Record Nr. UNIORUON00439418

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Titolo Vijayanagara / Burton Stein

Pubbl/distr/stampa Cambridge,: Cambridge University Press, 2005

ISBN 05-216-1925-4

Descrizione fisica XIX, 156 p : ill. ; 24 cm

Classificazione SI IV B

Soggetti INDIA - STORIA - PERIODO VIJAYANAGARA

Lingua di pubblicazione Inglese

Formato Materiale a stampa

Livello bibliografico Monografia

Record Nr. UNINA9910557223103321

Autore Gouw Arvin M

Titolo Applying Next Generation Sequencing and Transgenic Models to Rare

Disease Research

Pubbl/distr/stampa Frontiers Media SA, 2020

Descrizione fisica 1 online resource (119 p.)

Soggetti Medical genetics

Science: general issues

Lingua di pubblicazione Inglese

Formato Materiale a stampa

Livello bibliografico Monografia

Sommario/riassunto A rare disease is a disease that occurs infrequently in the general

population, typically affecting fewer than 200,000 Americans at any given time. More than 30 million people in the United States of America

(USA) and 350 million people globally suffer from rare diseases. Out of the 7000+ known rare diseases, less than 5% have approved treatments. Rare diseases are frequently chronic, progressive, degenerative, and life-threatening, compromising the lives of patients by loss of autonomy. In the USA, it can take years for a rare disease patient to receive a correct diagnosis. The socioeconomic burden for rare disease is huge. For those living with diagnosed rare diseases, there is no support system or resource bank for navigating financial, educational, or other aspects of having a rare disease. The purpose of this Research Topic is to bring together leading researchers, non-profit organizations, healthcare providers/diagnostic companies, and pharma/biotech/CROs in the field to provide a broad perspective on the latest advances, challenges, and opportunities in rare disease research.