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CLINICAL ASSOCIATIONS WITH SS-A ANTIBODIES IN CHILDREN.

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Antibodies to SS-A (Ro) are associated with primary Sjögren's syndrome, Systemic Lupus Erythematosus (SLE), subacute cutaneous LE with of without C2 deficiency and neonatal lupus. They can also be found in up to 1% of healthy individuals. Antibodies to SS-A are rare in childhood.

We retrospectively looked at clinical pictures of all children who had circulating SS-A antibodies determined with immunodiffusion (IF) and ELISA techniques in the year 1993. SS-A antibodies were detected in nine children, six girls and three boys. The majority of children were from Caucasian origin, two were Hindustan and one negroid. One of the patients was a neonate with a congenital heart-block (mother ANA and SS-A positive without clinical symptoms). In the other children the age of onset of clinical symtoms varied from 5 to 12 years. Two patients were diagnosed as SLE with secondary Sjögren's syndrome, two as SLE with nefritis (one with restrictive lung disease), three as undefined autoimmune disease (one with a chronic uveitis) and two boys with fatigue, myopathy and parotic swellling (Raynaud's phenomenon in one). In all patients fatigue was a predominant symptom. None of the patients complained of dry eyes. Antinuclear antibodies were positive in seven children, antibodies to dsDNA in four.

In conclusion, SS-A antibodies are associated with a limited number of clinical syndromes and may be present in spite of a negative ANA-IF test. Longtime follow-up is indicated as it may take years for autoimmune diseases to fully develop.

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HYPERTROPHIC OSTEOARTHROPATHY IN CHILDREN WITH LEUKEMIA

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Bone lesions are frequently encountered in children with leukemia as part of their disease or occasionally as consequence of therapy. While leukemic infiltration is a well known cause of bone pain in children, avascular necrosis and osteoporosis can also be responsible for these complaints.

The authors present two children with an unusual cause of bone pain in childhood leukemia, hypertrophic osteoarthropathy (HOA). This condition is well known in adults, but is rarely seen in children. The most frequently associated conditions in children are chronic lung, liver or heart disease and sometimes malignant or benign tumors. In a child with acute non-lymphoid leukemia (ANLL) FAB type M7 and in a child with common acute lymphoid leukemia (ALL) - both in the maintenance phase of therapy - bone pain developed. The radiographs showed cortical thickening and periosteal new bone formation along the diaphyses of the long bones. Bone scintigraphy remained negative. MRI showed no signs of residual leukemic infiltration. Both children were in remission at the time of detection of the bone lesions, and did not have any signs of chronic infection. Approximately three months later however - although the bone pain disappeared - the child with ANLL developed a myelodysplastic syndrome (MDS) and the patient with ALL a partial relapse with 12 % blasts in the bone marrow. The occurrence of HOA in children with leukemia, both ANLL and ALL, should instigate repeated bone marrow investigation since it might herald MDS or relapse.

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"PRIMARY CUTANEOUS MALIGNANT B-Cell NHL IN A INFANT"

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Cutaneous malignant Lymphoma in infancy is rare. The majority of these tumors present nonB - nonT cell phenothype. Recently, we encountered an infant with cutaneous malignant Lymphoma, which was a B-cell tumor in origin. The patient was a 23 month old female and she came to our Clinic on July, 1992. Seven months previously, her mother noticed a tumor on the back which was considered a sebaceous cyst and then an angioms. Her general conditions was good. Since the lesion increased, the baby got into Hospital. On physical examination a violet, dome-shaped, indolent and movable nodular cutaneous tumor was confined to a circumscribed area of the skin, in left subscapolar region, about 3x2 cm in size. Systemic lymphadenopathy and splenomegaly were not present. The tumor was radically removed and the histological diagnosis was: High grade non Hodgkin, diffuse lymphoma, non Burkitt, with small non clived cells, according to Working Formulation. The neoplastic infiltrate react with CD 45R, CD 45RA, CD 20, CD43 and failed to react with CD30, CD 45RO, confirming the B cell phenotype. Bone marrow aspirate revealed no evidecence of involvement by tumor cells. After staging investigations, following Murphy's criteris, the patient was treated with two blocks of therapy, according to AIEOP Non Hodgkin Lymphoma 92 Protocol (DEXA, MTX-MD, IFO, VP16,ARA-C + DEXA, CPM, MTX - MD, ADR).After 19 months, there is no evidence of local recurrence or dissemination.

