931 CHROMOSOMES, SYNDROMES AND PERINATAL DEATHS. Patrick M. MacLeod, Fred J. Dill and David F. Hardwick. (Spon. by Sydney Segal). University of British Columbia Medical School, Departments of Medical Genetics and Pathology, Vancouver, British Columbia

Detailed postmortum examinations including chromosomal studies were performed on 100 unselected babies delivered in the Greater Vancouver Regional Hospital District during a one year period. Multiple congenital anomalies (MCA) were noted in 23 cases. Of these, a recognizable pattern of genetic malformation was identified in 18 cases and a chromosomal abnormality in 3 other cases. Chromosomal cultures grew in 8/59 cases of intrauterine deaths, 7/24 intrapartum deaths and 14/17 early neonatal deaths. The risk of recurrence was high (2-25%) in 11/23 of those MCA cases. Amniocentesis for prenatal diagnoses is available to monitor subsequent high risk pregnancies. Attention is drawn to the significant (20%) amount of recognizable heritable disease in cases of stillbirths and perinatal deaths.

932 APPLICATION OF ULTRASOUND AND FETOSCOPY TO PRENATAL DIAGNOSIS. Maurice J. Mahoney and John C. Hobbins. Depts. Hum. Gen. & Obs. Gyn., Yale U., New Haven, CT

Anatomic description of the fetus in utero would permit diagnosis of many birth defects which at present are not defined by biochemical or cellular abnormalities. We have used ultrasonography for indirect definition and fetoscopy for direct viewing of fetal anatomy in the attempt to establish or exclude the presence of certain morphologic abnormalities in the second trimester.

Three pregnancies at risk for recessively inherited Ellis-van Creveld syndrome were examined with grey scale and real time (B scan) ultrasound to measure limb lenghts, and with fetoscopy to look for polydactyly. In two of the pregnancies limb lengths were normal. Fetoscopy showed a normal foot of the first fetus and a normal hand of the second. The first pregnancy has given birth to a normal term infant and the second continues without problem. In the third pregnancy a fetal hand with six digits was seen with the fetoscope; in addition, ultrasound lengths of a humerus and femur were significantly shorter ($p<.001$) than direct postmortem measurements from control fetuses. Ellis-van Creveld syndrome was confirmed after elective abortion.

We examined one pregnancy at risk for recessive tetraphocomelia (Robert syndrome) with ultrasound; limb lengths and movements were normal and the pregnancy continues. Ultrasound measurements of fetal kidney and bladder size were normal in three pregnancies where previous newborns had died with Potter syndrome and aplastic kidneys; two normal infants have been born and one pregnancy continues.

(Sponsor: Leon E. Rosenberg)

933 HEAD GROWTH IN SICK PREMATURE INFANTS-A LONGITUDINAL STUDY. Keith H. Marks, Ellen Moore, Kathy Gifford, Zvi Friedman, M. Jeffrey Maisels (Spon by Nicholas M. Nelson), Penn State Univ Coll Med, M S Hershey Med Ctr, Dept Ped, Hershey, PA.

Serial weekly measurements of somatic growth and head circumference were made for 10 weeks on 45 Caucasian infants. They had major neonatal problems - 32 (71%) required prolonged assisted ventilation. All were AGA, gestations ranged from 26-32 weeks ($\bar{m}$ 30.4±1.6 wks) and caloric intake from 102-130 cals/kg/day by the end of the 2nd week. During the first 7 weeks, coincident with their illness, the velocity of growth was significantly below that of the normal fetus with deviation away from and below the fetal growth curve. For head circumference, velocity was 0.2 cm/wk vs. 0.9 cm/wk for normal fetal growth ($p<.05$). During recovery over the next 3 weeks growth paralleled that of healthy preterm infants. Subsequently rapid "catch-up" growth in head circumference occurred. By comparison, 6 similar infants whose head circumference followed the intra-uterine growth curve had proven hydrocephalus. These results suggest that: (1) Growth in the sick preterm infant does not proceed at the same velocity as in the fetus and the use of growth charts based on cross-sectional fetal growth may be misleading; (2) Hydrocephalus should be suspected in a sick infant whose head circumference follows the fetal growth curve; (3) In spite of an adequate caloric intake, the energy distribution of the sick LBW infant appears to prevent new growth until the acute illness has resolved.

934 RESISTANT CARDIOMYOPATHY ASSOCIATED WITH MENTAL RETARDATION IN DUCHENNE MUSCULAR DYSTROPHY. Gilbert Martin and Kenneth Frankel. (Spon. by P. W. Wehrle). Univ. of Southern California Sch. of Med., Inter-Community Hospital and the Magan Medical Clinic, Inc. Depts. of Pediatrics and Pathology, Los Angeles and Covina, Calif.

