Commentary: Decoding genetic retinal diseases research in India

The authors Kumaragurupari and Mishra have made a wonderful and very timely effort by bringing the genetic retinal diseases (GRDs)-related research in India into current perspective.[1] In India, the burden of GRDs is disproportionate to the amount of research in GRDs.[2] A recent population-based epidemiological study from southern part of India found one of the GRDs, that is, retinitis pigmentosa, to result in a higher incidental visual impairment than the causes like diabetic retinopathy and age-related macular degeneration.[3] With this evolving trend and continuing inbreeding practices in India, as also relevant and successful forays into management of GRDs, the findings of the current study bring into light the daunting task faced by Indian researchers.

The article by Kumaragurupari serves as a definite guide for all the clinicians and basic scientists working on GRD. The authors have wisely assessed the journals where papers were published with respect to the impact factors as a marker for quality of research, and even the latter was chosen from the center point of the decade, thus partially negating the impact of the time trends on a bibliometric analysis. The use of existing research publications by the peer researcher is termed as dependability, which is one of the components in assessing the quality of research. The ranked number of citations attained by these publications

is another factor, which, if looked at as a function of time, guides on the impact or novelty of the manuscript. Use of citation index also guides on the transferability of the published research.[4,5] These factors must be a part of similar future endeavors.

The finding of lowest research on basic sciences in GRDs by Kumaragurupari in the current article is an opportunity for all the clinicians to introspect and improvise on the research related to basic science in GRDs.[1] This deficiency is attributable to heavy volume of clinical patient load, lack of training in post-graduation toward basic sciences, accessibility, and affordability of genetic testing by patients. Another most important reason is the genetic heterogeneity in GRDs, with very large number of genes already known for the diseases and new genes discovered very often in different populations across the world. This makes the basic research in genetics very much time consuming with a longer path. Apart from these, basic research also requires suitable animal models that are not available in many institutions in India. In this direction, the paper correctly extracted the role of leading institutions like Sankara Nethralaya and the ensuing collaborations, which appear to be mostly external and international. At the current and crucial juncture, there is a gap within India for internal collaborations toward GRDs and genome-wide association study (GWAS) assessment, the latter being entirely dependent on collaborations. Further, given the multiethnic nature of our population, research would become useful for the Indian society holistically only if such connections are developed and maintained over a long time. The current paper could not highlight this gap, but the authors acknowledged the lack of such insightful developments.

Probably, in relation to search methodology and use of natural language terms, diseases like diabetic retinopathy and macular degeneration, generally not a pivot for discussion on GRDs and following complex non-Mendelian patterns, came into forefront as the most highly researched GRDs. Kannabiran *et al*.[6] have recently reported on studies related to genetics of inherited retinal diseases, with special consideration of retinitis pigmentosa in India. It highlights the scope for investigating the genetics of inherited retinal dystrophies (IRDs) in an understudied population with high degree of consanguinity and inbreeding, which will lead to better identification of genes in Indian families. Moreover, understanding of the genetics helps in better counseling and future risk assessment in the upcoming generations, in addition to the possibility of applying newer therapies related to gene therapy.

In a much broader logical light, the manuscript by Kumaragurupari and Mishra defines the gaps and limitations of Indian research on GRDs or IRDs. With goals of affordable, appropriate, and timely care, Indian research needs to keep pace with research in other countries and focus on developing lasting national-level collaborations that ultimately seek a holistic care for the Indians.

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