Understanding the burden of endocrine and metabolic disorders in Prader-Willi syndrome: data from the Italian registry.

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Purpose: this study is a cross-sectional analysis of data from the Italian registry for patients with PWS, aimed at assessing the prevalence of endocrine and metabolic abnormalities in 539 patients with Prader-Willi syndrome (PWS), aged 0.0 to 61.6 years. Demographic and genetic data were also analyzed.

Methods: patients were recruited from 18 PWS referral centers participating in the Italian PWS registry. Each subject underwent comprehensive health screening as part of routine clinical care.

Results: the analysis of database revealed: (1) a reduction of obesity prevalence compared to previous data in the Italian population of PWS (51.0% versus 62.6%); (2) a high frequency of endocrine disorders (hypogonadism 53.5%, hypothyroid ism 16.1%, central adrenal insufficiency 5.9%, precocious puberty 5.6%); (3) 337 individuals were undergoing GH therapy (62.5%), including 286 < 18 years (91.4%) and 51 > 18 years (22.6%); (4) a high prevalence of altered glucose metabolism (27.4%), dyslipidemia (29.7%), hyperuricemia (10.4%), hypovitaminosis D (50.2%), osteoporosis (6.9%), metabolic dysfunction-associated steatotic liver disease (33.3%) and cholelithiasis (13.3%); (5) endocrine and metabolic abnormalities were more frequent among patients > 18 years; (6) patients > 18 years had a higher rate of paternal 15q11.2-q13 deletions and a lower rate of maternal uniparental disomy for chromosome 15 than younger individuals (61.1% versus 40.9% and 31.0% versus 46.0%, respectively; $p < 0.001$); (7) the median age at genetic diagnosis was lower in the younger group compared to those > 18 years (0.1 years versus 3.7 years; $p < 0.001$).

Conclusion: our findings confirm the high prevalence of endocrine and metabolic comorbidities in Italian subjects with PWS.