ANNUAL REPORT FOR 1969.

(Haemoglobinopathies and Chromosomal Defects)

During the period under report (1-1-69 to 4-12-69) suspected cases of haemolytic anaemias were referred for investigation. With the addition of quantitative estimation of haemoglobin A_2 included in the existing battery of tests for detecting thalassemias, it has been possible to evaluate atypical cases. In all 289 individuals were investigated in detail for abnormal haemoglobins including thalassemia. Of these 92 were probands, 96 males (father and male sibs of the proband) and 101 females (mother and female sibs of the proband). Following are the details of the cases detected which are of interest to us.

26 .. Thalassemia major

70 .. β-thalassemia trait

2 .. Hereditary persistence of Foetal Haemoglobin

3 .. Sickle cell anaemia

1 .. Sickle cell - thalassemia

15 .. Sickle cell trait

1 .. Haemoglobin E-thalassemia

2.. Haemoglobin E trait.

One family of speical interest was a case of βδ-thalassemia where the propositus showed ablsence of adult haemoglobin. This is being investigated in detail.

Samples for reference work continued to be received from Pathologists and Physicians outside Bombay. In all 17 such cases were investigated.

In collaboration with Nutrition Research Laboratories, Hyderabd, a survey was undertaken for abnormal haemoglobins in Onge Tribes in Andaman Islands. In 36 samples investigated, no abnormal haemoglobin was detected.

Blood samples were collected by Mrs. L. Ranandive from Bene-Israel Jews residing in Bombay and suburbs as a part of a project for study. Investigation for abnormal haemoglobins including thalassemia were done in 80 samples collected so far. Two instances of β-thalassemia trait were detected, of which one was confirmed by family study.

In view of the finding of β-thalssemia complicating leukaemia in two cases detected from those referred by T.M.Hospital, cases showing haemolytic episodes continued to be referred to us. Eleven such cases were investigated for presence of abnormal haemoglobin synthesis and presence of thalassemia.

Various equipments required for the W.H.O. Project on "Survey of Thalassemia in Western India" continued to be received. The last of the items was received on 3-12-69 thus completing the order. In veiw of this, the work on this project is just started.

Lectures: 1. "Haemoglobinopathies in India" at Seminar on Medicine, (Current topics in Internal Medicine), Post-graduate Medical Association, Poona, on October 17, 1969

2. "Asian Haemoglobinopathies" at Dept. of Pediatrics, J.J.Hospital, Bombay, for the UNICEF/WHO Course for Senior Teachers in Child Health, on October 30, 1969

Deputation: Attended the XXIV Joint Annual Conference of the Association of Physicians of India at Hyderabad, (January 22-25, 1969) and presented a paper.

Publications: Haemoglobin polymorphism in Indian Zebu Cattle.
S.N.Naik, P.K.Sukumaran and L.D.Sanghvi, HEREDITY (1969) 24:239-47.

Bone Changes in Congenital Haemolytic Anaemias - report of cases in Ceylon.
N.Nagaratnam and P.K.Sukumaran. Radiol. Clin. (1969) 38: 154-60.

Haemoglobin Variants and Blood Groups in Gujarati-speaking Lohanas in Bombay
P.K.Sukumaran, H.M.Bhatia and L.D.Sanghvi
(read at the XXIV Joint Conference of Association of Physicians of India, Hyderabad, 1969)

Participated, on invitation, in a Groups Discussion on "Haemoglobinopathies" at the above Conference.

Role of chromosomes in carcinogenseis has been the subject of many investigations in recent years. Chromosome abnormalities, specific and non-specific are common in neoplasms.

Chromosomal abnormalitiesin the form of morphology (Structural) and/or modal (numerical) are commonly known to occur. In the former can be observed (a) presence of marker chormosomes, ranging from one to at times as many as three, (b) dicentrics, dicentrics, (c) chromosomes with gaps or breaks and (d) acentric bits. Heteroploidy also is a common feature.

The work this year consisted of pleural effusion and peritoneal effusion materials obtained from the Chemotherapy Unit, Indian Cancer Society. Freshly obtained fluid was fixed and studied for chromosome counts and chromosome morphology. It is interesting to report that in the material obtained from a female aged 50 years, suffering from Adenocarcinoma Ovary, a definite marker chromosome was seen in most of the plates. Besides the marker, polyploidy along with dicentric chromosomes, and a few chrmatid breaks were also seen. With proper facilies recently obtained, work on leukocyte culture and the like are being undertaken. It is too early to report the findings.

P.K. Sukumaran.