father's cells showed decreased O_2 uptake below the average for normal males (P/B of 9 compared to normals of 11.83 ± 1.94).

It is suggested from these studies that leukocyte O_2 uptake may be of value in detecting asymptomatic carriers of CGD for more precise genetic counseling.

α -1,4 Glucosidase activity in Pompe's disease. IRA S. SALAFSKY and HENRY L. NADLER. *Northwestern Univ. Med. Sch., Children's Mem. Hosp., Chicago, Ill.*

α -1,4 Glucosidase (α -glu) has been shown to be absent in cultivated amniotic fluid cells (AFC), fibroblasts (FIB) and liver (LIV) of patients with Pompe's disease. In contrast, α -glu is present in the kidney (KID) and amniotic fluid (AF) of these patients. In an attempt to explain these differences, properties of α -glu including pH optima, turanose inhibition and heat inactivation were studied in controls (C) and Pompe's (P). The results are as follows:

	Specific Activity*		pH Optima		Turanose Inhibition†		Heat Inactivation‡	
	C	P	C	P	C	P	C	P
AF	5.4	14	6.0	6.0	47	51	7	8
AFC	5.1	0	4.0	—	92	—	100	—
FIB	5.9	0	4.0	—	90	—	100	—
LIV	9.3	0	4.0	—	94	—	100	—
KID	10.3	7.5	6.0	6.0	33	53	100	100

* umoles maltose hydrolyzed/min/gm protein.

† % inhibition by 0.03 M turanose.

‡ % activity remaining after heating at 45°C for 15 minutes.

No α -glu activity could be demonstrated in urine or maternal serum. α -glu in amnion had properties identical with FIB and LIV enzyme. Upon differential centrifugation of the cell-free amniotic fluid, cells and tissues, α -glu was found primarily in the 25,000 $\times$ g fraction. These data clearly indicate that the α -glu present in AFC, FIB and LIV have identical properties which are distinctly different from the α -glu in AF and KID. The α -glu in AF differs from that found in KID in its greater heat lability. These studies fail to identify the origin of the α -glu in amniotic fluid. The diagnosis of Pompe's disease *in utero* must rest on the demonstration of the α -glu deficiency in amniotic fluid cells.

Testicular feminization: Expression in sex skin fibroblast culture.

LEONARD PINSKY, SHREE MULAY, ROSANNA WEKSEBERG, and SAMUEL SOLOMON. *Lady Davis Institute, Jewish General Hosp., Royal Victoria Hosp., McGill Univ., Montreal, Que., Canada* (Intr. by Charles R. Scriver).

The inheritance and pathogenesis of human testicular feminization (TF) are unknown. The possibilities of X-linked recessive inheritance and primary target organ refractoriness to testosterone (T) could be studied optimally in serially subcultured cell strains. Fibroblasts derived from sex and non-sex skin of males, females and patients with TF were incubated with T-4- 14 C. T and its metabolites in the culture medium were separated by paper chromatography and identified by reverse isotopic dilution. The rate of T metabolism was much greater in sex than in non-sex strains. For both newborn foreskin and adult labial strains, androstenedione was the primary metabolite to T, however they differed in their rate of production of 5 α -androstenedione and andosterone. In a strain derived from the foreskin of a 7-year-old, 5 α -androstenedione, rather than androstenedione, appeared to be the major early metabolite. 5 α -dihydro-testosterone (DHT) was

a major metabolite in none of the strains. Sex and non-sex strains from two patients with TF were indistinguishable: Both had low rates of T metabolism. We conclude that: 1) *in situ* differences in T metabolism between normal sex and non-sex skin persist in their serially subcultured fibroblasts; 2) an expression of the TF gene is detectable in sex skin fibroblasts without reference to the rate of DHT formation; and 3) Lyonization of cloned heterozygous fibroblasts may prove that human TF is X-linked.

