Biological markers in medicine

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Biomarkers are naturally occurring molecules or genes by which specific physiological or pathological processes such as symptoms of a disorder or a disease can be identified. Biological markers can indicate disease characteristics or a particular response to a given medication. Identification of the dominant or co-dominant nature of a gene with a marker may help to differentiate between heterozygous and homozygous organisms. It also allows amplification of the desired sequence within the genome for comparison and further analysis. Genetic markers are genes or fragments of DNA that are linked to a certain location within the genome and can be used to identify individuals or species. An ideal DNA marker should include the following properties: a) highly polymorphic nature as polymorphism is measured for genetic diversity studies, b) frequent occurrence in genomic sequence, i.e. it should be evenly distributed throughout the genome, and c) easy accessibility and high reproducibility. The development of biomarkers requires the collaborative effort of medical, analytical and bioinformatics researchers. The role of bioinformatics is essential as it helps in building a relationship between previously established biomarkers and the one which is isolated for a particular research purpose through imaging techniques. The steps involved in biomarker discovery comprise experimental design, biomarker candidate screening and candidate validation. The applications of biomarkers are broadly categorised into diagnostic, prognostic and predictive. These biological markers can assess the risks, predict the appropriate drug doses, predict and monitor treatment response as well as analyse the chances of recurrence; therefore, they play a significant role in personalised therapy and cancer research. However, the limitations of biomarkers include issues with specificity, sensitivity and tissue accessibility. Hence, further research and development in this field would aid in overcoming these limitations and make biomarkers indispensable tools for clinical applications.

Keywords: Biomarkers, Cancer research, Genetics, Bioinformatics, Diagnostic tools

Citation:

Himanshi Bakshi. Biological markers in medicine. The Torch. 2021. 2(28). Available from: https://www.styvalley.com/pub/magazines/torch/read/biological-markers-in-medicine.