NEUROFIBROMATOSIS IN CHILDREN, Bernard D. King, Mary Terlaak King and Stella B. Kontras, Ohio State Univ. Coll. of Med., Children's Hospital, Dept. of Ped., Columbus.

The literature suggests that the presence of 2 of the following is necessary to diagnose neurofibromatosis (NFT): 5 or more cafe au lait spots > 0.5 cm. in diameter, positive family history or biopsy-proven neurofibromas. Fifty-one children seen between 1952 and 1972 at Columbus Children's Hospital meet these criteria. Eighty-six percent showed neurofibromas, 53% had a positive family history and 80% showed cafe au lait spots. Although 22% showed cafe au lait spots at birth only 50% of the cases had been diagnosed by age 5. Fifty-five percent had bone abnormalities primarily kyphoscoliosis. Seven of these patients required surgical correction. Twenty-five percent had facial neurofibromas and showed gross facial asymmetry.

Other significant findings include mental retardation in 16%, developmental defects notably growth retardation and delayed sexual development in 22%. Involvement of renal arteries was noted in one patient and an ovarian teratoma and sarcoma in 2 others. Recognition of the diagnosis of NFT early would allow genetic counseling, attention to complications such as skeletal and growth difficulties, and surveillance for tumors.

QUANTITATIVE EVALUATION OF ABNORMAL FEATURES IN SELECTED SYNDROMES by Marilyn Preus, F. Clarke Fraser, M.D. and Murray Feingold, M.D., Department of Medical Genetics, The Montreal Children's Hospital, Montreal, and The Center for Genetic Counselling and Birth Defect Evaluation, Boston Floating Hospital for Infants and Children, Boston.

Statements that certain syndromes are characterized by abnormal dimensions of facial features or body proportions are often subjective and lack statistical backing. We have compared the cephalic index, inter outer and inner canthal distances, interpupillary distance, ear height and length, philtrum length, angle at elbow, hand size, hand/finger ratio, internipple/chest circumference ratio and upper/lower segment ratio in normal children and patients with DeLange, Noonan, and XO Turner syndromes. Many comparisons demonstrate statistically significant differences. For instance, in DeLange syndrome, the digits are short, relative to the palm. In Noonan syndrome the ears are low set but not appreciably so in Turner syndrome. The ratio of sitting height to height is increased in Turner syndrome as is the internipple to chest circumference.

THE CLINICAL SPECTRUM OF CONGENITAL CONTRACTURAL ARACHNODACTYLY: A CASE WITH CONGENITAL HEART DISEASE. Edward H. Lipson, Chirane Viseskul, Jürgen Herrmann, (Intro. by. C.C. Lobeck), Univ. of Wisconsin Ctr. for Health Sciences and Med. School, Depts. of Ped., Path., Madison, WI 53706.

The concept of congenital contractural arachnodactyly (CCA) as a connective tissue disorder distinct from the Marfan syndrome is extended by the observation of a patient with CCA and congenital heart disease. An infant who had features of CCA including joint contractures, arachnodactyly, and "crumpled" auricles died neonatally of cardiac malformations. Postmortem examination showed an ASD, VSD, bicuspid aortic valve, and an interrupted aortic arch. Family history is remarkable in that the mother has arachnodactyly.

Two further cases of CCA and congenital heart disease in the literature included a VSD in one instance and an ASD in the other. In contrast, the most common cardiac lesions in infants with the Marfan syndrome are mitral and aortic regurgitation. The valvar incompetence appears to result from stretching of connective tissue including dilatation of the mitral ring and elongation of the chordae tendineae. This difference in cardiac involvement seems to reflect a difference in the underlying abnormality of connective tissue in the two disorders. Abnormalities of connective tissue in the Marfan syndrome are diffuse and often progressive, while those in CCA are "spotty" and usually non-progressive. The cardiac involvement in our patient suggests that the clinical spectrum in CCA may be broader than previously assumed.

