Affordable Neurogenetic Screening as a Powerful Tool for Beti Bachao

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Genetics is the study of genes and genetic variations in living organisms. Genetic variations in complex diseases can be tested using sophisticated molecular tools developed by scientists over the years. Despite the success of the human genome project, the potential of this technology for the *Beti Bachao* program remains underrepresented in government hospitals and medical institutes.

For example, Duchenne muscular dystrophy (DMD) is a fatal incurable disorder of dystrophin gene abnormality. It is inherited in the X-linked recessive fashion, thus affecting only the male child. This means that it does not manifest in female mothers because they possess the extra gene copy (XX) to shield the deleterious effect of the defective dystrophin gene. However, this is not the case in males where it expresses it, leading to the fatal disease. Interestingly, the disease also has a higher incidence in India, up to 1 in 1,400 as compared to global figures of 1 in 3,200, with bulk of the rehabilitation work (read alternative therapies) being carried out by non-government organizations such as Indian Association of Muscular Dystrophy in the North India and Molecular Diagnostics, Counseling, Care and Research Centre in South India. Several experts continue to insist that neurogenetic diseases are no longer rare, and therefore their social and economic impact is enormous. The X-linked recessive pattern of inheritance makes the females susceptible to becoming an invisible carrier of the disease, thus increasing the proba-

bility of her offspring to be affected, with up to 25% DMD sons, and 25% daughters as carriers, and 50% offspring remaining unaffected, if not screened promptly. The rural and/or urban poor women (especially, DMD mothers) who do not have access to affordable genetic tests in government hospitals remain uninformed about their impending vulnerability. Prevention options for the families are, therefore, critical for the management of this incurable genetic disease so that death and misery do not spread any further, and DMD mothers are not ostracized in the family or society. The consequences can be disastrous in our patriarchal society, especially when this DMD mother has to bear the pain of seeing her DMD sons die in her lifetime apart from being forsaken by her husband.

These women can be mapped for a follow-up so that there is complete integration into a socially secure environment. This is also required because the identification of female carriers of deletions/duplications of the DMD gene is crucial for preventing the birth of children affected by DMD. By ignoring the potential of molecular techniques in government hospitals/institutes of the region, known for female feticide, there is looming risk of unmasking the vulnerability of carriers of genetic disorders.

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This can be prevented by carrier screening, pre-natal screening, and pre-implantation manipulation. SAARC countries and African nations face similar challenges, where access to genetic testing is very expensive or limited. Developed nations face a different kind of challenge due to the high cost of genetic testing (up to USD 1,500), which Indian institutes provide for a mere USD 20. This is also an opportunity for globalizing India's genetic testing outreach and capacity building if the new Medical Education Commission, set to replace Medical Council of India, is represented equally by individuals from medical, scientific, patient groups, engineering, economics, and AYUSH backgrounds to achieve an integrated healthcare system. This can also save the deprived sections of the society, especially female carriers of affected dystrophin gene, from impending exploitation and ridicule.

Such free genetic screening measures for detecting female carriers can become an important aspect of *Beti Bachao* policy of Government of India, partly experimented by Joshi Foundation that organizes such health camps that seek to integrate genetics with modern and traditional medicine. These health camps are socially effective for poor carrier females (DMD mothers) who are otherwise left to bear the brunt of politics of referral to private centers.

Mandatory nationwide launch of genetic testing service units by scientists in the tertiary care hospitals can not only promote *Beti Bachao* campaign in a scientific manner, but also preserve the genetic database which can be exploited for bio terrorism. This can best be achieved by establishment of Centers of Integrative Medicine within the Medical Institutes.