DIFFERENTIATION OF TYPES 1, 2 and 3 G_{M1}-GANGLIOSIDOSIS IN SKIN FIBROBLAST CULTURE. Leonard Pinsky, Jack Miller, Bernice Shanfield. Cell Genetics Lab., Lady Davis Inst., and Depts. of Ped. and Biol., McGill University, Montreal.

Type 1 G_{M1}-gangliosidosis presents in early infancy with a pseudo-Hurler phenotype. Type 2 begins about 1 year of age with neural but little or no skeletal involvement and no visceromegaly. β-galactosidase (β-gal) activities were measured with the 4-methylumbelliferyl substrate in strains from two type 1 and four type 2 patients. In normal strains, pH optimum is 4.4 and 95% of original activity is destroyed after 2 min at 55°C. A minor component is very heat stable and has greater activity at pH 3.8 than 4.4. In type 1 strains, residual β-gal activity is less than 1% of normal, has a pH optimum around 3.7 and is very thermostable. In type 2 strains, residual activity is about 3% of normal, has a pH optimum around 4.4 and its thermostability profile is biphasic, the larger component representing the major heat-labile species of β-gal in normal strains. One of our type 2 strains was derived from a patient whose liver β-gal had a pH optimum around 6.5 with the p-nitrophenyl substrate. Another was from a patient whose liver β-gal had a similar pH optimum with the 4-methylumbelliferyl substrate. Thus, the residual β-gal activity in type 2 is tissue-specific. The type 3 cell strain is from a patient with a clinical course less severe than type 2. Its β-gal activity is several times greater than those of type 2 strains and it stains normally with the indigogenic histochemical substrate for β-gal.

PLANTAR HALLUCAL PATTERNS IN BLACK MONGOLS. Q.H. Qazi, C. Ganapathy, H.C. Mapa, Dept. of Pediatrics, Downstate Med. Ctr. Brooklyn, N.Y.

The plantar hullucal dermatoglyphic patterns were studied in 49 Black mongols compared to the patterns of 200 Black controls. The results of the study along with those of White controls and mongols (Walker 1958) are summarized in the Table.

	Pattern Frequencies			
	White		Black	
	Controls (500)	Mongols (200)	Controls (200)	Mongols (49)
Arch Tibial (A^t)	0.3	47.0	0.3	55.1
Small Loop Distal (l^d)	11.7	32.5	3.0	25.5
Large Loop Distal (L^d)	41.5	13.2	14.5	16.3
Vestigeal Loop Distal (v^{ld})	0.0	3.4	1.3	2.1
Loop Tibial (L^t)	9.9	1.8	19.8	1.0
Whorl (W)	31.7	1.8	57.3	0.0
Others	4.9	0.3	3.8	0.0

Pattern frequencies in Black and White controls are significantly different: l^d and L^d patterns are more frequent in White controls while W and L^t patterns are more frequent in Black controls. However, the frequencies of plantar hallucal patterns in White and Black mongols are not significantly different from one another. This observation suggests that the Down's anomaly produces similar dermatoglyphic changes in Black and White mongols independent of existing racial differences in the pattern frequencies.

THE CELL CYCLE AND CHROMOSOME ANOMALIES, Ian H. Porter and Betty Paul, New York State Health Department, Birth Defects Institute and Department of Pediatrics, Albany Medical College, Albany, New York.

Delayed growth, retarded intellect, premature senescence, shortened life expectancy, and other clinical signs which autosomal trisomy syndromes have in common and, perhaps, the increased susceptibility to malignancy, suggest a basic derangement of cellular regulation.

As patients with Down's syndrome have a decreased rate of DNA synthesis both in fibroblasts and in lymphocytes and as patients with trisomy 13, 18 and 21 have less than normal numbers of cells in different organs, the question arises whether these findings may be related to alterations in the cell cycle. We measured the length of cell cycles in patients with abnormal chromosomal constitutions by pulse labeling cultured fibroblasts and counting the proportion of labeled metaphases every 3 hours for 30 hours. Patients with trisomy 13, 18 and 21 and other chromosomal anomalies all demonstrated an increase in the total length of the cell cycle and particularly in the length of G_1.

