925 A NEW SYNDROME OF LETHAL MESOMELIC SKELETAL DYSPLASIA WITH OCULAR AND CARDIAC ABNORMALITIES

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We have seen two unrelated infants with a lethal form of mesomelic dwarfism, which to our knowledge has not been previously reported. Both cases had a flat face with low set ears and micrognathia, small rib cage, short bowed limbs with a maximal shortening of the middle segments, ulnar deviation of fingers of a normal length. One infant had a cleft palate. Microcornea and retinal dysplasia were found in the infant who survived longer. Both infants had congenital heart disease and neonatal respiratory distress and died at 1 and 30 days respectively. The radiograms of both infants showed among other abnormalities a separation of the dorsal and ventral ossification centers of the vertebral bodies by vertical radiolucent bars, severe shortening and bowing of the long bones with metaphyseal flaring, normal size of metacarpal and phalangeal bones. Differential diagnosis from other forms of lethal mesomelic dysplasias tabulated on the basis of the clinical and radiologic findings will be presented.

926 THE IMPORTANCE OF ULTRASOUND IN THE PRENATAL DETECTION OF CONGENITAL MALFORMATIONS.

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Congenital malformations are responsible for 15% of the neonatal deaths (Warkany & Fraser in Nelson's Textbook of Peds., 1975) The prenatal detection of some congenital anomalies may give the opportunity for an elective delivery with a forewarned medical and surgical approach to treatment. With the use of improved equipment for real time and gray scale ultrasound, it is possible to accurately evaluate details in the structure of fetal organs, e.g., the heart chambers, walls & valves; the kidney's collecting systems, ureters & bladder, etc. We report here a case of complex congenital heart disease with associated diaphragmatic hernia in which ultrasound was diagnostic during the 3rd trimester of pregnancy. The earliest possible treatment of infants with these disorders may be lifesaving. We diagnosed two cases of urinary tract obstruction and a case of urachal cyst. The early treatment of obstructive uropathy may save the infants from infection & renal failure. Hydrocephalus has been detected before viability (Freeman, et. al., Obstet & Gynecol, 1977) leading to a therapeutic abortion. The detection of this condition in the 3rd trimester & subsequent decompression of the fetal head may prevent an unnecessary Cesarean section. Since ultrasound is a noninvasive and an apparently safe technique, it should be considered as one of the primary tools for screening patients at high risk of congenital malformations.

927 ENDOCRINE STUDIES IN X-LINKED FAMILIAL PRECOCIOUS PUBERTY.

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The pathogenesis of this type of precocious puberty is not yet understood. Low urinary gonadotropins (Gn) have been reported but no RIA determinations are available.

Serum FSH, LH, testosterone (T)and timed urinary Gn, during awake(A) and sleep(S) periods, and 2 LH-RH challenges within 9 months interval were performed in 2 cousins with precocious puberty; an 11 9/12 yr old male with pubertal stage(PS)V and a bone age(BA)of 17 yr and a 7 6/12 yr old male with PS IV and a BA of 15 6/12. Both had low or undetectable values of serum and urinary FSH at any time and low baseline values of serum LH. Their response to the first LH-RH challenge was immature(early pubertal and prepubertal)for their PS. The second LH-RH challenge showed an appropriate response in the older patient with still an immature response for PS in the other. At this time A and S levels of serum FSH in the older were low whereas, LH and T were appropriate for PS. In the younger, levels of FSH and LH remained low with T levels in the range of adult men. The latter had 1.6 million spermatozoa in the seminal fluid. Urinary 17-KS, 17-OH, DHEA and serum estrone, DHEA-S and androstenedione were appropriate for the CA in both patients. The study suggests that familial precocious puberty is the consequence of a premature increase of testicular sensitivity to FSH and LH. Hypothalamic maturation and adrenarche are more in concordance with CA than with PS. Similar findings were recently reported in male patients with idiopathic precocious puberty.

