Role of genetic awareness and counselling in prevention of genetic disorders in Eastern Uttar Pradesh

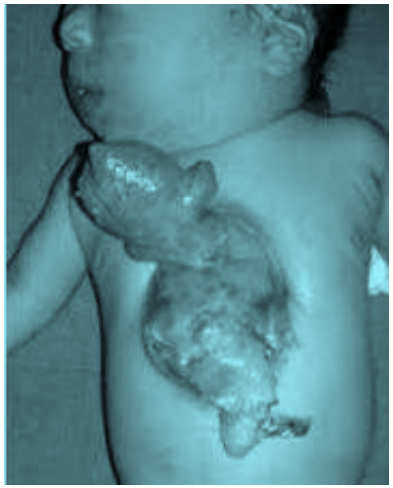
Prof Vandana Rai
Department of Biotechnology
VBS Purvanchal University
Jaunpur

Genetic Disorder

Disease or syndrome attributed to inherited genetic defects;

- Down Syndrome
- Fragile X Syndrome
- Neural Tube Defects
- Cleft Lip & Palat
- Thalessemia
- Hemoglobinopathies
- Schizophrenia
- Bipolar Disorder

https://www.istockphoto.com/photos/downsydrome?mediatype=photography&phrase=down%20syndrome&sort=mostpopular



https://en.wikipedia.org/wiki/Cleft_lip_and_cleft_palate

https://en.wikipedia.org/wiki/Neural_tube_defect

Global Scenario

- 3-5% of all births result in congenital malformation
- 0.5% of all newborns have a chromosomal abnormality
- 20-30% of all infants death are due to genetic disorders
- 11% of pediatric hospital admissions are for children with genetic disorders
- 10% of the chronic diseases (heart, diabetes, arthritis) which occur in the adult populations have a significant genetic component
- 50% of mental retardation has a genetic basis.

IC Verma,2000

India

- Congenital malformations and genetic disorders are the third commonest cause of mortality in neonates in Indian cities.

Genetic Disorder	Birth Per Year
Congenital Malformation	495000
G6PD Deficiency	390000
Down syndrome	21400
Beta Thalassemia	9000
Sickle cell Anemia	5200

IC Verma,2000

Factors influencing the high prevalence

- Consanguineous Marriages
- High Birth Rate
- Poor Governmental Support Facilities
- Lack of Expertise in Genetic Counseling

Preventive Strategies

- The affected child, once born, is the source of physical and emotional stress for his/her family members.
- The most plausible option is to use preventive strategies (there is no curative treatment for genetic disorders).
  - at macro level to cover the entire population and
  - at micro level for individual patients and their families

Preventive genetic medicine/community medicine

1. Base line epidemiological screening studies in the target population
2. Awareness campaign in the population about the target disorder
3. Genetic counseling of the patients and the family to make them aware about the impact of genetic disorder

Multispecialty Holistic Approach

The birth of affected child can be controlled by implementation of multispecialty holistic approach like-

- carrier screening,
- prenatal diagnosis,
- termination of affected fetuses
- genetic counseling,

This approach has been tried and found feasible in many developed countries.

Why Eastern Uttar Pradesh?

- The eastern UP population is very less explored genetically.
- Only few hospital based reports are available.
- There is a need for screening a large number of random samples as well as case samples.

Genetic screening

- Method of identifying individuals in a given population at high risk of having or transmitting a specific genetic disorder.

- Screening of rural Eastern UP population for following genetic/psychiatric disorders should be done

  - Down syndrome, Neural tube defects, Thalassemia, Hemoglobinopathies, Fragile X syndrome, Schizophrenia, Bipolar disorder etc.

- Screening rural population as well as genetic/psychiatric disorder patients for allelic variants of the following genes should be also done-

  MTHFR, MTRR, MTR, COMT, ACE, FMR1, beta-globin and alpha-globin etc.

Objectives of Human Molecular Genetics Group, VBS Purvanchal University, Jaunpur

- Generate an epidemiological database of this region.
- Aware the rural/affected population of eastern UP about the genetic defects.
- Counsel the family (which have an affected child/proband) with the help of a professional counsellor.

Awareness Programmes organized in Eastern UP

Health Awareness Campaign in Rural Women of Jaunpur District: Special Emphasis on Genetic Disorders

Genetic Awareness Campaign: Role of Folate and MTHFR, MTRR Polymorphism in Genetic Disorders.

