

important cause of morbidity and mortality

Multi-factorial Disorder

This result from a combination of multiple genetic and environmental causes. Many birth defects such as cleft lip and / or cleft palate as well as many adult disorders, including heart disease belong to this category⁴.

Ante-natal and post natal screening can help in early detection of genetic diseases which will help in timely health care. Various indications are as follows:

1. In *prenatal cases* fetal cells obtained by amniocentesis or chorionic villus biopsy for investigation are carried out at advance genetic centers. The common indications for such investigations are – Advanced maternal age (more than 34 years), A parent with a structural chromosomal abnormality (e.g. robertsonian translocation), Previous child with chromosomal abnormality and in patients who is carrier of an X-linked disease (to determine fetal sex).
2. In *postnatal cases investigations* are performed on peripheral blood lymphocyte culture for diagnosis and can be conducted at district hospital and medical colleges in urban and rural areas. The indications for this investigation are multiple congenital abnormalities, unexplained mental retardation, and suspected chromosomal abnormalities, suspected fragile X syndrome, ambiguous genitalia, certain cancers, infertility to rule out sex chromosomal abnormality and recurrent abortion (both parents must be evaluated to rule out carriers of balanced translocation)⁵.

The patients can be referred for molecular Analysis at specialized centers after primary screening for the diagnosis of genetic diseases by recombinant DNA technology:

GENETIC ETHICS

Genetic information provides a unique type of knowledge about an individual and his/her family, fundamentally different than a typically laboratory test that provides a "snapshot" of an individual's health status. The unique status of genetic information and inherited disease has a number of ramifications with regard to ethical, legal, and societal concerns⁶. The patient should be made to understand the implications of genetic tests and should be provided information regarding the benefits, the effectiveness, the risks and the alternatives.

AUTHOR'S EXPERIENCE

Working as in-charge of genetic laboratory functional under department of Anatomy at Government Medical College Nagpur, we investigated the suspected cases of chromosomal disorder referred from various clinical out patients department. The average patient seen per year was seventy (70) during 1990 of by syndrome, 14 cases of Turner's syndrome, 4 cases of Klein-felter syndrome, 4 cases of sex reversal like XY female and XX males and 4 cases of autosomal translocation. Later at NKP Salve Institute of Medical Science and Lata Mangeshkar Hospital Nagpur within a period of two months after establishing the cytogenetic lab in anatomy department, out of twelve (12) cases investigated, four (4) cases were positive and these

were diagnosed as Turners. Down's, translocation and XX males. As genetic lab facilities are not available in most of the medical colleges so also trained personnel are not available, neither standard guidelines are available to establish genetic lab in new medical colleges, this gave us impetus as the principal author along with my colleague in the department of Physiology who has been focusing his teaching on genetic control over cellular physiological functions to evaluate the need of fully functional genetic lab in a medical college.

The co-author who has been monitoring academic administration at NKP Salve Institute of Medical Sciences and Lata Mangeshkar Hospital, Nagpur feels that the medical curriculum needs modification with theoretical and practical exposure to basics in Genetic Medicine.

SUGGESTIONS

- 1) Due to India's large population, many rare disorders exist in India. These families can contribute to identification of genetic defects of rare genetic disorders. These are mostly seen in rural areas but remain undiagnosed due to lack of facilities and awareness. Identification of cases with possible genetic etiology by primary care physicians is essential so that these families can get facilities of special genetic tests; genetic counseling and prenatal diagnosis, the primary health physician and General Medical Officers should be imparted special training in identifying genetic disorders and be made aware of investigation modalities.
- 2) Medical curriculum for the student of medicine needs modification with more theoretical and practical exposure. These basic medical graduate doctors should be well updated with knowledge so as to be to identify genetic disorders and do the genetic counseling for further referral at specialized centers.
- 3) To address the lack of awareness in general population about genetic disorders, their screening and diagnostic tests, there should be awareness campaign.
- 4) Though the National Accreditation Board for Testing and Calibration Laboratories (NABL), has started accreditation of all laboratories including genetic labs Many private labs have taken up the diagnostic testing without any proper guidelines resulting in misdiagnosis. All genetic labs in the country should follow guidelines for certifying genetic diseases as per guidelines of NABL.

RECOMMENDED READING

- 1) **Verma I.C.** Burden of genetic disorders in India. *Indian J. Pediatr* 2001; Jan; 68(1):25.
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- 3) **Van den Daele, W.** The spectre of coercion: Is public health genetics the route to policies of enforced disease prevention *Community Genetics* 2006; 9; 40-49.
- 4) **Jorde Carey, Bamshed White.** *Medical Genetics. 3rd Edition; Elsevier Publication* 2003.
- 5) **Berg van den, M, Timmermans, D.R. M., ten Kate, L. P. et al.** Are pregnant women making informed choices about prenatal screening *Genetics IN Medicine* May/June 2005; 7(5):332-338.
- 6) **Holtzman N.A., Shapiro D.** Genetic testing and public policy *British Medical Journal* 1998; 316852-856

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