Evidence linking an extra Y chromosome to sociopathic behavior. LYTT I. GARDNER and RICHARD L. NEU. *State Univ. of New York Upstate Med. Ctr., Syracuse, N. Y.*

There is accumulating evidence that certain XYY males are more prone to sociopathic behavior than are XY males, and that the extra Y chromosome may be causally related to this behavior. Eleven studies were surveyed which the percentage of XYY were identified among selected male populations in prisons and mental hospitals. The men were selected for height (over 59 in.), having dangerous or violent propensities, or for being mentally subnormal. These percentages were contrasted with the findings of XYY males in several surveys of newborn populations. There was found a strikingly higher incidence (1.8 to 12.0 per cent) of XYY males in institutions than in the general populations as determined from newborn surveys (0.14 to 0.38 per cent). It could appear that only a small fraction of the total numbers of XYY males known to exist in the population are institutionalized sociopaths. The XYY sociopaths represent a numerically small subdivision of the large group of mostly normal XYY individuals (there is some indication that a nosologic classification is developing, since XYY males with abnormal genitalia appear to represent a discrete subgroup). The implications of these data are obvious in the counselling of parents of XYY children. It is especially important that the physician allay the fears created in parents by the numerous popular articles linking the XYY karyotype to criminality through a careful presentation of the data thus far at hand.

Perinatal expression of the Lewis and secretor blood group systems. MINERVA B. ARCILLA and PHILLIP STURGEON. *U. C. L. A. Sch. of Med., Los Angeles, Calif.*

Studies on expression at birth and on maturation of the secretor and Lewis blood group systems, as manifest on red cells and in saliva, have been carried out on a series of infants using three Lewis reagents: anti-Le^a, -Le^b and -Le^x.

Among adults, 20% are Lewis red cell type Le(a+b-x+) and 70% are Le(a-b+x+); both types are essentially absent at birth. A third type Le(a-b-x-) is found in both 10% of adults and in 10% of infants; 90% of newborn infants, however, have the unique type Le(a-b-x+). Infants of the latter type undergo a rapid maturation during the first week of life to the adult Le(a+b-x+) type. Then, if they are salivary non secretors of ABH, they remain that type throughout life. Whereas those who are secretors evolve during the ensuing four months to Le(a+b+x+) which is also a type practically unique to infancy; finally, by two years of age, they evolve to the most common adult type, Le(a-b+x+).

Salivary phenotypic expression of the genes involved in the above red cell types is found in soluble form but the state of development at birth is more advanced than on the red cell and the subsequent maturation is more rapid. Prematures show a relatively delayed maturation in the expression of these genes. Thus, unlike other blood group systems, the Lewis system is unique in infancy; it shows differential tissue expressivity and,

with growth and development, undergoes a systematic pattern of distinct qualitative changes.

The biological significance of the phenotypic expression of this blood group system during fetal life and of its well organized maturation process during early infancy remains obscure.

Evidence of interaction between the polymorphism of placental alkaline phosphatase and those of Rh and Abo systems. E. BOTTINI, P. LUCARELLI, P. PIGRAM, R. PALMARINO, G. F. SPENNATI, L. TERRENATO, and M. ORZALESI (Intr. by C. D. Cook). *Sch. of Sci. and Sch. of Med., Rome, Italy, and Yale Univ. Sch. of Med., New Haven, Conn.*

Placental alkaline phosphatase presents electrophoretic polymorphism with 6 common phenotypes determined by 3 alleles (PI^{s1} , PI^{t1} and PI^{l1}) at an autosomal locus. The electrophoretic phenotypes of alkaline phosphatase from 2000 consecutively delivered placentas of white and negro subjects were determined. The results were subdivided according to race and to the presence or absence of Rh and/or ABO feto-maternal incompatibility. In the group of ABO incompatible infants, a further subdivision was made according to the presence or absence of a positive direct Coombs test and/or jaundice. Within the compatible groups the frequency of allele PI^{s1} was significantly higher in "first born" than in "subsequently born" infants. The reverse was true in the incompatible groups. Among infants with ABO incompatibility, allele PI^{t1} was significantly more frequent in infants without evidence of isoimmunization than in those with a positive Coombs test and/or jaundice. These results suggest that placental alkaline phosphatase may play a role in the maintenance of the fertilized ovum in utero and that in this regard allele PI^{s1} may be more efficient than allele PI^{t1} . These results also suggest the possibility of an interaction between the polymorphism of placental alkaline phosphatase and those of Rh and ABO systems whereby, in the presence of feto-maternal incompatibility, allele PI^{t1} seems to provide an advantage when compared to allele PI^{s1} .

