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INCIDENCE OF CONGENITAL MALFORMATIONS AMONG NEONATES ADMITTED TO CAIRO UNIVERSITY NEONATAL UNITS

Thesis
Submitted for partial fulfillment of M.D. degree in pediatrics

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To my family and my patients

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List of Abbreviations

A.V. Canal	Atrioventricular canal
AFP	Alpha fetoprotein
ASD	Atreal septal defect
B. of ledd	Bands of ledd
CDH	Congenital diaphragmatic hernia
CHARGE	Coloboma of the eye, Heart, Atresia Choana anomalies, retardation of mental and somatic development microphillus, Ear anomalies or deafness.
CHPS	Congenital hypertrophic pyloric stenosis
CNS	Central nervous system
Confluent P.A.	Confluent pulmonary artery
CVS	Cardiovascular system
DNA	Deoxyriboneuclic acid
duod. atresia	Duodenal atresia
dup	Duplication
EB	Epidermolysis bullosa
FISH	Florescence in situ hybridization
H.I. anus	High imperforate anus
H.S.D.	Hürschsprung disease
HLA	Human leucocytic antigen
HPE	Holoprosencephaly
i(xq)	Isochromosome for Xq
IGUR	Intrauterine growth retardation
ILS	Isolate lyssencephaly sequence
Jeuj. atresia	Jeujenal atresia
L.I. anus	Low imperforate anus
LBW	Low birth weight

Malposition of G.V.	Malposition of great vessels
MIHV	Middle interhemispheric variant
MLPA	Multiple ligation probe Amplification
NTDs	Neural tube defect
p	Short arm of chromosome
PCR	Polymerase chain reaction
PDA	Patent ductus arteriosus
PFO	Patent foramen oval
PKU	Phenylketonuria
q	Long arm of chromosome
SHH	Sonic hedge hog
TOF	Tracheoesophageal Fistula
TR	Tricuspid regurge
VACTERL	Vater+Renal anomalies + cardiac
VACTERL-H	Vacterl-Hydrocephalus
VATER	Vertebral defect, Anal atresia, Tracheoesophagial fistula with esophageal atresia, Radial displasia
VSD	Ventricular septal defect

Introduction

A normal birth is always regarded as a logical, natural event. But for any reason, a deviation occurs, the malformation is received with a sense of fear and horror, but at the same time as an evincing example of the power of God **(Mazzola et al., 1990).**

Birth defects, congenital malformations, and congenital anomalies are all encompassing terms currently used to describe developmental defects present at birth (L. Congenitus, born with). Birth defects may be structural, functional, metabolic, behavioral or hereditary **(Moore and Persaud, 1993).**

An endless list of giants, dwarfs, conjoined twins, Cyclops and anencephalic creatures fill the literature of the sixteenth and seventeenth centuries. Numerous reports on semihuman beings were invented as the result of bestiality and sexual perversions. Mothers of deformed children were considered witches and burnt at the stake for having an affair with the devil **(Schenk, 1609; Licetus, 1634; Schot 1667 and Van Meekeren,1682**).

The consept that malformed children were to be taken as a warning was very popular not only in ancient civilizations but also later, in the period from the Middle Ages to the eighteenth century. It is, however, very interesting to note that this idea is still deeply rooted in some populations of Africa and Latin America **(Mazzola, et al., 1990).**

The population risk for medically significant birth defects is approximately 3% of all live-born infants. However, not all birth defects are detected at birth, for example, some forms of kidney disorders, congenital heart disease, and mental retardation are diagnosed later in life.

So by adulthood the percent will rise to 7% of the population **(Elias, et al., 2005).** Congenital anomalies cause 10% of all neonatal deathes **(Cassidy and Whiteman, 1996).**

Egypt's rapidly growing population has an estimated rate of natural increase of 2.9% **(Courbage and Khlat, 1993).** This population is highly inbred. There are no significant intercultural, multiethnic, and interreligious marriages, and only recently has migration been common among Egyptians.

Fetal death and adverse outcome of pregnancy are quite high among Egyptian's. Six percent of all still born and 16% of all pregnancies end in miscarriages. Miscarriages increase to 21% if there is a history of birth defect in a previous pregnancy. In a study of birth defects among Egyptian, it was found that 11% of birth defects were attributed to single-gene defects, 31% were chromosomal in origin, 54% were due to multifactorial etiology, and 4% were environmental. A history of using teratogenic substances was found in 17% of cases of birth defects compared to 3% among the general population **(Hashem, 1980).**