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928 X-LINKED HYPOGONADISM, GYNECOMASTIA, MENTAL RETARDATION, SHORT STATURE AND OBESITY. A NEW SYNDROME.

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Five male members in four generations of the same family presented with hypogonadism, micropenis, mental retardation and short stature. The adults had gynecomastia, obesity and normal size hands and feet. Four had small head, with narrow forehead in two. History of neonatal hypotonia with difficulty in feedings was obtained in two. A variety of structural abnormalities of the hand and other skeletal defects were observed in some. Chromosomal studies including banding were normal. Serum LH and FSH were normal in three adults (12.3 and 4.5 mIU/ml; 9.0 and 7.0 mIU/ml and 22.5 and 18.1 mIU/ml). Serum testosterone levels were low (90 ng/dl; 54 ng/dl and 75 ng/dl) and responded to hCG with at least doubling of the initial value. Testicular biopsies in two adults showed patchy involvement with tubular shrinkage, folding of the basement membrane, thickening of the tunica propria and loss of germinal epithelium in some areas. Other tubules were less affected and showed spermatogenesis. Leydig cells were present. The findings suggest partial hypogonadotropic hypogonadism. Their phenotype and the abnormalities in their sexual development are most likely the result of a developmental anomaly of the CNS. The mode of inheritance, gynecomastia and absence of small hands and feet suggest that this disorder represents a new syndrome resembling but distinct from that described by Prader and Willi.

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NEONATOLOGY

929 THE ROLE OF EXCHANGE TRANSFUSION (ET) WITH SETTLED CELLS ON ALTERING MORTALITY IN VERY LOW BIRTH WEIGHT (VLBW) INFANTS < 1100 GMS WITH SEVERE RESPIRATORY DISTRESS SYNDROME (RDS) AT BIRTH.

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In infants of VLBW with severe RDS, in spite of supportive care including ventilation and ET, mortality remains high. The purpose of this study is to report the therapeutic benefits achieved by employing ET with fresh blood anticoagulated with citrate phosphate dextrose and allowed to "settle" to an Hct. of 55-60%. Thirty-nine infants 750-1100 gms. with severe RDS requiring an $FiO_2 > 60\%$ for $PaO_2 \leq 60$ mmHg within the first few hours of life were divided into 2 groups matched for weight and gestational age. All infants received supportive care including ventilation. Group I consisted of 20 infants 991 $\pm$ 104 g. who were exchanged within the first 8 hrs. of life with settled cells in addition to receiving supportive care. Group II consisted of 19 infants 929 $\pm$ 118 g. who served as controls. Mean hematocrit prior to exchange transfusion was 45 $\pm$ 1.86% and increased to 51 $\pm$ 1.9% post ET ($p < 0.03$). In non-exchanged infants the hematocrit was maintained at 45 $\pm$ 2.9% with boost transfusions. Twelve out of 20 infants exchanged survived (60%) while there was only 1 (5%) survivor in the non-exchanged group. These results indicate that early exchange transfusion with settled cells presumably by improved tissue oxygenation, increases the survival rate of VLBW infants with severe RDS and is a useful adjunct to supportive therapy.

930 DECREASING MORTALITY AND MORBIDITY IN INFANTS OF DIABETIC MOTHERS.

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The care of diabetic mothers and their progeny with modern techniques and methodology has led to a striking change in appearance and a marked decrease in mortality of 104 infants in our institution over the past decade. Now appropriate for gestational age infants (AGA) comprise approximately one half of these infants in contrast to one third previously. Mortality has fallen from 10% to 1.9%. A decrease in the incidence of hyaline membrane disease, hypoglucosemia, hypocalcemia, and neonatal asphyxia has accompanied these changes; yet, the incidence of malformations increased. Mortality and morbidity will be discussed in relation to gestational age and size of infants and maternal classification of disease. Determination of fetal age and condition and control of maternal diabetes and neonatal intensive care have been major responsible factors in achieving a mortality very near the total neonatal mortality.