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Francesco Schettini, Rosa Penza, Paola Giordano, Giovanni C. Del Vecchio, Gabriella Aceto, Maria Altomare, Domenico De Mattia. DETERMINATION OF von WILLEBRAND FACTOR AND FACTOR XIII IN CHILDREN WITH SCHONLEIN HENOCH SYNDROME. Dipartimento di Biomedicina dell'Età Evolutiva - University of Bari - Italy.

In 16 children (7 M and 9 F, aged between 4 and 11 years) with Schonlein Henoch Syndrome (SHS) at the onset, we have studied the levels of von Willebrand Factor Antigen (vWf:Ag) and Factor XIII activity (F XIII) in relation to the severity of clinical symptoms and the immunological parameters: IgA, C3, C4 and circulating immune complex (CIC).Arthral, abdominal and renal symptoms (except purpura) were scored from 0 to 3 and their mean values resulted respectively 1 ± 0,82; 1,13 ± 0,96; 1,38 ± 1,26 (total: 3,5 ± 1,59). The vWf:Ag resulted increased in 7 patients,the F XIII resulted decreased in 6. In all children we found high levels of IgA, while the C3 and C4 levels were normal.The CIC were elevated in 11.The total clinical score was positively correlated with vWf:Ag, IgA and CIC ($p<0.05$). The abdominal score was positively correlated with vWf:Ag ($p<0.05$); while the renal one with vWf:Ag ($p<0.05$) and CIC ($p<0.001$).

Lastly vWf was correlated with IgA and CIC ($p<0.05$) and as in other systemic vasculites its level was increased in the acute phase of SHS probably in reference to immunomediated endothelial cell damage. In conclusion vWF levels are a good marker of SHS severity.

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T LYMPHOCYTE DIFFERENTIATION IN PRETERM NEONATES

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We have defined T lymphocyte (lph) membrane receptors (Rc) and glucocorticoid receptors (GR) in peripheral blood of 40 neonates with 31-36 weeks gestational age (GA) and in cord blood of 42 term infants, using monoclonal antibodies (Ortho Diagnostic Systems, USA), Lippman-Barr technique incorporated. The results of our studies are presented in the table.

PATIENTS	TERM	31-33 weeks GA		34-36 weeks GA	
CD	Cord blood	Peripheral blood (%)			
		Day 7	Day 30	Day 7	Day 30
CD3	54,2	29,2	44,1	36,8	51,6
CD4	37,6	23,1	38,2	26,9	38,7
CD8	14,2	10,8	19,8	18,2	18,6
CD2	38,4	33,3	51,3	36,6	55,8
Young cells (CD1,6,9,10)	6,2	32,1	46,8	19,2	40,3
Rc IL-2	abs.	12,1	14,9	8,1	12,9
GR	6286±1045	14204±3543			

We conclude, that immaturity of T lph functional receptors can be the basis of clinical immune deficiency.

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ICF SYNDROME

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ICF syndrome (Immunodeficiency, Centromeric instability, in particular of chromosomes 1 and 16 and Facial dysmorphisms) was described in 1988 by Maraschio et al. (J Med Genet 25:173-180).

Here we present another case of ICF syndrome. The boy was born at 38 weeks, small for gestational age (2055 gram, 45 cm). During the first year he had a failure to thrive and a slight psychomotor retardation. At the age of 10 months, he suffered from RS bronchiolitis. Immunological studies revealed an agammaglobulinaemia (all immunoglobulins were absent). B cells were normal, as were T cell markers (CD_2 58 %, CD_3 56 %, CD_4 36 %, CD_8 17 %). In vitro responses of peripheral blood cells to PHA and PWM were normal. He had a typical face. Cytogenetic studies revealed a normal 46 XY male karyotype, although half of the cells showed alterations (e.g. gaps, breaks, translocations, centromeric fusions, multiradial figures) in the centromeric regions of chromosomes 1 and 16.

During immunoglobulin substitution he did not suffer from infection but he ceased growing, probably due to malabsorption.

Untill now, 15 patients with ICF syndrome are recognized. All but one had facial anomalies. All had immunodeficiency varying from IgA deficiency to SCID. A review of these patients will be given.