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PSEUDOTUMOR CEREBRI IN ASSOCIATION WITH SEVERE HEMOLYTIC ANEMIA AT PRESENTATION OF SYSTEMIC LUPUS ERYTHEMATOSIS (SLE). R. Davidson,* L. Frankel, D. Crisp,* W. Allen,* S. Bassion.* Scott and White Memorial Hospital, Texas A&M University School of Medicine, Temple, Texas.

Accurate diagnosis of SLE is often difficult, since the diagnosis is capable of involving any organ system; yet no single test exists for specific diagnosis. The patient (pt.) must have a constellation of symptoms and lab values which meet the 1982 diagnostic criteria for SLE. Two young girls, ages 11 6/12 and 13 9/12 years, presented with similar, unique clinical findings. Both pts. presented with sudden onset of fatigue, headache and vomiting. Physical exam revealed tachycardia, pallor and bilateral papilledema with small splinter hemorrhages adjacent to the fundal blood vessels. Neurologic exam and brain tomography revealed no abnormalities. The first pt. also displayed mild hepatosplenomegaly. Visual field testing confirmed full fields with a peripapillary scotoma consistent with papilledema. Neither pt. demonstrated joint, skin, cardiovascular or renal involvement. Specific laboratory findings are outlined:

LAB	Patient #1	Patient #2
Hbg/Hct	5.9/16.1	4.8/17.1
Retic Count	7.2	28.5
Coombs	negative	positive
Bili T/D	7.6/3.0	2.2/0.1
ESR	95	150
CSF pressure (mm H_2O)	340	450
Antineuronal abs. in CSF	Not done	IgG 435 units IgM 104 units
ANA	1:5120	1:5120
CH50	156	254

We conclude: pseudotumor cerebri presenting with severe hemolytic anemia in the initial presentation of SLE may result from a combination of the following: the severity and rapidity of the hemolytic anemia, and the involvement of antineuronal antibodies in the central nervous system.

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A NEW KINDRED OF HEREDITARY ELLIPTOCYTOSIS (H.E.) WITH A SHORTENED SPECTRIN α CHAIN. D. Dhermy,* M.C. Lecomte,* M. Garbarz,* C.Féo,* C. Galand,* O.Bournier,* H. Gautero,* and P. Boivin. INSERM U160, Hôpital Beaujon Clichy; INSERM U299, Kremlin-Bicêtre, France.

The major determinant of erythrocyte membrane shape and stability is a proteinaceous meshwork named membrane skeleton, composed mainly of spectrin (Sp), actin and protein 4.1. Sp is a heterodimer composed of two chains α and β (Mr = 240,000 and 220,000 respectively) which associate head to head to form tetramers and higher oligomers. Few cases of H.E. with Sp association defect were found to be related with a truncated Sp β chain (3 reported kindreds), or a shortened Sp α chain (1 reported kindred). We studied a French family of HE with a shortened Sp α chain, similar to that described previously by Peter Lane (J. Clin. Invest. 1987,79, 989). Obvious elliptocytosis without hemolysis was found in the proband and in 3 out of 6 studied siblings as well as in the mother. The mutant Sp was characterized as a new band migrating between α and β bands on SDS-polyacrylamide gel electrophoresis. This new band was a shortened α chain (Mr = 234,000) for the following reasons: (i) it reacted more strongly than β chain with an anti Sp antibody having an higher affinity for α chain and it did not react with a monoclonal anti β chain; (ii) it was not phosphorylatable; (iii) the amount of normal α chain was decreased by an amount equivalent to the shortened α′ chain. The percentage of α′ chain, the consequences on Sp self-association (appreciated by the increase in dimer percentage in 4°C extract, normal 6% ± 2) and the ghost mechanical stability are shown in table.

Family members	Percentage of mutant spectrin $\alpha'/\alpha+\alpha'$	Percentage of spectrin dimers in 4°C extract	Ghost mechanical stability % of normal
Suz. H. (mother)	37	12.6	10
Jea. L. (proband)	25	11.5	50
Col. L. (sister)	21	8	55
Ala. L. (brother)	20	6.6	22
Ann. L. (sister)	17	10	42
And. L. (brother)	0	4.4	130
Suz. L. (sister)	0	5	130
Pau. L. (sister)	0	2.4	70

Sp-4°C extract was electrophoresed in 2 dimensions (non-denaturing electrophoresis following by SDS electrophoresis). Sp dimers resolved in two bands on non-denaturing gel, the fastest band being composed of the abnormal α′β dimer. Tryptic digest patterns of 37°C Sp extract failed to show any modification, even in the mother who displayed the highest amount of α′ chain in the membrane extract (33%).