Genetic Disorder Awareness Campaign in Purvanchal University for Women Students

Screening

- Ethical Clearance, IEC, VBS PU, Jaunpur
- An informed consent
- A brief clinical record
  - age
  - ethnic group
  - history of past illnesses

Methodology

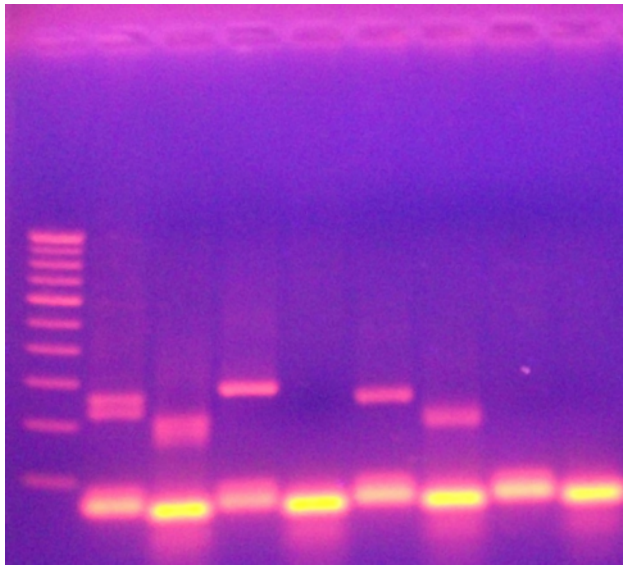
Blood Samples

- Patients with Mental retardation (Male & female)
- Relatives of patients
- Individuals randomly selected from the local Population
- Control sample (Healthy individuals without any family) History of mental retardation

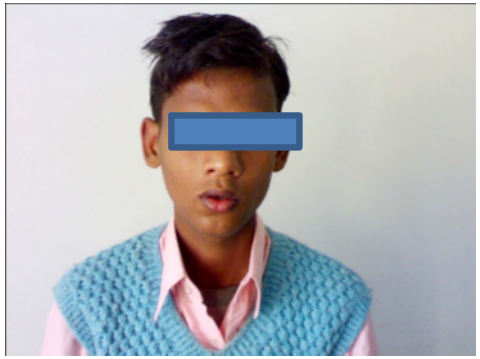
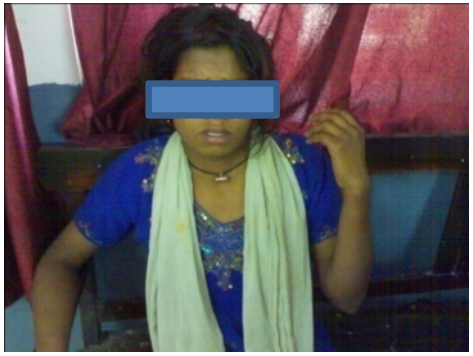
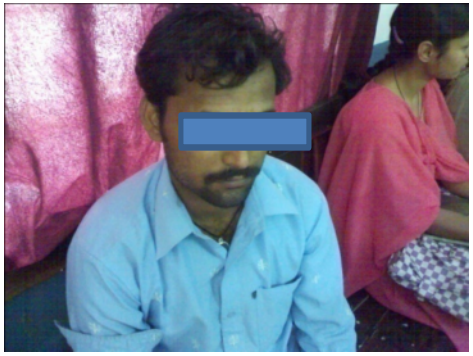
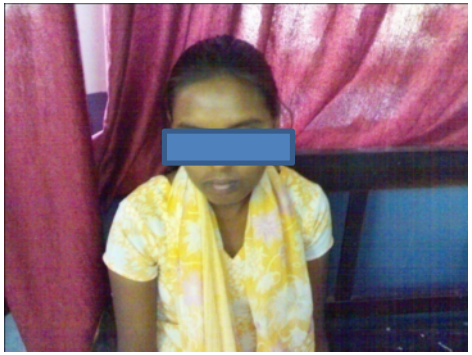
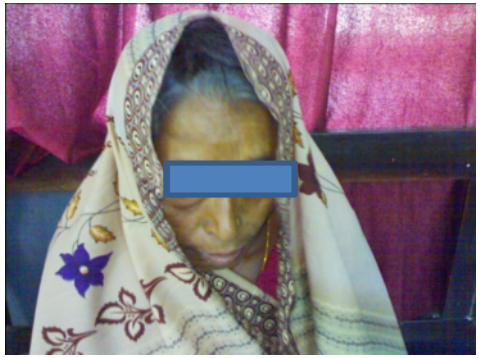
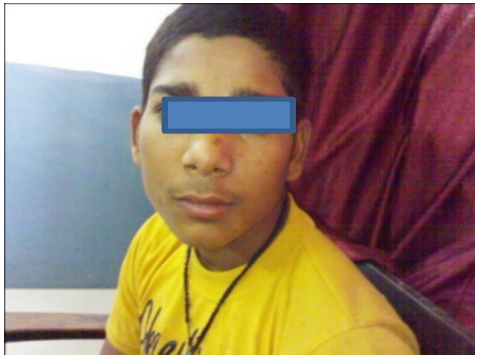
Genomic DNA Extraction

