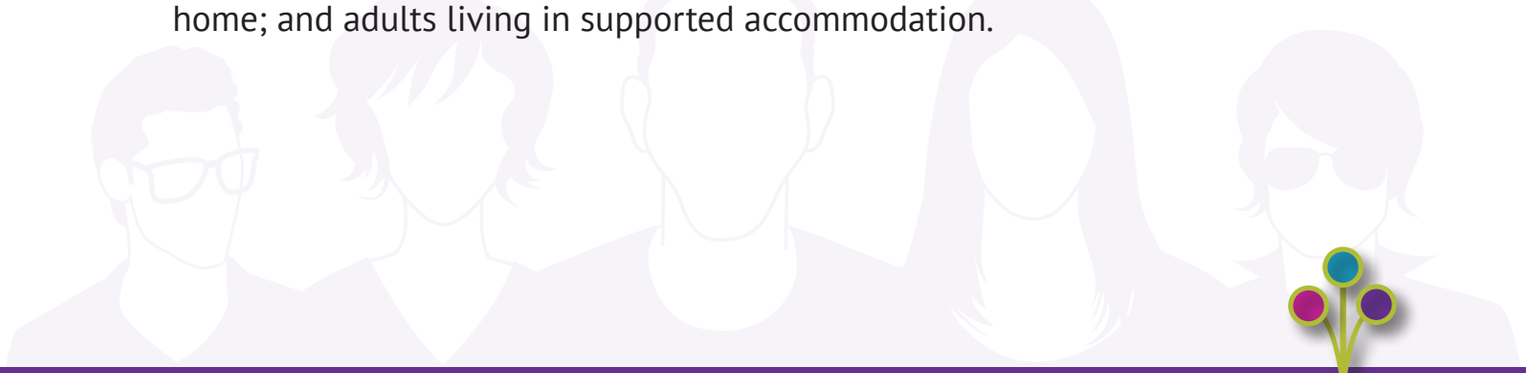
PWSAI is an organisation founded by parents in the late 1980's as a support group for individuals with PWS and their families in Ireland.

PWSAI strives to raise awareness and understanding of Prader-Willi syndrome (PWS) and to enhance the choice and quality of care, education and support for people with the condition and their families and carers. PWSAI also provides support for these people by providing services such as promoting awareness and information, including conferences and seminars, and promoting best practice standards of care for those with the syndrome. The Association also strives to enable earlier diagnosis and encourage and support research into PWS.
See https://pwsai.ie/.

About the National Survey

The PWSAI National Survey was designed and compiled to assess the needs of people with PWS and their families in Ireland.

The results are based on a survey that was designed by experts in the field and those who care for people with PWS. It included input from 61 primary carers throughout Ireland and represents approximately 60% of all people living with PWS in Ireland today. People with PWS included in the Survey are divided into five groups: 0-4 years; 5-10 years; 13-17 years; adults living at home; and adults living in supported accommodation.



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Executive Summary

- People with PWS struggle with an insatiable appetite (hyperphagia) which, if left uncontrolled, can lead to extreme obesity. This poses a significant threat to the health of people with the syndrome and places their wellbeing in jeopardy from a number of conditions.
- PWS affects approximately 350,000-400,000 people worldwide.
- PWS is a multisystemic random genetic disorder of chromosome 15.
- The only centre in Ireland that tests for PWS is the National Centre for Medical Genetics, which diagnoses 5-6 new cases of PWS each year, but this may be an underestimation of the true figure.
- There is currently no cure for PWS and because of the complexity of the condition care is required by a range of medical specialists from different disciplines, depending on age. Among infants the top 5 specialists attended are paediatric endocrinologists, paediatricians, dentists, respiratory specialists and ophthalmologists. In the 5-12 years age group, this also includes the need for gastroenterologists, orthopaedic surgeons and psychiatrists.
- PWS places a tremendous strain on the families of those with the syndrome as people with PWS often require round-the-clock supervision and care.
- Approximately 90% of Irish children with PWS have received growth hormone replacement therapy in the form of daily injections.
- In the adult population with PWS, 93% require help with managing their healthcare needs, such as taking medication as prescribed and 88% need support managing money.
- Overnight admissions to hospital were common across all age groups in the PWS population. For example, 40% in the 0-4 age group and 80% of 13-17 age group has had a hospital admission.
- In the 13-17 years age group, 40% of those with PWS attended a psychiatrist. Among adults with PWS, 44% living at home attended a psychiatrist, however this rose to 78% among those living in supported accommodation.

