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LEARNING RESOURCE REVIEW

METAGENE 3.0 - A KNOWLEDGE BASE FOR INBORN ERRORS OF METABOLISM

Inborn errors of metabolism are generally considered to be complex bio-medical issues. Their complexities arise from diverse clinical presentation and symptoms, for genetic heterogeneity results in a wide spectrum of phenotypic manifestations. This is also an area of clinical science which is undergoing rapid progress as a result of advances in basic medical sciences. Sir Archibald Garrod initiated the brilliant studies of alkaptonuria, which culminated in his Croonian Lectures in 1908, followed by his monograph *Inborn Errors of Metabolism,* which appeared in 1909 and in a modified form in 1923. Around 1960, inborn errors of metabolism were *rediscovered* and slowly converted to molecular diseases. Subsequently, there has been a marvelous union of molecular genetics and metabolism so that we have now an extensive database on both the metabolic basis of inherited diseases and the hereditary basis of metabolic diseases.

In such a scenario, Metagene 3.0 software provides a knowledge base for inborn errors of metabolism, specially addressed to pediatricians, geneticists, endocrinologists, biochemists, laboratory physicians, pharmacologists and students involved in diagnostic and research work on genetic metabolic disorders. This knowledge base gives an easy and quick access to comprehensive information about 340 metabolic diseases, important differential diagnoses, associated clinical and laboratory findings (more than 1000 traits), about 600 metabolic findings and some relevant publications. Such a knowledge base is likely to improve the diagnostic yield for at least two reasons. Firstly, it gives a quick access to associated information. Secondly, it provides a 'watch-dog function' so that all possible metabolic diseases associated with a set of typical symptoms receive consideration. This software also provides a separate database for storing clinical and metabolic data available to the users. The data used have been stored in twenty tables which are connected in a user-friendly network. Thus, the knowledge database and patient database can be accessed rapidly and easily. This software embodies a commendable attempt by two very experienced authors towards improving the diagnostic yield in the area of inborn errors of metabolism. After running through the demo copy of the software, I am convinced that, although it does not intend to replace textbooks available on the subject, authors have indeed instilled their knowledge and experience in this creation in such a way that it will be of great help to metabolic laboratories, clinicians, researchers and students involved with patients having inborn errors of metabolism. Similar knowledge based user-friendly software for other human disorders will also be of great help.

Technical details: Metagene, edited by Dr. von G. Frauendienst-Egger and Prof. F.-K. Trefz. Wissenschaftliche Verlags-gesellschaft mbH, Birkenwaldstr. 44, D-70191 Stuttgart, Germany. Operational requirements: 80386 IBM compatible PC with Microsoft Windows 3.11 or higher, or Windows 95, and 30 MB hard disk space. Data carrier: 1 CD-ROM. Price: DM 990/- + VAT. Annual updates: DM 100/- + VAT.

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