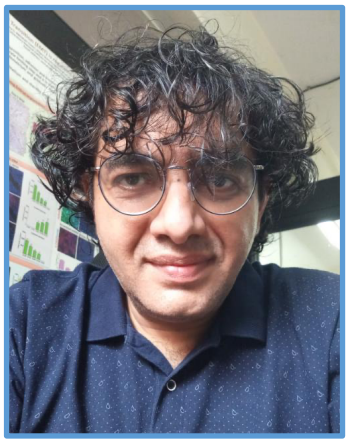
Pre-implantation genetic testing and morally responsible eugenics



Dr. Deepak Modi

National Institute for Research in Reproductive Health, Mumbai, India
E-mail: deepaknmodi@gmail.com
Associate Editor

Dr. Modi is a Professor and Scientist at the National Institute for Research in Reproductive Health, Mumbai, India. He works in the field of reproductive biology.

Dr. Rajender Singh

Central Drug Research Institute, Lucknow, India
E-mail: nainrs@gmail.com
Associate Editor

Dr. Singh is Senior Principal Scientist at the Central Drug Research Institute and a Professor at the Academy of Scientific and Innovative Research (AcSIR), India. Dr. Singh works in the field of reproductive biology with specific interests in sperm RNAs and epigenome.

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Polymorphism journal wishes all its readers a happy and prosperous new year. The journal is now four years old and entering its 5th year. We express our gratitude to the esteemed editorial board, the reviewers and our authors whose enthusiasm has kept it going. The journal has published several articles and a few special issues in diverse fields including Genes and Oral Health, Genetic and Genomics of Cancer and Ancient DNA. We are at a fast pace of establishing ourselves as one of the leading journals publishing polymorphism research.

In its regular issues, the journal has seen a surgency of articles in the specialty of preimplantation genetic testing. Discovered a decade ago by Prof. Alan Handyside, it was applied to screen the embryos for its genetic sex in couples with a history of X-linked disorders. The idea was to identify XX embryos in couples undergoing assisted reproduction and only transfer these embryos so that the couples conceive a female child always free of the disease. Since then, the field has expanded exponentially and preimplantation genetic testing (PGT) is being increasingly applied in the reproductive medicine. From its very invasive nature where biopsy of a few cells is required for collection of DNA, we now report the possibility of using cell free DNA in the culture medium for screening of genetic aneuploidy in in vitro cultured embryo, making the process truly non-invasive. The first live birth by non-invasive PGT screen is reported in the current issue of Polymorphism (http://www.peerpublishers.com/index.php/snp/article/view/72).

From its original applications in embryo sexing for medical reasons, in the current scenario PGT is increasingly used for testing embryos for social reasons. In 2020, the journal reported the birth of healthy twins in a case of Hypochondroplasia (http://peerpublishers.com/index.php/snp/article/view/46). It is a form of skeletal dysplasia characterized by macrocephaly, short stature, stocky build, disproportionately short arms and legs, broad and short hands and feet and caused due to mutations in the *FGFR3* gene with an autosomal dominant inheritance. The infertile couple was given a choice who opted for testing their embryos for FGFR3 mutations. Of the six embryos the couple had, four were genetically healthy and free of the said mutation. Transfer of the two embryos resulted in live births of two babies who are disease free. This is the first case report from India and second in the world. This case is interesting as for not just its rarity but it demonstrates the power of the technology in giving couples a new hope of having a healthy baby without the ordeal of undergoing abortions. However, hypochondroplasia is a mild disorder and many patients do not think of themselves as disabled. Also, the reproductive fitness of the patients with hypochondroplasia is not compromised. Thus, in its strictest sense, it is not a condition qualifying for the termination of pregnancy. However, some parents or would-be parents might consider short stature as a significant physical, emotional, and/or social disability and the associated developmental delay is bothersome to many couples. In such instances, prenatal diagnosis and abortion my not be socially or ethically acceptable, but PGT provides an excellent option.

The second case report in the ongoing issue of Polymorphism is the application of PGT specifically for the creation of genetically matched siblings who will act as a donor in the future (http://peerpublishers.com/index.php/snp/article/view/83). In this case, the couple had a child with thalassemia major who could be cured by replacement of the bone marrow from a healthy matched donor. Herein, the couple opted to

produce a second sibling who was not just thalassemia free and chromosomally healthy but also had the same HLA profile as that of the sibling. The idea was that the younger sibling will act as a donor for the older sibling. By applying a series of genetic tests on several 5-day old blastocysts, a single HLA matched euploid embryo (out of 16 tested) that was heterozygous for the thalassemia mutation was identified and transfer of this embryo resulted in the birth of a healthy baby. This saviour sibling will be the future donor of bone marrow cells at the age of two years. In both these cases, the application of the genetic technologies is now not the prevention of the birth of a genetically abnormal and non-viable baby or the birth of a baby with a severely compromised life conditions, rather, the purpose here are socially driven reasons and we are now entering newer realms of eugenics. While in the first case, the would-be parents are given an option of having a baby free of a perceived disability, the second case is that of creating a baby who will be born with an inherited pressure of altruism. Since in neither case, there is destruction of life (abortion of foetus), both these are within the ethical norms. However, in both instances, surplus embryos which potentially lead to the birth of healthy babies will be discarded. As a society, we will have to think of socially and morally relevant guidelines to handle such cases. While couples should not be deprived of having healthy babies with desirable qualities even if they are for aesthetic or altruistic reasons, the creators of such embryos, mainly the IVF clinics, must protect the rights of the embryos. We are unclear what happens to the surplus embryos in such instances. There will be embryos with FGFR3 mutations which can give a dwarf, but otherwise a healthy baby. The non-HLA matched healthy embryos will be perfectly healthy babies if implanted and born alive.

Historically, the eugenics movement is thoroughly discredited on ethical, moral and scientific grounds, but the social need of genetically improving humans remains relevant. The emergence of new genetic technologies and their applications often demand fresh debate. Can eugenic ideas be dissociated from the evils of the past and pursued through renewed means? Against the selection of individuals with desirable traits in the classical eugenics, a new, morally responsible eugenics must be considered on its own terms. Whether these case reports represent a "new" form of eugenics need to be asked. Without asking these questions, the ethics of genetic technologies and the new eugenics will be far from settled. What we need is a framework towards the same. It is the time when geneticists, bioethicists, counsellors, doctors, embryologists and couples should come together and discuss the framework which must be applied to "new" eugenics where the patient is a mass of cells whose fate is to be decided in the most responsible way without compromising the principles of eugenics. It's us who will have to pave the way for the future generations and a society which is healthy yet responsible. We congratulate and thank the authors of both these papers for reporting their interesting cases and throwing light on the growing dimensions of eugenics.

We once again wish our readers a Happy New Year and look forward to their renewed support in making this young journal a success.