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Titolo	Rare Diseases : Integrative PPPM Approach as the Medicine of the Future // edited by Meral Özgüç
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Collana	Advances in Predictive, Preventive and Personalised Medicine, , 2211-3509 ; ; 6
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Soggetti	Biology - Technique Cytology Pediatrics Diseases Biological Techniques Cell Biology
Lingua di pubblicazione	Inglese
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Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	National Plans on rare diseases -- Biobanking for rare diseases -- Impact on personalized medicine -- Emerging Technologies for Gene Identification in Rare Diseases,- Personalized Medicine for Hereditary Deafness -- Mitochondrial Diseases -- Enzyme Replacement Therapy in Lysosomal Storage Diseases -- Complexity of Genotype-Phenotype Correlations in Mendelian Disorders: Lessons from Gaucher Disease -- Rare Cancers -- Adeno-Associated Virus Gene Therapy and its Application to the Prevention and Personalized Treatment of Rare Diseases -- Induced pluripotency for the study of disease mechanisms and cell therapy.
Sommario/riassunto	This book focuses on predictive, preventative and personalised medicine (PPPM) and how it is related to the healthcare of rare diseases. Readers will discover how advanced rare diseases healthcare provides an excellent "proof-of-principles" for the personalisation of healthcare

systems on a global scale. Chapters look at national plans for rare disease, at biobanking, gene identification, rare cancers, virus gene therapy, induced pluripotency for cell therapy amongst other topics. There is a chapter dedicated to personalised medicine for hereditary deafness, and another exploring the complexity of genotype-phenotype correlations. Specific diseases such as Fabry's, Gauchers and mitochondrial cytopathies are highlighted and we look at enzyme replacement therapy in lysosomal storage diseases. This work is part of a series, produced with the involvement of the European Association for Predictive, Preventive and Personalised Medicine. The series focusses on the concept of an integrative medical approach by PPPM. This volume is dedicated to all aspects related to the prediction, prevention and personalised treatments of rare diseases, and in doing so it explores developments relevant to all medical branches. The authors cover ethical considerations, the creation of a robust platform for professional communication, synergies with patient organisations, "doctor-patient" collaboration and a new philosophy of integrative medicine by PPPM. This volume serves as a reference source for scientific and medical centres in the field and can be used both at medical curricula and graduate level in the life sciences. Those who place a special emphasis on healthcare promotion and innovations intended to combat rare diseases, save the affected lives and enhance life quality will all find this book of great value.

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