71 A SYSTEM FOR AUTOMATED DIAGNOSIS OF HEREDITARY DISEASES IN CHILDREN

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A computer consultation service has been made at this institute to provide differential diagnoses of multiple hereditary diseases in children, including rare syndromes. Formal descriptions were obtained for 1200 monogenic and chromosomal disease entities. Symptomatology was classified into obligatory, major and minor. The descriptions specify time of onset, mode of inheritance, biochemistry of disease. Mathematically, the system was "precedence"-based. A diagnosis required inputs like disease description, symptom pattern, retrieval specifications (direct retrieval, denial, both). The system employs a IBM-370 computer with software in /1(O) language. Remote terminal interaction was obtained by using distant telephone transmission of data. A clinical trial demonstrated the efficacy of the automated information diagnostic system and its usefulness in pre-laboratory identification of disease through its phenotype.

72 THE DEVELOPMENT OF ADRENERGIC SYSTEMS IN CHILDREN WITH CONGENITAL HEART DISEASE

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In children with Congenital Heart Disease (CHD) the responsiveness of the cardiovascular system to catecholamines differs intra- and interindividually to a great extent. Since this might be due to a dysfunction of the adrenergic system the following investigations were carried out in 18 infants with and without cyanotic CHD. Tissue from the right cardiac auricle was taken during heart surgery. Radioligand studies, using ^{125}-I-Cyanopindolol were carried out on these tissues to estimate the density and affinity of the β-adrenoceptors (β-R). The proportion of β_1- and β_2-receptors was determined by means of displacement curves using selective β_1- and β_2-antagonists.

1.) The average β-R density was lower in infants with cyanotic CHD than in those without cyanosis. The ratio of β_1- and β_2-R appeared to be identical in both groups.
2.) Excessive release of noradrenalin, and even more so of adrenalin, was found. These catecholamine levels were ten times higher than in controls. Infants with cyanotic CHD had significantly higher levels compared to those suffering from CHD without cyanosis.
3.) A linear correlation was found between catecholamine levels and β-R density.

The conclusion can be drawn that infants with CHD have an increased activation of the sympathetic nervous system. The long-term exposition of myocardial β-R to catecholamines evidently leads to receptor desensitization.

73 PULMONARY ARTERY DEVELOPMENT: NORMATIVE ANGIOGRAPHIC DATA

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Successful surgical treatment in children with congenital heart disease and hypoplastic pulmonary arteries depends on the size of the pulmonary artery tree. For this reason we determined normative angiographic data from cineangiograms of 51 infants, children and adolescents without heart disease (n=16) or with cardiovascular malformations which had no hemodynamic relevance (n=35).
Systolic and diastolic diameters of the pulmonary artery anulus, pulmonary artery sinus, pulmonary artery trunc, as well as those of the right and left pulmonary arteries were determined. The corresponding systolic and diastolic diameter values were averaged in order to obtain a mean diameter of each vascular segment. All measurements were correlated with the body length and the body surface area (BSA). A root exponential function $[\sqrt{y=a(1-e^{-bx})+c}]$ was used, the regressions coefficients of which can be explained by physiological means. In this function c^2 is the theoretical minimal diameter and $(a+c)^2$ the theoretical maximal diameter of a cardiovascular structure; in this way the growth of the cardiovascular dimensions could be well defined mathematically and correlated strongly (r=0,99) with body length and BSA. Different exponents were found for the pulmonary artery anulus, sinus, trunc as well as right and left pulmonary arteries.
These data on normal values for the angiographic measurements of the pulmonary artery anulus and arteries at different age are useful for the identification of patients with hypoplasia of the pulmonary artery tree and serve as a valuable guide for cardiac surgery.

74 α_1-PROTEINASE INHIBITOR PHENOTYPES IN PRETERM NEWBORNS.

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α- proteinase inhibitor (α_1PI) phenotypes were determined by isoelectric focusing in polyacrylamide gel in 305 preterm newborns and 109 their mothers, special attention being paid to the presence of Z allele responsible for deficiency of this protein. The newborns were divided in four groups according to anthropometric data: 1) appropriate for gestational age (AGA), 2) small for birth weight (SBW), 3) small for birth length (SBL), and 4) small for gestational age (SGA). Prevalence of Z allele (2,95%) in the total group of preterm newborns did not differ significantly from its prevalence in population study (1,73%). Z allele was more prevalent (5,45%) in preterm newborns with infections, birth defects (5,88%) and with combination of these pathological conditions (6,14%). Especially high prevalence (15%) of Z allele was encountered in SBW newborns with infections. We suggest that prematurity and α_1PI deficiency (phenotypes PI MZ and PI ZZ) are high risk factors for neonatal morbidity.

75 SIGNIFICANCE OF α_1-PROTEINASE INHIBITOR DEFICIENCY IN ETIOPATHOGENESIS OF LIVER DISEASES IN CHILDREN.

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α_1-proteinase inhibitor (α_1PI) phenotypes were examined in 27 children with chronic persistent hepatitis (1st group) and in 70 newborns and infants with protracted icterus and neonatal hepatitis (2nd group). 1442 adults and children in population study served as controls (3rd group). Prevalence of phenotype PI MZ was equal to 7.41% in the 1st group, 4,29% in the 2nd group, and 3,47% in the 3rd group. Prevalence of phenotype PI MS in the groups studied was equal to 1,87%, 2,86%, and 3,33%, respectively. α_1PI phenotype PI ZZ responsible for α_1PI deficiency was not encountered in the 1st group. Its prevalence in the 2nd group reached 5,71% and in the 3rd group equaled to 0,07%. Thus, the cause of hepatitis in newborns and infants may be attributed to α_1PI deficiency approximately in 6% of cases. Hepatitis related to α_1PI deficiency was characterized by persistent jaundice, hepatomegaly, elevated serum level of conjugated and unconjugated bilirubin and hemorrhagic syndrome.

76 PATIENT TRIGGERED VENTILATION IN NEONATES

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Patient triggered ventilation (PTV), the delivery of positive pressure inflation initiated only by the infant's respiratory efforts, has been assessed in 32 infants of gestational age range of 24 to 36 weeks. A conventional newborn ventilator has been modified, such that in CPAP mode the manual breath control can be triggered by the infant's respiratory efforts to deliver a single positive pressure inflation. The triggering sensor is a pneumotachograph placed between the endotracheal tube and the ventilator circuit, this detects changes in flow and triggering occurs if the inspiratory flow exceeds 0.4 l/min. Inflation time during PTV mode was limited to 0.4 sec or less. Initially PTV was associated in the majority of infants with improvements in oxygenation ($p<0.01$) when contrasted with conventional ventilation. PTV was maintained till final extubation in 20 infants without complications. One infant only developed pneumothorax. In the remaining 11 infants, who tended to be less mature ($p<0.01$), PTV had to be discontinued after only a few hours. Predictors of failure of PTV at 1 hour were both a lack of improvement in oxygenation and a relatively slow triggering rate when related to gestational age. We conclude that PTV is a useful alternative for preterm neonates, but at present its use should be restricted to those more mature than 28 weeks gestational age.