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POSTER PRESENTATIONS

Educational, Psychosocial and Societal Aspects of Genetics: Screening *(continued)*

1850

Screening for the fragile X syndrome: Impact of genetic counseling on reproductive choices of women at risk for having an affected child. G. Turner, H. Robinson, S. Laing, S. Sherman*(1). Prince of Wales Children's Hospital, Randwick, N.S.W., Australia, (1) Emory University, Atlanta, GA.

During the last 10 years we have screened 15,000 intellectually handicapped individuals in New South Wales, Australia for the fragile X syndrome in order to provide genetic counselling for the parents at risk. We followed up on 400 women who had been counseled to determine if this had altered their reproductive decisions. Of these, information on 380 females was available including year of birth, age at counseling and follow-up, level of intellectual functioning, carrier risk, and sex, affection status and year of birth of children born before and after counseling. For each case, the number and type of children born during the period between counseling and follow-up was compared to the children born to a matched control during the same age period but who had not had counseling as yet. We found that intellectually normal women significantly reduced their expected number of affected offspring: fewer of these women had children compared to controls and those who did had fewer offspring and/or chose to terminate fetuses which were at a high risk for being mentally retarded. In contrast, borderline normal/mildly handicapped women were similar to their matched controls; thus they did not obviously change their reproductive choices based on carrier risk information.

1851

A STUDY OF PALMAR DERMATOGLYPHICS AND ABO BLOOD GROUPS.

M.S.Subhadra* & K.N.Mohanlal, Department of Anatomy, Medical College, Thiruvananthapuram, Kerala,India,695 011.

300 medical students(150 girls and 150 boys) of 18-20 yrs of age and of Kerala origin are studied in relation to their palm prints and ABO blood groups. Dermatoglyphics methods used are those of Cummins and Midlo(1961). The results are analysed on the basis of sex, bimanual differences and blood groups, qualitatively and quantitatively. The loops are more numerous (63.6%) than whorls (31.7%) and arches (4.7%). Bilateral homologous patterns are seen most frequently on 5th digit. The 3 standard indices are compared with other reports on Indian and Mongoloid populations. Pattern Intensity Index(PII) and Furuhata's index show lower values, while Dankmeijer's index displays a very high value. Total Finger Ridge Count(TFRC) and ATD angle are significantly higher in males but AB ridge count is found identical in both sexes. Significant bimanual differences are not observed. Correlation with palmar Dermatoglyphics pattern and ABO blood groups are rather irregular.

1852

Screening in a heterogeneous population: The American public. DE Wagener*, JA Reidy, KK Steinberg, ATL Chen, GM McQuillan. Centers for Disease Control, Hyattsville, MD, USA.

Molecular genetic assays reveal great heterogeneity in the American Public. Any screening program in such a heterogeneous population would be largely uninformative without detailed knowledge of genotype distributions in population subgroups. The Cell Bank of the 3rd National Health and Nutrition Examination Survey (NHANES III) will offer investigators DNA samples from nationally representative populations. NHANES III (1989-1994), conducted by the Centers for Disease Control, will survey 20,000 people, at least 12 years of age. Data include subject's health and nutritional history, medical and dental examination, and a battery of physical and biochemical tests. Sera and mononuclear cell samples are being stored. This presentation focuses on the impact of statistical properties (sensitivity, specificity, and predictive value of positive tests) on the goal of establishing a mass screening program in the U.S. population. Because predictive value is directly proportional to the disease frequency and inversely proportional to marker frequency, non-national, special study populations that oversample diseased individuals may underestimate the proportion of false positive findings in the general population.

1853

Aneuploid induction in mouse bone marrow by Tripterygium Hypoglaucum (Level.) Hutch. X. Wang*, RM Zhou, ZJ. He, S. Cao. Dept. of Biology, Yunnan Normal Univ., Kunming, Yunnan, PR China.