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HEMOLYTIC ANEMIA DUE TO HETEROZYGOSITY FOR Hb C, α^+-THALASSEMIA AND HEREDITARY XEROCYTOSIS. A.E. Felice, M. Abraham,* D. Bockman,* V. C. McKie,* and M. Greenberg* (Intr. by F.A. Garver) Comprehensive Sickle Cell Center, Depts. of Cell and Molecular Biology, Anatomy, and Pediatrics, Medical College of Georgia; and Hemoglobin Research Laboratory, Medical Research Service, Veterans Administration Medical Center, Augusta, Ga.; and Division of Pediatrics, Memorial Medical Center, Savannah, Ga.

Microcytosis among Black children without significant anemia is often due to α^+-thalassemia, or Hb C trait or combinations of the two. Among these, we identified a 4-year-old boy with a history of pallor, intermittent icterus, and frequent hospitalization because of chest infections. On examination he was small and under weight for his age and presented with splenomegaly at 4 cms below the left costal margin. Hematological evaluation of propositus and family members gave data summarized below.

	Sex Age	Hb g/dl	MCV fl	Hb Type	Dense Cells	Pitted Cells	α Genes	Xero-cytes
Prop.	M-04	8.6	70	AC	14.3%	12.9%	-α/αα	++
Brother	M-06	10.7	71	AC	3.1%	-	αα/αα	+
Sister	F-10	10.5	77	AA	6.1%	-	-α/αα	+
Mother	F-30	10.0	77	AC	6.1%	8.0%	-α/αα	+

Osmotic fragility gave decreased hemolysis in standard saline dilutions. Morphological examination revealed microcytosis, anisocytosis, many target cells and bizarre large empty oval cells with small quantities of Hb at the poles reminiscent of Xerocytosis (Xerocytes). The identification of anomalous erythrocytes with increased density and morphological abnormalities in the sister who had normal hemoglobins suggested an additional red cell (membrane) abnormality seggregating in addition to Hb C and α^+-thal traits in this family and indicates avenues for the identification of rare conditions such as Xerocytosis among children with microcytosis.

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SURVIVAL OF PHOSPHOFRUCTOKINASE-DEFICIENT ERYTHROCYTES IN A CANINE MODEL. U. Giger. Section of Medical Genetics, University of Pennsylvania, Philadelphia, PA

Inherited muscle-type phosphofructokinase (PFK) deficiency in dogs is characterized clinically by chronic compensated hemolysis with intermittent severe hemolytic crises. Severely reduced erythrocyte PFK activity (8-22% of control) results in low erythrocyte 2,3-diphosphoglycerate (DPG) levels thereby increasing the hemoglobin-oxygen affinity and the alkaline fragility of canine erythrocytes. Consequently, severe intravascular hemolysis can be caused by hyperventilation-induced alkalemia. Survival of PFK-deficient erythrocytes and the effect of various therapeutic modalities were studied in this animal model. The apparent half-life ($T_{\frac{1}{2}}$) of ^{51}Cr-labeled PFK-deficient erythrocytes during steady-state conditions was 4.1±0.4 days (m±SD,n=3) compared to normal canine erythrocytes' $T_{\frac{1}{2}}$ of 22.1±2.2 days (n=3,$p<0.01$). After splenectomy, the survival of PFK-deficient erythrocytes was not significantly improved ($T_{\frac{1}{2}}$=4.6±0.5 days). Acetazolamide, administered orally in an attempt to lower the intracellular erythrocyte pH, caused a metabolic acidosis, but failed to prolong survival of PFK-deficient erythrocytes ($T_{\frac{1}{2}}$=4.3±0.6). However, administration of acetazolamide prior to episodes of hyperventilation induced by exercise or high environmental temperature was effective in preventing acute hemolytic crises. In the autohemolysis test, addition of glucose was less effective in reducing the degree of lysis of PFK-deficient erythrocytes compared to control cells. In contrast, incubation of blood from PFK-deficient dogs with adenosine, inosine, or ATP markedly lowered the degree of lysis, suggesting that PFK-deficient cells utilize these substrates. Changes in DPG content and alkaline fragility of PFK-deficient erythrocytes were studied after CPDA-1 anticoagulated blood was incubated with various substrates. The addition of phosphate and pyruvate or phosphate in combination with dihydroxyacetone, inosine or alanine increased the DPG content of PFK-deficient erythrocytes and reduced their alkaline fragility. In conclusion, acute hemolytic crises in PFK-deficient dogs were prevented by acetazolamide administration and erythrocyte DPG concentrations were restored in vitro by alternative substrates used in transfusion medicine.