[BIOINFO 2023]

Advancements and prospects of PubCaseFinder, a generic name for three services related to rare and genetic diseases

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Recent advances in the field of life sciences have led to the widespread adoption of genome analysis, resulting in the rapid accumulation of genomic and clinical data obtained from daily clinical practice (i.e., real-world data). This rich data is highly useful for a variety of purposes, including personalized medicine and clinical research in the field of rare and genetic diseases. Because of over 10,000 different types of rare and genetic diseases, each with a small number of patients, physicians often struggle to gain sufficient expertise. As a result, the diagnosis and treatment of these diseases are often challenging. To address this challenge, we have developed and have been operating three services: DiseaseSearch (https://pubcasefinder.dbcls.jp), CaseSharing (https://pubcasefinder.dbcls.jp/casesharing), and PanelSearch (https://pubcasefinder.dbcls.jp/panelsearch). These services related to rare and genetic diseases are collectively called PubCaseFinder. DiseaseSearch has been in operation for seven years as a service to help physicians make clinical diagnoses and has now been widely used by many hospitals and universities both domestically and internationally. However, it currently relies solely on public databases and cannot incorporate real-world data. To collect real-world data, we have developed CaseSharing, a case information management service, and PanelSearch, a service that provides curated gene panels to facilitate the diagnosis of rare and genetic diseases. CaseSharing can be used without user registration, and the case information collected by the user is saved in the user's local environment. We plan to develop a centralized database to collect CaseSharing data in the future. Panel Search includes 9,998 gene panels constructed by automatically collecting disease-causing genes for each disease. We are developing a function that allows clinical professionals to curate these automatically constructed gene panels. We believe that by seamlessly combining these three services, we can provide more accurate clinical decision support. In this presentation, we will provide an in-depth overview of each service, address current challenges, and outline our future direction in this field of explosive data growth.