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Nota di contenuto	About the Editors -- Special Issue: Genetics of Prader-Willi Syndrome -- Single-Case Study of Appetite Control in Prader-Willi Syndrome, Over 12-Years by the Indian Extract Caralluma fimbriata -- Venous Thromboembolism in Prader-Willi Syndrome: A Questionnaire Survey -- The Global Prader-Willi Syndrome Registry: Development, Launch, and Early Demographics -- Early Diagnosis in Prader-Willi Syndrome Reduces Obesity and Associated Co-Morbidities -- Defining Mental and Behavioural Disorders in Genetically Determined Neurodevelopmental Syndromes with Particular Reference to Prader-Willi Syndrome -- Age Distribution, Comorbidities and Risk Factors for Thrombosis in Prader-Willi Syndrome -- Prader-Willi-Like Phenotype Caused by an Atypical 15q11.2 Microdeletion -- Food and Non-Food-Related Behavior across Settings in Children with Prader-Willi Syndrome -- Clinical Observations and Treatment Approaches for Scoliosis in Prader-Willi Syndrome -- The Potential Role of Activating the ATP-Sensitive Potassium Channel in the Treatment of Hyperphagic Obesity -- Growth Trajectories in Genetic Subtypes of Prader-Willi Syndrome -- The Gut Microbiota Profile in Children with Prader-Willi Syndrome -- A 24-Week Physical Activity Intervention Increases Bone Mineral Content without Changes in Bone Markers in Youth with PWS -- Genetic Subtype-Phenotype Analysis of Growth Hormone Treatment on Psychiatric Behavior in Prader-Willi Syndrome -- Pharmacogenetic Testing of Cytochrome P450 Drug Metabolizing Enzymes in a Case Series of Patients with Prader-Willi Syndrome.

Sommario/riassunto

Prader-Willi syndrome (PWS) is a complex genomic imprinting disorder associated with a spectrum of medical, cognitive, behavioural, and psychiatric problems and is also the most common cause of life-threatening obesity that can be effectively treated with hormone therapy and restricted diet, if detected early. PWS is usually caused by the loss of the paternally inherited 15q11.2-q13 region and abnormal expression of genes within that region and beyond. While some genotype-phenotype correlations with delineation of clinical characteristics and natural history have emerged when comparing the three main molecular classes of PWS (maternal uniparental disomy (UPD) 15, imprinting centre defect, and deletion of paternal 15q11-q13), better awareness and informative biomarkers are still needed. These could facilitate early diagnosis, counseling, prognostic testing, as well as patient stratification for clinical trials, to improve outcomes for the affected children and their families. This Special Issue will comprise reviews and original research articles focused on the recent advances of genetics/genomics, testing, and epigenetic processes along with clinical description, co-morbidities, and natural history of PWS. Current and future directions with focus on improved screening, diagnosis, and treatment will be addressed in this rare neurodevelopmental genetic imprinting disorder influenced by the PWS genetic subtypes.
