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THE 7p PARTIAL TRISOMY SYNDROME. Judith P. Willner, Sophie Paciuc, Steluta Cristian, Lillian Y.F. Hsu, Ramon Murphy, and Kurt Hirschhorn, Dept. of Peds., Mt. Sinai Sch. Med., New York, N.Y.

Various partial trisomy syndromes have been described since the advent of the chromosome banding techniques. However, a documented case of partial trisomy 7p has, as yet, not been reported. We have recently detected this aberration in a four month old female infant who presented with the following anomalies: asymmetric cranium with widely patent anterior fontanelle and metopic suture, deep widow's peak, hypertelorism, bilateral choanal atresia, prominent nasal bone, low set, rotated ears, maxillary hypoplasia, high arched palate, micrognathia, arachnoidactyly with contractures of the interphalangeal joints, congenitally dislocated hip, and vertical talus. The patient's banded karyotype revealed a 46,XX,Sp+ complement. With G, Q, and R banding, a 46,XY,t(5;7)(p15;p15) karyotype was found in the father. Studies on the mother were normal. Therefore, it is evident that the patient is trisomic for the distal short arm of chromosome 7: 46,XX,-5,+der(5),t(5;7)(5qter→5p15::7p15→7pter) pat.

The finding of this translocation will allow us to diagnose an affected child prenatally with amniocentesis in the subsequent pregnancies of this couple and in those of other translocation carriers in the paternal family.

Of further interest is the potential for using this translocation for regional gene mapping of genes known to be on chromosome 7 both by dosage effect and by somatic cell hybridization.

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BLOOD FLOW MEASUREMENTS IN CONGENITAL VASCULAR ABNORMALITIES OF THE LOWER LIMBS.

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An understanding of the vascular dynamics in limbs that are the site of congenital vascular abnormalities is of importance in understanding the etiology of these syndromes and planning management. Limb blood flow has therefore been measured by venous occlusion plethysmography in 28 patients with mixed vascular abnormalities of the lower limbs. The average blood flow in 17 patients with multiple diffuse arteriovenous fistulae was 12.7 ml/100g/min. (S.E. of Mean 3.4) compared with 3.8 ml/100g/min in the normal contralateral limbs. (S.E. of Mean 0.5). In 11 patients with radiologically confirmed Klippel-Trenaunay Syndrome the average blood flow was 8.1 ml/100g/min. (S.E. of Mean 1.7) compared with 3.0 in the normal contralateral limbs (S.E. of Mean 0.8).

A linear correlation was found between the measured blood flows and the extent of abnormal lengthening found in the affected limbs. ($r=0.75$).

These results support the view that abnormal arteriovenous communications may be present in limbs affected by Klippel-Trenaunay Syndrome where the lesions appear clinically and radiographically to be purely venous.

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VARIATIONS IN FINDINGS FROM NEONATAL AUTOPSIES
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Prominent lesions in 312 neonatal autopsies performed in our service between 1965 and 1970 inclusive were compared with those from 170 necropsies from the last 6 years. Autopsy rates were 90% and 83%. During these two periods 54.5% and 49.5% of deaths respectively occurred in infants less than 1000 gms. Incidence of babies of 500 gms. or less has remained over 10%. Predominant lesions in both periods have been hyaline membrane disease (HMD), pneumonia and intracranial hemorrhage (ICH). HMD has not decreased lately, but there is a higher incidence of early lesions and 40% of the infants were under 1000 gms. Pneumonias have diminished (34% to 26%), as also have omphalitis, septicemia and meningitis. However, necrotizing enterocolitis and systemic candidiasis have only been observed in recent years. ICH has also decreased from 30% to 20%. There has been a higher proportion of congenital malformations (excluding patent ductus arteriosus) from 9% to 15.8%. The most striking changes have been related to certain pulmonary lesions. Bronchopulmonary dysplasia present in 10 autopsies in the last 4 years was not seen previously, and complications due to extrapulmonary air have risen from 2% to 12.5%. Pulmonary hemorrhage has increased slightly. Traumatic lesions have been infrequent throughout the period of observation.

