

EMPOWERING INTELLIGENT TRANSFORMATION THROUGH GENOMICS TO ADDRESS HUMANITY'S GREATEST CHALLENGES

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Mankind's greatest challenge lies in issues pertaining to health, environment, and nutrition. All these are heavily interdependent and trying to resolve anyone would need a careful evaluation of the other two. Besides, the dynamic nature of all the three components makes the issue of resolving them even more challenging. Fluctuations and disruptions in these components are inadvertently getting incorporated due to growing social needs, which is an inherent property of the human mind. Developing strategies to build an appropriate AI-based model to monitor the impact of changing environment, nutrition, and health status, individually and in combination, would be the need of the hour. On the hindsight, one would also wonder, are there better and simpler read outs of these three components, which can act as biological indicators for health, environment, and nutrition. The best biological indicators of these components lie in one's genome.

Genome testing can be the best available point of care testing for diagnosis and therapy, which can increase the confidence and reliability and would also minimise frequent interventions in the name of testing. In a conventional clinical establishment, one has to undergo a multitude of tests, but a genetic test may aid in minimising and streamlining these tests. However, to achieve this, one needs to build a mature dataset and then develop AI-based prediction tools to minimise the random tests.

Present AI prediction models based on genomic data has limited takers as it is not a representation of global genome profiles, and the clinical genomic screening is often restricted to clinical exome, which is again an offshoot of a regional bias enriched with European dataset. Besides, the clinical exome is targeted to address the issue of therapeutic intervention. Therefore, if one has to better understand the possible biological readouts of gene-environment-nutrition interaction, it would be ideal to consider the whole exome for a resource crunch establishment, while the real advancement or precision towards understanding the gene-environment-nutrition interaction would come from a whole genome study.

One may ask what purpose will this whole genome data provide. It is quite well established that one progresses towards a disease state when there is a shift in the homeostatic balance. This homeostatic balance is maintained by an internal feedback mechanism. The threshold for the homeostatic balance and the feedback mechanism is governed or determined by the inherent variations in an individual's genome. Human biology has to be interpreted from the point of view of the interactome. Emerging evidence from nutrigenomics projects or gene-environment interaction based on physical exercise, meditation, etc., indicates that we have inherent feedback mechanism which can aid in the prevention of a majority of the disease conditions. Growing evidence on the implication of epigenetic genes, metabolic genes, and immunological genes in common diseases like mental disorders, neurodegenerative disorders, metabolic disorders, developmental disorders etc., are a clear indication in this direction. Therefore, if we determine what genomic feature defines the threshold of homeostatic balance and controls the internal feedback mechanism, we can possibly prevent the onset of the disease or its pathological process. There is tremendous scope in developing an AI-based prediction model for predicting or preventing the onset or the pathological process of any disease.

It is pertinent to believe that the DNA is life and rest is all translation. If one can model the background and basis of these translations, then it is possible to predict the benefits and risks of changing environment and nutrition. Emerging newer domains of OMICS technologies are indeed the best possible way to assess the role of genomics. These emerging technologies are posing greater challenges to computational hardware and software developments to handle or analyse the data. On one side, we have the issue of technological developments in the generation of OMICS data, while on the other side, we have rapid advancements in the analysis of data. Despite of all the developments, the analysis of genome still remains a challenge. Basic analysis requires a quality check, alignment, annotation, detection of SNPs, insertion/deletion, copy number variation, de novo mutations, which can take several hours to days as one has to integrate multiple pipelines into one. Besides, the gap between knowledge and expertise is further widening, which makes the tasks even more difficult. Therefore, there is a tremendous need to develop simple, easy-to-use AI-based tools which can aid in narrowing the gap. **Lenovo's Genomics Optimization And Scalability Tool (GOAST) 3.0, powered by Intel® Xeon Scalable processor** indeed makes a significant step in this direction where basic annotation of the genome can be completed in less than 10 minutes. **This solution from Lenovo** also makes a good attempt for a simple-to-use format that needs minimal training. However, intelligent transformation of health-environment-nutrition data and developing a precise AI-based feedback mechanism using genomics would be the way forward. This, in turn, will aid in refining diagnostic, therapeutic interventions and in-situ modulation.
