Ontology Based Approach for Clinical Diagnosis

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As a solution for the difficulties of clinical diagnosis by a physician, there are number of MDDSS have been developed such as POEMS, DXPLAIN, HELP, POSSUM, LDDB, OMIM etc. Also there are number of search routines developed such as syntactic feature vectors and semantic search routines. As well the technologies used to develop MDDSS are Fuzzy logics, neural networks, statistics are used for differential diagnosis. A novel generic mathematical model into differential diagnosis of patients with complex genetic diseases is proposed. Instead of the traditional way of analyzing the gene mutations to identify the diseases in gene testing, this differential diagnosis is achieved by genotype-phenotype correlation through a common computer science concept called Ontology. The publicly available biomedical and biological ontology: Human Phenotype Ontology was used to retrieve the expert knowledge of medical domain. It might not be a fine method to identify the diseases with genotype (analysis the mutations) of a dedicated gene because any disease can be caused by interacting genes or however complex behavior of gene functions or any other exceptional way. Therefore it is better to consider phenotypes of a disease. Then this approach is maps the patient genotype into semantic behavior called ontology, representation of the relationships of signs and symptoms (phenotypes) and then calculates the patient's differential diagnosis by using its mathematical model. The ultimate goal of this novel approach is to deliver the diagnostic report only for a selected gene of a patient by mapping process without analysis the genetic mutations.

If the design and implementation of the novel differential diagnosis systemis considered, it consists with two generations respective to the time and also consist with two modules respective to the structure. Initially when it will be used by a physician, diagnostic system has no knowledge of the diagnosis behavior of that physician therefore after inserting the genotype of the patient, physician should select the corresponding phenotypes by observing the patient. Then after some diagnosis times, the system acquires the physician's knowledge. It is the second generation of the system. It means when the genotype is given to the system, it gets the final diagnosis results.

Key words: *Diagnostic, Diseases, Genotype, Phenotype, Symptoms*