PCR amplification

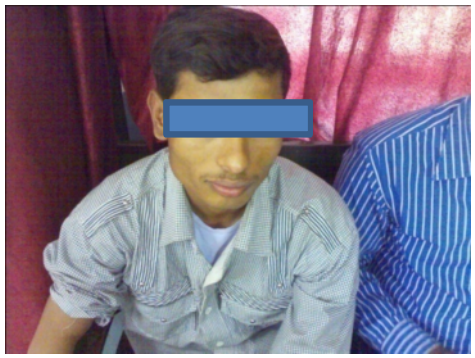
Electrophoresis

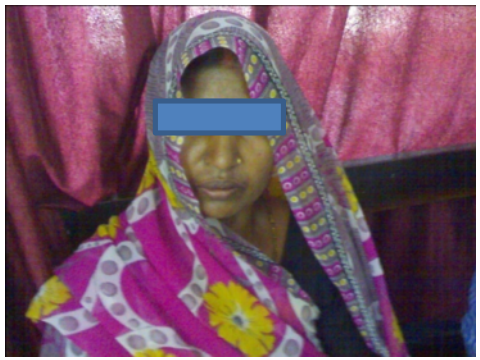
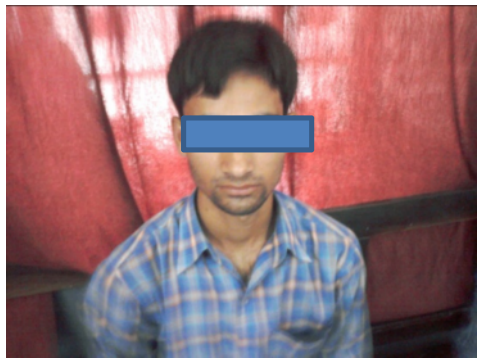


Schizophrenia Patients

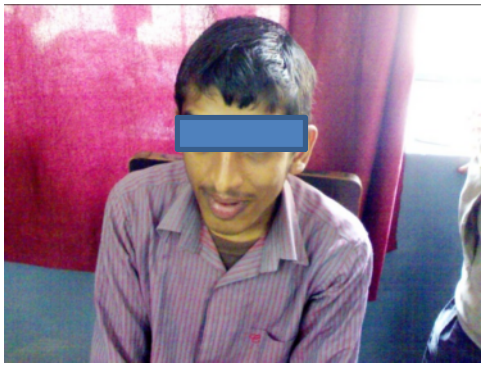
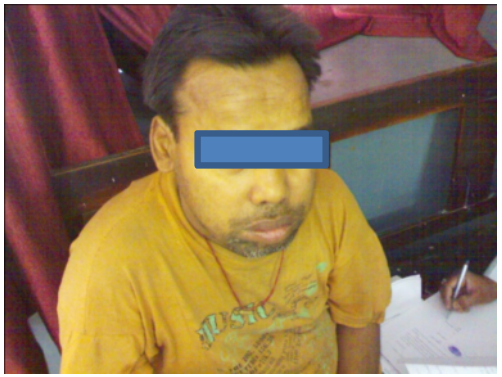
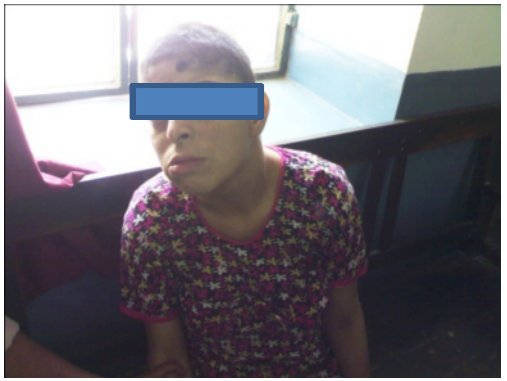







