Ecologic Study of Nutritional Status of Ecuadorean Children

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The nutritional status of children is the product of many ecologic factors, including social, cultural, economic, educational, and emotional, as well as the biologic determinants. A descriptive study has been conducted to evaluate the relative importance of these variables to the nutritional status of rural and urban Ecuadorean children. Two hundred twenty-four families were evaluated. One hundred ninety-four of the families studied constitute the present investigation.

Significant differences were observed between urban and rural populations in terms of maternal age and maternal education. The average ingestion of animal proteins is much lower in the rural populations. Nutritional status is inferior in the rural children.

Multiple regression equations revealed a strong correlation between the nutritional status of the children and the ingestion of animal proteins. However, in specific population groups, a relationship was also evident between vegetable protein and nutritional status. Sociocultural environmental variables were significantly correlated with nutritional status; however, these variables differed in ranks of importance among the various rural and urban population groups.

It can be concluded that the nutritional status of the rural children is substantially less than that of the urban study group. The ecologic factors vary in relative importance among these study groups and appear to be specific in their impact on the nutritional status.

Net Acid Balance in Marasmic Infants during Recovery from Diarrheal Dehydration

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The net acid balance technique of Lennon *et al.* (1966) permits the simultaneous measurement of acid production and excretion. Seven marasmic infants were studied during the first 6 days of treatment for diarrheal dehydration. As a control, five well nourished infants were studied during recovery from dehydration.

Acid excretion was higher in marasmic infants, with significant differences on the third and sixth days. These differences are due to higher excretion of NH_4 and titratable acidity. Acid production of marasmic infants was lower, with significant differences on the third and fourth days. These differences are due to lower excretion of SO_4 and higher absorption of "combustible anions." Net acid balance was lower in marasmic infants with significant differences on the first and third days. Cumulated net acid balance was significantly lower in marasmic infants during all 6 days of study.

These findings suggest that, in marasmus, mechanisms of disposal of acid load are effective and are able to cope with acid loads generated during diarrheal episodes.

Malnutrition and Adoption—Two Variables in Infant Development

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It is difficult to differentiate the effects of nutritional and psychologic factors during the recovery of malnourished children in the adoptive home. Our investigation attempted to differentiate the nutritional from psychologic stimuli in severely malnourished children who were institutionalized before being adopted by families. Twenty-six children were placed for adoption after hospital-

ization in the Dr. N. H. Sbarra hospital. All of this study group had suffered from second or third degree malnutrition at the time of admission to the hospital. The mean age at hospitalization was 5 months and the mean stay in the hospital was 6 months. The physical and developmental progress was compared with a control group of 17 well nourished children of similar age at hospitalization and adoption. During the period of hospitalization, all infants received an individualized psychomotor stimulation program along with an adequate nutritional intake. Weight, supine length, language, and adaptive behaviour and developmental quotient were assessed at admission, at the time of adoption, and 6 and 12 months after adoption.

All malnourished infants demonstrated a rapid catch-up growth during nutritional rehabilitation in the hospital and during the first months after adoption. There was a significant reduction in rate of psychomotor development in both the malnourished and the control groups during hospitalization despite the stimulation plan. However, the effects of hospitalization on psychomotor development were no longer evident after 6 months of adoption. These observations indicate the independence of nutritional recovery from psychomotor development and confirm the importance of a stable family environment in the rehabilitation of infants.

Chlordiazepoxide Effects on Learning Ability of Euthrophic and Marasmic Rats

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The objective of this research is to demonstrate that emotion plays a role in learning of the undernourished rat. In other words, the high anxiety level of undernourished subjects acts negatively upon problem solving in different learning situations. Marasmus was induced in rats by increasing the litter size from birth until weaning. The reactivity and anxiety state of rats was modified using tranquilizer, chlordiazepoxide, 5 mg/kg body wt ip, administered 30 min before each test. Their response was evaluated in a Hebb-Williams maze.

