

## Genetic Lab - A Necessity in Medical Colleges and District Hospitals.

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**Abstract:** In view of increase turn over of patient's suspected of genetic diseases for basic investigation in existing genetic labs the author with their experience in screening genetic diseases and so also while imparting teaching regarding genetic disorders to medical students opine regardign situational need to start genetic facility at district hospital and all upcoming medical colleges in India. The basic investigations may be done by setting a cyto-genetic lab with minimal essential requirement and the help of biochemistry lab in the hospital. Authors also suggest the need to modify the curriculum of medical education by including detail aspects of medical genetic at undergraduate level so that a basic medical graduate doctor will have sufficient knowledge in diagnosis and giving genetic counseling at urban and even rural setup so that the patients with genetic disorder after screening will be referred to specialized genetic centre. Analysis of the present screening status in the country with a basic insight regarding genetic diseases has been highlighted.

**Key Words:** Genetic diseases, Screening Counseling, Medical Science Colleges, District Hospital

### INTRODUCTION

To describe the challenges facing India regarding the appropriate clinical use of genetics in health care systems in countries where infant mortality rate has fallen to a range where genetic and congenital disorders contribute substantially to ongoing handicap and early mortality, is a challenging task. India, like other developing countries, is facing an accelerating demographic switch to non-communicable diseases. Due to the high birth rate in India a very large number of infants with genetic disorders are born every year, almost half a million, with malformaiton and 21,000 with Down syndrome. In a multi-centric study on the causes of referral for genetic counseling the top four disorders were repeated abortions (12.4%), identifiable syndromes (12.1%), chromosomal disorders (11.3%) and mental retardation (11%). In a more recent study in a private hospital the top reasons for referal were reproductive genetic (38.9%)—comprising prenatal diagnosis, recurrent abortion, infertility and Torch infection—mental retardation +/- multiple congenital anomalies (16.1%) down syndrome (9.1%), thalassemia / haemophilia (8.8%), and muscle dystrome/spinal muscular atrophy (8.4%).

A recent study carried out in three centers (Mumbai, Delhi and Baroda) on 94,610 newborns by using a uniforma showed a malformation frequency of 2.03%, the commonest malformtions are neural tube defects and musculo-skeletal disorders. The frequency of Down syndrome among 94,610 was 0.87 per 1000 or 1 per 1150. Screening of 112,269 newbornns for aminoacid disorders showed four disorders showed four disorders to the commonest-tyrosinemia, maple syrup urine disease and phenylketonuria<sup>1,2</sup>. Epiendemiologists and geneticists claim that genetics has an increasing role to paly in public health policies and programs in the future. Within this perspective, genetic testing and screening are instrumental in avoiding the birth of children whiht serious, costly or untreatable disorders<sup>3</sup>.

### GENETIC LAB FACILITIES IN INDIA

The genetic lab facilities are available in various metropolitans' citis in India. Few well established labs which conduct investigation and research are

1. *Genetic Dignostic Centre*, Mumbai The Centre provides genetic counseling, cytogenetic test from blood chiruib villous sampling amniotic fluid cell cultures, Fetal blood sampling Products of

conception among other The centre also offers training courses for individuals interested in pursuing cytogenetics as a profession and also provides guidance for setting up of the new laloratory.

2. The *cancer cytogenetic laloratory* at Tata Memorial Hospital is engaged in basic and applied research programmes in malignancies.
3. Some of the main subjects of study in *medical genetic department of SGPGI*, Lucknow are clinical delineation of new multiple malformation syndromes, not yet reported in the medical literature.
4. *Centre for DNA Fingerprinting and Diagnostics (CDFD)*, Hyderabad is engaged mainly in provinding human cytogenetics for a variety of disorders such as chromosomal defects in struture and in number.

But our primary concern is that these basic investigation facilities for genetic diseases should be available in all existing and newly opening medical colleges and district hospitals.

### CLASSIFICATION OF GENETIC DISEASES

Genetic diseases are classified as: Each human is estimated to have approximately 30,000 to 40,000 different genes. Aterations in these gense or in combination of them can produce genetic disorders. These disorders are classified mainly into three groups:-

#### *Single Gene Disorder*

It is to alteration in single gene. Often called Mendelin conditions, well known examples include Cystic fibrosis, Sickle cell disease, Haemophillia the 2003 on line edition of Mckusicks *Mendelian Inheritance in Men* lists more than 14,000 known monogence traits of these more than 13,000 are located on autosomes, 778 are located on the X chromosome, and 43 are located on the Y chromosome.

#### *Chromosomal Disorders*

In the entire chromosomes (or large segment of them) are missing, duplicated or otherwise altered. These disorders include diseases such as Down syndrome and Turner's syndrome. Chromosomal abnormalities are for a significant proportion of genetic diseases occurring in approximately one of every 150 live birth. They are the leading known cause of both mental retardation and pregnancy loss Chromosomal abnormalities are seen in 50% of first trimester and 20% of second trimester spontaneous abortions. Thus they are an

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 category<sup>4</sup>.

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)<sup>5</sup>.

*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 concerns<sup>6</sup>. 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. Downs, 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**

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