SAMPLE:

2nd Asia Pacific PWS Scientific Workshop &Conference 2012
Scientific Conference Abstract Submission

STOMACH NECROSIS: A LETHAL COMPLICATION OCCURRING IN SLIM INDIVIDUALS WITH PRADER-WILLI SYNDROME

Suzanne B. Cassidy, MD[1], Ann O. Scheimann, MD, MBA[2], Merlin G. Butler, MD,PhD[3], Moris Angulo, MD[4]; James Loker, MD[5], Patrick Kendell[6], Janalee Heinemann, MSW[7], Norma Rupe[7], David A. Stevenson, MD[6]

[1]Department of Pediatrics, Division of Medical Genetics, University of California, San Francisco, CA, USA; [2]Johns Hopkins Hospital, Baltimore, MD; [3]Section of Medical Genetics and Molecular Medicine, Children's Mercy Hospital, University of Missouri- Kansas City, MO, USA; [4]Department of Pediatrics, Winthrop-University Hospital, Mineola, NY, USA; [5]Bronson Methodist Hospital, Kalamazoo, MI, USA; [6]Department of Pediatrics, Division of Medical Genetics, University of Utah, Salt Lake City, USA; [7]Prader-Willi Syndrome Association (USA), Sarasota, FL, USA.

INTRODUCTION: People with Prader-Willi syndrome (PWS) have hyperphagia that leads to obesity if uncontrolled. Their well-documented ability to consume very large amounts of food without feeling discomfort is due, at least in part, to their characteristic high pain threshold and high vomiting threshold. Control of food intake combined with exercise can prevent or treat the obesity, and many affected individuals today are not obese. However, the hyperphagia cannot yet be controlled medically. We here report the occurrence of unexpected mortality in adults with PWS, most of whom are slim, who develop gastric dilatation and necrosis sometimes leading to rupture and death, often following an eating binge.
METHODS: In recent years there have been a number of efforts to identify the various causes of death in individuals with PWS. The Prader-Willi Syndrome Association (USA) is conducting a large-scale questionnaire survey of families.
RESULTS: Thus far, their database contains a total of 178 deaths, of which 6 individuals had suspected or documented gastric necrosis and rupture. Age range is 17-46 years. All were relatively slim. There were 4 males and 2 females. An additional 7 individuals had a suggestive clinical course but no confirmatory documentation. Typically, there was an episode of binging associated with a holiday or special event followed several hours to a day later by complaints of stomach pain and sometimes evident abdominal distention and/or vomiting. Since complaints were not strong, they were often ignored. Death was sudden with the cause being sepsis when documented.
DISCUSSION: Wharton et al. (1997) reported 4 adults (3 F, 1M) with PWS having acute gastric dilatation and necrosis, all of whom were slim. One died. Four others had dilatation without necrosis. In an international series of 27 people with PWS who died, Schrander-Stumpfel et al. (2004) reported two adult males (one obese) died with abdominal pain, one of whom had autopsy-proven gastric dilatation. Gastric dilatation and necrosis also occurs in people with anorexia nervosa following an eating binge. The pathophysiology is controversial, with both vascular occlusion and thinning of the mucosa in a shrunken stomach as proposed explanations. In individuals with PWS, abdominal complaints and dilatation should prompt rapid evaluation and consideration of possible gastric necrosis and rupture, especially following a binging episode in those who are slim or have lost weight over a long period, whether or not complaints seem adequately strong.