Thus, it would appear that the addition of a particular chromosome has a *general* effect on the length of the cell cycle leading to slowing of the rate of cell proliferation and consequently to growth retardation and, perhaps, also a *specific* effect on the differential mitotic rate at certain developmental stages at times when increased activity of particular cells is required, leading to specific congenital anomalies.

EARLY CHILDHOOD DEVELOPMENT OF FOUR BOYS WITH 47,XXY KARYOTYPE. Arthur Robinson, Mary Puck, Katherine Tennes, Kathleen Bryant, Univ. of Colo. Med. Ctr., Denver, Colo.

Infants identified at birth in an epidemiological study of sex chromosome anomalies are enrolled in a voluntary long-term evaluation program. Case histories of the first four boys in the series with 47,XXY karyotype are presented. They have been followed from birth for six to nine years with regular and frequent physical and psychological evaluations. Close cooperation with the children's parents was maintained to provide families with supportive consultation. Results demonstrate that their development so far clearly falls within normal range. Minor deviations in motor, speech, and emotional development suggest a common underlying pattern, although four cases are too few on which to establish a relationship between karyotype and phenotype. The data, however, suggest that symptomatology which has been reported in selected children with a 47,XXY karyotype may be strongly dependent on factors other than the chromosomal constitution, and that appropriate environmental sustenance may minimize elevated risks due to the marked genetic defect.

DERMATOGLYPHICS AND SYSTEMIC LUPUS ERYTHEMATOSUS (SLE), Q.H. Qazi, S.M. Fikrig, E.M. Smithwick, E.R. Grimes and D. Kaplan, Dept. of Pediatrics, S.U.N.Y., Downstate Med. Ctr., Brooklyn, New York

Dermatoglyphic patterns were studied in 32 black female patients with SLE and compared to 100 black female controls. The digital pattern frequencies are shown below.

	Digital Pattern Type Frequency			
	UL	RL	W	A
Patients	224 (70%)	7 (2.2%)	72 (22.5%)	17 (5.3%)
Controls	588 (58.8%)	17 (1.7%)	313 (31.3%)	82 (8.2%)

Patients with SLE, compared to the controls had a significantly increased frequency ($p<0.005$) of ulnar loops and decreased frequency ($p<0.005$) of whorls. No significant differences were found in pattern frequencies in hypothenar, thenar, third and fourth interdigital areas and summed atd angles. The abnormal dermatoglyphic pattern frequencies noted in the present study is not sufficient to aid in the diagnosis of SLE but the association may represent a prematal and/or genetic characteristic which may contribute to the understanding of SLE.

DIRECT CHROMOSOMAL LOCALIZATION, ISOLATION, AND CHARACTERIZATION OF A SPECIFIC HUMAN GENE. Roy D. Schmickel and Mechthilde Knoller. Univ. of Michigan Med. Sch., Dept. of Ped., Ann Arbor.

Although genetic diseases are usually described in terms of enzyme function and structure, further understanding of genetic mechanisms in the human will also depend upon the ability to physically isolate and characterize specific human genes. We have used thermal fractionations, molecular hybridization and sequential equilibrium density gradients to purify the human genes of 28S and 18S RNA - the nucleic acid components of ribosomes. The genetic sequence was isolated as a rRNA/DNA hybrid which was homogenous as demonstrated by a sharp melting transition at 80° in 0.1 x SSC. Linkage of 28S RNA and 18S RNA genes on the same isolated sequence was demonstrated by isolation of 28S RNA/DNA hybrids, followed by 18S hybridization with that molecule. The specific gravity of the hybrid (1.745) indicates that 30% of the DNA sequence is complimentary to 28 & 18S RNA. The remainder is probably accounted for by transcribed precusor molecules and nontranscribed "spacer" regions. The chromosomal localization of rDNA was possible by a method in which we added ^{3}H-Uridine to human cells auxotrophic for uridine to achieve rRNA of very high specific activity and purity. In situ hybridization of this rRNA with chromosomes demonstrated that the ribosomal genes were located only on the D and G group chromosomes. The potential for studying fine chromosomal structure by this technique is demonstrated by the absence of ribosomal RNA genes on a D/D translocation chromosome.