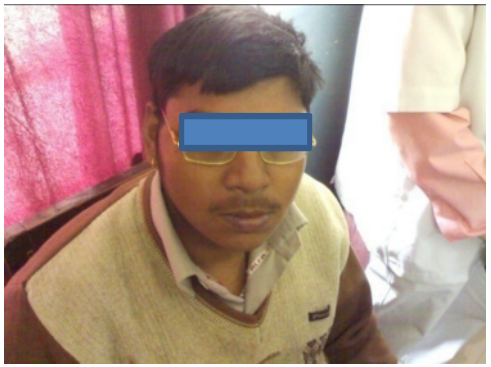
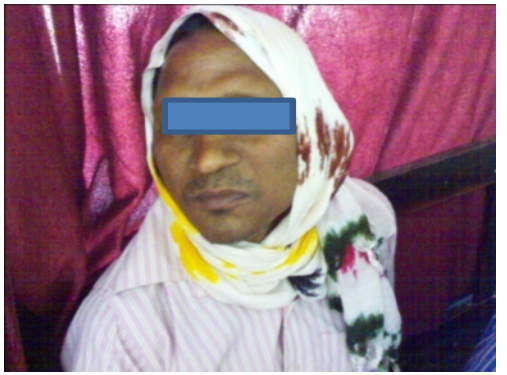
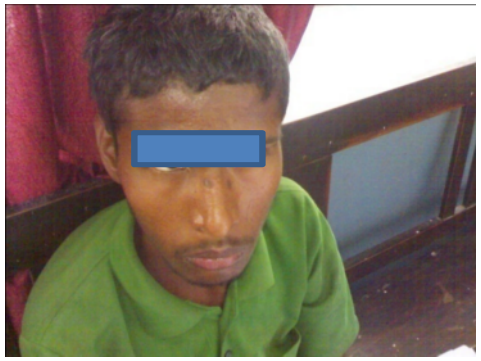
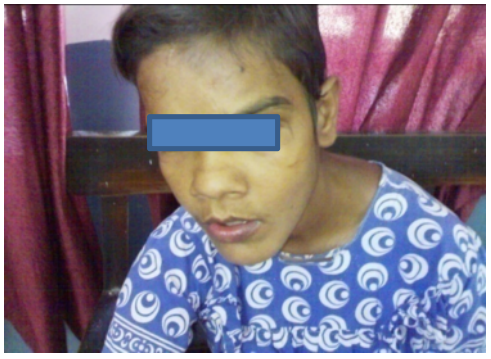
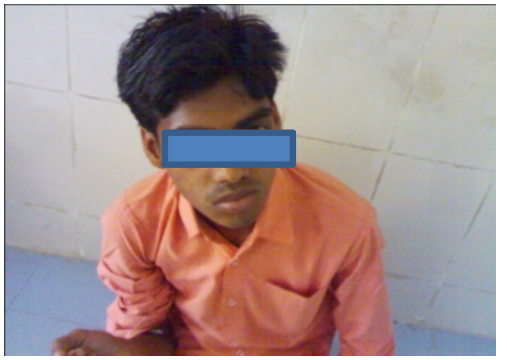
Depression Patients



