CHAPTER 18

Biomarkers and Personalized Medicine

I. INTRODUCTION

Biomarkers include proteins, peptides, cells, and genetic markers. Genetic markers encompass single genes, small collections of three or four genes, and large collections (arrays) of genes. Typically, when a gene is used as a biomarker what is actually measured is the mRNA expressed by the gene. However, in the case of gene amplification and chromosomal abnormalities, what is measured is the gene itself. For genes that encode polypeptides, the term "gene" refers to the combination of regulatory sequences plus sequences encoding the polypeptide. Genetic markers include classical mRNA, as well as micro-RNA (miRNA) (1,2,3,4). Common biomarkers include low-density lipoprotein (LDL) cholesterol, used for assessing risk for atherosclerosis (5) prostate-specific antigen (PSA), used for assessing risk for prostate cancer (6,7) and human estrogen receptor-2 (HER2), used to assess risk for breast cancer (8).

Most laypersons have seen lists of biomarkers on printouts from their annual medical checkup. These printouts include biomarkers, as well as other parameters, such as blood cell counts and electrolytes. Blood cell counts and electrolytes are not considered biomarkers because they are clinically important in their own right, and are not used primarily to represent or predict any future-arising medical condition. In other words, an extremely low red blood cell count is, by definition, *anemia* (9). Moreover, a low red blood cell count has the following utility – it means that you have anemia right now; not that you are at increased risk for getting anemia some time in the next 5 years.

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Clinical Trials
DOI: 10.1016/B978-0-12-391911-3.00018-9