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EDITORIAL

Genomic Beyond Rare Diseases

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For decades, the field of genetics was viewed primarily through the lens of rare, Mendelian disorders. The diagnostic odyssey moved from basic metabolic workups to single gene testing, and eventually to Whole Exome and Genome Sequencing. However, as we stand on the height of a new era in medicine, it is evident that the utility of genomics has expanded far beyond these traditional boundaries. We are witnessing a transition where genomics is becoming a cornerstone of up-to-date healthcare for the broader population.

The modern application of genomics is transforming precision medicine across multiple disciplines. In pharmacogenomics (PGx), for example, the impact is immediate and tangible. In a pilot study of 512 patients at a tertiary care center, >90% were found to carry an actionable allele (1), highlighting the critical need for genetic profiling to prevent adverse drug reactions and ensure the "Right Drug Dose Now". Beyond pharmacology, genomic profiling is revolutionizing oncology through liquid biopsies and targeted immunotherapies, and redefining microbiology by treating the microbiome as a "new organ" via metagenomics.

Furthermore, the scope of genomics now encompasses complex disease risks. Through Polygenic Risk Scores (PRS), we can now stratify populations based on their susceptibility to widespread conditions such as Type 2 Diabetes, inflammatory bowel disease, schizophrenia, and cardiac disorders. This shifts the medical focus from reactive treatment to proactive, personalized prevention, balancing genetic risks with lifestyle interventions.

For Saudi Arabia, the implications of this shift are profound. Our unique population structure, characterized by specific incidence rates of common genetic disorders, necessitates the creation of robust, local "Big Data" genomic databases. Relying on international data is insufficient, we must prioritize data sharing and basic research within the Kingdom to capture population-specific insights (2).

To realize this vision, we must leverage Artificial Intelligence (AI) and deep learning to handle the scale of data generation. However, technology alone is not enough. We must address the "elephant in the room": infrastructure and cost. "Making Genomics Easy" requires investment in manpower, training programs, and accredited lab facilities capable of high-throughput operations.

The ultimate goal is accessibility. By streamlining operations and embracing local operational authority, we aim to bring the cost of sequencing down. This democratization of data will ensure that genomic medicine is not a luxury for the few with rare diseases, but a standard of care for the many.

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