

Causes of Death in Prader-Willi syndrome: Prader-Willi Syndrome Association | USA 40-year mortality survey

Merlin G. Butler, Ann M. Manzardo, Janalee Heinemann, Carolyn Loker, and James Loker: *Genetics in Medicine* (2016).

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While we celebrate exciting new discoveries and therapies for PWS, we also continue to expand our understanding of the significant challenges. Knowledge is power – even when the topic is distressful and sad. This study was sponsored by PWSA | USA and is the largest published summary on mortality worldwide.

As a part of the bereavement support program, PWSA | USA has gathered information since 1973 on deaths as reported by families. This data was first summarized in 2005 in a paper written by the mortality committee. In 2016, a publication by Dr. Butler and colleagues extensively reviewed the data from 1973-2015, with a priority of describing more specifically the causes of death.

The article states that 486 families reported a death to PWSA | USA making this the largest study of death in Prader-Willi syndrome. These individuals were between the ages of 2 months and 67 years. 70% of these deaths occurred in adulthood. Of the 486, 312 had a cause of death provided and 36% died “of unknown cause.” Not all records provided enough detail; for example, listing “obesity” as a cause of death (7%).

Respiratory (breathing) failure was the most common cause of death (31%) in PWS in both adults and children. Heart issues were the second leading cause at 16%, primarily in adults due to obesity-related right heart failure rather than to coronary disease. These are not surprising findings when uncontrolled weight has long been the greatest challenge in the syndrome.

A surprising 10% of deaths were due to GI problems, and GI related death occurred at all ages. This finding is the reason we have become very vigilant about gastric problems – obstructions, motility, perforations. Prior to analyzing this information, physicians were unaware of how common or lethal gastric problems could be for persons with PWS. With this new knowledge, we have developed the GI Chart to provide to doctors and emergency rooms, included in the red Medical Alert handbook.

Choking was more common in males, in part due to rapid eating when sneaking food but is also complicated by lack of saliva and the ineffective swallowing we know is often part of the syndrome. Another surprising finding was the 7% incidence of pulmonary embolism. We are now paying more attention to the problems of blood clots and why those might be so common.

In looking at the trends over time, lifespan has increased through the decades for women, but curiously not so for the men. Men were more likely to have behaviors such as binge eating, leading to choking or stomach perforation. Accidental and/or hyperphagia-related deaths appear to disproportionately impact younger males, possibly owing to impulsive characteristics or higher activity levels.

Deaths in childhood are more likely due to respiratory failure, aspiration, infection and choking rather than obesity related factors. Cardiac disease, pulmonary embolism, accidents, sepsis, and obesity-related complications were primarily seen in adolescents and adults.

The article noted that early diagnosis and prevention of obesity are key factors in preventing premature death in PWS. These include close monitoring and supervision of food access and quantity of food to avoid choking and stomach rupture. Not included in the article was that 80% of the 147 individuals with known height and weight were obese at the time of death.

PWSA | USA will continue to collect this important information. We offer our thanks and deepest sympathy to the families who have suffered the loss of a loved one. Their courage in contacting us is deeply appreciated.

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