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GROWTH HORMONE AND MORTALITY IN PRADER-WILLI SYNDROME

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INTRODUCTION

Prader-Willi syndrome (PWS) is a unique condition associated with lack of normal expression of paternal alleles in a highly imprinted region of chromosome 15q11-13. Involvement of regions encoding small nucleolar RNA (snoRNA) clusters HBII52 and, perhaps more crucially, HBII85, have been identified as particularly associated with phenotype expression.^{1,2} Minimum birth incidence has been recently estimated at ~1 in 20,000 to 30,000 and population prevalence at ~1 in 50,000 to 80,000.³⁻⁵ Fewer than 1% of cases are due to inherited mutations.

Affected individuals suffer from excessive body fat independent of weight, marked deficits in muscle mass and function, growth failure with adult short stature, osteoporosis, scoliosis, hypogonadism, acromicria, neurodevelopmental delay, hyperphagia, and cognitive defects.⁶ A variable deficiency of induced growth hormone (GH) secretion and more consistently-observed low insulin-like growth factor (IGF)-I levels are characteristic and may play a role in pathophysiology.⁷

In 2000, after nearly 15 years of favorable clinical experience, recombinant human (rh)GH became the first and, to-date, only pharmaceutical agent specifically approved for treatment of PWS. The FDA labeling states: "...for long-term treatment of pediatric patients who have growth failure due to PWS" and European

From The Editor's Desk

Yes, *GGH* has a new look to acknowledge a new era with our new sponsor, INSMED. Also, with this change we welcome 2 new distinguished colleagues to the editorial board, Dr. Roberto Lanes from Caracas and Dr. Martin Savage from London. Biographical sketches highlighting their wonderful credentials are available on the journal's website. The above mentioned changes have brought about a renewal of the journal's scope and mission that we hope will be appreciated and relished by the readers.

In this issue the lead article deals with an important current dilemma—the treatment of individuals with PWS with rhGH. Dr. Phil Lee was invited to review the mortality risks to these patients and his paper clearly presents the current status and issues. Although the data are not sufficient to fully determine patients' risks, with or without this therapy, the review was necessary. The lead article facilitates an understanding of the facts as they now stand and therefore it aids in formulating the clinical choices that need to be made when treating PWS. There clearly is a need to gather additional scientific information for a precise risk analysis in PWS that will lead to well substantiated recommendations.

The abstracts and editorial comments are also very timely, all dealing with a variety of subjects that affect patients and help elucidate important pathophysiological mechanisms ie, growth in Noonan syndrome and the role of Obestatin, a new hormone that opposes Ghrelin, among other important contributions. Altogether this issue constitutes another very successful issue of *GGH*; enjoy it and thank our new sponsor for their commitment to continuous medical education.

Sincerely,
Fima Lifshitz, MD
Editor-in-Chief