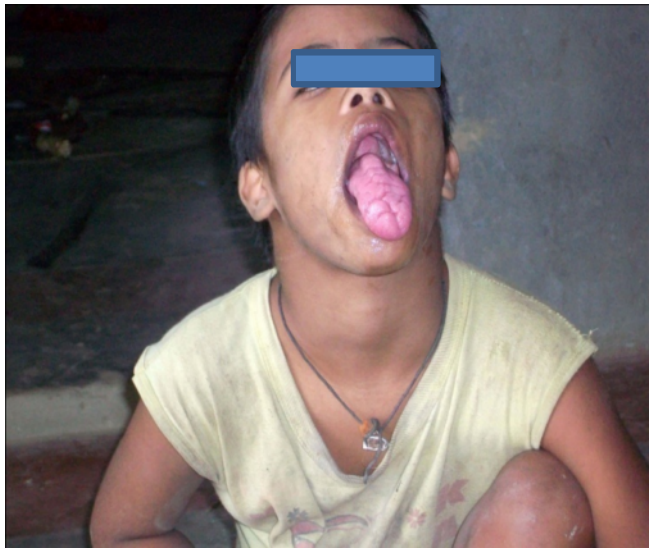
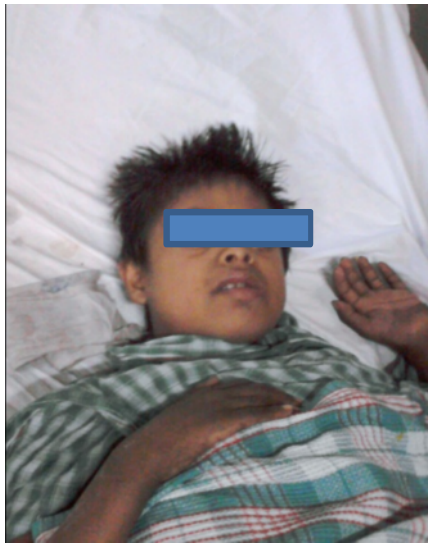
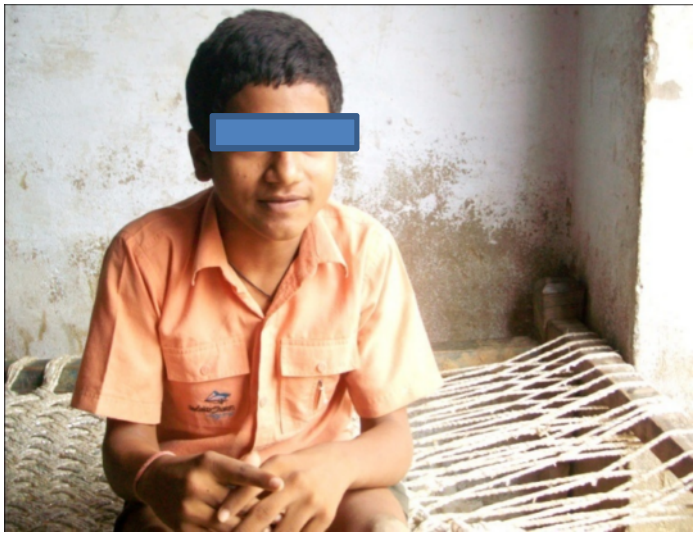





Mentally Retarded Patients

Down's Syndrome Patients

Collaboration

Collaboration/tie up should be done at different levels-

- primary health centers,
- District hospitals ,
- nearby medical college hospitals and
- Primary schools
- Sarva Shiksha Abhiyaan, Government of India

Genetic Counseling

- **Genetic counseling is a *communication process that deals with*** the human problems associated with the occurrence or risk of occurrence, of a genetic disorder in a family.
- The process aims to help the individual or family to:

understand:

- the diagnosis, prognosis and available management
- the genetic basis and chance of recurrence
- the options available (including genetic testing)

choose:

- the course of action appropriate to their personal and family situation

adjust:

- to the psychosocial impact of the genetic condition in the family

Counseling

Once the frequency of mutation is identified, the genetic counseling for high risk couples /women is the most urgent medical advice to be imparted.

Counsel the women and family.

- To improve the life quality of the proband (affected child).
- To reduce the financial burden of the parent because they go to quacks for treatment and waste their money,
- To encourage the parents of affected child to send their child to special School/rehabilitation centers.

Significance

- This type of studies will certainly help to
  - Prevent the recurrence of an affected child by advising women to go for prenatal diagnosis at nearby diagnostic center. (Women who are already having an affected child will have a 20-25 folds increased risk for subsequent child).

- Women with the family history of genetic disorder will be advised and encouraged to visit primary health centers for free check up and to take medical care for risk prevention (folic acid).

Need of the Hour

- A nation wide network of genetic centers capable of providing diagnosis, Counseling and pre-natal Diagnosis
- Development of trained manpower