

**STUDY THE EFFECT OF  
SOME ENVIRONMENTAL FACTORS  
ON DOWN'S SYNDROME**

**By**

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**A Thesis Submitted in Partial Fulfillment of the  
Requirement for the Doctor of Philosophy  
In  
Environmental Science**

**Department of Medical Science  
Institute of Environmental Studies & Research  
Ain Shams University**

**2006**

## ABSTRACT

Based on several studies linked the occurrence of Down's syndrome (DS) to maternal exposure to certain exogenous and endogenous factors, we evaluated the socio-demographic features of 20 families with DS with consanguinity analysis. Also, we evaluated certain associations of some maternal factors which might affect the outcome. In this study, we estimated serum levels of zinc, selenium and thiamine as well as amino acids profile in blood and urine of the 20 children with DS as a base of nutritional intervention. With the advances of molecular cytogenetics and development of fluorescent in situ hybridization technique (FISH technique), we did a comparative diagnostic study between the ordinary karyotype (G banding) and FISH technique as tools of diagnosis. We found a significant difference regarding both maternal and paternal age being higher among cases than the control group ( $P < 0.05$  for each), while study of gender, age spacing, birth order, social class and consanguinity revealed a non significant association ( $P > 0.05$  for each). Maternal habits (tea, coffee, alcohol intake and smoking) and maternal chronic diseases were insignificantly associated with DS pregnancies ( $P > 0.05$  for each). Although there were insignificant associations of maternal exposure to a variety of exogenous factors, lack of supplementation with folic acid tablets before and during pregnancy showed a highly significant association among mothers of children with Down's syndrome ( $P < 0.0001$ ). Also there were insignificant associations with manifestations of bad obstetric history. Preterm and delivery with Caesarean section were significantly associated among cases ( $P < 0.05$  for each). We found a

highly significant decrease in serum levels of zinc, selenium, and thiamine among our cases with DS compared to the control children ( $P < 0.0001$ ), while the amino acids profile was normal among both cases and control children. Also, on performing karyotyping using (G banding) we found that all our patients are of the nondisjunction trisomy 21. On the other hand, FISH technique failed to prove diagnosis in two cases out of our twenty cases. We found no evidence of seasonality of births among our patients to explain the nondisjunction process.

In conclusion, the positive associations of some maternal factors with DS have to be considered -with the maternal age- as minor factors which may contribute to Down's syndrome. The decrease in serum levels of zinc, selenium and thiamine should be considered on nutritional intervention for those children with DS. Also, diagnosis of trisomy 21 should be based on the ordinary karyotype.

## *ACKNOWLEDGEMENT*

*My profound thanks and gratitude to Prof. Dr. Mohammed El-Sawi, Head of Human Genetics Unit, Faculty of Medicine, Ain Shams University; for his great support, continuous encouragement and valuable remarks. He was interested in this work and enriched me with constructive observations. His help and kindness is greatly appreciated.*

*My great thanks and all my respects to Prof. Dr. Mohammed El-Khafif, Professor of clinical biochemistry and Dean of the Institute of Environmental Studies and Research, Ain Shams University; for his kind care, encouragement and guidance. He enriched me with his great experience and continuous help throughout the work. His meticulous supervision will be always kept in mind.*

*I am very grateful to Prof. Dr. Nagwa Abdel-meguid, Head of the Department of Research on Children with Special Needs; National Research Centre; for her kindness and support. Her advices were part of the stem of this work,*

*In this occasion we should not forget late Prof. Dr. Neamat Hashim, Professor of Pediatrics, Ain Shams University; as what she had mentioned regarding the seasonality in Down's syndrome thirty years ago, is going to be discussed in this thesis.*

## **GLOSSARY**

### **Amniocentesis**

A technique for obtaining a sample of fetal cells.

### **Amniocytes**

The fetal cells floating in the amniotic fluid.

### **Aneuploid**

A chromosome number that is not an exact multiple of the haploid number.

### **Apoptosis**

A common process throughout life that helps the body to get rid of cells it doesn't need.

### **Autosome**

Any chromosome which is not a sex chromosome (X or Y).

### **Brachycephaly**

Head shape which is flattened at the back.

### **Chromosome**

Thread-like, darkly staining bodies within the nucleus, made up of chromatin and DNA, which carry the genetic information.

