742 LIPOPROTEIN CORRELATIONS IN PARENTS AND THEIR ADULT AND PEDIATRIC PROGENY.

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Parent-adult progeny(>20yrs old)(P-AP), and and parent-pediatric progeny (6-19 yrs old)(P-PP) correlations for plasma low and high density lipoprotein cholesterols(C-LDL,C-HDL) were assessed in randomly recalled white subjects in the LRC Princeton school family study to compare intrafamilial lipoprotein associations among family members still sharing and no longer sharing a common household environment. Father(F)-son(S), F-daughter(D),mother(M)-S, M-D C-LDL and C-HDL correlations in the 75 P-AP pairs and 245 P-PP pairs, (*p<.05,**p<01), were as follows:

	F-S C-LDL	F-D	F-S C-HDL	F-D	M-S C-LDL	M-D	M-S C-HDL	M-D
P-AP	.416*	.441**	.012	.236	.068	-.197	.345	.303
P-PP	.384**	.426**	.116	.215*	.333**	.106	.240*	.361**

For C-LDL, F-S and F-D relationships were significant and comparable for both P-AP and P-PP pairs; M-S and M-D C-LDL correlations were less consistent than for F-S or F-D, and were significant only for M-S in the P-PP pairs. For C-HDL, in contrast to C-LDL, there were no significant P-AP relationships. For C-HDL, in P-PP pairs, the M-S, M-D, and F-D relationships were significant. The general similarities in C-LDL relationships in P-AP and P-PP pairs, particularly in F-S and F-D, suggest a lasting genetic influence on C-LDL. The loss of significant P-AP C-HDL relationships, when compared to those in P-PP suggests a substantial differential environmental effect when common households are no longer shared.

743 PRENATAL IDENTIFICATION OF AN ADRENOLEUKODYSTROPHY HETEROZYGOTE

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Adrenoleukodystrophy (ALD) is an X-linked disorder associated with the accumulation of saturated very long chain fatty acids, particularly hexacosanoate (C26). Cultured skin fibroblasts show an increased ratio of C26 to C22 fatty acid (Annals of Neurol. 7:542, 1980). Fibroblast clones from ALD heterozygotes show two types of cells, one with a normal C26 to C22 ratio, the other with a ratio similar to the ALD hemizygote (Migeon,B.R. et al. Am. Journal of Hum. Genetics 32:157A, 1980). We have studied a large kindred which included heterozygotes for both ALD and an electrophoretic variant for G6PD, and found that in this kindred the ALD trait was associated with the G6PDA variant. One woman known to be ALD heterozygote by clonal analysis and G6PD type A had an elective abortion for reasons unrelated to ALD. A female fetus was found to have the G6PDA electrophoretic variant, and the fetal liver contained 5-10 times more C26 than did controls. Duplicate cultured amniotic fluid cells had a C26 to C22 ratio of 1.23 and 1.75 compared to 0.167 $\pm$ .094 (SD) in 16 control cultures. Ability to identify prenatally the ALD heterozygote makes it likely that it will be possible to achieve the prenatal identification of the ALD hemizygote.

744 PARTIAL TRISOMY 8q,

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Full and partial trisomies have been well delineated. A male infant who died within 72 hours following an apneic episode showed clinical features of trisomy 8: abnormal facies, cloanal atresis, congenital heart disease, hypospadias, broad thumbs and toes, and tracheoesophageal fistula.

Cytogenetic studies performed on the child and parents revealed the mother to be a reciprocal translocation carrier, [46,XX,t(8;13)(q11;q34)]. The child's karyotype was 46,XY,-13, +der(13),t(8;13)(q11;q34)mat. The father and a 4 year old female sibling were chromosomally normal. The maternal grandparents were not available for karyotype analysis.

The family history is noteworthy. The mother is said to have had a sibling who died within the first year of life with central nervous system malformations. Several generations (maternal side) in whom central nervous system defects and other malformations occurred associated with death within the first two years of life. These data suggest instances in which a syndrome similar to that of the propositus may have occurred.

