11 DIAGNOSIS AND TREATMENT IN CHILDHOOD LEUKAEMIA.

T. Révész, D. Schuler, J. Keleti, M. Vargha, and Hungarian Study Group for Childhood Leukaemia. Dept. of Pediatrics, Semmelweis Univ. of Medicine, Tüzolto, Budapest, Hungary.

545 leukaemic patients have been diagnosed by the Leukaemia Study Group since 1971. Retrospective analysis of the initial morphological picture in ALL indicated a poorer prognosis for patients with acute undifferentiated leukaemia /L_2 in the FAB classification/. Patients with poor initial prognostic criteria /high WBC, mediast. mass. etc/ are treated more vigorously since 1975. However, survival results in this group are still far from being satisfactory. Overall results for ALL patients show a steady improvement. Interestingly, there was a considerable difference between various leukaemia centres in the country as far as their treatment results are concerned, in spite of uniform protocols. Currently a randomised trial is on the way to assess two different maintenance schemes following uniform remission induction regimens. Survival data are analysed using the MRC computer program.

12 AUTOSOMAL CHROMOSOME MARKERS IN A CASE OF BONE MARROW TRANSPLANTATION IN A SEVERE APLASTIC ANEMIA.

J. Fraisse, F. Freycon, G. Pouyau, C.P. Brizard, J.J. Ravussin, Dept. of Pediatrics and Lab. of Cytogenetics (CTS), Hop. Bellevue, Saint Etienne, France.

A bone marrow transplantation ($3,6 \times 10^8$/kg nucleated cells) was performed in a seven years old girl with a severe aplastic anemia, from her sister HLA A B identical and areactive MLC. Engraftment was successfull but with a severe cutaneous chronic GVHD. Donor's RBC markers (E and Duffy antigens, PGM1, GPT) are present in the recipient.

Cytogenetics investigations were performed on peripheral blood conventional staining and R banding did not show any variant but Q banding showed up heteromorphism of centromeric heterochromatin.

The patient had formerly one brillant n° 3 (level 5) her sister did not have this marker but two n° 13 (level 3). After the graft no more cells with a bright n° 3 were found in the patient but the two n° 13 were present.

When no sex chromosomes can be used, autosomal markers are to be emphazised with several staining methods.

13 CONGENITAL MEGALOBLASTIC ANEMIA.

L.Ulukutlu, Y.Tangün, M.Bilger, M.Fràter. Dept. of Pediatric Hematology and Oncology Cerrahpaşa and Istanbul Medical Faculties, Univ. of Istanbul and Univ. Children's Hospital Zürich, Turkey and Switzerland.

Megaloblastic bone marrow transformation was observed in a 7 years old girl, showing pallor since age 2. Marked lowering of the serum B12 level and gastric hypoacidity were demonstrated on her second consultation 6 years later for general lassitude and pallor. The transcobalamin II level was found to be on the upper border-line of the normal and the total transcobalamin showed a marked increase in the subsequent controls.

No parietal cell antibodies could be detected. Folic acid level was low in the serum but on the border-line in the red blood cells.

Urine findings were normal.

14 HEMOPHILIA IN FEMALE PHENOTYPIC PATIENTS

A. Orsini, H. Perrimond, I. Juhan-Vague, M.C. Lagrange, M. Mattei, J. Orsini-Roubin. Service de Pédiatrie et d'Hématologie Pédiatrique, Hôpital d'Enfants de la Timone, Marseille - France.

The authors discuss a case of A 6 year old phenotypic female with true hemophilia. The infants caryotype is 46 X Y, the female phenotype probably explained by gonadal dysgenesis.

Diminished factor VIII in a female phenotypic patient should first be suggestive of VON WILLEBRAND'S disease. After elimination of this diagnosis, two situations are possible :

1 - Pseudo feminine hemophilia in a patient with only one X chromosome such as seen in TURNER'S syndrome (45 X), testicular feminisation syndrome (46 X Y) and in gonadal dysgenesis with a X Y caryotype.

2 - True feminine hemophilia could be observed either exceptionnaly in homozygotic or more frequently in heterozygotic girls with complete gene expression, explained by early inactivation of the two X chromosomes.

In both cases, clinical and laboratory signs resemble that of typical hemophilia, which is easily differentiated from VON WILLEBRAND'S disease.

15 POST-NATAL DEVELOPMENT OF Hb A_2*.

F. Schettini, M.I. Capalbo, D. De Mattia, A. Pascazio, A. Campana. Institute of Child Health, University of Bari, Italy.

The development of Hb A_2 levels was determined by micro-chromatographic method in a cross-sectioned study of 152 normal infants aged from 1 to 390 days. The results were expressed as per cent of total Hb and as mg/dl. The ratio between Hb A_1/Hb A_2 was also calculated.

Hb A_2 increased from the 1st month of life and the mean adult values were reached at 151-180 days. The absolute concentration of Hb A_2(mg/dl) increased from the birth reaching the adult levels at 331 - 390 days of life. The linear regression equations showed that Hb A_2 increased significantly with the age only from 1 to 180 days of life ($r = 0.933$). After this period the correlation coefficient was not significant ($r = 0.429$). The mean increase in relation to age was of $0.0144 + 0.0005$ per cent/day and of 1.354 ± 0.079 mg/dl/day.

Therefore Hb A_2, as per cent, was fully developed in the first 6 months in healthy children. Instead the subsequent variations of absolute Hb A_2 levels were related to physiologic modifications of Hb concentration because the ratio Hb A_1/Hb A_2 after the age of 6 months was constant.

*Supported by a grant from CNR(contract 78.02282.04).

16 RED CELL ATPASES IN VITAMIN D DEFICIENCY RICKETS.

M.Alexander, C.Heyder-Bruckner, F.Vertongen and I.M. Mandelbaum. Clinique Pédiatrique et Laboratoire de Chimie Médicale, Hôpital Universitaire St.Pierre, Brussels, Belgium.

A shorter red cell life span has been described in children with vitamin D deficiency rickets. This anomaly does not seem to be related to a hyposideremia. Also, the enzymes of the glycolytic pathway and the intraerythrocytic ATP are normal. On the other hand, hypophosphoremia and hypocalcemia are regularly found in rickets. In order to investigate the effect of these modifications on the red cell membrane, a study of the red cell ATPases was undertaken.

1. (Na-K) ATPase was measured in 12 rachitic children and 9 controls. (Na-K) ATPase values were significantly higher in the patients than in the controls (ouabain sensitive ATPase : $p<0,05$; ouabain insensitive ATPase : $p<0,01$). In this group, rachitic children also had lower Hb and serum iron. In contrast no correlation was found between (Na-K) ATPase and P or Ca serum values.
2. (Ca-Mg) ATPase was studied in 16 rachitics and 7 controls. The values obtained do not differ significantly.

It is concluded that anemia in vitamin D deficient children is not due to a lack in these red cell membrane enzymes. The higher values obtained for (Na-K) ATPase in rachitic patients are probably related to anemia and hyposideremia. Hypophosphoremia and hypocalcemia seem to play no evident role in the origin of the anemia Wether a deficiency of some other extracellular substance could interfere with normal red cell life span remains hypothetical.