1. **Ethics clearance from your institutional EC.**

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| There is no Ethics committee at our institute. The authors are solely responsible for authenticity of data & research undertaken, and ethics related issues. |

1. **Details of how the literature search was done?**

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| Electronic databases such as PubMed, Google Scholar were searched to collect data on key ethical and social issues in laboratory medicine focusing on molecular diagnostics. The literature related to Infectious Disease Molecular testing and/or Molecular Oncology and/or clinical genetic testing and/or genetic counselling and ethical and/or social issues of these Molecular services, performed in India; was collected. The issues mentioned in the Result section of the article were identified through literature survey. The knowledge of some suggestions in the Discussion and Conclusion section is also from literature survey. |

1. **How the five doctors were selected?**

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| We searched all the major Government & Private Hospitals, and Medical Institutes in Delhi where the services of different areas of Molecular Diagnostics namely, Diagnostics of Infectious Disease, HLA typing, Genetics and Genomics, Department of Biochemistry, Laboratory Medicine, Community Medicine, Fetal Medicine & Medical Genetics, were present. Hospitals were contacted to check whether the departments were functional or not. Six major Hospitals were identified and selected where these departments were existing and operational. Concerned Doctors were contacted and out of these six, five responded positive and they were interviewed to gain insight into the ethical & social issues and possible ways to deal with them. |

1. **What questions were asked in the interviews?**

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| 1. What types of tests are performed in your laboratory related to Molecular Diagnostics (e.g. genetic, paternity, infectious disease, so forth)? 2. Since molecular diagnostics is an emerging field in your opinion what are the common ethical dilemmas/ problems faced in your daily practice? 3. Can you give some incidences for the same 4. Do you think adhering to these ethical issues is important for Diagnostician? 5. Let’s say there is a patient with terminal or pre-terminal diagnosis, do you know if practices like hiding the information from a patient about a in an effort to bolster their spirit or attitude, are being followed in our country? 6. What is your opinion on the practice that is it acceptable to cover up or avoid revealing a mistake if that mistake would not cause harm to the patient? 7. What you think is it acceptable to discuss patient information in situations that did not fully protect their privacy, e.g., socially or while talking with doctors about subjects not related to the patient? 8. Is it acceptable to break patient confidentiality if you know that a patient’s health status may be harming others? 9. Do you think Education of ethics to the health professionals is important? 10. Do you think Education to masses about various advancements in Science and taboos should be given at elementary level education system? 11. Setup of ethic committees in all health organization can help solving the problem? 12. Do you have an ethics department? 13. Do you think Ethics must change depending on society? 14. In comparison to developed nations where do you think India stands in terms of ethical policies 15. What common ethical dilemmas you face and challenges you face dealing with various issues? 16. Set of Ethic rules must change depending on technology? 17. Are there any ethical issues specific to molecular diagnostics that cannot be seen in other healthcare departments? 18. Do Ethic rules change depending on conflicts? 19. Do you treat a patient potential of having a communicable disease differently? 20. How stringent is our system in maintaining privacy and confidentiality of patients? 21. If a patient has a communicable disease e.g. AIDS but he fails to disclose the information, it is definitely an issue for the doctor’s health, when information is sought merely for the protection of the doctor the search for information becomes perverse, unethical and immoral. 22. Practice of paternalism is followed? 23. The situation in which the assumed father is not the biological father. Whether to disclose the information and if so to whom. 24. Testing of someone who has not reached the age of majority for a condition that generally has onset in adulthood and for which no effective prevention is available. Whether it is acceptable to override the child’s future autonomy (for immature minors); whether testing will cause harm to the child (for immature and mature minors). 25. Whether it is acceptable to test competent individuals without informed consent; whether the needs of a third party can ever outweigh those of the person being tested. 26. In case of monozygotic twins, whether the right of one twin to know his/her genetic status outweighs the right of the other twin to not know. 27. Whether the right of a relative not to know his/her genetic status outweighs the right of the person wanting a genetic test. 28. Whether parents have the right to produce a child for the purpose of saving another child; the potential for harm to the children. 29. Whether the client has a right to confidentiality in circumstances in which there is considerable potential for harm to known relatives if information is not shared. 30. How do you tackle incidental findings? 31. Since we are living in a country which has highest number of people living below poverty line, so what do you think more investment should be made in advancements in genomics and related fields or better providing basic healthcare for better living standards, what should be our utmost priority? 32. Some argue that the diagnosis of a future terminal illness, with no available treatment, should be kept from the patient. True or false? 33. Sometimes due to religious believes people reject certain medical procedures so what stand a diagnostician takes at that point? 34. What are your suggestions to tackle various ethical and social issues in the field? 35. What are your suggestions for government to tackle such issues? |

