PWSA UK
PRADER-WILLI SYNDROME ASSOCIATION UK

Information for CQC and Commissioners of Care

Prader-Willi Syndrome (PWS) is a rare life-long 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 because of emotional and social immaturity
- 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

Prevalence and life expectancy

Estimates of prevalence range from 1:15,000 (USA estimate) to 1:52,000 (UK estimate)

Around 1200 people with PWS of all ages are known to the PWSA UK, but we estimate there are at least 2000 people in the UK with PWS. Of those known to us, approximately 50% are aged 18 or over. Although life expectancy is increasing due to better understanding and management and control of access to food, the PWSA UK knows of only about 6 people who are aged 60 or over.

Management of PWS – its impact on care

There is currently no cure or appropriate medication to treat the excessive eating 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 obesity. Early death in the person's teens, 20s or 30s is not uncommon. However, with restricted access to food, the person can live well into middle age and beyond. The oldest known person with PWS died at age 74 - she had lived in an institution for the greater part of her life.

Currently the only way to prevent life-threatening obesity in PWS is to control access to food and to keep the person on a lower calorie diet – not easy when the person is hungry most of the time.

Hence, people with Prader-Willi Syndrome (PWS) have a set of special needs which do not fit readily into the usual requirements for people with learning and/or physical disabilities. These needs constitute a duty of care and reflect the person's best interest.

Recommendations to accommodate these needs have been formulated across the PWS community worldwide, and operate regardless of usual social norms and practices in any country. These include:

- Restricted access to food - for some this may mean locked kitchens and food cupboards and supervision when out and about
- (Usually) A lower daily calorie intake than other people
- Emphasis on healthy foods - very few treats and snacks
- Structured meals and eating times
- Restricted locations as to where eating is allowed
- (Often) Restricted access to money
- Well-trained staff with a very good understanding of PWS and how to manage the challenging behaviour which often accompanies it (see below)

In addition, many people with PWS appear more able than they actually are. Some may test outside the learning disability intellectual spectrum but, because of immature emotional and social skills are unable to function in everyday life at their intellectual level. There are similarities with people with autism spectrum disorders.

It is very unusual for someone with PWS to live completely independently and, where this occurs, early death from life-threatening obesity or vulnerability to sexual and financial abuse are ever-present risks.

Supported living is an option for people with PWS, but only where there are adequate levels of care with staff trained in caring for people with PWS.

Deprivation of Liberty Safeguards (DoLS) are commonly used in caring for people with PWS.

Challenging behaviour

Challenging behaviour is common, regardless of the degree of learning disability. 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

One feature of this challenging behaviour is its unpredictability, with swift mood changes occurring. It is this unpredictability which makes it essential to have access to support to contain challenging behaviour available at all times.

Mental Capacity

Although a person with PWS may have the capacity to make decisions about many things, their capacity around food, money and health can questioned. Read more about Mental Capacity and PWS

Money

Many people with PWS are good with money and enjoy shopping. However, access to money without supervision or restrictions may not work in their best interests. Money is likely to be spent on food, hence increasing weight, as well as not being used to purchase other necessities or to pay bills.

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. Both children and adults 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

Organisations providing care for people with PWS

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. The PWSA UK maintains a list of these providers – contact us for details.

Information for social workers

Information for residential care and supported living staff

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