

## Myths and realities of Personalized Medicine by ฉนญัจฉ์ตน์ บั้งเล็ง

### **What impact is personalised medicine currently having on your chosen disease? (Neurological track)**

Within the neurological track, there have been many genes associated with Alzheimer's disease, such as CR1, CLU, PICALM, ACE, CST3 and the infamous APOE. Ten years ago the disease would be described by its effects such as dementia, its physical characteristics such as amyloid plaque and tau tangles in the brain and personal individual characteristics like gender, age, and weight and life style like exercise, education, anti-inflammation drugs taken.



Many factors were known to influence Alzheimer's but much was not explained as to why some people would get the disease and others wouldn't. While much is still not understood, understanding the individual's genetic profile has significantly narrowed the knowledge gap.

The genetics of early onset Alzheimer's is fairly well known. When one of several genes that are involved in APP, Amyloid Precursor Protein, are modified, individuals develop Alzheimer's between 30-60 years old. This is a dominant trait. The parent with the mutation has a 50% chance of passing it to their offspring, who will probably then develop Alzheimer's early.

Since people with the APOE4 allele develop Alzheimer's more frequently and earlier, researchers screen for APOE4 and preferentially select APOE4 research subjects.

### **Will personalised medicine switch the focus from disease treatment to prevention?**

Personalised medicine will switch the focus of disease treatment to disease prevention and prevention is much better than cure. I'm looking at this from the viewpoint of being both a patient and a medical who developed breast cancer. In the United Kingdom chronic disease is proving to be a great burden to the National Health Service which is growing massively as time goes on, not least because we are living longer and modern medicine is helping us to survive into old age. However, 'what it is the quality of our lives in old age?'. If personalised medicine, rather than just treating disease, focuses on

preventative measures then an individual's life will be of better quality and they are less of a burden on their families or societies - reaching old age in a generally 'fit and well' state.

The National Health service in the UK is free at the point of access and is funded by the tax payer- insurance companies are not involved in this public institution. If it were then there might be a greater push for bringing effective preventative measures into the NHS primary care system. As personalised medicine focuses on an individual's genetics and genomics then it would be an important foundational step to establish these baseline parameters for each individual patient as the starting point in individualising preventative measures- most notably when considering lifestyle factors and how they can enhance or negatively affect a particular individual's health. We know that lifestyle factors can influence an individual's chances of developing cancers. Smoking, obesity, hormonal imbalances, exposure to toxins in the environment etc, can all be linked to cancer development. Preventative measures have greater effectiveness if people are managed as individuals and not 'en masse'- though the latter is better than no preventative measures at all.

So looking at my own health situation, had there been an early focus on a personalised (preventative) medical approach to wellbeing, I would have taken heed of the following and very likely have avoided developing breast cancer. My disease treatment instead involved endure very exhausting and traumatic therapies that followed (which included surgery, chemotherapy, radiotherapy, hormonal therapy and targeted monoclonal antibody therapy). Focus on MY disease prevention with Personalised Medicine (notably with genetic/genomic profiling) would have highlighted the following:

Exercise- knowing which type of exercise would bring me greatest benefit, I could have led to better results, greater physical fitness overall and enhanced motivation- exercise improves immune response in fighting infections and preventing cancer.

Neutrigenomics- knowing which type of diet suits my genome best and which micronutrients I have a risk of poorly metabolising would have allowed me to streamline my dietary intake to better suit my metabolism.

Metabolism - my genomic variation with regard to oestrogen metabolism and detoxification I believe contributed to my developing an oestrogen dependent type of breast cancer. If I had know about this personal tendency to build up more toxic oestrogen metabolites then I would have reconsidered use of oral contraception and HRT. I also would have looked more closely at the whole of the Steroidogenesis pathway and been more alert to my psychological stress levels which through 'progestogen steal' and increased cortisol levels also increased my risk of breast cancer. Sleep deprivation was also more harmful to me (in the light of genomic testing) and avoidable.