Metabolic research critique

Article Critique: The diagnosis of inherited metabolic diseases by microarray gene expression profiling This paper presents the study made by Hernandez et al. published in 2010 Orphanet Journal of Rare Diseases which is entitled The diagnosis of inherited metabolic diseases by microarray gene expression profiling. The authors of the article explored the gene characteristics of 68 patients with an array of inherited metabolic diseases through the method of profiling cultured fibroblasts.

Review of Prior Works

The article presented a short background of the study which is more library research based but there is no literature review which can be related to the fact that it is a relatively short article. According to the paper, there are approximately 300 accounted types of inherited metabolic diseases (IMD) and there is a continuous increase and the authors of the article perceived the importance of early identification of the presence of IMD in the system of the patients. The challenge is that there is a higher percentage of diagnosis during 15 years and above than in the first year of life but this can make the different since early diagnosis is important. The subjects have indications of possible IMD undertaken (both confirmed and suspected).

Research Objective

The research objective is stated on the last part of the background of the study. It is to be able to establish a gene expression signature to be able to improve detection of IMD earlier than normal diagnostic techniques.

Research population

The subject of the research or the population had been described in the methodology clearly. These 68 patients' human skin fibroblast cells became the subjects of the research after being cultured and tested for Mycoplasma

infection. One criticism regarding the population can be related to the number of subjects. Based on the rules of scientific research, it is important to have a higher number of subjects to be able to improve validity.

Methodology of the Paper

The methods used in the research had also been stated and organized clearly. This part of the paper is one of the longest and most specific part of the study with clear distinction of the fact that the paper is technical and that most of the readers are peers who can relate to the subject matter and the techniques presented such as the RNA extraction, the determination of the microarrays, the gathering and analysis of the mycroarray data, and finally the PCR and sequencing analysis.

On defining concepts, terms and variables

In relation to the technical presentation of the methodology, the different concepts, terms and variables are defined in a manner that only the related parameters and values are given and not defined in a layman’s perspective. It can be considered that the authors’ peers comprise the majority of the readers of the article and the journal. Examples of the said terms are the genes used for PCR analysis which are names of sequences presented with the assumption that the readers already encountered the said terms. One positive point though are the parenthetical phrases pointing to references for additional reading which can help non-science majors to have further information gathering.

Variables

The research includes variables such as the Principal Components of different diseases such as lysosomal storage disorders, purine and pyrimidine disorders, peroxisomal disorders, urea cycle defect, fatty acid

oxidation disorders, mitochondrial disorders, carbohydrate metabolism defect, and other unknown diseases. These components are analyzed through 2 methods namely Robust multiarray averaging (RMA) and Factor analysis for robust microarray summarization (FARMS). The variable are independent of one another since the analysis of the microarray compared different components to one another to determine signatures of IMD.

Results

The results of the research are highly specified which is a positive aspect of the research. Although this is the case, the information given is also highly technical thus limiting the understanding of the research to the peers in the scientific community. The researchers presented specific ways to better determine results related to the IMD. One point the needs improvement is the coverage of the research, due to the large number of IMD to be studied; there is information overload as well as the need for specificity in terms of study of different types of metabolic diseases.

Research Limitations

The limitations of the research are related to the high number of IMD needed to be covered while having low sample or population size of fibroblasts. Another limitation is the dependence to present designated standards for comparative analysis of Principal Component Array (PCA).

Threats to Validity

Different threats to validity is also related to the number of the samples used which is considered low as compared to standard requirements for scientific and qualitative research. There is also a need to have a part that focus on validity but the research recognizes the needed for further research.

Relevance to PT Practice

The different types of inherited metabolic diseases can affect the totality of the physical functions of an individual, thus, it is connected to PT practice. Usually, IMD limits the capability of patients to be given different forms of PT treatments.

References

Hernandez, MA, Schulz R, Chaplin T, Young BD, Perrett D, Champion MP, Taanman JW, Fensom A, Marinaki AM. The diagnosis of inherited metabolic diseases by microarray gene expression profiling. Orphanet Journal of Rare Diseases 2010; 5 (34).