The control group, well nourished rats receiving injections of 9% NaCl committed 107 ± 22.7 errors in the 12 problems of the Hebb-Williams maze. This group showed the best performance. The well nourished group treated with chlordiazepoxide had a total error score of 196.9 ± 42.2 . The marasmic group receiving 9% NaCl injections committed 196.9 ± 55.9 errors, and the marasmic group with chlordiazepoxide, 148.56 ± 33.4 errors. Variance analysis gives an $F = 8.68$ which is significant ($P < 0.01$). This result supported the hypothesis pointed out by Levitzky-Barnes that high anxiety level of undernourished subjects acts negatively upon problem solving. The marasmic group that received chlordiazepoxide injections committed less errors than the marasmic subjects injected with 9% NaCl. This difference was significant at a level of $P < 0.01$.

Balanced Translocation 46,t(13;18) (q21;q21) with Abnormal Phenotype: Possible Evidence for Position Effect in 18q21

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Individuals with balanced chromosomal translocations generally do not have an abnormal phenotype, presumably because of unchanged content of genetic material. Thus, the finding of apparently balanced translocations in phenotypically abnormal individuals is surprising. This may be due to coincidence or may be related through some undetected chromosomal mechanism (small deletions or insertions at the breakpoints). However, even without chromosomal imbalance a translocation might produce an abnormal phenotype through a position effect. Position effects are known in other species, but the existence of this chromosomal

anomaly has not been proven in man. The detection of position effects might be possible in man by the analysis of apparently balanced translocations and a search for correlations between specific breakpoints and phenotypic abnormalities. A dysmorphic and mentally retarded boy has been found presumably carrying a *de novo* balanced translocation which involves the long arms of chromosomes 13 and 18. Breakpoint mapping the long arms of the chromosomes 13 and 18 has revealed presumptive evidence for a position effect in 18q21.

Possible Evidence of Y Chromosome in Testicular Tissue of Patients with True Hermaphroditism and Karyotype 46,XX

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It is widely accepted that male determinant genes must be present, at least in primordial germinal cells, in order to determine testicular differentiation. However, exceptions have been reported, such as the existence of XX in apparently normal males and in the majority of true hermaphrodites. Several theories have been advanced to explain the existence of testicular tissue in the absence of a Y chromosome, but they have not been confirmed cytologically. The present work attempts to detect the presence of a Y chromosome by the quinacrine fluorescence technique in histologic sections of gonads from three patients with true hermaphroditism and somatic cell karyotypes 46,XX. Typical fluorescence of the Y chromosome was found in the testicular tissue from the three patients, indicating the existence of the Y chromosome in gonadal tissue. These findings strongly support the hypothesis that a Y chromosome is necessary for testicular differentiation.

Partial Trisomies and Deletions of Chromosome 13

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With the advent of new banding techniques there have been increasing numbers of patients with trisomies and deletions for specific portions of chromosome 13 discovered. Some authors have suggested a preliminary phenotypic mapping of chromosome 13. We have studied five patients with trisomies and deletion of parts of chromosome 13 using banding techniques and we have attempted a karyotype-phenotype correlation. 1) *Deletion 13q31 → 13qter*: karyotype 46,XX,del(13)(13pter → 13q31:) is associated with mental retardation, craniofacial asymmetry, microcephaly, wide forehead, short philtrum, large mouth with protruding and wide spaced superior incisors, prominent and wide nasal bridge, large ears, hypertelorism, retinoblastoma of left eye, and kyphoscoliosis. 2) *Deletion 13q31/13q32/13q33 → 13qter*: karyotype 46,XY,13r is associated with growth and mental retardation, large eyes, epicanthus, hypertelorism, frontal bossing, high arched palate, short philtrum, protruding and wide spaced superior incisors, simple low set ears, hypoplastic fingernails, posterior prominence of heel, deep crease of hallux, dorsiflexed first toe, and a third toe longer than the other toes in both feet. 3) *Trisomy 13q12-13qter*: karyotype 46,X,+t(Y;13)(q11;q12)(Ypter → Yq11:13q12 → 13qter:) is associated with trigonocephaly, craniostenosis with prominent metopic suture, epicanthus, colobomata of left iris, high arched palate, dysplastic low set ears, postaxial symmetric polydactyly, abnormal flexion of fingers, hypoplastic fingernails, and bilateral club feet. 4) *Trisomy 13q13 → 13qter*: karyotype 46,XX,t(5;13)(p14;q13)(13qter → 13q13::5p14 → 5qter)pat. is associated with growth retardation, microcephaly, craniostenosis, sloping and prominent forehead, protruding nasal bridge, short palpebral fissures, bilateral cleft lip, cleft palate, short neck, abnormal flexion of fingers, simian crease, clinodactyly of fifth fingers, bilateral club feet, and duplication of fifth toe with syndactyly in the right foot. 5) *Trisomy 13pter → 13q12*: karyotype

