Preface

The Department of Animal Genetics, Madras Veterinary College, although primarily concerned with studies on the Genetics and breeding of livestock, began in 1960 to interest itself in Human Genetics as this type of work was not pursued in the country. While the students of Veterinary course received a year's instruction in Genetics, the Medical students hardly learnt anything of this subject. Naturally the importance of Genetics to the medical field had not been realized and the initiation of studies in Human Genetics in the Veterinary College may well be considered a starting point. And so, when I joined the Department in 1960 as a research fellow of the University of Madras, this branch of investigation was assigned to me and I was asked to look into association between blood groups and diseases in man. I had naturally to turn to the city hospitals for my material and found that although data were in plenty they were scattered and entirely mixed up. There was considerable reticence on the part of the medical profession to give me the required help. This is understandable because there are so few of them here and there is so much for them to do by way of alleviating pain and distress that it is impossible for any actively engaged medical practitioner to interest himself in research of this type. There were even some who said my study was not purposeful. But with an adamant zeal I went into the hospital records for the previous five years and gathered all the data I could glean in regard to blood group status and disease conditions. This would account for the disease nature of the diseases dealt with in this thesis.
As these studies were under way, the availability of blood group sera became restricted as a result of the breaking out of hostilities in the country and so blood typing had to be abandoned and the studies continued in other directions. Thus the determination of taste sensitivity, sex chromatin status and palmar dermatoglyphics was undertaken among a certain class of patients. In 1964, I was initiated into the techniques of culturing peripheral leucocytes for chromosome characterization. This turned the tide in that the medical profession was now completely on my side and they began referring all kinds of cases to me for karyotyping and I found myself quite inundated with material. While co-operation from the practitioners was possible through perseverance, the eliciting of co-operation from patients has been a most formidable task. The average Indian patient is quite amenable to his doctor, but when it comes to donating blood for a smear or for a culture or tasting a substance for its bitterness or providing a palm print for study, he will just not agree unless the medical officer says it has a bearing on his disease and is likely to result in relief. As for photographs of patients or their bodies, they were well nigh impossible. It is against this background that the author would like this work to be judged. It is the result of a sustained effort to gain some insight into the genetic aspects of diseases in India and it is certainly the first effort of its kind made in a systematic manner.

The thesis comprises six chapters. Chapter I deals with the ABO blood group status among patients suffering from duodenal ulcer, gastric ulcer, peptic ulcer, stomach cancer, echinophoria,
epilepsy, mental retardation, cancer of cervix, fibroid uterus, prolapse of the uterus, pre-eclamptic toxemia, cholelithiasis, cancer of rectum and cancer of breast. For the reasons stated earlier, familial blood group status could not be determined, but the distribution among the affected individuals is set against the distribution in 10,000 healthy control subjects and association between blood groups and disease status is assessed. The data have been statistically analysed in more than one way.

Chapter II describes a study of taste sensitivity to PTC in 347 schizophrenic and mentally retarded patients of both sexes. Using seven concentrations of PTC, not only taste sensitivity but also taste acuity has been determined for the patients as well as for 704 healthy control subjects. The results have been subjected to statistical analysis and discussed in detail. Supporting graphs and histograms are provided.

In Chapter III are set out results of a study on the occurrence of sex chromatin in buccal mucosal cells and of drumstick and sessile bodies in the polymorphonuclear leucocytes of 329 schizophrenics and mentally retarded patients of both sexes ranging in age from 5 to 60 years. These patients are essentially the same in whom blood group status and taste sensitivity had been studied earlier. A control series of 346 healthy individuals of both sexes, and of the same age range has also been examined for comparison. The findings are discussed with reference to contemporary work.
As an offshoot of this study, an investigation into the incidence of sex chromatin, drumsticks and sessile bodies in the newborns was undertaken because the situation in these seems different from that in adults. A description of this investigation and its results is provided in Chapter IV.

Chapter V deals with palmar dermatoglyphics among schizophrenic patients. 316 psychotic patients, the same group that was employed in previous studies, were palm-printed and the prints studied for digit-wise pattern size, pattern type distribution and "atd" angle. An equal number of healthy controls were also similarly dealt with. Mean values, standard deviation and coefficient of variation have been determined. Graphs depicting the results are provided.

In Chapter VI will be found details of chromosomal studies undertaken in 60 schizophrenics, 13 mentally retarded cases, 4 cases of Down's syndrome, 20 cases of primary amenorrhea, 3 cases of secondary amenorrhea, 3 cases of sterility and one case of Klinefelter's syndrome, besides a miscellaneous collection of disease conditions as they presented themselves for study. Clinical histories are provided wherever it has been possible to obtain them, and the account is supported by ten plates. The results of each study are discussed in the light of relevant and recent knowledge. Continuous pagination has been adopted for the thesis but separate summary and bibliography is provided for each chapter.