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CESAREAN SECTION BIRTH AS A FETAL STRESS. Bruce D. Ackerman and Mark J. Felberbaum, Downstate Medical Center of SUNY, Matmonides Med. Center, Brooklyn, NY. Umbilical artery (UA) blood gas values were analyzed for 31 infants born by cesarean section. The mean values for pH, PCO₂, PO₂ and BD were 7.23, 56mm Hg, 18mm Hg, and 9.2mEq/l. Values were similar for infants with and without labor or fetal distress. For 10 infants born by elective repeat cesarean section (ERCS) under spinal anesthesia (Sp An) the mean values were similar to those in 5 infants born by ERCS under general anesthesia. For those born under Sp An, the interval from Sp An to birth varied from 13 to 32 minutes. There was a very strong relationship between length of this interval and severity of the fetal metabolic acidosis. This interval was 24 minutes or longer for 5 of 6 infants with a BD of more than 9.0mEq/l. Fetal metabolic acidosis was unrelated to maternal blood gas values. A transient drop in blood pressure occurred for 7 or 10 mothers following Sp An. The drop was quickly reversed by intravenous ephedrine. Comparison of the present data with known values obtained in early labor by the Saling technique, and with data for in utero values in the fetal monkey suggest that cesarean section birth constitutes a metabolic stress similar to that caused by labor and vaginal birth. Metabolic acidosis probably occurs during normal labor as a result of decreased uterine blood flow. It is plausible that circulatory adjustments due to Sp An, even when hypotension is pharmacologically reversed, may cause a reduction in flow similar to that caused by labor.

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NONHYPEROXIC RETROLENTAL FIBROPLASIA
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Retrolental fibroplasia has traditionally required the combination of prematurity and exposure to high ambient oxygen concentrations. However, cases of RLF in stillborn infants as well as an infant near term never given supplemental oxygen have been reported. Two recent papers have emphasized an increased incidence of RLF in populations of preterm infants with respiratory distress syndrome who received exchange transfusions.

This report describes two infants near term who received exchange transfusions for hyperbilirubinemia and subsequently developed severe RLF in the absence of environmental hyperoxia and clinical respiratory distress. The case histories, clinical courses and retinal photographs will be presented. The possible etiologic role of the shift of oxygen-hemoglobin affinity caused by transfusions with adult erythrocytes with increased oxygen availability to the retinal vessels and tissues will be discussed.

The medical and legal implications of our cases and the recent reports are many. For one, the levels of oxygen considered safe for neonates receiving exchange transfusion may have to be re-evaluated. Secondly, serial thorough ophthalmologic evaluations in any neonate receiving adult erythrocytes by exchange transfusion or multiple transfusion replacement is crucial. Finally, the traditional relationship of hyperoxia and RLF must be expanded and re-evaluated to include other parameters like the hematologic status of the neonate and should include appreciation that this may be a phenomenon independent of environmental hyperoxia.

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EFFECT OF CAFFEINE ON CONTROL OF BREATHING: J.V. Aranda*, M. Mazzarelli*, L. Tamas*, M.J. Serysels*, and J. Milic-Emili. (Spon. by E. Colle) Roche Developmental Pharmacology Unit and Depts of Physiology, Pediatrics & Pharmacology, McGill University; Dept of Newborn Medicine, Montreal Children's Hospital, Montreal, Quebec, Canada.

Caffeine is used in the treatment of apnea in low-birth-weight infants despite inadequate data on the physiological basis for its efficacy. The effect of increasing doses of caffeine (10-70mg/kg) on pulmonary ventilation (\dot{V}_E), mean inspiratory flow (V_T/T_I) and tracheal pressure generated at 0.5 sec ($P_{0.5}$) after the onset of inspirations against occluded airways at functional residual capacity was studied in pentobarbital (35mg/kg) anesthetized cats, breathing various gas mixtures under steady state conditions. During room air and 50% O₂ breathing (balance N₂), increasing doses of caffeine caused a progressive increase in \dot{V}_E with corresponding increase in $P_{0.5}$ and V_T/T_I without changing T_I/T_T (ratio of inspiratory time to total cycle duration) indicating that caffeine primarily affects inspiratory drive. Increasing doses of caffeine also caused an incremental decrease in end-tidal PCO₂, causing a negative feedback on inspiratory center drive. When PCO₂ was maintained at control (pre-caffeine) levels, the increase in \dot{V}_E , V_T/T_I and $P_{0.5}$ at all caffeine levels was about 3-fold greater than non-is^o-CO₂ conditions. Thus, the concomitant hypoxia greatly masked the inspiratory drive by caffeine.

We conclude that caffeine markedly potentiates the effect on inspiratory drive. The therapeutic efficacy of caffeine in apnea is best explained by increase in inspiratory center output.