



Panelists (l. to r.) Dr. (Mrs.) S. K. Nair, Dr. S. M. Merchant, Mr. P. K. Sukumaran, Dr. G. S. Mutalik, Dr. L. D. Sanghvi, Dr. K. G. Nair, Dr. R. A. Pai and Dr. S. M. Bhandarkar.

Knowledge of Genetics Vital For the Physician

BOMBAY

AN understanding of genetics can help the physician to better recognise, treat and prevent genetic diseases which today number more than 1,800, said the panelists who took part in a discussion on "Genetics and Medicine" here recently.

The panel discussion was held as part of the First Annual Conference of the Indian Society of Human Genetics and was chaired by Dr. G. S. Mutalik, Joint Director of Medical Education and Research, Maharashtra, and Founder-Secretary of the Indian Society of Human Genetics.

The panelists were Dr. S. M. Merchant (Professor of Pediatrics, Jerbai Wadia Hospital for Children, Bombay), Dr. K. G. Nair (Director-Professor of Medicine, K.E.M. Hospital, Bombay), Dr. (Mrs.) S. K. Nair (Tata Blood Bank, J. J. Group of Hospitals, Bombay), Dr. S. M. Bhandarkar (Endocrinologist, K.E.M. Hospital, Bombay), Dr. R. A. Pai (Indian Agricultural Research Institute, Pusa, Delhi) and Mr. P. K. Sukumaran (Indian Cancer Research Institute, Bombay). Dr. L. D. Sanghvi, President of the Indian Society of Human Genetics, was the expert.

In his introductory remarks Dr. Mutalik remarked that genetic disease can be represented by a spectrum with a broad part consisting of common diseases and a narrow sector consisting of pure Mendelian entities. Increasing number of genetic diseases have been recognised and documented. From 412 known genetic diseases in 1958 the number had increased to 1487 in 1966 and 1876 in 1971. In addition to the rare genetic syndromes which are inherited according to Mendelian Laws, genetic factor plays a significant role in common diseases, like obstructive emphysema, asthma and hypertension.

Significant advances have been made in the management and prevention of genetic disorders. Physicians therefore need to know more about genetics so that they can diagnose the disease, understand the prognosis and carry out treatment on scientific lines, Dr. K. G. Nair said. Detailed history taking often provided the first clues to diagnosis, clues such as consanguinity which are an important factor in transmission of genetic disorders in our country.

Dr. Sanghvi described the 3 broad categories of genetic conditions:

- 1) Diseases and defects inherited by simple genetic mechanisms. This includes dominant conditions such as malignant retinoblastomas, recessive conditions such as phenylketonuria and sex-linked diseases such as muscular dystrophy.
- 2) Chromosomal aberrations such as Down's syndrome,

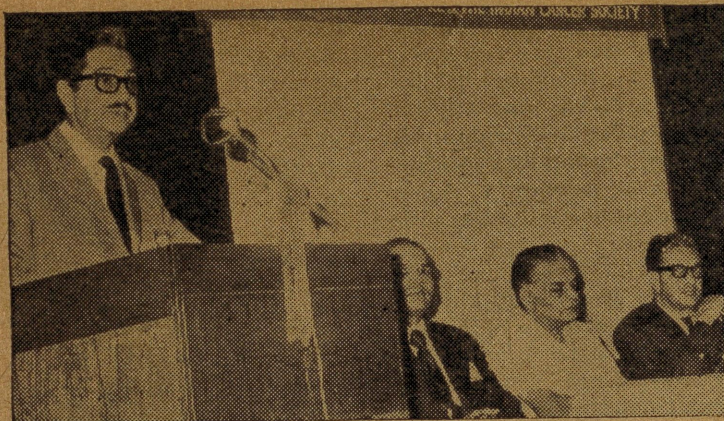
and
3) Congenital malformations, e.g. Fallot's tetralogy.

The normal human being has 46 chromosomes (23 pairs) grouped into 7 categories. The aberrations in this pattern, found in some cytogenic disorders, were described by Mr. Pai. Today techniques are available that can accurately pinpoint these abnormalities, he said.

Usually, the pediatrician is the

first to suspect a genetic abnormality. Some of the commonly encountered disorders in pediatric practice were mentioned by Dr. Merchant. These included achondroplasia, osteogenesis imperfecta, vitamin D resistant rickets (autosomal dominant) albinism, cystic fibrosis, galactosemia (autosomal recessive) and congenital deafness, hemophilia, G6PD deficiency (sex linked). The commonest chromosomal aberration was Down's syndrome.

With knowledge of genetics it is possible to predict, to some extent, the recurrence rate of certain diseases in certain families, Dr. Merchant added.



Dr. D. J. Jussawalla inaugurating the course on continuing education on oncology. Seated (l. to r.) Dr. A. R. Mehta and Dr. A. Srinivasan (both of Tata Memorial Hospital) and Mr. H. N. Sethna (Chairman of the Atomic Energy Commission).

