Single Nucleotide Polymorphism

SNP and future of genetic epidemiology

This research article mainly focuses on the uses of polymorphism technologies in the field of epidemiology, where it can be used to detect the factors responsible for a specific disease and what should be we focusing on regarding this technology in the near future (for them, for us it's probably the present or past already). I am mentioning the key takeaways from the paper for you guys' convenience.

[I took help of AI to summarize and rearrange the takeaways, so if you guys' still have any confusions about the contents, please do ask away]

Key Takeaways

1. Genetic Polymorphism and SNPs

- Polymorphism refers to genetic loci where the most common allele occurs at a frequency of less than 99%.
- SNPs are the most common type of polymorphism, resulting from single base mutations, and are found approximately every 300–1000 bases in the genome.
- Other types of polymorphism, such as microsatellites and insertion/deletion variants, are also discussed for their roles in population studies.

2. Traditional Uses of Polymorphism

- Historically, polymorphisms have been used for meiotic mapping or linkage analysis to identify genes associated with traits or diseases.
- Challenges faced with the techniques based on ***polymorphism*** include:
    - The neutral theory of evolution, which posits that much sequence variation is not phenotypically significant.
    - The multifactorial nature of most traits, making it difficult to isolate individual genetic factors.
    - Methodological issues in linking sequence variation with phenotypic variation.

Applications of SNP Technologies

1. **Gene Discovery and Mapping**
    - SNPs can help refine linkage analyses and identify candidate genes associated with diseases.

2. **Association-Based Candidate Polymorphism Testing**
    - Testing specific SNPs for associations with diseases or traits can provide insights into genetic predispositions.
3. **Diagnostics and Risk Profiling**
    - SNPs may be used to develop diagnostic tools or risk profiles for diseases based on genetic predispositions.
4. **Prediction of Responses to Environmental Stimuli**
    - This includes responses to drugs (pharmacogenomics), diet, or other environmental factors.
5. **Homogeneity Testing and Study Design**
    - SNPs can help assess population structure and ensure homogeneity in epidemiological studies.
6. **Physiologic Genomics**
    - Understanding how genetic variation influences physiological processes and contributes to phenotypic diversity.

Advantages and Challenges of Using SNPs

- **Advantages:**
    - High abundance across the genome.
    - Stability over generations.
    - Potential for high-throughput genotyping using advanced technologies like microarrays.
- **Challenges:**
    - Difficulty in linking specific SNPs to phenotypic traits due to confounding factors.
    - The need for robust study designs to avoid false associations.
    - Variability in SNP frequency across populations, which may limit generalizability.