

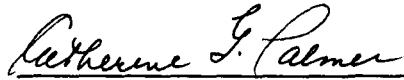
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CYTOGENETIC STUDIES OF MOSAICISM

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Submitted to the faculty of the Graduate School
in partial fulfillment of the requirements
of the degree
Doctor of Philosophy
Department of Medical Genetics
May, 1987

Accepted by the Graduate Faculty, Indiana University,
in partial fulfillment of the requirements of the degree
of Doctor of Philosophy.

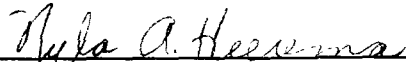


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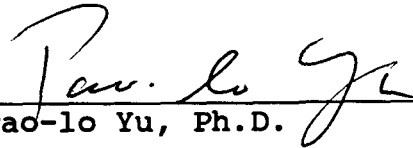


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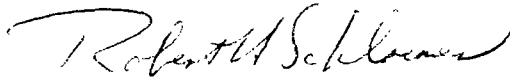
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ABSTRACT

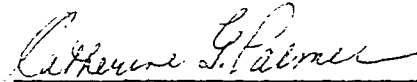
Cytogenetic Studies of Mosaicism

This dissertation has addressed several problems related to the diagnosis of mosaicism. Mosaicism has always implied a clone of more than one cell with the same abnormality. Because chromosomal abnormalities can be present in fibroblasts and not peripheral lymphocytes, tissue limited mosaicism and the culture parameters that affect its detection were examined. The significance of loss of chromosomes from single cells of leukocyte cultures and the occurrence of translocations in single cells of such cultures were also addressed.

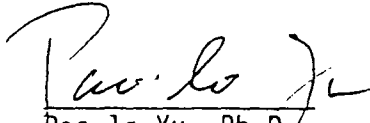
Fibroblast karyotype analysis of 43 individuals with multiple congenital anomalies, but with normal peripheral lymphocyte karyotypes, showed that 5% of these individuals have tissue limited mosaicism, i.e. chromosome abnormalities which were detected in fibroblast cultures that were not detected in lymphocyte cultures.

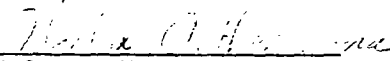
The second portion of this dissertation considered the significance of single cell abnormalities detected in lymphocyte cultures of 171 individuals referred because of a history of habitual abortion and 505 individuals referred for other reasons. There is a greater frequency of individuals with single cell chromosome loss, and single cell structural abnormalities, among habitual abortion referrals as compared to all other referrals. Among females, there is a greater frequency of individuals experiencing X chromosome loss in the habitual abortion group. When structural abnormalities occurring in single cells were considered, there was a greater frequency of individuals with all types of single cell structural abnormalities, in particular single cell translocations in females and deletions in males, among those individuals referred because of a history of habitual abortion.

The significance of these findings is discussed.


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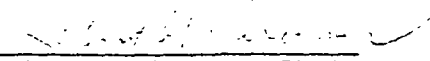

Robert Schloemer, Ph.D.

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