Information for Social Workers and Care Managers

Prader-Willi Syndrome (PWS) is a rare lifelong genetic condition which is present from birth. Caused by an abnormality on chromosome 15, it affects the functioning of the hypothalamus. Main characteristics of the syndrome are:

- Hyperphagia (excessive appetite, overeating) – beginning around 2 -4 years
- Hypotonia (low muscle tone) – from birth
- Hypogonadism (immature sexual development)– from birth
- Short stature, compared with other family members
- Immature emotional and social development
- Learning disabilities ranging from severe to borderline. A minority of people with PWS do not have a measured learning disability but will not function at the level of their measured IQ
- Challenging behaviour, ranging from mild to severe (regardless of whether or not the person has a learning disability). A significant number of individuals with PWS also have a diagnosis of Autistic Spectrum Disorder or Attention Deficit Disorder.

Treatment and management of PWS

There is currently no cure or appropriate medication to treat the excessive eating and food-seeking which is one of the major characteristics of the syndrome. The severity of this feature varies between individuals and may vary across time in any single individual. Low muscle tone, and an abnormal body composition in which there is more fat mass than lean muscle (even in individuals who are not overweight), means that a child or adult with PWS needs fewer calories than someone of the same age to maintain a healthy weight. Excess calorie intake can quickly result in significant and life-threatening obesity.

Currently the only way to prevent life-threatening obesity in PWS is to control access to food and support the person to keep to a lower calorie diet – not easy when the person is hungry most of the time. Deaths at a very early age from obesity-related complications are unfortunately not uncommon.

Find out more about weight management in PWS

https://www.pwsa.co.uk/information-for-families/dietary-management-and-exercise

Low muscle tone can be partly helped by growth hormone treatment, which is licensed by NICE for children with PWS. Children on growth hormone treatment generally grow taller than they might otherwise have done and have improved muscle tone.

Sex hormone treatment may be given to teenagers and adults of both sexes to enable them to become more sexually developed, but fertility is extremely rare. Worldwide there are only four reported cases of women with PWS having a child (none in the UK) and none of a man with PWS fathering a child.

Challenging behaviour is common in both children and adults. Typical behaviours are:

- Temper outbursts (tantrums, rages), sometimes including aggressive verbal or physical assaults
- Food seeking and/or stealing
- Stubbornness and resistance to change, argumentativeness
- Perseveration (asking the same question, or reverting to the same subject over and over again
- Compulsive behaviour and insistence on routine
- Attention-seeking and manipulative behaviour
- Skin-picking
- Lying and blame-shifting

In teenagers and adults with PWS mental health problems may also emerge. The help of a CAMHS team, behaviour therapist, psychologist or psychiatrist should be engaged if these issues are evident.

Families

Studies carried out on the effect of PWS on families have revealed significant stress levels affecting all members of the family – even more than those which affect other families with a child or adult with special needs.

A USA study found that "Families/mothers/siblings with children with PWS showed poorer perceived quality of life compared with general inpatient and outpatient samples of children with complex health conditions. Families/mothers/siblings with children with PWS reported difficulties in family functioning, communication problems, and an increased number of conflicts. They appeared to be experiencing significant behavioural distress symptoms, with higher than average levels of depression and feelings of isolation, anger and worry. Ninety-two per cent of the siblings indicated moderate-to-severe symptoms of PTSD. Compared with normative populations, siblings demonstrated poorer quality of life, with mothers perceiving more significant deficits in the sibling."

For further information about working with families and the stresses placed on families by PWS, see also Social Work Interventions: Advocacy and Support for Families (From "Management of Prader-Willi Syndrome (3rd Edition) – Ed: Merlin G Butler, Phillip DK Lee, Barbara Y Whitman"). This is from the USA, but much of it is also relevant to UK families.

