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Low birth weight infants: evidence of decreased genetic variability

Some 40% of birth weight variation can be attributed to genetic factors, half of them being fetal and the other half maternal. "Normal variation" would give a 10% contribution to the small-for-dates population. The distribution of phosphoglucomutase 1 (PGM₁), adenosine deaminase (ADA), erythrocyte acid phosphatase (ACP₁) and adenylatekinase (AK) polymorphisms was studied in 91 low birth weight infants (LBWI). ACP₁ and ADA gene frequency distribution in LBWI showed a significant increase ($p < 0.01$ and $p < 0.05$) of the proportions of the more common alleles (P^b and ADA¹) and a reduction of heterozygotes as compared to adults or unselected newborn. The presence of multiple molecular forms of enzymes may protect the delicate balance of the developing zygote against the deleterious effects of environmental variations. Therefore the present data suggest that the (low) degree of heterozygosity for enzyme polymorphisms may represent a significant part of "normal variation" contribution to the low birth weight population.

56 ANDREW R. WILKINSON*, GEORGE A. GREGORY*, RODERIC H. PHIBBS* (Introduced by P. Johnson) Cardiovascular Research Institute and Depts. of Pediatrics and Anesthesia, University of California, San Francisco, U.S.A. Continuous Oxygen Saturation Recording during resuscitation of asphyxiated newborn infants.

We measured Oxygen Saturation (SaO₂) continuously in 29 severely asphyxiated infants (mean Apgar score 3.5 at 1 min), birthweight 1.05 - 2.7, mean 1.86 Kg. A new dual lumen umbilical arterial catheter containing fiber optics was positioned in the lower aorta. Red light transmitted down one fiber optic is reflected by the blood and the signal converted by the instrument to a digital reading of SaO₂. The other lumen is used to sample blood and measure pressure. Electronic calibration is made before insertion. Reliability was shown by comparison with SaO₂ measured in a cuvette oximeter (I.L. 182) $r = 0.98$, S.D. 2.46, $n = 139$. The time constant is 5 secs. and rapid changes in SaO₂ are seen during resuscitation. Immediately after the catheter is inserted changes in SaO₂ show the efficiency of assisted ventilation in improving blood oxygenation. Optimal levels of positive end-expiratory pressure and inspired oxygen concentration are quickly determined while blood sampling is reduced. In asphyxia and acidosis SaO₂ reflects oxygen content more accurately than PaO₂. The relationship between SaO₂ and PaO₂ for these infants shows that hyperoxia may be avoided if SaO₂ is kept below 96%.

57 Familial neonatal pulmonary haemorrhage associated with defects of the urea cycle

Four families are reported in which nine neonatal deaths occurred out of thirteen pregnancies. Pulmonary haemorrhage was found at necropsy in all nine patients. In three families citrullinaemia was responsible and in one the diagnosis was found to be arginino succinic aciduria. The diagnoses were made on the basis of hepatic enzyme assay, together with urinary and plasma amino-acid analysis. The dead infants were born near or at term and all were in excess of 2.6 kg (range 2.6-3.8 kg). The unusual finding of multiple deaths occurring in mature infants within a family should alert the neonatologist to the possibility of an inborn error of metabolism involving the urea cycle. The deaths typically occurred during the first five days of life in infants considered healthy at birth and who became hypothermic subsequently and developed diffuse neurological signs. As the pattern of deaths is particularly distressing to the parents, early investigation of suspicious cases may avoid repetition in subsequent pregnancies. Biochemical and necropsy data will be presented together with speculation as to the causes of the pulmonary haemorrhage which were not due to a generalised coagulopathy.

58 M.J. Pollitzer*, A.K. Morgan*, E.O.R. Reynolds, L.P. Soutter* and D. Parker*. Depts. of Paediatrics and Medical Physics, University College Hospital, London, England. Continuous comparison of in vitro and in vivo-calibrated transcutaneous oxygen tension (tcPO₂) with arterial oxygen tension (PaO₂) in infants with respiratory illness.