DIABETES MELLITUS, SHORT STATURE AND JOINT STIFFNESS -- A NEW SYNDROME. Arlan L. Rosenbloom, Ante Grgic and Jaime L. Frias. Univ. of Fla. Col. of Med., Dept. of Ped., Gainesville.

Three unrelated patients, 2 boys 18 1/2 and 19 years old and a girl aged 17, have stable insulin-dependent diabetes mellitus, short stature and joint stiffness beginning 8 to 13 years after the onset of diabetes. The older boy, after 16 years of diabetes, has small retinal hemorrhages, hypertension, azotemia and proteinuria. The younger boy developed diabetes at 14 months, has punctate retinal hemorrhages with no proteinuria, and the girl has proteinuria but no retinal changes; neither of these 2 has azotemia or hypertension. Stature of the 2 boys, who have attained final height following a normal adolescent spurt, is 17 and 25 cm. less than that of their fathers. The girl has familial delayed maturation and is near the end of an adolescent growth spurt with predicted final height 14 cm. less than maternal stature. Joint stiffness involves limited extension and flexion of interphalangeal, metacarpophalangeal, wrist and ankle in all 3, neck and trunk in 2, toes and elbows in 2 and knee and hip in one. The girl has pes cavus. There is no pain or disability and no progression of joint stiffness after the 6 months to 1 year period of its development. Slight but definite improvement in mobility has been noted in 2 during 2-3 years of follow-up study. Roentgenograms show no intrinsic or periarticular abnormalities. Skin is thickened and was unusually difficult to cut; biopsies were studied by light and electron microscopy and for collagen turnover in tissue culture.

AN INFANT WITH THE 4p- PHENOTYPE CARRYING A FAMILIAL t(4p-; 19p or q+) TRANSLOCATION. Richard L. Neu, Roger J. Shott, Lytt I. Gardner. State University of New York, Upstate Medical Center, Department of Pediatrics, Syracuse, New York.

To our knowledge, no translocations between the short arm of a number 4 chromosome and a number 19 have been reported. The case presented here is that of an infant with clinical features strongly suggestive of the 4p- syndrome. At the time of the patient's birth the mother was 34 and the father 36 years of age. Physical examination revealed a male with a large bilateral cleft palate, abnormal anterior fontanelle, abnormally-shaped ears, hypertelorism, a small penis with third degree hypospadias, a single testicle on the right side, stubby fingers and toes, and bilateral simian creases. There was constant seizure activity. Chromosome studies revealed what appeared to be a translocation of 4p material to an F group chromosome; subsequent autoradiographic and G-banding studies demonstrated a t(4p-; 19p or q+) translocation. The patient died at seven weeks of age following a seizure episode. The patient's phenotypically normal mother, maternal grandmother and an uncle were also found to have the same translocation. The patient's aneusomy could be explained using a model involving three chromosome breaks during ßogenesis resulting in the loss of a critical part of the translocated 4p material.

NON-RANDOM LATERALITY OF MALFORMATIONS IN PAIRED STRUCTURES. Bill S. Schnall and David W. Smith. Univ. of Washington Sch. of Med., Dept. of Ped., Seattle.

An evaluation of over 20,000 cases, encompassing 17 separate unilateral defects, revealed a statistically significant non-random propensity for sidedness for each aberration of morphogenesis studied. Some anomalies were strikingly left versus right sided, such as post-axial polydactyly (3:1) and cleft lip (2:1); while others were predominantly right sided, such as fibular aplasia (1:2) and radial aplasia (1:2). Of special interest was the left sided incidence of hemiatrophy versus the right sided predominance of hemihypertrophy. The propensity of certain potentially cryptic anomalies, such as dislocation of the hip and renal agenesis, to be left sided can be of practical relevance. This non-random preference for sidedness also exists for several teratogenically induced defects as well as in both normal and abnormal morphogenesis in several other mammals studied.

These findings imply subtle developmental differences between the two sides for each tissue, allowing one of the sides a greater liability for a particular type of defect in morphogenesis.