Adults with PWS

Some adults with PWS appear more able and articulate than is actually the case, and will tell outsiders that they know all about diet and are able control their own food intake – this is almost universally not the case. Unfortunately, because of this, some have been placed in supported living with minimal support hours, or even in an independent situation, with a subsequent life-threatening weight increase, and/or vulnerability to exploitation from members of the public.

Mental capacity around food and finances can be an issue, particularly where the person is generally high- functioning. Sometimes a Mental Capacity Assessment may be the only way to limit the person's access to food. Very rarely this has gone to the Court of Protection.

Unfortunately most of the case laws are not yet in the public domain, but in those unpublished cases which are known to the PWSA, it has been deemed that the person does NOT have capacity around food. The only one that has been published, which reports that the person DOES have capacity, can be seen at http://www.courtofprotectionhub.uk/cases/fx-2017-ewcop-36

Several organisations specialise in providing residential care and supported living for PWS, with particular emphasis on limiting access to food, calorie-controlled diets, and management of challenging behaviour. Funding is sometimes provided via joint health and social services funding and, occasionally, where other issues may be present, by health funding only, but generally funding is provided by social services. The PWSA UK maintains a list of specialist providers, but does not make recommendations – contact us for a copy of this list.

The PWSA UK publication *Beyond the Veneer: A Guide to the Essential Features of Residential Care and Supported Living for Adults with Prader-Willi Syndrome* is available to purchase and is a useful guide to what needs to be in place to effectively care for someone with PWS.

It is extremely rare for an adult with PWS to be in full time paid employment, even when they have the intellectual capacity to do a particular job. The tendencies to food-seeking and stealing, sleepiness, and rage outbursts may all affect their overall abilities. However, part-time paid work or voluntary work is quite possible, and helps the person's self esteem by providing them with a way of contributing to their community. Find out more in our Best Practice Guide for Employers of People with Prader-Willi syndrome

Unusual features and potential risk factors in PWS

Body temperature abnormalities
An individual with PWS may be unaware when they are too hot or too cold, or eat or drink very hot food or liquids. Hyper and hypothermia have been reported. Hyperthermia may occur during minor illness and after anaesthesia. Fever may be absent despite serious infection.

High pain threshold
The majority of people with PWS have a high pain threshold which may mask fractures and serious internal injuries. Individuals may have difficulties localising pain.

Lack of vomiting
Vomiting rarely occurs in those with PWS. Emetics may be ineffective. The presence of vomiting, particularly when the individual has a history of rarely vomiting, may signal a life-threatening illness.

Skin-picking
Many individuals with PWS may pick and small wounds or spots, intensifying and increasing the wound, and thus the potential for infection.

Easy bruising
Many individuals with PWS bruise easily and, because of their high pain threshold and/or cognitive problems, may be unable to say how they came by the bruise. A few individuals may falsely accuse others of assault (but in other cases the accusation may be true).

Full list of risk factors

Life expectancy and causes of death

Whilst good management has extended the life expectancy of people with PWS, it is still shorter than the general population. The oldest person we know of died at the age of 74, and the oldest we know who is currently still living is 64 – one of only a handful of people with PWS who are aged over 60.

Sadly, death can occur early in PWS, with people dying in their early 20s and 30s. Most common causes of death are:

- As a result of obesity (usually):
    - Heart disease
    - Diabetes complications
    - Breathing problems
- Septicaemia (cellulitis, skin-picking)
- Pneumonia (often sudden onset)
- Choking
- Stomach necrosis
- Cancer (relatively rare, although increasing as life expectancy increases)

A recent study in the USA found that 77.9% of deaths were unexpected, and 72.1% were sudden. This may relate to the high pain threshold common in PWS, so that the person does not experience or report severe symptoms until it is too late.

Medical alert in other languages

Important information about some of the more unusual features of PWS which medical personnel and others should be aware of, with translations into several languages

Further information

PWSA UK has a wide range of information and knowledgeable staff who can help with enquiries.

Training is also available to organisations working with people with PWS.

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