47,XX,+del(13)(13pter → 13q12:) is associated with mental retardation, microphthalmia, enophthalmia, colobomata of iris and glaucoma of left eye, bilateral preauricular dimples, imperforate anus, rectovulvar fistula, and double left calyceal system.

These observations permitted phenotypic mapping of the partial trisomic and monosomic segments in cases 1, 2, 3, and 5, which was not possible in case 4 because the patient also had a partial monosomy of chromosome 5. The deletions of the short arm of chromosome 13 (case 2) and long arm of chromosome Y (case 3) were considered to have no significant functional effect.

Tay-Sachs Disease Heterozygote Selection

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The authors determined the serum hexosaminidase A levels in 197 subjects (106 males and 91 females); 174 were adults and 23 were children aged from 20 days to 15 years; 13 of the children were of Jewish origin, the other 10 children were non-Jewish. Of the total 197 subjects, 94 (51 males and 43 females) were Jews whose ancestors lived in Western and Central Europe (Ashkenazi). The other 103 subjects (53 males and 50 females) had no Jewish ancestors.

The serum hexosaminidase A levels were determined following the method of O'Brien *et al.* (1970) modified by Kaback (Methods Enzymol., 28: 862 (1973)). In those cases where the results were doubtful, especially in the pregnant women (6), the hexosaminidase A levels were also tested in the leukocyte, according to the method described by Kaback and Zeiger (Advan. Exp. Med. Biol., 19: 13 (1972)). The results are presented in Table 1.

Table 1. Serum hexosaminidase A in Jews and non-Jews

Hexosaminidase A serum (% A of total)	Children (23)		Adults (174) ¹	
	Jewish (13)	Non-Jewish (10)	Jewish (81)	Non-Jewish (93)
>40% (normal homozygotes)	9	8	57	83
30-40% ("doubtful" cases)	5	1	14	9
<30% (heterozygotes)	0	1	10	1

¹ In the pregnant women in which results from 15.6-35.0% of serum hexosaminidase A were found, the leukocyte tests showed levels within the limits of normal homozygotes.

Comments: The high incidence of heterozygote carriers of Tay-Sachs disease in Ashkenazi Jews (1:30), emphasized once more by this investigation, justifies the population screening for these heterozygotes, particularly among Jews, in order to offer genetic counseling and try to prevent further cases of Tay-Sachs disease.

Anti-Human Growth Hormone (HGH) Antibody Determination in HGH-treated Patients

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Determinations of anti-HGH antibody were performed by radioimmunoassay in 57 patients with pituitary insufficiency who had been treated with human growth hormone (HGH). Antibodies were not detected in 38 patients, 10 had low antibody titers (1:15-1:120), 4 medium titers (1:120-1:960), and 5 high titers (of 1:960). In 42 patients, antibody titers could be correlated with growth velocity during HGH treatment. An adequate growth velocity was observed in 25 of 27 patients with negative titers, 5 of 7 with low titers, 3 of 4 with medium titers, and 1 of 4 with high