- Respondents also reported delay in pubertal development among 40 per cent of people with PWS.
- The survey reveals a delay in receiving growth hormone treatment across all age groups of individuals with PWS.
- Hyperphagia is one of the greatest challenges for those with PWS and the people who care for them. This was reported in 80% of people over 5 years old.
- Among those with PWS living at home, carers reported an increased financial burden associated with orthotics, specialised prams, travelling to appointments, home modifications, specialist equipment and medications, among others. This was described as 'an extreme negative impact' in 10% among the 0-4 years age group, 13% in those aged 5-12 years, 20% in the 13-27 years age group, and 29% in adults with the condition.
- Caring for a person with PWS strongly affects the carer's participation in the workforce. In 70% of households caring for a person with PWS aged 13-17, one partner had given up paid employment and in the remaining 30%, the main carer had reduced his working hours.
- 50% of those with PWS aged 5-17 years has no access to a medical card.
- Aside from the practical burdens of caring for someone with PWS, the emotional and day-to-day stress is considerable. The emotional burden was reported as 'significant' or 'extreme' in terms of the negative impact on quality of life among 70-80% of respondents.
- There was also a 'significant' or 'extreme' negative impact on family relationships among those caring for people with PWS, particularly among those caring for the 13-17 years age groups, at 60%. This was 44% among those caring for children aged 8-12 years.
- In siblings of the 13-17 years age group, 50% of parents reported that having a sibling with PWS negatively impacted the sibling's social life in a 'significant' or 'extreme' way. A negative impact on mental health for this group of siblings was reported in 50% of families, with 26% reporting a significant or extreme impact.

Summary of main findings

Early life and development

- There is an increasing awareness of PWS. Children born with PWS in the years 2012-2016 received a diagnosis within four weeks, on average. This compares to an average of six weeks for those born between 2005-2011 and 19 weeks for those born with PWS between 1999-2003.
- The most common medical conditions affecting infants with PWS are low muscle strength and tone (hypotonia) (90%) and sleeping disorders (80%).
- 87% of infants born with PWS were admitted to an intensive care unit.
- 50% of infants required feeding via nasogastric tube.

Physical health and wellbeing

- In the 5-12 year-old group, children with PWS also have to deal with scoliosis (50%); pronated or 'flat' feet (44%); visual difficulties (50%); and sensory difficulties (43.8%). In the 13-17 years age group, sleep disorders (90%) and mental health difficulties (23%) were added to these health challenges.

Physical health and wellbeing

- People with PWS have an increased risk of mental health problems, including anxiety disorders, depression, bipolar illness and psychosis.
- 60% of adults with PWS were reported to have been diagnosed with a mental health difficulty.
- In terms of managing emotions and feelings in those with PWS, carers reported that this was most challenging in the 5-12 and 13-17 years age groups.
- Carers reported significant difficulties in dealing with a range of challenging behaviours. Most common were skin picking (58%), repetitive questioning (63%), obsessions/compulsions (53%) and non-compliance (48%).

Education and employment

- Of the 43 individuals with PWS who had undergone IQ testing, 65% were deemed to have a mild learning disability, followed by 26% with a moderate learning disability.
- Only one adult with PWS is currently in paid employment.
- 63% of school-age children aged 5-12 years with PWS attend a mainstream educational setting. This drops to 30% for those aged 13-17 years.
- The majority of people with PWS in Ireland leave school with no qualifications.

Residential and respite needs

- Only 6% of those caring for children aged 5-12 years said they had access to respite care. This increased to 40% in adolescents but decreased to 25% for adults living at home.
- 70% of families with teenagers and 75% of families with adults reported a need for overnight respite care.
- More than 40% of adults with PWS require assistance with everyday tasks such as showering, brushing their teeth and using the toilet. This dependence increases in adulthood and the teenage years.
- 16 of the people with PWS surveyed will need supported accommodation within the next 5 years (2021).

Impact on families

- There is a significant negative impact on those who care for people with PWS in terms of financial burden and employment status.
- There is a considerable emotional and physical burden on those who care for people with PWS.
- There is a significant negative impact on the mental health and social life of the siblings of people with PWS.
- The emotional and psychological burden of caring for a person with PWS has a significant negative impact on family relationships.

Key recommendations

- Those with PWS and their carers need not only support from medical professionals, but also people with expertise in dietary support and psychological wellbeing to help prevent obesity and the health risks that go along with it.
- More resources and teams with a range of expertise are necessary to get timely access to medical care to prevent avoidable disability.
- There is an urgent need to set up a number of mental health teams for adults and children/adolescents with expertise in rare disorders, including PWS.
- Specialist medical, psychiatry and multidisciplinary supports should be provided to community teams so services can be delivered locally.
- There is a pressing need for more PWS specific residential accommodation that can provide appropriate environmental controls in a caring, family-like environment.
- Appropriate PWS specific respite care is urgently required to support parents and relatives in their role as caregivers. Respite is an important part of management to help support the person with PWS to live at home as long as they can and to prevent carer burnout.
- 'Most importantly, people with PWS are individuals with their own set of needs, interests, likes and dislikes. Therefore, it is essential that services are person-centered and take account of the perspectives of people with PWS. This calls in the future for greater facilitation of the involvement of people with PWS in service planning.'

For more information, resources and supports for people with PWS and their carers and families, please visit: **www.pwsai.ie**

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