PERSONALIZED MEDICINE IN 2020

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Over the past decade, powerful genotyping tools have allowed geneticists to look at common variation across the entire human genome to identify the risk factors behind many diseases. Two striking findings will define the study of disease for the decade to come. First, common genetic variation seems to have only a limited role in determining people's predisposition to many common diseases. Second, gene variants that are very rare in the general population can have outsized effects on predisposition.

For example, rare mutations that cause the elimination of chunks of the genome can raise the risk of diseases such as schizophrenia, epilepsy or autism by up to twentyfold. Some researchers view these major risk factors as aberrations. My guess is that as more genomes are sequenced, many other high-impact risk factors will be identified.

If so, here's one confident but uncomfortable prediction of what personalized genomics could look like in 2020. The identification of major risk factors for disease is bound to substantially increase interest in embryonic and other screening programmes. Society has largely already accepted this principle for mutations that lead inevitably to serious health conditions. Will it be so accommodating of those who want to screen out embryos that carry, say, a twentyfold increased risk of a serious but unspecified neuropsychiatric disease?

Some advances will be relatively uncontroversial, such as the development of tailored therapeutic drugs based on genetic differences that are otherwise innocuous. Others will be transformational, such as the identification of definitive genetic risk factors that provide new drug targets for conditions that are often poorly treated such as schizophrenia, epilepsy and cancers. Over the next decade millions of people could have their genomes sequenced. Many will be given an indication of the risks they face. Serious consideration about how to handle the practical and ethical implications of such predictive power should begin now.