Course on Continuing Education In Oncology Held

BOMBAY

EVERY MEDICAL COLLEGE hospital should set up a department of oncology and universities should establish chairs in oncology, Dr. D. J. Jussawalla, Director of the Tata Memorial Centre and Hon. Founder Secretary and Managing Trustee of the Indian Cancer Society, said here recently. He was addressing the opening session of the course on Continuing Education in Oncology.

"As a disease entity, cancer cuts across all the regional specialities currently practised in various branches of medicine and surgery. An oncologist is thus a true hybrid who is expected to have sound knowledge not only of general medicine and surgery, but also of radiotherapy, chemotherapy and nuclear medicine", said Dr. Jussawalla.

Mr. H. N. Sethna, Chairman of the Board of the Tata Memorial Centre and Atomic Energy Commission, in his inaugural address said that the course on Continu-

ing Education in Oncology was significant in that research workers and clinicians were joining hands to impart specialised knowledge to the practising doctors and teachers in medical colleges throughout India.

Held under the joint auspices of the Tata Memorial Centre and the Indian Cancer Society, the course comprised of 16 scientific sessions and was attended by about 120 delegates. As many as 55 specialists in the field participated in the course.

What are the effects of genetics on the endocrine system? These could be categorised into 3 groups, said Dr. Bhandarkar. The first group comprised of clearly identifiable biochemical disturbances, e.g. discreet deficiency of the thyroid enzyme or the pituitary growth hormone.

The second group is that of cytogenic disorders or chromosomal aberrations where the brunt falls on the gonads, e.g. as in Turner's syndrome, and the third group, perhaps the largest, consisted of endocrinological disorders with genetic overtones e.g. diabetes.

Referring specifically to hemoglobinopathies, Mr. Sukumaran said that the incidence of sickle cell anemia and thalassemia was fairly high in certain communities, e.g. the adibasis, and it was necessary to find out the magnitude of the problem before preventive measures could be undertaken.

It is generally agreed that preventive measures constitute the most effective approach to the control of genetic disorders. Some diseases can be detected as early as the 12th week of pregnancy by transabdominal amniocentesis. In the advanced countries, amniocentesis is performed during the second trimester as part of management of genetically high risk pregnancies followed by selective abortion if the fetus is found to carry the suspected disorder.

Another measure of importance in prevention of genetic disorders is genetic counselling, i.e. assessing for parents the chances of having another affected child.

Dr. (Mrs.) Nair felt that clinicians looking after the affected children are best suited to investigate the family history and to calculate the risk. She outlined the role of the trained social worker in genetic counselling and stressed the need to arrive at a very definite diagnosis. Having a family planning clinic close to the counselling centre is a help, she added.

Summarising the discussions, Dr. Mutalik said that the large number of genetic diseases which can be recognised today by amniocentesis in the prenatal period, has made way for precise genetic counselling and prevention of the birth of defective individuals. Though treatment is not a strong suit in medical genetics disease, certain principles have already emerged as guidelines to treatment. Elimination diets as in phenylketonuria or in galactosemia, supplementation diets as in orotic aciduria, supply of the missing gene products as AHG in hemophilia, substrate competition

India Offers Wide Scope For Genetic Studies

BOMBAY

INDIA, with its divergent population groups and ancient social customs, offers a rich field of study to the population geneticist, Dr. L. D. Sanghvi said in his presidential address at the First Annual Conference of the Indian Society of Human Genetics.

The 3-day conference, which was inaugurated by Maharashtra's Health Minister, Dr. Refiq Zakaria, was attended by delegates from all over the country.

Dr. G. S. Mutalik, Secretary of the Indian Society of Human Genetics, in his report traced the efforts made to bring together groups of scientists interested in the field of human genetics, which culminated in the first national congress on human genetics held in Poona in 1970. This congress was the first step towards the foundation of the present society. In view of the tremendous impact of the science of human genetics on all sciences in general and on medicine in particular it is time that a full-fledged institute of human genetics be founded in the country, he said.

Presidential Address

Geneticists agree, said Dr. Sanghvi, that the genetic burden carried by every society is maintained either by recurrent mutation or selective advantage of certain genes or genotype in specific environment. Most of the chromosomal aberrations are new occurrences, and both the parents in such cases are normal. On the other hand, hemoglobinopathies, particularly sickle cell anemia and thalassemia, are maintained by selective advantage of heterozygotes in malarial environment.

It is not yet known what proportion of genetic load is maintained by mutation and what proportion is maintained by selective advantage.

This problem can be investigated in India which has a unique population structure, Dr. Sanghvi said.

He also referred to the age old occupations still in practice and to the custom of marriage between blood relations which is specially common in South India.

In such a context, it would be well worthwhile to undertake population genetics studies to help differentiate between the genetic loads maintained by mutation and selection, Dr. Sanghvi observed.

One of the highlights of the conference was the seminar on recent trends in population genetics in India.

as in oxaluria, avoidance of certain drugs acting on defective genetic metabolic pathways as in porphyria and preventive measures such as colectomy in certain forms of intestinal polyposis are some of the examples of application of this knowledge.