### **Clinodactyly**

Small inward curving little finger.

### **De novo**

Occurring for the first time, rather than inherited.

### **Epicanthic fold**

Fold of skin at the inner canthus of the eye. Particularly common in Down's syndrome but can be seen as a normal variant in some unaffected children.

### **Fibroblast**

The precursor cells which are found in connective tissue.

**Gamete**

Ovum and sperm.

**Gonadal**

Referring to the gonads. Ovaries and testes.

**Incidence**

The incidence of a genetic disorder is the number of persons who are born with this disorder in a specified group per year.

**Interferon**

A substance produced inside the body in response to viral infections.

**Karyotype**

An arranged pictorial presentation of the chromosomes of a cell.

**Kerala**

An Indian city.

**Lymphocytes**

A sub-group of white blood cells.

**Mean value**

Is the sum of observations divided by the number of observations.

**Meiosis (meiotic)**

The type of cell division which occurs in gamete formation, with halving of the somatic number of chromosomes so that each gamete is haploid.

**Microcephaly**

Small sized head

**Mitochondria**

Structures in each cell that convert molecules into energy.

**Mitochondrial inheritance**

A type of inheritance – also called maternal inheritance – that applies to genes in mitochondrial DNA.

**Mitosis (mitotic)**

The type of cell division which occurs in replication of somatic cells.

**Oocyte**

Haploid female gamete.

**Phalanx / phalanges**

The bones in the fingers and toes.

**Prevalence**

The prevalence of a genetic disorder is the total number of persons who have this disorder in a specified group at a given time.

**Robertsonian translocation**

A translocation between two acrocentric chromosomes, involving fusion at the centromeres.

**Sample Range**

Difference between the highest and lowest values.

**Sandal gap (sandal sign)**

A gap between the big toe and the other four toes.

**Standard deviation**

The square root of the variance.

**Sonolucent**

Allowing ultrasound to pass through without refraction. Giving a dense black appearance on an ultrasound scan, implying fluid filled.

**Telomere**

Specialized DNA sequence located at the end of chromosomes.

**Trait**

A character

**Translocation**

Transfer of chromosome material from one chromosome to another. If there is exchange of genetic material from one chromosome to another, this is called a reciprocal translocation.

**Trisomy**

Three, rather than the usual two copies of a chromosome.

**Variance**

Measure of spread of observations about the mean.

**Zygote**

Fertilized ovum



## LIST OF ABBREVIATIONS

<b>AA</b>	-----	Atomic absorption
<b>AAI</b>	-----	Atlanto-Axial Instability
<b>AAS</b>	-----	Atomic absorption spectrophotometry
<b>AFP</b>	-----	Alpha fetoprotein.
<b>ASD</b>	-----	Atrial septal defect
<b>ATP</b>	-----	Adenosine Tri Phosphate
<b>CDC</b>	-----	Center for Disease Control and Prevention
<b>DNA</b>	-----	Deoxyribonucleic acid
<b>DSCR</b>	-----	Down's Syndrome Critical Region
<b>FDA</b>	-----	Food and Drug Administration
<b>FISH</b>	-----	Fluorescence in situ hybridization
<b>GC</b>	-----	Gas chromatography
<b>GLC</b>	-----	Gas-liquid chromatography
<b>GSC</b>	-----	Gas-solid chromatography
<b>GSH-Px</b>	-----	Glutathione peroxidase
<b>hCG</b>	-----	Human chorionic gonadotropin
<b>HMPS</b>	-----	Hexose monophosphate shunt
<b>IgG</b>	-----	Immunoglobulin G class
<b>IDDM</b>	-----	Insulin dependent diabetes millitus
<b>I Q</b>	-----	Intelligence quotient
<b>MMWR</b>	-----	Morbidity and mortality Weekly Report
<b>Mt DNA</b>	-----	Mitochondrial DNA
<b>OC</b>	-----	Oral contraceptives
<b>PAP A</b>	-----	Pregnancy associated protein A
<b>PDA</b>	-----	Patent ductus arteriosus
<b>SOD</b>	-----	Superoxide dismutase
<b>TLC</b>	-----	Thin layer chromatography
<b>UNSCEAR</b>	-----	United Nation Scientific Committee on the Effects of Atomic Radiation
<b>VSD</b>	-----	Ventricular septal defect

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