Tripterygium Hypoglaucum (Level.) Hutch (THH) is a Chinese herb which has been used broadly for the treatment of various human immune diseases. The medicine block mitosis and germinal cell developing both in human and rat. Our study employed three cytogenetic methods, i.e., C-mitotic (CM) effects, micronucleus (MN) and chromosomal aberration (CA) analyses to investigate the aneuploidy inducing effects of THH in mouse bone marrow cells after the animals were treated with THH (crude water-extracts). The doses used in i.p. administration were 5-14.3g/kg.

To analyse CM effects, 5 treated and 5 control males were used per dose group. The mitotic index (MI), frequencies of C-mitosis and anaphase were considered synchronously. In MN and CA tests, 5 males and 5 females were included in each dose group (with 1 control per sex). 1000 polychromatic erythrocytes (PCE) and 50 metaphases were analysed per animal for the presence of the micronuclei and chromosomal structure aberrations, respectively.

THH showed CM effects significantly accompanied with increases of MI and the frequencies of C-mitotic cells as well as decreased frequencies of anaphase ($p \leq 0.01$-0.05). The frequencies of induced micronucleus in PCE were significantly higher than that of the controls at two dosage groups (1, 14.3g/kg). A dose response was found at 24h sampling interval. THH showed no clastogenic effects in CA tests. It is concluded that THH may contain some aneugenic component. Most of micronuclei induced by THH may be formed from lagging chromosomes. Clinical use of THH should be more serious.

1854

Chromosomal and metabolic screening on mentally retarded school children in Taiwan. K. Wuu* (1), S. Li(2), M. Chao(3), K. Hsiao(1) and T. Wang(4). (1) National Yang Ming Medical College, Taipei, (2) Chung Shan Medical College, Taichung, (3) Kaohsiung Medical College, Kaohsiung, and (4) National Taiwan University Hospital, Taipei, Taiwan, Republic of China.

For the purpose of exploring the possibility of implementing a nation-wide screening program for chromosomal and metabolic abnormalities of mentally retarded school children in Taiwan, a pilot study was carried out from 1988 to 1990 in Taipei, Taichung, and Kaohsiung to detect patients with phenylketouria (PKU), galactosemia, homocystinuria, congenital hypothyroidism (CH) and chromosome abnormalities (CA). A total of 1,248 children was screened, 961 of them had IQ 50-75, while the remaining 287 had IQ < 50. The results are as following:

IQ	No. Children Examined	Incidence of PKU	CH	CA
50-75	961	3(0.3%)	3(0.3%)	69(7.2%)
< 50	287	3(0.3%)	0	44(15.3%)
Total	1,248	6	3	113

Of the six PKU patients, five were classical PKU and one was atypical PKU due to dihydropteridine reductase deficiency. Among the 113 patients with chromosome abnormalities, 68 were numerical and 45 structural. It is evident that the more severe the mental retardation, the higher incidence of PKU and chromosome abnormalities was observed.

1855

Consumer attitudes toward thalassemia screening: heterozygotes of multi-ethnic origin. J.Yuen*, Y.E. Hsia. Kapiolani Med. Ctr. U. Hawaii, Honolulu, USA.

Among 1073 adult α and β thalassemia heterozygotes over age 18, who were diagnosed and counseled in a grant-funded multi-ethnic screening project in Hawaii, 804 were surveyed about their perceived value of the screening. We interviewed 38 by telephone and sent 1-page questionnaires to 766.

We received responses from 392 [48.7%]: 39 were Laotian, 187 Filipino, 123 Chinese, and 43 were of mixed ethnicity; 286 [73%] were married, 96 [25%] single, 10 widowed or divorced.

A similar proportion of married and single subjects felt glad they were tested, had families tested and were concerned about passing an affected gene to their children. Over 70% thought our brochures and reports were clear, almost 65% had discussed the results with their doctors.

	Married [N=286]		Single [N=96]	
Glad to be Tested	221	[77.3%]	74	[77.1%]
Partner was Tested	218	[76.2%]	27	[28.1%]
Family was Tested	174	[60.8%]	60	[62.5%]
Afraid of Passing Gene	184	[64.3%]	61	[63.5%]
Brochure was Clear	203	[70.1%]	70	[72.9%]
Results were Clear	205	[71.7%]	78	[81.3%]
Discussed With Doctor	182	[63.6%]	61	[63.5%]