Duchenne Muscular Dystrophy (DMD) is a primary degenerative disorder of skeletal muscle which affects the myocardium in 50-95% of patients. Cardiac involvement varies from characteristic ECG changes (asymptomatic carriers) to terminal cor pulmonale in affected males. Early heart failure is rare and appears to be caused by an unusual pattern of myocardial fibrosis. Adjacent to areas of fibrosis there are hypertrophic, atrophic fibers surrounded by small islands of fat. This pattern is noted in the epimyocardium. The coronary arteries demonstrate a peculiar intimal pattern of smooth muscle degeneration in close proximaty to areas of epimyocardial fibrosis. Mental retardation occurs in up to 54% of patients with DMD. The association is thought to be genetically determined but not necessarily X-linked. Four patients with severe myocardopathy and DMD were studied. Three patients were mentally retarded. All patients had early heart failure resistant to usual cardiac medications and all expired within 6 days. These patients suggest that in the spectrum of DMD there is a syndrome of early severe refractory heart failure, mental retardation and epimyocardial fibrosis that is determined l by an yet unknown linked genetic factor.

935 GLYCEROLURIA, PSYCHOMOTOR RETARDATION, SPASTICITY DYSTROPHIC MYOPATHY, AND OSTEOPOROSIS IN A SIBSHIP. Edward R. B. McCabe, Mary Anne Guggenheim, Paul V. Fennessey, Donough O'Brien, Barbara Miles, Stephen I. Goodman. University of Colorado Medical Center, Department of Pediatrics, Denver.

We have identified two brothers who appear to represent a previously undescribed disease. Their clinical problems include poor somatic growth (less than 3rd percentile), moderately severe psychomotor retardation, generalized spasticity, a non-paralytic esotropia, severe generalized osteoporosis resulting in pathologic fractures, and a "wizzened" facial appearance. At ages 18 months and 4 years there is no evidence of a degenerative course. There is no demonstrable renal, thyroid, parathyroid or nutritional problem. Histochemical studies of muscle demonstrate a strikingly dystrophic process. Routine and EM studies of bone show nonspecific osteoporosis. Gas chromatography-mass spectroscopy of urine indicates elevated glycerol concentrations in both patients.

In a survey of an institutionalized, mentally retarded population, four patients were noted to excrete glycerol in their urine (Ann. Med. Exp. Fenn. 45:90, 1967). Three exhibited spasticity but no additional clinical information was presented. Our patients appear to represent a previously unrecognized, apparently genetic syndrome, characterized by psychomotor retardation, spasticity, dystrophic myopathy, osteoporosis, and glyceroluria.

936 CLINICAL AND BIOCHEMICAL EFFECTS OF VITAMIN D (D) ON PREGNANT RABBITS AND THEIR OFFSPRING. Douglas Mehlhorn, Gary M. Chan, John J. Buchino, Kevin E. Bove, Jean J. Steichen, Lori Abrams, Reginald C. Tsang. U of Cincinnati.

Maternal and fetal D homeostasis has not been studied during ingestion of large amounts of D in pregnancy. Twenty New Zealand white rabbits were mated and divided equally into one of five D_2 treatment groups receiving 10^5 u, $5x10^4$ u, 10^4 u, 10^3 u or placebo IM during pregnancy. At term, all newborns were delivered by C-section. There were 2 maternal deaths (from 10^5 and $5x10^4$ D group) and 18 abortions (6 of 10^5 D, 12 of $5x10^4$ D). The rest of $5x10^4$ D group failed to conceive. Increased maternal D was related to poor maternal weight gain ($r=-.47$, $p<.05$). At term, 10^4 D group had significantly higher total Ca, 17.2±0.2mg% (mean±SEM) vs 13.6±1.0 pre-study (paired t, $p<.05$); the $5x10^4$ D and 10^4 D had higher Ca (16.4±.5 and 17±0.2) vs controls, 13.3±0.96 (t test, $p<.05$). The other 3 groups were not different. Increased maternal D was related to decreased maternal-fetal gradient of D ($r=.58$, $p<.01$). Newborns from 10^4 D group had higher ionized Ca (Orion SS-20), 6.21±.28mg% vs 5.23±0.06 in controls ($p<.001$). Newborn 25-OH D (modified Belsey's) was lower in 10^5 D and 10^3 D, 25±1ng/ml and 23±1 vs 30±3 in controls ($p<.10$). Newborn ascending aortas (modified step-sectioning technique) showed focal proliferative supravalvular intimal lesions in newborns of rabbits given D but not in controls. Thus, high D ingestion in rabbits during pregnancy adversely affects maternal weight gain, fetal death and fetal Ca, maternal-fetal gradient of D, newborn D levels, and produces histological changes in the newborn aorta.