185 ALPHA-1-ANTITRYPSIN DEFICIENCY AND EMPHYSEMA IN YOUNG CHILDREN. Michael F Hird, Anne Greenough, Giorgina Mieli-Vergani, Alex P Mowat. Dept of Child Health, King's College Hospital, London, UK

Alpha-1-antitrypsin deficiency (AlATD) in children has previously only been associated with liver disease whereas in adults emphysema is a frequent manifestation. We demonstrated children with AlATD are hyperinflated compared to children with other liver diseases and controls. We have now investigated if in children with AlATD hyperinflation persists and if persistent hyperinflation unresponsive to bronchodilator (BD) therapy (emphysema) was more common in children with AlATD than children with extrahepatic biliary atresia and healthy controls. Serial measurements of functional residual capacity (FRC) were made over at least 6 months. Children were persistently hyperinflated if their FRC was >120% of that predicted for height on at least 3 occasions. Measurements were made before and after BD, a change in FRC of >8% indicated bronchodilator responsiveness. 262 measurements were made on 54 patients with liver disease (mean age 6 yrs), 28 had AlATD. Their results were compared to those from controls of similar age. No control had an FRC >120% and their median FRC on 3 serial measurements was 93%, range 77-100%; 101%, 74-110% and 98%, 70-113%. Only 5 children, 4 with AlATD had persistent hyperinflation unresponsive to bronchodilator (emphysema), 2 were <5 years. We conclude emphysema may be associated with AlATD in early childhood.

186 RAFFINOSE AND STACHYOSE FREE DIET REDUCES GALACTOSE-1-PHOSPHATE IN ERYTHROCYTES FROM GALACTOSEMIC PATIENTS

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Most of the galactosemic patients, (GalP), in spite of a strictly galactose (gal) free diet, have elevated galactose-1-phosphate (gal-1-P) levels in their erythrocytes (EC), thought to be due to endogenous production of gal. Substantial amounts of bound alpha-linked galactose are ingested as oligosaccharides stachyose (S) and raffinose (R) from cereals, beans and potatoes. This gal is thought not to be absorbed, because no α-galactosidase exists in the intestine but liberated by bacterial α-galactosidase activity may cause flatulence by formation of CO_2 and hydrogen. Six GalP, age between 5 and 20 years, - after one week of recorded regular diet (R + S = 800 - 2000 mg/die) - received for another week a diet without S and R (<10%) but with the same amount of proteins, fat and carbohydrates. Then a recorded regular diet (R + S = 800 - 2500 mg/die) was reassumed. Gal in the serum and gal-1-P in EC was measured at the end of each period. Each GalP was taken as his own control. Gal-1-P in EC was 2.6±0.8 and 4.1±2.1 mg/100 ml. In five of six patients gal-1-P was significantly reduced at the end of the S and R free diet (1.5 + 0.9, $p<0.05$). The values before and after the diet were statistically not different from each other ($p<0.5$). Our results suggest that some of the G-1-P in the EC of GalP may originate from intestinal absorption of gal liberated by bacterial α-galactosidases. S and R diet, however, cannot be maintained for prolonged time because of lack of fi-

Maternal Phenylketonuria /PKU/

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Social and family status aswell as intellectual level of 34 PKU girls being in reproductive age was analyzed. The analysis included history of 15 pregnancies. There vere 4 abortions, 2 pregnancies not treated ended with birth of malformed offspring, 3 with low phe diet introduced again during first trimester of pregnancy and 3 treated before the conception and finalyzed with birth of normal healthy newborns. The course of treatment of pregnant women was satisfactory, they tolerated the diet well, and had low blood phe concentration. In all of them drop of phe level at the end of pregnancy was observed. Control of biochemical parameters revealed normal levels of plasma proteins, minerals and most of trace elements, urea and creatinine. Therapeutic problem was iron deficiency anemia which needed to be supplemented. Offspring from the gestations treated with low phe diet were born on time with normal Apgar score. Their psychomotorial development does not rise any objections. The oldest child is 5 years now and his IQ is 100.

Intellectual Outcome in Congenital cytomegalovirus infection.

Stuart Logan and Catherine Peckham.

188 Congenital cytomegalovirus (CMV) infection is now the commonest congenital infection in the UK. 35,000 newborn infants in three London hospitals were screened for the presence of congenital (CMV) infection. One hundred and three congenitally infected infants were identified. These children and their matched controls are being followed until 5 years of age.

Four congenitally infected children have major neurological handcap. A further 6 children have sensorineural hearing loss (unilateral in 2 cases), two in association with minor motor abnormalities.

It has been suggested that apparently asymptomatic children with congenital CMV infection are at risk of subtle intellectual impairment. Griffiths assessments on the children in this study at 2 years of age revealed no difference in developmental quotients between cases and controls. At five years the children complete Wecshler Pre-school and Primary Scales of intelligence tests. Results of those tests on the 70 asymptomatic cases and their matched controls who have reached 5 years will be presented.

Vertical transmission of HIV infection: The European Collaborative Study

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189 Participating centres include: Padua, Genoa, Valencia, Madrid, Barcelona, Brussels, Amsterdam, Berlin, Stockholm, Edinburgh and London

The European Collaborative Study is a prospective study in which children born to women known to be HIV positive at or before the time of delivery are being followed from birth. The main objectives are to determine the rate of vertical transmission and to describe the natural history of paediatric HIV infection. Children are examined using a standard protocol at regular intervals and each time a blood sample is taken. Infection is determined by the persistence of antibody beyond 18 months, the clinical presence of AIDS or the detection of virus or p24 antigen.

By the end of March 1990, 564 children were enrolled. The rate of vertical transmission based on the 248 children over 18 months was 25.5%. 16 of the 59 antibody positive children have been diagnosed with AIDS. Prognosis for antibody positive children is poor and mortality high. Early onset and rapid progression of symptoms poses problems for treatment.

GLUTATHIONE SYNTHESIS VELOCITY IN PRETERM BABIES.

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Red cells previously depletioned of glutathione incubated with amino acid precursors can be used to evaluate glutathione synthesis velocity (GSV) and thus, indirectly the measurement of cystathionase activity which renders indispensable for the step from cystathionine to cysteine. This can be achieved substituting methionine with NAcetylcysteine (NAC). Thus we do not need to use other tissues from living infants. We determined GSV at different GA and adult controls. Healthy newborns were grouped after GA:A(24-32wk); B(33-36wk) and C(term). Half of the red cells sampled were incubated with glutathione aa precursors (x10 plasma concentration); the rest with NAC instead of methionine. GSV determined by method of Brigelius.

GROUP		with aa (nmol/1'/g of tissue)		with NAC
A(n=10)	**	6.58 ± 2.66	**	24.09 ± 5.70
B(n=16)		18.65 ± 5.83	*	24.90 ± 4.02
C(n=13)	*	23.13 ± 2.07	NS	25.64 ± 1.74
Adults(n=16)		26.37 ± 4.01	NS	27.87 ± 3.34

Simple regression: GSV(aa): R=0.67 $p<0.01$; GSV(NAC): R=0.42 NS

1)Glutathione synthesis in prematures is lower than in term infants and adults. 2) In prematures when using NAC instead of methionine, GSV increases suggesting an impaired synthesis of cysteine. 3) Enzyme cystathionase matures probably along gestation.