Masterclasses

M9.2 Clinical Genetics and Genomics – Application and Beyond 09:00 Room 221

Genetics and Genomics beyond Rare Diseases – The Future is Now

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Diseases caused by genetic or genomic abnormalities were once managed by rare individuals with highly specialised medical expertise. With the rapid expansion in knowledge in human genomics and the availability of powerful investigational tools, the use of genetic and genomic tests are now infiltrating many branches of medicine. For example, the risks of Steven-Johnson syndrome and toxic epidermal necrolysis could be mitigated by avoiding carbamazepine prescription in Asian individuals with HLA-B*1502 allele. Warfarin doses could be titrated according to the person's CYP2C9 and VKORC1 genotypes. Tyrosine kinase inhibitors are the first line drugs for patients with non-small cell lung cancer harbouring sensitising epidermal growth factor receptor mutations. Maternal blood DNA analysis allows for sensitive and specific, yet non-invasive, screening of fetal chromosomal aneuploidies. Non-invasive DNA-based screening of fetal single gene diseases which are individually rare but relatively common collectively, is now scientifically feasible and provides an opportunity for in utero or neonatal therapy. Circulating tumour-derived DNA testing has been shown in a prospective study to be effective in the detection of early nasopharyngeal carcinoma among asymptomatic community participants, with a demonstrable reduction in mortality. These are just some of the examples whereby DNA testing on relatively large target populations has demonstrable clinical benefits. Evidently, the incorporation of genetic and genomic information in medical care, aka. genomic medicine, is becoming a daily routine in many areas of medical practice now. How may we, as healthcare professionals, better equip ourselves to embrace this new paradigm?

(Supported by the Research Grants Council of the Hong Kong SAR Government under the Theme-based research scheme (T12-401/16-W) and (T12-403/15-N)).