## 5

THE POSSIBLE ROLE OF LOW ERYTHROCYTE SUPEROXIDE-DISMUTASE ACTIVITY ON THE OXIDATIVE HEMOLYSIS IN THE NEWBORN INFANT.

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Previously communicated data demostrated the possible role of the variations of superoxide-dismutase (SOD) on the susceptibility of newborn infants erythrocytes to the hemolysis. Wider experience on the determinations of the SOD activity in the erythrocytes of newborn infants allowed to detect newborn infants with a particularly low SOD activity. Among more than 80 tested newborn infants only 4 cases had SOD activity less than 40% of the mean adult value. All of these four cases had hyperbilirubinemia, decrease of red blood cell count in the first weeks of life and increased sensivity of the erythrocytes to the APH "in vitro".

The mean value of SOD activity in the healthy newborn infants was significantly higher than in the newborn infants with jaundice or anemia: 145.5±42.5 (S.D.) $\mu g/g$ Hb and 128.8±42.7$\mu g/g$ Hb: $P<0.05$.

## 6

IMERSLUND-GRASBECK ANEMIA.

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A follow-up study has been performed of the 10 patients originally described by Imerslund in 1959, and 4 additional patients, 8 males and 6 females aged 2-41 yrs. The patients have a hereditary, selective malabsorption of vitamin $B_{12}$, probably due to a block in the transfer of $B_{12}$ from the enterocyte to the blood, and with intact secretion of intrinsic factor and HCl in the stomach.

On $B_{12}$ treatment the patients were clinically well with normal neurological status. Hgb, r.b.c. and PCV were in the high normal range. Serum $B_{12}$ was normal, while serum and red cell folates were in the lower normal range. Proteinuria was present in 12 patients. These excreted on an average 750 mg protein/24 hrs (range 300-1500 mg), mostly of low molecular weight. Two thirds of the protein was albumen. The proteinuria is predominantly glomerular in nature, but the presence in the urine of light chains indicates an affection also of the proximal tubules. Creatinine clearances were normal, inulin clearances moderately decreased, PAH clearances definitely reduced indicating reduced renal blood flow. Kidney biopsies were performed in the two oldest patients (aged 35 and 41 years). Light microscopy did not disclose anything abnormal, but electron microscopy showed mild signs of glomerulonephritis with an increase of mesangial cells and matrix and a thickening of Bowman's capsule.

## 7

IMMUNODEFICIENCY IN DOWN'S SYNDROME. ANTIBODIES TO E.COLI AND RABBIT ERYTHROCYTES AT DIFFERENT AGES.

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Titers of "natural" antibodies to E.Coli O antigens of different serotypes (O1.K1.H7; O2.K1.H4; O4.K3.H5; O6.K2 ac H1; O15.K14.H4; O75.K?.H5) and to rabbit red blood cells (RaRBC) were determined in 82 subjects with Down's syndrome (DS) and 76 mentally retarded but chromosomally normal controls. Age ranged from 1 to 50 years; subjects with DS and controls were matched for sex, age and socio-environmental conditions. Titers of both antibodies assessed by hemagglutination, were significantly lower in subjects with DS 1 to 5 years old. E. Coli antibodies transiently increased to normal values in subjects with DS during the second 5 years of life, thereafter rapidly declining to levels significantly lower than those observed in controls. The RaRBC antibody titer in subjects with DS remained significantly lower up to the age of 15; due to the rapid age-dependent decline of RaRBC antibody titers in controls, the significance of the difference between the 2 groups disappeared at later ages. These data can be taken as further evidence for the existence of a congenital immunodeficiency in DS.

## 8

NUCLEOSIDE-PHOSPHORYLASE DEFICIENCY, AUTOIMMUNE HAEMOLYTIC ANAEMIA AND SELECTIVE T-CELL DEFICIENCY.

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Recently three patients with a severely defective T-cell immunity and no measurable activity of purine nucleoside phosphorylase (NP) were described. We had the occasion to observe a 22 months old girl with NP deficiency who was referred to us because of haemolytic anaemia which proved to be due to IgG and IgM incomplete warm autoantibodies against red cells. A CMV infection was diagnosed and a bilateral infiltration on chest roentgenogram was evident 4 weeks after admission. The NP activity of the red cells and granulocytes was absent. The parents are both heterozygous for the NP deficiency as well as both grandmothers, whose mothers are half-sisters. T-cell deficiency was demonstrated by negative skin tests to PHA, Varidase and Candida, by absence of E-rosette forming cells, total impairment of in vitro mitogenic response and mixed-lymphocyte reaction. The percentage of surface Ig-bearing lymphocytes and all serum immunoglobulin levels were normal. The clinical history will be fully discussed.

## 9

RELATION OF ZINC WITH LYMPHOCYTES IN PEDIATRIC HODGKIN'S DISEASE

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The recent experimental studies indicated that zinc deficiency may cause thymus hypoplasia and cellular immune deficiency. This inspired us to investigate the relationship between zinc and lymphocytes in pediatric Hodgkin's disease. The serum zinc levels, total lymphocyte counts, coutaneous reactivities to three intradermal antigens and the invitro lymphoblastic transformation response to PHA were evaluated in 24 children with Hodgkin's disease and 20 control cases.

Serum zinc level was measured by atomic absorption spectrophotometer (Perkin Elmer M 103) in Hodgkin's cases found to be significantly low in all patients, regardless of the clinical stage and histopathological pattern. Our preliminary results also disclosed a relationship between serum zinc levels and the lymphocyte abnormalities in Hodgkin's disease.

## 10

CLINICAL, IMMUNOLOGICAL AND CYTOGENETIC FEATURES OF $Ph^1$ POSITIVE CHRONIC GRANULOCYTIC LEUKAEMIA IN CHILDHOOD.

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Adult ($Ph^1$ positive) chronic granulocytic leukaemia was diagnosed in eight of 123 (6%) consecutive new cases of childhood leukaemia. Four patients presented as typical CGL and four in blast crisis; two of the four chronic cases have since developed blast crisis. Morphological characterization of the blasts in these six acute cases suggested a myeloblastic crisis in one and a lymphoblastic crisis in five. Study of 'membrane markers' indicated one myeloid, four lymphoblastic and one mixed lymphoid/myeloid crisis. Remission induction was easily achieved in two of the five lymphoblastic cases; one child relapsed rapidly and the other remains well eighteen months after conversion but serial cytogenetic studies show persistence of the $Ph^1$ chromosome.

We conclude that adult CGL in childhood presents frequently as blast crisis and may even mimic 'good risk' ALL. Response to treatment cannot be predicted by morphological or immunological characteristics.