

A Cytogenetic Study of Children with Congenital Anomalies

Thesis submitted by

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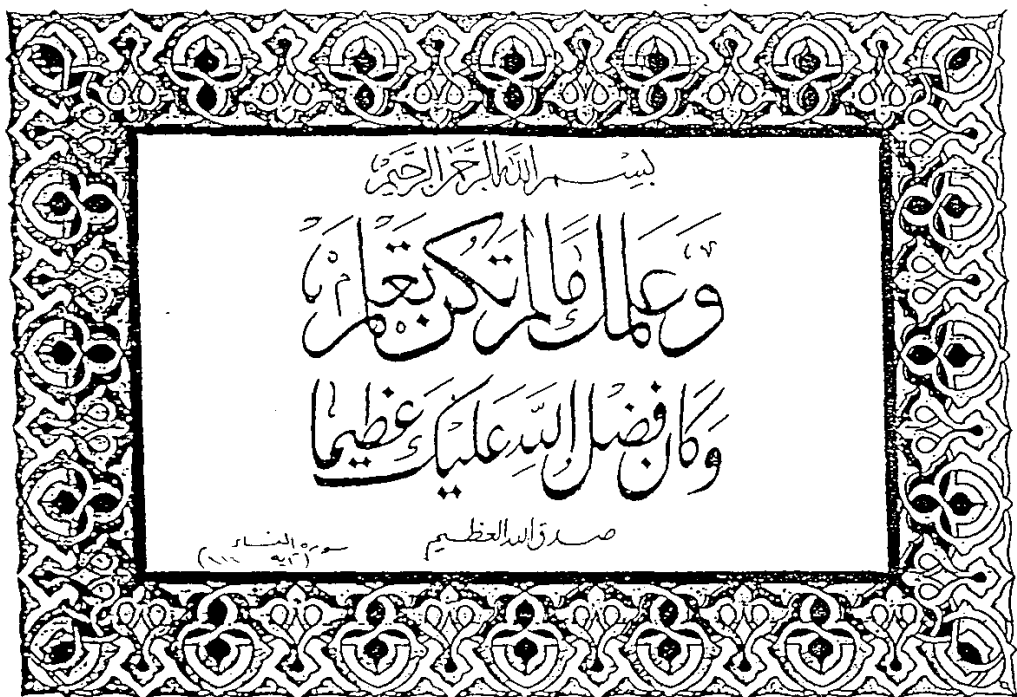
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The Pre-Master Studies

1. HISTOLOGY
2. PHYSIOLOGY
3. INVERTEBRATES
4. HISTOPATHOLOGY
5. ENGLISH LANGUAGE
6. STATISTICS

Abstract

A cytogenetic study of children with congenital anomalies

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Aim: To study the different factors leading to congenital anomalies or associated multiple congenital anomalies and cytogenetic pattern in each type.

The study was conducted on 250 cases (200 cases with multiple congenital anomalies and 50 cases with isolated limb anomalies). All cases were collected from the out patient clinic of Genetic Unit, Pediatric Department at Ain Shams University Hospital, and El-Sahel Teaching Hospital.

Patients under study were subjected to a full history taking, family pedigree study, clinical examination, cytogenetic study, sister chromatid exchange and lastly dermatoglyphic analysis.

The paternal consanguinity was highly significant in the patients with recognizable genetic syndromes. Increased the maternal age in chromosomal group and patients with recognizable genetic syndromes. Cytogenetic analysis revealed numerical and structural aberrations. Karyotyping for mothers patients revealed centromeric separation in some chromosomes, and partial monosomy in chr. 6 (6q-).

Sister chromatid exchange frequency increased with statistically significant in mothers of patients with multiple congenital anomalies due to teratogenic effect. Dermatoglyphic analysis revealed increased total ridge count with peculiar dermal pattern in some groups compared to the control group.

Keyword: Limb, Congenital Anomalies, Chromosomes, Sister Chromatid Exchange, Dermatoglyphic, Consanguinity, Teratogenes.

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