Towards a genomics revolution (GS paper 3 ,UPSC IAS mains ,Sci & Tech)

India has the scientific resources for genetic research – all it needs is the vision at the national level to leverage them
In 1865, Gregor Mendel discovered the two laws of inheritance that are now named after him. Almost 90 years later in 1953, the work of James Watson, Francis Crick, Maurice Wilkins and Rosalind Franklin, deciphered the structure of the molecule – DNA – that stores our hereditary information and gets transmitted from parents to children over generations.
Personalised medicine
At this point, in principle, the prospect of building individualised medicine based on the precise information stored in each human's DNA (their genome) had come into view. But the human genome has around 3 billion base pairs and in 1953 it wasn't possible to imagine extracting genetic information on the molecular scale and of this collective size.

Technological advances in sequencing methods have made the possibility glimpsed 60 years ago a reality today. Already by 2001 the human genome project and its private competitor, Celera Genomics, showed that an entire genome could be sequenced.

Since then the cost of doing so has plummeted – currently it is something like $1000 per person and becoming cheaper – and the age of genomics-informed medicine is now within sight. Perhaps this will also make interventional treatments feasible, in the not too distant future, thanks to the revolutionary advances brought about by the discovery of new gene-editing techniques, such as CRISPR.

Surveying Indian variation

What implications do these developments have for India and are there deliberate choices that would shape this coming future more advantageously for the country and its people? Are there strengths that India can bring to this task? To gain fully from the genomics revolution, India needs to collect information about the genetics of its population and train manpower capable of interpreting it. The information that is needed has to come from a large and sustained collection of data – fully sequenced individual genomes along with medical histories for the individuals who volunteer for this effort.

This kind of longitudinal study is what would allow actual physical manifestations relevant to health, e.g. specific illnesses, to be related to features in the genome. To pick an ambitious but not impossible number, a data bank that collects this kind of information on one million Indians over the coming decade would be a feasible effort of the right magnitude. We note that the China Kadoorie Biobank has been studying half a million people since their recruitment in 2004-2008. As India is much more genetically diverse – with something like 5,000 ethno-linguistic and religious groups (castes and others), all of which probably have some degree of genetic distinctiveness – it needs a larger survey to do justice to all Indians.

The genetic distinctiveness of different Indian groups is in part the result of endogamy. While we cannot know the full impact of endogamy in advance of a proper survey, some recent research has shown that endogamy is very likely to be medically significant. Castes are not just “of the mind”. The genetic implication of this is that there are likely to be many recessive diseases stemming from single genes specific to individual groups that can be identified.

Decreasing disease burden

This knowledge could then also be quickly applied to the task of managing diseases in these groups as well as be used for genetic counselling that could reduce their incidence in

future generations. As an example elsewhere, the founder group of Ashkenazi Jews have almost eliminated Tay-Sachs disease from their population by such means. Looking ahead a bit more, with large samples the technique of “genome-wide association studies” that compare genomes of cases and controls could be used to identify genetic risk factors related to common diseases (such as heart disease that stem from many genes) that affect the health of many more individuals. We would like to emphasise that much of this is simply a question of applying existing methods and could all be done fairly quickly.

This is a good point at which to note that such a survey of Indian genetic diversity will be an important asset, beyond disease genetics. The data collected as part of these efforts will also help to uncover the basic biological function of genes and their interactions, which are not yet fully understood. This knowledge will be useful to humanity worldwide and also offer India a chance to claim a piece of the global medical and scientific frontier.

As a large part of the enterprise would be the application of information technology or “bio-informatics”, the prospects of establishing viable commercial enterprises with synergies to existing IT champions are also promising.

What then is to be done?
As things stand there is certainly progress under way. There has been path-breaking work in using genomics to shed light on Indian history, a small number of hospitals are using genetic information to help patients, and there is at least one private sequencing company in India. But all of this activity is on a much smaller scale than needed and is currently not generating the manpower required to equip the next generation of medical and research activities in the area. What is needed is a coherent push at the national level that involves government, academic institutions, the existing health-care industry, the IT industry and the nascent biotechnology

industry. This coherent push should aim to set an ambitious but realistic objective of creating an Indian genetic data bank, to promote academic programmes that train scientists, technicians and doctors in this area and to create a regulatory framework that promotes broad objectives for both public and private sectors without being self-defeating.

The fact is that both genetic data and biological samples are easily transported across borders and if Indian regulation is shortsighted, it will simply cause Indian genomics to move abroad to places such as Singapore. In this context it is worth mentioning that the GenomeAsia 100K Initiative based in Singapore plans to sequence 100,000 Asian genomes, including some from South Asia. While this is eminently worthwhile as it will provide a broader pan-Asian set of data, it would be important to make similar investments at a national scale quickly to avoid the situation that this is one of the only enterprises to which Indians can turn to.

All in all, the time is ripe for India to begin its own genomics revolution. The technical understanding and will needed to launch this is present in India’s scientific leadership, in medicine and in industry. What is needed is a vision and leadership at the national level to leverage this and seize the day. Nothing less than the very health of the nation is at stake.

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