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EDITORIAL

Genomics in Saudi Arabia call for data-sharing policy

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Traditional genetics are becoming genomics in the era of big data. Saudi Arabia has a specific population structure with high rates of consanguinity and high incidence of population-specific autosomal recessive disorders. T he estimated rate of consanguineous marriages in Saudi Arabia is around 52%–58% (1,2). Two studies of large, whole-exome cohorts conducted in Saudi Arabia [Alfares et al. (3); Monies et al. (4)] reported high rates (84%) of autosomal recessive disorders with homozygous disease-causing variants; compound heterozygous variants account for 5% and non-recessive disorders account for only 11%, making them comparatively less prevalent. As our understanding of genetic disorders advances, underlying genetic defects at the molecular level are being unveiled by many genetic clinics, local and international diagnostic laboratories, and research centers. However, testing laboratories generate a huge amount of data by many means, including advanced clinical genetic testing, single nucleotide polymorphisms arrays, whole-exome, and whole-genome sequencing targeting specific populations. Since such data are fragmented by individual or among institutions, one of the goals of the Saudi Human Genome Project (SHGP) is to assemble a Saudi population-specific database by combining and fi lling in gaps in data (however, access to the SHGP database is still limited). Similarly, curated data are now being generated by many other local institutions for diagnostic or research purposes.

A fundamental principle in health ethics is that all physicians should have access to the best available medical information and patients should receive equal treatment. Since most genetic disorders have as yet no treatment, data sharing could prevent severe outcomes by lowering the incidence of genetic disorders, keeping a child with chronic illness from burdening families or communities, and minimizing the economic impact of genetic disorders in terms of the high costs of management and treatment. Data sharing would lead to clinical and research advances, enhancing health care, providing accurate data on the carrier frequencies of the most commonly encountered disorders in Saudi Arabia, and ultimately providing better variant curation and classification. This would lead to better utilization of preventive measures like parental testing or pre-implantation genetic diagnostics and could thereby lower the incidence of genetic disorders in Saudi Arabia. Data sharing in a clinical setting could also allow carrier testing for well-known familial variants of any lethal or chronic genetic disorder.

However, privacy and security are always concerns, since human genomic information is sensitive and potentially identifiable (5). In Saudi Arabia, local data protection and sharing policies are either insufficient or lacking due to diverse institutional policies or sources of funding. Immediate and urgent efforts are required to establish countrywide and robust genomic data-sharing Policeis to allow data to be exchanged among institutions without

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restriction and thereby reduce the burden that inherited disorders place on the community. Data sharing will also improve the research efforts regarding bio-marker detection, computational biology, and treatment discovery. Ultimately, sharing genomic data is essential to improve health care and save lives.

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