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Implementation of pharmacogenetics in patient care

Piatkov Irina
Blacktown Hospital, Australia

Interactions between substances metabolised through the same metabolic pathway and variations in enzyme activity should obviously be considered in drug response interpretation. However, despite the accumulation of pharmacogenetic data, the medical application of genotyping is still a subject of debate. One of the reasons is the fact that phenotype characteristics of pharmacogenetics testing are confusing and sometimes, in our opinion, are used in inappropriate ways. Biomedical laboratories provide a variety of pharmacogenetics tests. During the last few years drug manufacturers are placing a new warning into the drug's prescribing information. This warning cautions medical practitioners of the reduced effectiveness of drugs in patients who are poor metabolisers. However, a heterozygote genotype, which is usually considered as an intermediate metaboliser, with some prescribed drug combinations can also create an 'organic' poor metaboliser. It causes confusion for clinical practitioners and consequently adds to the uncertainty in the clinical application of pharmacogenetics. Our data also demonstrates that the coincidence of multiple loss-of-function heterozygote CYPs polymorphisms producing diminished enzyme activity is rare in any population. Their significance is important though, as it dramatically alters patients metabolising capacity and produces a poor metaboliser phenotype which often results in severe effects arising from the toxicity. As per the latest scientific development, it is undeniable that pharmacogenetics testing in combination with other parameters will help to avoid adverse reactions by providing information about an individual's drug metabolising capacity, thereby identifying the patients for whom a drug would be safe and also potentially lowering overall healthcare costs. However, precise definitions of phenotype and genotype characteristics are needed to avoid confusion in medical practice. The phenotype and genotype terminology in pharmacogenetics testing interpretation will be discussed. The following will be included in the presentation: Data from our previous projects; data from our laboratory testings and patient individual cases.

Biography

Irina Piatkov, PhD, is a Senior Hospital Scientist in Charge of the Blacktown Molecular Research Laboratory at the University of Western Sydney Clinical School and Research Centre, Blacktown Hospital, Western Sydney Local Health District. The laboratory supports research and provides tests in the area of personalised medicine, mostly in pharmacogenetics. As an interdisciplinary centre for scientific research and teaching, it is involved in projects for different hospital departments. She has received her PhD in1986 and started her scientific career in the former USSR. Since that time, she has been working in diagnostic, teaching and research projects in clinical biochemistry and molecular genetics in USSR, Canada and Australia.

irina.piatkov@health.nsw.gov.au