tcPO₂ simultaneously recorded by Dräger and Radiometer electrodes on the abdominal skin was compared for 6h-periods with aortic PaO₂ recorded by a Searle electrode in 10 newborn infants to test the accuracy of estimation of PaO₂ by tcPO₂. The infants' mean birthweight was 1997 (range 768-4560)g and gestation 33 (26-40)w. 5 needed mechanical ventilation. The skin-electrodes were heated to 44°C and calibrated in vitro and then in vivo. Half-hourly points were taken for analysis from the simultaneous records of tcPO₂ and PaO₂ over the range 20-120 mmHg. Using in vitro calibration values, the relation between tcPO₂ given by the Dräger skin-electrode and PaO₂ was $tcPO_2 = 0.79 PaO_2 + 6.85$ ($r = 0.86$, $p < 0.001$) and after in vivo calibration $tcPO_2 = 1.02 PaO_2 - 0.46$ ($r = 0.96$, $p < 0.001$): the slopes of these regressions were different ($p < 0.001$). For the Radiometer electrode $tcPO_2 = 0.98 PaO_2 + 1.07$ ($r = 0.92$, $p < 0.001$) after in vitro calibration and $tcPO_2 = 1.00 PaO_2 - 1.69$ ($r = 0.94$, $p < 0.001$) after in vivo calibration: these regressions were almost identical. We conclude 1. After in vivo calibration both skin electrodes gave a very accurate estimate of PaO₂ for 6h. 2. The Radiometer electrode gave as satisfactory an estimate of PaO₂ after in vitro as after in vivo calibration. 3. The Dräger electrode gave a significantly less accurate estimate of PaO₂ after in vitro than after in vivo calibration.

59 M. OBLADEN⁺, F. BRENDLEIN⁺, T. MENZEL⁺ (intr. by H. BICKEL) Univ.-Kinderklinik Heidelberg, GFR: Phospholipid patterns and surface activity of neonatal tracheal effluent.- Tracheal aspirates were analyzed in 150 newborn infants when therapeutic suctioning of the endotracheal tube was required. Dynamic surface tension measurements of hystereses (HYS) and minimal surface tension (γ_{min}) were performed. Using 2-dimensional TLC and Phosphorus-determination (P), the following phospholipids were identified: Sphingomyelin (SPH), Phosphatidylcholin (PC), Phosphatidylserin (PS), Phosphatidylinositol (PI), Phosphatidylglycerol (PG). Six patterns characteristic for the infant's clinical condition were found: 1. NORMAL TERM PATTERN: Wide HYS, $\gamma_{min} < 10$ dyn/cm, PC 61%, PI 10%, PG 10% P, L/S ratio 4.88, PI/PS-ratio 1.59. 2. RDS-EARLY STAGE: Narrow HYS, $\gamma_{min} > 20$ dyn/cm, PC 41% P, PI 12% P, PG absent, L/S-ratio 1.51, PI/PS-ratio 0.93. 3. RDS-RECOVERY STAGE: Narrow HYS, $\gamma_{min} 10-20$ dyn/cm, PC 59% P, PI 18% P, PG absent, L/S-ratio 5.5, PI/PS-ratio 3.2. 4. ACCELERATION (preterm infants with chronic prenatal stress like prolonged rupture of membranes): Wide HYS, $\gamma_{min} < 10$ dyn/cm, PC 62% P, PI 14% P, PG 4% P, L/S-ratio 4.6, PI/PS-ratio 2.8. 5. RETARDATION (delayed surfactant maturation like maternal diabetes): Narrow HYS, $\gamma_{min} > 20$ dyn/cm, PC 46% P, PI 11% P, PG 1.9% P, L/S-ratio 1.48, PI/PS-ratio 0.9. 6. INDUCTION (preterm infants after maternal glucocorticoid administration within 24 and 72 hours before birth): Wide HYS, $\gamma_{min} < 15$ dyn/cm, PC 59% P, PI 13% P, PG 7% P, L/S-ratio 4.94, PI/PS-ratio 2.51. Tracheal phospholipid composition changes after birth and during RDS. The presence of PG excludes RDS with a specificity of 0.84, its absence proves RDS with a sensitivity of 0.94.

60 W. ENDRES*, I. LEWANDOWSKI*, J. SCHAUB, F. HÖPNER*, F. JEKAT* and G. HOLLMANN*. Departments of Pediatrics and Pediatric Surgery, University of Munich; Chemical Institute, Oberhausen; City Hospital of Pediatric Surgery, Bonn-St. Augustin; Germany. Total parenteral nutrition in infants and children after abdominal surgery using a "breast milk adapted" solution of amino acids and hypertonic glucose solutions.

24 infants and 12 children were investigated. Normal intake of calories was achieved by infusion of glucose in increasing amounts from 9 to 20 g/kg/day. The 1st group received an amino acid solution (PE) based on the "potato-egg protein". The 2nd group was treated with a solution (BM) based on the relative content of amino acids in breast milk. The 3rd group received only glucose. The patients of the 3rd group had lowered plasma levels of Thr, Ser, Pro, Gly, Ala, Ileu, Leu, Tyr and Orn. In the groups receiving amino acids slightly increased plasma levels of Thr, Ser, Pro, Gly, Ala, Met, Phe, Orn and Arg were noted. Among the children nitrogen balance was more positive in the 1st group (PE) than in the 2nd group (BM). The supply of amino acids together with glucose had no influence on the blood glucose levels. The mean blood glucose level varied between 125 and 180 mg/dl. However, 2 infants sporadically had blood glucose levels up to 500 mg/dl.