1. **Separate details of findings in the literature survey and interviews.**

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| All the findings of the literature survey and interview are included in the manuscript under Result and Discussion & Conclusion section.    The issues included in the Result section were identified through literature survey and the opinions of the experts.   1. **Issue from literature survey:** The high cost of molecular diagnostics and its relevance for Indian scenario: Looking at the lack of even basic healthcare facilities the question comes whether the country should focus on channelizing resources for the otherwise costly area of molecular diagnostics. But at the same time if a developing country like India focuses on providing only the basic amenities to the public, it is good for the people, helping them leading a life with some respect and dignity, but another fear that overrides is that taking a step back from genomic sciences will create a gap between developed and developing nations. The question of priority, competing for needs and rights & obligations come into picture.   **Experts view:** diagnostics is an inevitable and very important part of the healthcare system, unless and until a proper diagnosis is made regarding any disease whether rare or common, proper treatment and cure cannot be provided. Since the diseases which have their roots in the genetic composition can be effectively be diagnosed by molecular diagnostics, therefore, the country should invest in this field for right diagnosis and treatment & management of such diseases.   1. **Issue from literature survey**: Issue for continuance as a medical professional in case of disorder:Diagnosing a medical professional with a disorder which might affect his/her functioning in future, gives rise to the question as to for how long the concerned professional be allowed to continue in the profession. The health and proper mental balance of medical professional are important for proper treatments of his/her patients. Whether there are any provisions for periodic health check-ups of medical professionals currently existing in India or not.   **Experts view**: As per expert views, the competence of medical professional diagnosed, even in the early stages, with such diseases may be compromised. It is suggested to get tested for Neurological disorders after the age of 40 years to ensure that they are able to perform surgery effectively. Currently, no such provision or use of molecular diagnostics is done for the medical professional in India or elsewhere in the world.   1. **Issue from literature survey:** Issue of age for informed consent:The issue of informed consent is important in the field of molecular diagnostics because the information revealed by these diagnostic tests can be life changing for an individual but taking informed consent also poses challenges, in case of a minor the consent for molecular diagnostic is taken from the parents/guardians, but in case of adult (18 years or above) the consent of the individual is sought, here the question comes why the consent age is kept at 18 as the cognitive development of an individual between the age of 15-17 is same as that of a person at the age of 18 years. So the question of informed consent or conveying the result of the test to the individual should be dependent upon age or cognitive capacity of the individual. 2. **Issue from literature survey:** Issue of conveying the results to the parents for prenatal tests:A case came across during the study where the brother of a pregnant woman was affected with Duchenne Muscular Dystrophy (DMD) and died at the age of 20 years. During the prenatal diagnosis of the fetus it the pregnant lady was diagnosed as a carrier of DMD but this information was not disclosed to the couple as the only female fetus can be the carrier of DMD. So conveying the information about the fetus being the carrier of DMD would have disclosed the sex of the fetus and since conveying the sex of the fetus is prohibited in India, this information was concealed from the parents rather they were informed that the fetus was healthy and did not suffer from DMD. The couple was not satisfied with this information and insisted on knowing the carrier status if any, as they did not want a baby which would be a carrier of the DMD mutation. The parents argued that a carrier female would face difficulty in finding a husband and would subsequently require invasive, inconvenient, and costly tests during pregnancy. The laboratory refused to disclose the information requested by the couple because Indian law is clear that the sex of the fetus should not be disclosed, except in cases where the information relates to the diagnosis of a genetic disease. This left the couple in dilemma and stress till the baby was born and adding up on the cost for further analysis later. Such situation requires flexibility in the law for the emotional and economic stabilization of the expecting parents.   **Expert views:** Sex selection is a crime in India because of the social stigmas attached to the birth of girl child. Conveying the information whether the fetus is a carrier of certain disease or not may be done at the cost of conveying the sex of the fetus. But provisions may be kept for revealing the carrier status for certain disease even if it amounts to convey the sex of the fetus.   