Rare Disease databases- a comprehensive review

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ABSTRACT

Rare diseases are those diseases which occur in a small number of populations. Orphan drugs are given to the patients suffering from rare diseases. Pharmaceutical companies will not get profit from the manufacturing of orphan drugs. But the manufacturing of orphan drugs is important for the patients suffering from the rare diseases. In this work, the author has focused on the databases that are useful for the rare disease analysis.

KEYWORDS

Rare diseases, orphan drugs, database, CADD.

INTRODUCTION

Biological databases play an important role in discovering the lead molecule [1]. The databases can provide information regarding the gene information, total number of amino acids in a presents, pathways to which drug responds, protein drug interactions, posttranslational modifications and genome and proteome of the pathogenic sources [2,3]. They also provide the information regarding the numbers of entries present in the database. Advanced search provides the exact information when a specific keyword is typed [4]. It excludes raw as well as non redundant data. Moreover, it can also provide the open reading frames in a gene. It can also tell us whether the proteins are in secondary or in tertiary structure. Secondary protein structure includes alpha- helix, beta sheets, and extended strands[5,6]. With the help of Swissmodel, the scientists can perform homology modeling and study the tertiary structure of the protein. Clustal W performs multiple sequence alignment. Multiple sequence alignment also helps to find the evolutionary relationships [7]. Prosite database identifies us to motifs and functional domains in the protein structures. BLAST is a powerful tool for finding sequence homology and sequence comparison. NCBI is the most common used database for finding the genes, proteins, SNP and medical literature [8,9]. The screen of NCBI is shown as under-

In this paper, the author has worked on the usage of databases that can be utilized for rare disease research work.
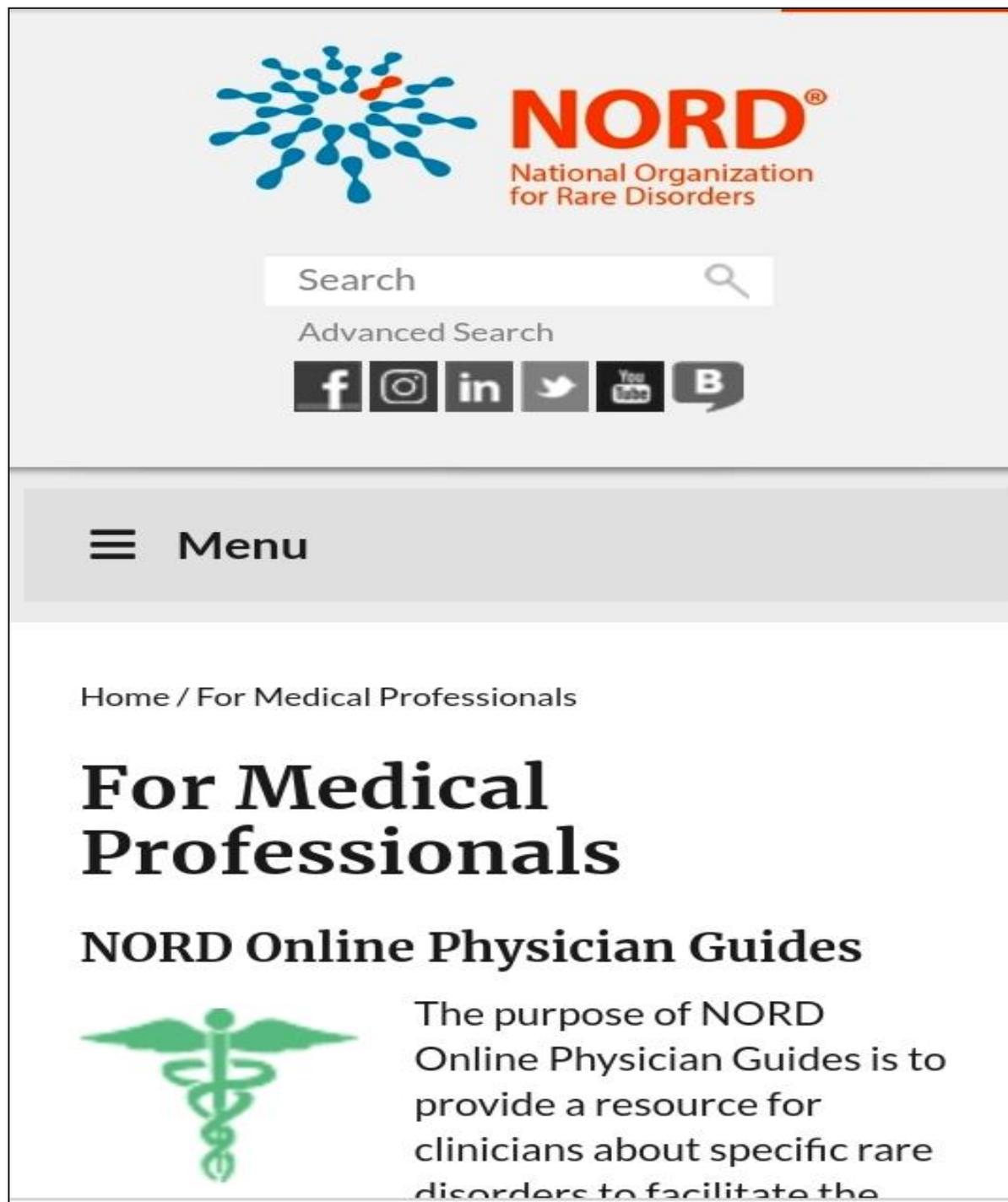
MATERIALS AND METHODS

Various databases have been discussed in this paper. These databases are extremely useful in the rare disease research.

Orphanet: It is a database which is dedicated to rare diseases [10]. With the help of Orphanet database, we can search the disease name in the search box and get the required information from the search box.

OMIM: Online inheritance in man is a database in NCBI. It is an exclusive database for studying the genetic orders [11-12]. With the help of OMIM, the researchers study the genetic pathways of the disease in detail. OMIM is linked with several databases suchas PMC and other web resources. Rare diseases can be studied in detail with the help of OMIM.

Gene Reviews: This database is a part of NCBI. In this database, various genetic polymorphisms can be studied. Moreover, it provides the important information the gene as well as regarding the genetic counseling [13].

NORD: It is a US leading organization and it stands for national organization for rare diseases. It provides latest information regarding the rare diseases [14]. It is extremely useful for the patients as well as researchers. It contains a database which has more than 1200 diseases incorporated into it. Disease includes Aarskog Syndrome, Wyburn-Mason Syndrome, XYY Syndrome, Yellow Fever and Zollinger-Ellison Syndrome. It also organizes various events which counseling to the patients regarding the rare diseases [15]. The snapshot of NORD is shown below:

Conclusion:

In the present work, the author has focused on the databases showing details regarding rare diseases. The information regarding the rare diseases will provide the path for drug discovery. Author sincerely hopes that this work will be beneficial for those scholars who are working in the field of rare diseases.

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