43 DEFICIENCY OF PEROXYSOSES, CATALASE AND CYTOCHROME B: A GENETIC DEFECT OF OXYGEN METABOLISM?

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A 14 months old male child failed to thrive, had severe mental retardation and liver cirrhosis. Besides serum iron elevation to 168 µg/100 ml no metabolic abnormalities were found. A brother had died at 3 months of age under similar conditions. Mitochondria from liver biopsy were studied. Content and redox state of respiratory chain components, oxidative phosphorylation and $K_1/2O_2$ of cytochrome aa₃ were measured by O_2 polarography and spectroscopy according to BRAUSER, 1968. Succinate respiration was normal. There was subnormal coupling of oxidative phosphorylation. The ratio of cyt b/cyt aa₃ was reduced. Oxygen affinity of cyt aa₃ was normal. In isolated intact lymphocytes normal succinate respiration and a reduced cyt b content was found. Like in ZELLWEGER's syndrome peroxisomes/electron microscopy/, catalase and peroxidase activity/histochemistry and spectroscopy/ are lacking. Smooth endoplasmic reticulum, however, is present. It is suggested that lack of peroxisomes and mitochondrial defects are related, since mitochondrial H_2O_2 production occurs in the cyt b region of the respiratory chain.

44 EFFECTS OF ACUTE BLOOD VOLUME CHANGE ON PLASMA RENIN AND ALDOSTERONE IN THE NEWBORN

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Increase activity of the renin/aldosterone system in the neonatal period is now well established in both animals and man, but the control mechanisms involved are poorly understood. We have therefore monitored plasma renin activity (PRA) and aldosterone concentration (PALdo) in 8 newborns undergoing 12 exchange transfusions. PRA and PALdo were measured before and at 5, 10, 15 and 30 min after the withdrawal or injection of 7 ml blood/kg birthweight immediately before exchange transfusion. Mean PRA increased to 140% and decreased to 60% of the resting values following withdrawal or injection of blood respectively. Mean PALdo values did not change significantly during the same period. Thus the renin-angiotensin system in the newborn infant is responsive to changes in blood volume. The absence of PALdo changes may be due to the shortness of the observation period.

45 CIRCADIAN CHANGES OF LUNG RESISTANCE (R_L) AND DYNAMIC COMPLIANCE (C_1 dyn) IN HEALTHY AND ASTHMATIC CHILDREN

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Circadian rhythms were demonstrated for many biological parameters. The aim of this study is to determine the circadian changes of lung mechanical factors in children. R_L and C_1 dyn. are measured by esophageal catheter technique four times during a 24 h period (07⁰⁰, 11⁰⁰, 16⁰⁰, 22⁰⁰) in synchronized/sleep: 21⁰⁰ 07⁰⁰/ children. In 7 healthy children a rhythm is detected for R_L and C_1 dyn. / $p < 0.05$. Optimal values are observed during the day / R_L min. 1630, C_1 dyn. max. 11⁰⁰/. The data are analyzed with cosinor method. B sympathomimetic and vagolytic bronchodilators modify circadian changes of R_L and C_1 dyn. These data suggest a circadian rhythm in bronchomotoricity which can be related to changes in neurovegetative activity. In asthmatic children /symptom free/ a similar pattern is observed for R_L and C_1 dyn changes. Circadian rhythms of lung mechanical factors in asthmatic condition may be related to circadian rhythms of cortisol, catecholamines, immunological factors to explain the maximal frequency of attacks during the night.

46 THE AIRWAY RESISTANCE IN NEWBORNS

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The airway resistance was measured in normal newborns and in low birth-weight newborns without or with a difficulty with respiration. Measurements were performed with aid of the additive method, separately during inspiration and expiration. In newborns without a difficulty with respiration relations of the airway resistance or the airway conductance to the body weight and the body length, the frequency of breathing, the tidal volume and the flow rate were studied. In low birth-weight newborns suffering from a respiratory disturbance were found either higher or lower values of the airway resistance than in those without any troubles. The lowest values were found in newborns suffering from IRDS. The reason of this unexpected finding will be discussed.

47 PROGNOSTIC VALUE OF BLOOD GAS TENSIONS IN CYSTIC FIBROSIS

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Blood gases and acid-base balance in 106 children suffering from cystic fibrosis were studied during ten years. 68 of the patients survive, 38 died. Based on 670 examinations in surviving children /group A/ and 507 examinations in those who died /group B/, the prognostic value of hypercapnea was evaluated. The first examination showed hypercapnea in 18 per cent of the group A and in 43 per cent of B. During the first months of life high pCO_2 values were found in both groups. In older children hypercapnea is an alarming sign showing the necessity of intensive care. It was proved practically in all examinations made in the affected children during the last month of their life. Metabolic alkalosis caused either by vomiting or by excessive loss of electrolytes in the sweat can be considered as an unfavorable complication being hypoventilation. It was found - either isolated or combined with respiratory acidosis - in 3 per cent of cases.

48 RESPIRATORY FUNCTION IN CHILDREN WITH NEUROMUSCULAR DISORDERS

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A comprehensive range of lung studies are being applied to children with various neuromuscular disorders. In addition to conventional assessment by spirometry and plethysmography in older children, infants under 1 year can also be studied in the infant whole-body plethysmograph. Diaphragmatic function is assessed by gastric and oesophageal balloons which record transdiaphragmatic pressure during breathing. Regional lung function can be assessed by isotopic nitrogen (^{13}N). To date 17 children have been assessed. These have included Duchenne dystrophy, spinal muscular atrophy, limb girdle and Becker type dystrophy, nemaline myopathy, mitochondrial myopathy, dermatomyositis and polyneuritis. In addition to the well recognised restrictive types of defect associated with intercostal weakness and diminished lung volume, some cases have had diaphragmatic dysfunction, even in the absence of intercostal weakness, and others an apparent central failure of respiratory control and loss of sensitivity to CO_2 . Since respiratory failure is the commonest life threatening problem in neuromuscular disorders, recognition of these different mechanisms is important in the proper management of these cases.