

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  $PI^{11}$ ) 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 fetomaternal 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 fetomaternal 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  $\mu$ g/24 hrs) but responded to human chorionic gonadotropin (HCG) with a ten-fold increase in UT. The other had castrate levels of LH ( $\geq 100$  mIU/ml) and adult male levels of plasma T (208 m $\mu$ 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.