Introductory comments—Special section: Prader–Willi syndrome

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Invited Comment

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The Prader–Willi syndrome (PWS) was originally described in nine patients by the Swiss group of physicians, Prader et al. [1956]. The paper was republished and translated in the recent book "Management of Prader-Willi Syndrome" by Butler et al. [2006], which is reviewed in this issue of the *Journal* [Thomson, 2007]. In the book, the authors include the photographs of these three original authors, a nice feature of the translation. On seeing the original now, the reader is struck by the brevity of such a seminal publication. After the original paper, there was little attention devoted to what came to be known as the Prader–Willi syndrome until the paper by Hall and Smith [1972]. Since that article, there has been increasing interest in the investigation of all aspects of this important syndrome. Remarkably, advances in our knowledge of PWS have paralleled the evolution of new concepts in the field of Medical Genetics: The recognition by Ledbetter et al. [1981] that a **microdeletion** was present in about 50% of patients with PWS (the original microdeletion syndrome); the discovery that PWS involved **imprinted genes** in the critical region; the recognition of the phenomenon of **trisomy rescue** related to the occurrence of uniparental disomy in a proportion of patients; the utilization of **DNA methylation studies** to make the diagnosis of PWS in the clinical setting; delineation of the genes in the **critical region** of 15q11–13; the improvements of outcome of obesity and linear growth in children with PWS treated with growth hormone; the development of **guidelines for health supervision** and anticipatory guidance; the use of **multidisciplinary teams** in the comprehensive care of patients with PWS [Cassidy and Allanson, 2005; Butler et al., 2006]; the establishment and growth of international **patient advocacy** and genetic support groups for PWS [www.pwsausa.org, 2007]; and the **collaboration** of families, healthcare professionals, and scientists in advancing knowledge of the condition [Cassidy and Allanson, 2005; Butler et al., 2006]. Given the national focus on the "obesity epidemic" in the U.S., interest in PWS has also occurred because of the potential of being able to generalize knowledge from the uncommon syndrome to the common population problem.

This issue of the *Journal* includes a Special Section: The Prader–Willi Syndrome. The series is led by an important paper by Butler and Bittel [2007], and comprises 11 other papers, including Scientific Abstracts presented at the International Meeting in 2005, and a book review on the Third Edition of the 'Management of Prader-Willi Syndrome' [Thomson, 2007].

The advances in our knowledge of PWS presented here, along with the advances of the last 25 years of basic science and clinical research, have radically changed the manner in which we approach the biology, cause, diagnosis, and management of PWS. Ultimately, this knowledge improves the care of our patients.

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Prader-Willi syndrome (PWS) is a highly variable genetic disorder affecting multiple body systems whose most consistent major manifestations include hypotonia with poor suck and poor weight gain in infancy; mild mental retardation, hypogonadism, growth hormone insufficiency causing short stature for the family, early childhood-onset hyperphagia and obesity, characteristic appearance, and behavioral and sometimes psychiatric disturbance.Â Prader-Willi syndrome (PWS; OMIM #176270) is a complex neurogenetic disorder that affects 1 in 15,000 children, with 400,000 cases diagnosed globally (1). PWS is a contiguous gene disorder caused by paternal loss of the maternally imprinted Prader-Willi syndrome (PWS) is a genetic disorder that occurs in approximately one out of every 15,000 births. PWS affects all sexes with equal frequency and affects all races and ethnicities. PWS is recognized as the most common genetic cause of life-threatening childhood obesity. What causes Prader-Willi syndrome? PWS is the result of an abnormality on chromosome 15. There are three ways that this can happen. Prader-Willi Syndrome. NORD is very grateful to Merlin G. Butler, MD, PhD, Director, Division of Research and Genetics, Director, KUMC Genetics Clinic and Professor of Psychiatry & Behavioral Sciences and Pediatrics, University of Kansas Medical Center, for assistance in the preparation of this report. Synonyms of Prader-Willi Syndrome. Prader-Labhart-Willi syndrome. PWS. Willi-Prader syndrome. General Discussion. Summary. Prader-Willi syndrome (PWS) is a genetic multisystem disorder characterized during infancy by lethargy, diminished muscle tone (hypotonia), a week suck and feeding difficult Prader-Willi syndrome is a rare genetic disorder that results in physical, mental and behavioral problems, including a constant sense of hunger.Â Other features of Prader-Willi syndrome appear during early childhood and remain throughout life, requiring careful management. These features may include: Food craving and weight gain. A classic sign of Prader-Willi syndrome is a constant craving for food, resulting in rapid weight gain, starting around age 2 years. Constant hunger leads to eating often and consuming large portions. Unusual food-seeking behaviors, such as hoarding food, or eating frozen food or even garbage, may develop.