745 A PENTA X FEMALE (49,XXXXX): A RESULT OF PARENTAL MOSAICISM?

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Penta X syndrome is a rare disorder since less than 10 cases have been reported. This communication describes an additional case of pentasomy X in a three-week old white female infant born at full term to a 22 year old mother. The birth weight was 3000 grams. Abnormal findings in the patient were hypertelorism, micrognathia, rather small and drawn back ears, simian creases, bilateral clinodactyly and congenital heart disease (tetralogy of fallot). The phenotype of this infant was distinct from that seen in Down's Syndrome. There is no family history of birth defects or other congenital anomalies. One to four Barr bodies were observed in her buccal smear analysis. Cytogenetic analysis of the peripheral blood and skin by Giemsa banding revealed a 49,XXXXX karyotype with no mosaicism or structural abnormality. Both parents and a 2 year old male sibling were also analyzed by C- and G-banding. The sibling had a normal karyotype. However, the mother's karyotype showed 8 percent 47,XXX, and the father's karyotype 6 percent 47,XXY along with a normal genetic complement. The majority of polysomies are due to double nondisjunction in oogenesis, but in this case either one or both parents may have contributed toward the anomaly.

746 A DE NOVO Y/Y TRANSLOCATION WITH XO MOSAICISM IN A VIETNAMESE INFANT,

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A two-day old Vietnamese infant with ambiguous genitalia was referred to us for cytogenetic evaluation. He was the 2680 gram product of a full term pregnancy to a 20 year old mother. There was no family history of usage of drugs or birth control pills, or congenital anomalies. Physical findings were normal except that the genitourinary examination revealed a 3 cm long phallus with bifid scrotum, hypospadias, bilateral palpable gonads, normal kidneys, and midline ovoid structure consistent with an infantile uterus. There was no physical evidence of a vagina. Blood chromosome analysis by G-banding revealed two cell populations: 48 percent with 45,XO and 52 percent with 46,X,t(Y;Y)(p11;q12). The abnormal chromosome resembled a C-group chromosome in conventionally stained cells since the Y chromosome in Oriental populations normally has a large amount of heterochromatin. The heterochromatic segment involved in the translocation was confirmed by Q- and C-banding. The father's karyotype was normal, and the centromeric index of his Y chromosome was used to confirm our findings. The predominance of testicular tissue is due to the presence of complete euchromatic segments of the Y chromosome while the ambiguity of the genitalia may have resulted from the XO mosaicism.

747 UTILIZATION OF GENETIC SERVICES AMONG PREGNANT WOMEN IN AN URBAN CLINIC POPULATION,

Donna O'Hare, Milagros A. Benedicto

The Maternity, Infant Care-Family Planning Projects of New York City is a Title V and XIX funded program that provides prenatal and family planning services to women in 10 clinics located in high risk areas in New York City. Genetic counseling and screening are offered routinely to pregnant women in need of such services.

A 2-year review of records of randomly selected women 35 and over was conducted in order to assess the need for genetic studies for advanced maternal age among the approximately 10,000 pregnant women registered annually for prenatal care in the Project.

In 1978 and 1979, 4.5% of women registered for prenatal care were 35 years of age and older. 165 records were audited which represented 21% of the total target population. The records were reviewed: for 1) age of gestation at registration; 2) provider compliance with agency policy to inform those targeted patients, less than 20 weeks gestation at registration, of the increased risk of Down's Syndrome and the availability of prenatal diagnosis; 3) frequency of patient acceptance of the offered referral for genetic counseling and studies. Of those eligible for genetic studies, approximately 50% accepted the referral. 900 were 35 years or over and 600 of these were eligible for genetic studies by gestational age. Of the 600, only 300 accepted the referral.

On the basis of our experience, approximately 30% of the target population accepted genetic studies for advanced maternal age.