Commentary

iMedPub Journals
www.imedpub.com

Journal of Rare Disorders: Diagnosis & Therapy
ISSN 2380-7245

2022
Vol.8 No.11:81

# Challenge of Current Biomedicine Is To Identify Curative Therapies for Every Disease in a Personalized Way

**Darby Schmidt***

Department of Science and Technology Studies, Faculty of Sciences York University, Toronto Ontario, Canada

***Corresponding author:** Darby Schmidt, Department of Science and Technology Studies, Faculty of Sciences York University, Toronto Ontario, Canada, E-mail: schmidtdar44@gmail.com

**Received date:** October 12, 2022, Manuscript No. IPRDDT-22-15134; **Editor assigned date:** October 14, 2022, PreQC No. IPRDDT -22-15134 (PQ); **Reviewed date:** October 24, 2022, QC No. IPRDDT -22-15134; **Revised date:** November 02, 2022, Manuscript No. IPRDDT -22-15134 (R); **Published date:** November 11, 2022, DOI: 10.36648/2380-7245.8.11.81

**Citation:** Schmidt D (2022) Challenge of Current Biomedicine Is To Identify Curative Therapies for Every Disease in a Personalized Way. J Rare Disord Diagn Ther Vol.8 No.11:81

## Description

Case studies in rare disease small molecule drug discovery are covered in this overview, with a focus on the use of new technologies and novel target approaches. Examples of case studies include repurposing, inducement of alternative splicing, stop codon read through, allosteric activation, and covalent modification. Biomarkers, inducible pluripotent stem cells, and rare disease animal models are all highlighted in this review. Scenario studies can be used as a tool for policy exploration in situations where futures are uncertain and technological developments stand to have significant effects on healthcare systems. We show how to adapt firm-based and policy-oriented scenarios for health policy and explain the significant differences between the two. There are five stages to our method. Focuses and potential obstacles are identified through in-depth interviews with a wide range of stakeholders. A set of scenarios is created by analyzing the interview data to determine the most important factors that will cause problems in the future. In a focus group with relevant health policymakers and advisors, the scenarios are used. Our scenario study on coverage decisions for drugs for rare diseases in Canada serves as an illustration of this strategy. The scenario method's extension and application, as well as our own experiences with it, are the primary focuses of this section. We show how scenarios can be used to find targets for robust health policy strategies that will be useful no matter what happens in the future.

## Genome Sequencing

According to the results of our post-focus group survey, the vast majority of participants found the scenario focus group to be useful and was able to identify additional obstacles related to drugs for rare diseases. From a health sector perspective, to estimate the cost-effectiveness of Genome Sequencing (GS) for diagnosing suspected rare genetic diseases in critically ill infants and non-critically ill children. Although Complex Genomic Rearrangements (CGRs) are known to cause disease, routine genetic screening frequently overlooks them. In order to identify CGRs, it is necessary to (i) simultaneously identify copy number variants (CNVs), (ii) Phase multiple cis breakpoint junctions, and (iii) find and resolve structural variants (SVs) within repeats. We demonstrate how new sequencing techniques and cytogenetics can be used to gain insight into CGRs' genomic architecture. We also look at the molecular characteristics and CGR patterns that have been discovered by studying constitutional genomic disorders. People who are interested in studying CGRs, determining their clinical relevance and frequency, and determining their impact on rare genetic diseases can learn a lot from these data. A significant number of people with Rare Diseases (RDs) have experienced severe mental health issues. However, there is insufficient focus on the psychological Quality of Life (QoL) of RD patients. While there is evidence to support the benefits of social support and social activity on recipients' mental health and overall Quality of Life (QoL), no comparable study has examined the impact of both on RDs' psychological QoL. The purpose of this study is to examine the relationship between psychological QoL and social activity among people with RDs and social support. The most difficult part of modern biomedicine is figuring out individualized treatments for every disease so that everyone can benefit from them.

However, in order to achieve this, we need to fully comprehend the disease mechanisms that will guide the development of novel treatments and novel approaches. The constraints are greater for Rare Diseases (RDs), which collectively affect 300 million people—roughly 10% of the world's population—but only a small number of individuals (less than one in every 2,000).This is due to:1) RD physiopathology is poorly understood;2) Clinical trials are severely limited by the low number of participants;3) Pharma has little commercial interest;4) When certain drugs are made available, not everyone who needs them can afford them because of their high cost. This article discusses orphan drug designation, drug repurposing, break-down into theratypes (as is currently the case for cystic fibrosis), and novel precision-medicine-based approaches as potential solutions to these obstacles. Research groups must put in a lot of effort in order to develop decision models that can evaluate interventions for rare diseases. This is especially true when it comes to gathering and synthesizing information in order to parameterize the model. The knowledge gathered in an ontology can be reused in this article to automatically generate decision tree models for various interventions and contexts. An unusual clinical manifestation of gestational choriocarcinoma is

subarachnoid hemorrhage, a potentially fatal medical emergency. We report a rare case in which a 31-year-old primigravid woman presented five weeks after giving birth with a cerebral oncotic aneurysm that had ruptured. Despite the absence of neurological deficits following endovascular embolization, the patient was quickly readmitted with symptoms of puerperal endometritis, including fever, abdominal pain, and fetid lochia.

## Single-Arm Treatment Trials

She was later given the diagnosis of metastatic choriocarcinoma after undergoing a thorough diagnostic investigation. Despite being in septic shock due to Escherichia coli bacteremia, the prognosis was favorable when multiagent chemotherapy was started early. The educational services, teachers, and other school staff face difficulties when educating children with rare diseases. Based on information provided by families, educators, and children with rare diseases, this study sought to identify these obstacles. The social-critical paradigm was used as a theoretical framework for a qualitative study. There were 43 participants in this study. Information was gotten through top to bottom meetings and center gatherings and investigated with talk examination. A total of ten categories were identified that influence an educational environment that is healthy, inclusive, and equitable: Coordination, diagnosis, official recognition, accessibility, absences, homework, autonomy, personnel resources, and peer support. The individual child's health condition, the level of family empowerment, the availability of resources, and the commitment of the school and health care providers all play a role in the process of providing appropriate education and care. One of the obstacles to obtaining the necessary adaptations is either the delay or the absence of a precise diagnosis.

There are many obstacles to overcome when conducting clinical research on patients with rare diseases. In addition, rare diseases represent a significant global burden of morbidity and a significant area of unmet medical need. The gold-standard randomized controlled trial design is frequently unsuitable for these small, frequently geographically dispersed populations, which is one of the most common challenges in clinical trial design for rare disease populations. Therefore, real-world data are especially important in the context of rare diseases because they can be used as a comparator in single-arm treatment trials and to support drug submissions to regulatory agencies. We present a recent case study of the successful use of external controls in the Neurofibromatosis type 1 (NF1) population and discuss the potential benefits and drawbacks of external controls for the regulatory approval of drugs for rare diseases. Although caregivers of rare cancers have recognized the significance of gaining benefit and meaning, this has not yet been investigated. We wanted to identify factors associated with finding benefit and meaning-making in providing care for patients with rare cancers and to characterize the unmet needs and experiences of caregivers of patients with Erdheim-Chester Disease (ECD) and other Histiocytic Neoplasms (HN).