Growth hormone treatment in adults with PraderWilli syndrome: an update Graziano Grugni & Alessandro Sartorio.

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Introduction: Prader-Willi syndrome (PWS) is a rare disorder caused by the lack of expression of paternal genes on chromosome 15q11.2-q13. The clinical picture of PWS is characterized by neonatal hypotonia, hyperphagia, obesity, altered body composition, cognitive impairment, behavioral disturbances, short stature, and multiple endocrinopathies, including growth hormone (GH)/IGF-I axis dysfunction.

Areas covered: this narrative review addresses the current state-of-the-art of recombinant human GH therapy (rhGHT) in adults with PWS, focusing on its effects on body composition, muscle strength and exercise capacity, cardiovascular and respiratory function, endocrine and metabolic parameters, bone health, and psychological aspects.

Expert opinion: available data demonstrated the positive effects of rhGHT on the body composition of GH-treated subjects. This observation is significant, as improving body composition has been shown to increase muscle strength and exercise tolerance. Overall, rhGHT appears to improve both cardiorespiratory function and psychological outcomes. However, most of the studies are uncontrolled and shortterm. Therefore, longitudinal trials evaluating the long-term effects of rhGHT are recommended to confirm these findings. Since the beneficial effects of rhGHT appear to be independent of the presence of GH deficiency, we believe that its approval should be considered in adults with genetically confirmed PWS without testing for GH secretion.