85 CLINICAL SYMPTOMS OF MUCOVISCIDOSIS /MV/ IN GENE CARRIERS. IS INCOMPLETE MV A HETEROZYGOUS STATE?

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It has frequently been suggested that MV gene carriers are not completely healthy, and that cases occur among them exhibiting clinical symptoms of MV of variable severity. Without an appropriate examination method, this could not be proved so far. We recently found that the bromide index is suitable for demonstration of MV heterozygous state. With some modification, the blood: sweat bromide ratio after per os NaBr administration is determined with an ion-selective electrode. With this method, MV gene carrying was confirmed in 24 children with chronic diseases of the respiratory tract. Such an MV genotype of the children was supported by similar examinations on the parents. The clinical symptoms were similar to those of classical MV, with varying characteristics and severities. In 5 of the 24 definitely heterozygous MV cases the sweat Cl⁻ concentration was above 60 mEq/l. These corresponded clinically to Schwachman's incomplete MV criterion. Apart from the above cases the MV heterozygous state was confirmed in two neonates considered pseudopositive.

86 CARBENICILLIN HALF-LIFE IN CHILDREN WITH EARLY DIABETES MELLITUS

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It has been shown earlier by the authors that the glomerular filtration rate /GFR/ and the penicillin clearance are increased in short-term childhood diabetes. Recently, GFR and carbenicillin half-life /T/2/ were determined in insulin-treated children with a diabetes duration of 1-5 years, and in a healthy control group. An inverse relationship, between carbenicillin T/2 and GFR was found in both groups. The GFR was significantly higher, and the carbenicillin T/2 significantly lower in diabetic children than in healthy controls. There was no correlation to blood sugar levels. It is suggested that the accelerated elimination of penicillin and carbenicillin in early diabetes necessitates higher dosages of these drugs for an adequate therapeutic effect.

87 STUDY OF ARYLSULPHATASE -A ACTIVITY IN A METACHROMATIC LEUKODYSTROPHIC PATIENT AND FAMILY MEMBERS

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Clinical and laboratory data are reported on metachromatic leukodystrophy of delayed infantile type in a five and a half years old female child from a marriage between cousins of first degree. Metachromatic staining sulphatide excreted in the urine was confirmed by cresyl violet staining urinary sediment. There was no arylsulphatase activity in the urine. Quantitative glycolipid thin-layer chromatography of the urine showed the characteristic sulphatide accumulation. Metachromatic granules were detected in fibroblast cultures. Lysosomal arylsulphatase-A was determined on serum and wbc homogenizates. The enzymatic activity of the leukocyte homogenizate of the patient was 8.9% of the normal value /homozygote/; corresponding values in the mother and father were 47.7%, and in the paternal grandfather 27.7% /all three heterozygote/. The course of the heredity of the disease could be well followed by examination of the specific enzymatic activity of the family members. Lysosomal arylsulphatase-A examinations are of great importance in the diagnosis of demyelination syndromes.

88 VISUAL OBSERVATION AND ELECTRONIC MONITORING OF MOTOR ACTIVITY IN NEWBORN INFANTS

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The spontaneous motor activity of newborn infants was electronically monitored using a sensing unit, placed in the bed or incubator. The sensor is an inductive-capacitive circuit, which allows monitoring of motor activity without attachment of cables or electrodes to the infant. The signal from the instrument and the coded visual observation of the motor activity was simultaneously transferred to a tape recorder. In a few cases the motor activity of the infant was also filmed. The correlation between the visual observations and the electronic records of motor activity was analyzed. The signals from the sensing unit are synchronic to the observed moments. The instrument presents a semiquantitative display of the motor activity pattern, and is superior to a method based on ordinary visual observation, which is inaccurate in prolonged studies. Different patterns of motor activity in newborn infants will be described and the application of motor activity studies in pediatric research and clinical work will be discussed.

89 THE STEM CELL COMPARTMENT OF THE BONE MARROW - ATTEMPTED EXHAUSTION BY STIMULATION OF GRANULOPOIESIS, LYMPHOPOIESIS AND ERYTHROPOIESIS

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The question of stem cell competition in the lymphocyte-transitional cell compartment of the bone marrow was investigated by subjecting guinea pigs to combined granulo-, lympho-, and erythropoietic stimulation. By a standard technique, absolute counts were made from the bone marrow of 1/normal animals, 2/ after typhoid vaccine /TAB/, 3/ of hypoxic guinea pigs, 4/ after TAB following hypoxia, and 5/ after hypoxia following TAB. High numbers of stem cells were found to accumulate in the bone marrow after TAB stimulation alone. In the groups receiving both stimuli, there was a fall in the number of transitional cells and lymphocytes which, however, did not reach the level of significance as compared to untreated control animals. It was concluded that the stem cell reserve of the bone marrow was large enough to meet the needs of combined hematopoietic stimulation.

90 THE BONE HISTOLOGY IN THE SYNDROME OF MEDULLARY STENOSIS OF TUBULAR BONES, DWARFISM AND HYPOCALCEMIA

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The pathogenesis of this syndrome has not been elucidated. The present case-a 12 years old boy-demonstrated dwarfism, stenosis of medullary spaces of bones, intracranial calcifications and permanent hypocalcemic hyperphosphatemic tetany. Thyroxine and growth hormone levels were normal. Plasma parathormone was elevated. The response to parathormone was normal: a significant rise in serum Ca, a fall in serum phosphate, an increase in renal phosphate clearance, and an increase in urinary proline and OH-proline. Microradiographs of cross sections from the fibula shaft showed normal bone structure and density. However there was remarkably little remodeling activity: only one or two forming osteones in the entire section and only one resorption cavity. This is the first child with this syndrome in whom the bone is examined.