Public Health Genetics – a Breast Cancer Perspective

With the rapid expansion of knowledge in the realm of genomics, there is no doubt that we are moving towards an era of personalized medicine, with more individualized disease prevention strategies and therapeutic options. Hence, the intricate interplay between molecular genetics and public health is being increasingly recognized, and this will be discoursed in the context of one of the commonest diseases – breast cancer.

Our knowledge about breast cancer genetic factors these days is generated through different genetic epidemiological study approaches, from genetic linkage studies, to candidate gene case-control studies and genome-wide association studies (GWAS). This led to the identification of breast cancer genetic factors of different penetrances and prevalences (Turnbull & Rahman, 2008).

Enlightening as they are, novel molecular discoveries confer minimal benefits unless they are translated into practical use. However, with emphasis on evidence-based medicine these days, there is a whole layer of complexity involved in the bench-to-bedside translation, which can be a protracted process. Hence, of the many breast cancer associated genetic factors known to-date, only a handful is widely available for genetic testing.

And while breast cancer genetic testing is becoming increasingly common, due to the high cost involved and limited public funding available in Australia, not all individuals who are concerned about inherited breast cancer risk are offered genetic testing. To ensure cost-effectiveness and equity of access to genetic testing services, various models, such as BRCAPRO and BOADICEA, have been introduced to facilitate selective genetic testing on individuals most likely to be affected by genetic mutations.

Also, as we advance towards more personalized medicine, cancer control will adopt a fresh new attitude. It appears that it is no longer appropriate to view cancer prevention as a ‘one-size-fits-all’ public health effort and apply the same prevention strategies to the population at large. With better understanding of genetic basis of breast cancer and hopefully more accurate cancer risk prediction, it seems inevitable that there will be a gradual shift towards more individualized cancer prevention strategies, based on one’s unique genetic make-up, and this is going to have a huge implication in the improvement of resources allocation (Pharoah et al, 2008).

In short, public health genetics is an emerging field of great importance, involving integration of genomic knowledge into public health research, policy and practice for disease prevention and management.