1. **Issue from literature survey:** Issue of conveying the carrier status: Molecular diagnostics presents effective techniques and tools to determine whether a person is a carrier for a particular disease or not. But the question is whether this should be conveyed to the concerned individual or not that she/he is the carrier of the disease or not; or whether such tests may be done at all or not. There is no consensus on whether to withhold such information on a minor until he or she is of a consenting age to receive the data personally. Some policies discourage genetic testing of asymptomatic minors for adult-onset conditions. Molecular diagnostics enable us to know what future holds for us in terms of health. If preventive measures are available, the predictive knowledge seems helpful but what if there are no effective preventive options of the probable diagnosis.   **Expert views: N**o tests should be done to determine carrier status and if at all future onset of a particular disease is diagnosed with the help of molecular diagnostics then the information has to be withheld till the person attains legal age of consent. Also, there is an agreement among genetic specialists that children should not be tested to determine their carrier status. The only case in which genetic testing on children is possible is when predictive tests may have beneficial medical effects for the children. If children are aware of their illness from an early age, they may suffer detrimental effects such as the feeling of humiliation and frustration. If nothing can be done right at the moment then it is better to wait till the child gains maturity to make an informed decision.   1. **Issue from literature survey:** Issue of revealing the information in case of non-curable disease: Diagnosis of a future terminal illness where no treatment is available should be withheld from the patient. There is a dilemma about conveying the results of such diagnosis as it could result in depression resulting from loss of hope which may result in suicide; in case the results are not conveyed then the patient will treat himself/herself as normal and will not get aware of the fact that he/she has limited time and will not be able to plan for their loved ones/family as per the time.   **Expert views**: If a person has reached the legal age of consent then it is his/ her right to know about the situation whether good or bad, moreover, it is necessary for proper management of the remaining life with the help of counselling.   1. **Issue from literature survey:** Issue of privacy in genetic but non-infectious disease:Privacy has always been an issue in the field of diagnostics, this assumes additional importance in case of molecular diagnostics as molecular diagnostics enables to predict future onset of many serious diseases. So the question comes whether the result of such diagnosis should be conveyed to the blood relatives of the concerned person or not as they may also be at equal risk of developing the similar disease due same genetic makeup. Revealing the results out might affect an individual’s career or marriage related prospects. E.g. determining the possibility of breast cancer in a woman might endanger future career or marriage prospects of a woman.   **Experts view**: Patient’s consent should be respected in such cases, if the patient does not want the results to be disclosed even to blood relatives, and then these should not be conveyed.   1. **Issue from literature survey:** Issue of privacy & confidentiality in infectious disease: In case of molecular diagnostic done for the infectious disease the question of whether or not the health worker should inform the identifiable third parties associated with the patient, assumes significance. On the one hand, notification to third parties about a patient’s health status would breach the patient's right to confidentiality. On the other hand, failure to warn could conflict with the innocent third party’s right to life - which many would say is more important than the incautious patient’s right to confidentiality.   **Expert views**: local authorities should be informed timely in case of infectious diseases like dengue, in case of infectious diseases like Syphilis where the associated third party is at risk, they must be informed.   1. **Issue from literature survey:** Issue of collective or individual decision making: India is a community-based society where family members play a prominent role in decision making of an individual. Here individual although has its own independent identity, he/she is also intimately integrated with extended family, caste group, and geographical locality, necessitating a holistic approach in the matters where individual’s consent is mandatory for decision making. This gives rise to dilemma whether the individual’s family should be involved before taking a decision on the issues related to disclosing the results or even to conduct the tests. In certain cases family members are involved in decision-making process. E.g. a pregnant woman carrying a child with a genetic disorder often creates a stigma for the mother while actually there is no fault of hers.   **Experts view:** In such cases, proper counselling of entire family is important to avoid any such issues.   1. **Issue from literature survey:** Issues related to religious Beliefs: Religious beliefs may affect attitude to medical genetic services particularly related to abortion or prenatal diagnosis or premarital screening. In such circumstances the decision of the individual may be treated as final.   **Experts view:** Counselling of the concerned person should be done to explain the benefits of such diagnostics and practices but nothing can be done against patients will.   1. **Issue from literature survey:** R & D investment in the field of molecular diagnostics: Rare diseases include auto-immune disorders, rare infectious diseases, and rare cancers; India lacks in proper facilities for diagnostics of many diseases. For the diagnostics of many such diseases, the samples are sent abroad resulting in huge delay and waiting time. There is a high dependency on import for devices which can be used for molecular diagnostics leading to unaffordability and unavailability to the masses. Thorough Research & Development investment & Policy framework for indigenous development and manufacturing of such devices should be created. Some areas in the country are clutches of certain genetic diseases which need molecular diagnostics for proper identification and treatment, e.g. Sickle cell anemia is prevalent in tribal areas in Madhya Pradesh. Each state in the country should create a mapping of the region-wise prevalence of such diseases. 2. **Issue from literature survey:** Issue of Coverage of molecular diagnostics under medical claim and Commercial availability of tests: Molecular diagnostic techniques are directly available to the consumers in western countries where an individual can directly approach any center for such services but in India such practice’s haven’t advanced enough & most of the times doctor’s prescription is required if one needs to undertake such tests.   **Expert views:** For making these tests directly available to the individual literacy and awareness level in the country has to be increased. In western countries, the medical insurance covers the cost of such tests but in India, these tests are not covered under medical claim, thus no check at the need and the demand of testing is made.  Lastly, **measures to avoid Moral Distress, Social Stigma & Discrimination were included again from the knowledge of experts, and literature searches in the Discussion & Conclusion section**.   1. Although there are set rules and guidelines in hospitals and medical institutions with regard to ethical practices but a separate dedicated section/department needs to be created headed by experts from the related domain capable of dealing with ethical issues arising out of molecular diagnostics as this is an emerging area and sometimes complex situations are faced because of the results coming out of such diagnostics. 2. An employee hired for molecular diagnostics should undergo proper orientation program to deal with situations arising out of molecular diagnostics. They should be provided with ample literature related to ethics in the context of molecular diagnostics. Behavior-based interviews should be organized to assess their capability to deal with people by giving certain situations. There should be a complete background check to assess ethical and moral orientation of the employee before hiring. 3. The process for dealing with ethics violations should be streamlined. Most of the Hospitals and research setups have ethical committees to look after ethical issues faced by the organization, which generally comprises of Doctors and healthcare workers. But hospitals should appoint professionals in ethics who could help their colleagues in their day-to-day concerns on ethical issues. These ethical professionals should be available to clinicians who can confidentially talk with them as and when needed. 4. It is suggested to get a complete medical diagnosis, based on molecular diagnostics for neurological diseases, to analyze health of professionals and to check their competence to take decisions and perform surgeries after the age of 40. 5. In India direct selling of molecular diagnostics services are advancing where a person can approach them directly for molecular diagnostics without requiring the prescription of doctors; such practices for a country like India should be discouraged as people are not literate enough to understand the consequence of such tests or to properly interpret the results. 6. State Governments should collect Epidemiological data with respect to genetic and infectious diseases specific to geographical areas covered under them. This will help in focused research and clinical trials with respect the local disease. These state Governments can then organize awareness, outreach and education programs with respect to ethical issues emerging out of molecular diagnostics with such diseases specific to that area. 7. Prenatal screening during the permissible period of abortion or new-born screening should be made mandatory to reduce the future burden of disease on child, parents, and society. 8. Specialized centers focused on research, diagnosis, treatment, and counselling should be set up. 9. Specialized courses such as PhD. in genetic counselling should be promoted and more and more genetic counsellors should be recruited in specialized centers. 10. As per experts views present health insurance doesn’t cover genetic medical conditions completely. Complete health insurance should be provided to patients with genetic medical conditions including diagnosis, treatment, and care. 11. The government may think of providing financial aid for specific cases, for diagnosis/ treatment from abroad, if no option is available in India. 12. Investment in the research on genetic diseases and development of devices needed for diagnostics should be promoted. At present most of the data available for research in this area does not belong to India. Most of the drugs and treatment techniques are developed outside India. Indigenous research can lead to cheaper techniques and easy availability. Special incentives can be provided to the Indigenous market for promotion. 13. Internet-based networks can be formed where Researchers, Doctors, and other health professionals can exchange and share views on ethical and social issues related to molecular diagnostics for smoother and better system in the society. |