Gonadal function in mosaic XO or XX/XY Turner's syndrome. THOMAS MOSHANG, JR., H. LAWRENCE VALLET, CARLOS CINTRON, ALFRED M. BONGIOVANNI, and WALTER R. EBERLEIN. *Univ. of Pennsylvania Sch. of Med., The Children's Hosp. of Philadelphia, Philadelphia, Pa.*

Gonadal function in four patients with mosaic XO or XX/XY Turner's syndrome was reviewed. Luteinizing hormone (LH) levels were determined by double antibody radioimmunoassay. Urinary testosterone (UT) and plasma testosterone (T) levels were determined by either double isotope or competitive protein binding methods. Three patients had ambiguous genitalia and exploratory surgery revealed only medullary elements in the gonads. Two virilized at puberty and were studied prior to surgery. One had normal LH levels (11 mIU/ml), a normal excretion of UT (0.5 μ g/24 hrs) but responded to human chorionic gonadotropin (HCG) with a ten-fold increase in UT. The other had castrate levels of LH (≥ 100 mIU/ml) and adult male levels of plasma T (208 m μ g%) which did not increase with HCG. The third patient at age 4½ years was found to have an undifferentiated streak and, contralaterally, a testis and epididymis. The fourth patient, 14 years of age, has no evidence of any adolescent development.

Prophylactic gonadectomy in mixed gonadal dysgenesis has been recommended because of the increased incidence of gonadal

malignancy. Our patients support the recommendation for early surgery. It appears that waiting until post-puberty serves no purpose because if the gonads are to function at all, they are more likely to produce androgens.

Screening of newborns for sex chromosomal abnormalities by a fluorescent technique. ARNOLD GREENSHER, ROBERT GERSH, DAVID PEAKMAN, and ARTHUR ROBINSON (Intr. by William Hathaway). *Univ. of Colorado Med. Ctr., Denver, Colo.*

The importance of screening newborn infant populations for abnormalities of sex chromosomes has been demonstrated. We report an improved method of identifying Y chromosome abnormalities using the affinity of this chromosome for the fluorescent dye Quinacrine Hydrochloride. Cells obtained from Wharton Jelly of the umbilical cord were used. Also, these cells exhibit fluorescence of the Barr body in female cells, a hitherto undescribed phenomenon. This body differs significantly from the fluorescent Y body in its size, form and intensity of staining. Touch preparations of a freshly cut section of the cord were prepared and were stained with Quinacrine Hydrochloride. Blind sex determination was carried out on slides prepared from the cords of 249 infants. All of the 120 females were correctly diagnosed by the fluorescence of the Barr body. One hundred male specimens were correctly identified by the presence of a fluorescent Y body and absence of a fluorescent Barr body.

Three hundred fifty males have been screened for abnormalities in the number of Y chromosomes and thus far none have been found. A sequential staining technique using Quinacrine Hydrochloride and Carbol Fuchsin on the same specimen verified the identity of the fluorescent Barr body with the conventional Barr body. In addition to the obvious power of this technique in screening newborns for sex chromosomal abnormalities we believe it may be of great use in the rapid diagnosis of sex in utero.

Identification of human translocation chromosomes by quinacrine fluorescence patterns. W. R. BREG, D. A. MILLER, P. W. ALLDERDICE, and O. J. MILLER. *Southbury Training Sch., Southbury, Conn., Yale Univ. Sch. of Med., Div. of Med. Genetics, New Haven, Conn., and Columbia Univ., Coll. of Phys. and Surg., N. Y., N. Y.*

Quinacrine stained human chromosomes fluoresce with distinctive patterns when exposed to UV light. These patterns are detectable visually in high contrast photographs of metaphase figures and permit the identification of each chromosome.

A variety of cases with translocation chromosomes have been studied by this method in our laboratories and the chromosomes involved identified with much greater accuracy than previously possible. Included are the following translocations: (3p-; 18q+), (5p-; 14q+), (5p+; 10q-) [preliminary], (13q13q), (13q14q), (14q21q), and (21q21q). The fluorescence patterns are distinctive enough, for instance, to differentiate the t(14q21q) from the C-group chromosomes and a t(13q13q) from the chromosomes no. 3.

This method now provides a rapid means for distinguishing cytologically between t(21q21q) and t(21q22q). Also, of significance is the relative ease with which the 14q+ chromosome in the cells with a t(5p-; 14q+) can be recognized. That a D-group chromosome was involved in this translocation was previously shown only with great effort (deCapoa et al. *Am. J. Hum. Genet.* 19, 586, 1967). It should now be possible to differentiate more readily between the several possible abnormal karyotypes and the normal one in the progeny of such a translocation carrier.