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

C. BERGADA AND R. COCO *Division of Endocrinology, Hospital de Ninos, Buenos Aires, Argentina*

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

ROBERTO COCO AND GRACIELA M. DEL REY *Laboratory of Cytogenetics, Division of Endocrinology, Hospital de Ninos, Buenos Aires, Argentina*

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, microophthalmy, enophthalmy, colobomata of iris and glaucoma of left eye, bilateral preauricular dimples, imperforate anus, rectovulvar fistual, 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

B. J. SCHMIDT, A. J. DIAMET, R. NISHIMURA, AND M. WAKAMOTO *Departamentos de Pediatria da Fac. Med. Sorocaba-PUC-Sao Paulo e Neurologia da F.M.U.S.P., Sao Paulo, Brazil*

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

M. FAHRER, J. HEINRICH, M. RIVAROLA, AND C. BERGADA *Division of Endocrinology, Hospital de Ninos, Buenos Aires, Argentina*

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