DERMATOGLYPHICS AND AMBIGUOUS GENITALIA IN EGYPT

Thesis

Submitted for partial fulfilment of Master degree in Pediatrics

by:

EMAN MOHAMED ABDEL MAKSOUD

M.B.,B.ch

618.920042

F. M

under the supervision of:

Prof. Dr. RABAH MOHAMED SHAWKY

Professor of Pediatrics and Genetics Ain Shams University

Prof. Dr. MOHAMED SALAH EL-DIN EL-KHOLY

Professor of Pediatrics Ain Shams University

Dr. MOHAMED ABDEL ADL EL-SAWY

Assistant Professor of Genetics Ain Shams University

> Faculty of Medicine Ain Shams University 1993

بسم الله الرحمن الرحيم

" هل آتى على الانسان حين من الدهر لم يكن شيئا مذكورا { انا خلقنا الانسان من نطفة أمشاج نبتليم فجعلنالا سميعا بصيرا"

صدق الله العظيم

سورة الانسان آية ٢,١



To my mother, who suffered a lot during this work, with love.

ACKNOWLEDGMENT

I would like to express my sincere gratitude and hearfelt thanks to Prof. Dr. Rabah Mohamed Shawky, Prof. of Pediatrics and Denetics, Sin Shams University for her giving me the precious chance to work under her supervision and for her guidance and encouragement that enabled me to fulfil my part in this research.

I am sincerely indepted to Prof. Dr. Mohamed Salah El Din El Kholy, Prof. of pediatrics, Sin Lhams University for his expert assistance and fruitful criticism throughout this work.

May words can express my deepest thanks to Dr. Mohamed Abdel Adl El Sawy, Assistant professor of Denetics. Ain Shams University for his kind human teaching soul, and sharing in every step of this research to put it on an objective scientific basis.

CONTENTS

Inroduction and aim of the work	
Review of Literature	
(I) Dermatoglyphics	
Introduction and historical review	
Embryogenesis of epidermal ridges	
Development of flexion creases	
Dermatoglyphic pattern configurations	
Subclassification of the interdigital patterns	
Congenital malformations of dermatoglyphics	
Methods of recording derrmatoglyphics	
Frequency of dermatoglyphic traits in normal populations	
Some rare traits of finger and palmar dermatogyphics	
Factors affecting dermatoglyphics	
Sex chromosomes influence on dermatoglyphics	
Dermatoglyphics and clinical disorders	
Assessment of dermatoglyphics as a diagnostic tool	
(i) Normal sexual differentiation	
Gonadal primordia	
Chromosomal sex	
Role of H-Y antigen	
Ovarian and testicular endocrine function	
The primitive genital ducts	
The primitive external genitalia	
Phenotypic differentiation	
Male development	
Mechanisms by which androgens virilize the male embryo	
Female development	
Control of gonadal secretions from conception to adulthood.	
(ii) Ambiguous genitalia	
Introduction and definition	
Chromosomal abnormalities	
Gonadal abnormalities	
Phenotypic abnormalities	-
Evaluation of a newborn infant with ambiguous genitalia	

Gender assignment	93
Subjects and methods	97
Results and discussion	
XX - Group	100
XY - Group	111
Discussion	119
Summary	126
References	128
Arabic summary	

LIST OF FIGURES

Fig (1) Diagrammatic representation of ridged skin structure.	6
Fig (2) Types of fingertip patterns.	11
Fig (3) Diagram of palmar dermatoglyphic pattern areas.	12
Fig (4) Thenar/first interdigital patterns.	12
Fig (5) Diagrams of ridge arrangements in the interdigital areas.	13
Fig (6) Hypothenar patterns.	14
Fig (7) Diagram demonstrating the nomenclature of palmar triradii.	15
Fig (8) Diagram showing the numerical values used to designate	
termini of palmar main lines.	16
Fig (9) Classification of the C line of the palm into four modal types.	17
Fig (10) Finger ridge count.	19
Fig (11) Position of the axial triradius.	20
Fig (12) Numerical values used in deriving the main line index.	21
Fig (13) The transverse palmar crease.	22
Fig (14) Types of "loop" patterns in the interdigital areas.	24
Fig (15.a) Methods of taking finger and palm prints.	30
Fig (15.b) Taking prints in infants.	30
Fig (16) T.S through the lumbar region of an embryo.	52
Fig (17) Enzymatic profile of the pathway of steroid hormone	
synthesis in the ovary and testis.	58
Fig (18.a) Cross section through the trunk of 4-5 weeks old embryo.	59
Fig (18.b) Cross section through a slightly older embryo.	59
Fig (19.a) Diagram of the urogenital system in 6-week old embryo.	60
Fig (19.b) Diagram of the urogenital system in a later stage.	60
Fig. (20) Successive stages in the development of the urinary	
bladder and the definitive urogenital sinus.	62
Fig (21) The indifferent external genitalia at about the 4th week.	62

Fig (22) The indifferent external genitalia at about the 6th week.	62
Fig (23) Development of the genital tract.	63
Fig (24) Mechanisms by which androgens act to virilize	
the male embryo.	68
Fig (25) Scheme of proposed biochemical mechanism	
of genital differentiation.	71
Fig (26) Embryogenesis of the male and female phenotypes.	73
Fig (27) Successive stages in the development of the urethra	
of the male.	74
Fig (28) External genitalia in the male at the 10th week.	74
Fig (29) The external genitalia in the newborn male.	74
Fig (30) L.S to show the different stages of development of the	
uterus and vagina.	75
Fig (31) Steps in vaginal development.	75
Fig (32) The external genitalia in the female at about the 9th month.	75
Fig (33) The external genitalia in the female at the time of birth.	75
Fig (34) The consequence of events in intrauterine sexual	
differentiation in relation to hormonal environment.	76
Fig (35) Evaluation of a newborn infant with ambiguous genitalia.	94

LIST OF TABLES

Table (1) Numerical values used to designate termini of palmar main lines	16
Table (2) Clinical disorders and associated dermatoglyphics.	47
Table (3.a) Errors in testosterone synthesis or action.	89
Table (3.b) The syndromes of androgen resistance.	92
Table (4) Laboratory tests employed in the differential diagnosis of patients	š
with ambiguous genitalia.	95
Table (4.a) Clinical and laboratory findings in 46-XX patients.	99a
Table (4.b) Clinical and laboratory findings in 46-XY patients.	110a
Table (5) Percent distribution of the digital patterns in XX patients and	
female control.	103a
Table (6) Percent distribution of the digital patterns in XX patients and	
male control.	103a
Table (7) Bilateral percent distribution of digital patterns in XX patients	
and female control.	103b
Table (8) Bilateral percent distribution of digital patterns in XX patients	
and male control.	103d
Table (9) Mean, standard deviation and standard error of the digital	
ridge counts in XX patients and female control.	103f
Table (10) Mean, standard deviation and standard error of the digital	
ridge counts in XX patients and male control.	103f
Table (11) Percent distribution of Hypothenar and Thenar pattern	
in XX patients and female control.	106a
Table (12) Percent distribution of Hypothenar and Thenar pattern	
in XX patients and male control.	106a
Table (13) Bilateral percent distribution of palmar patterns	
in XX patients and female control.	106b

Table (14) Bilateral percent distribution of palmar patterns	
in XX patients and male control.	106c
Table (15) Bilateral percent distribution of patterns of the interdigital areas	
in XX patients and female control.	106 d
Table (16) Bilateral percent distribution of patterns of the interdigital areas	
in XX patients and male control.	106e
Table (17) Percent distribution of modal types of A, C and D main lines	
in XX patients and female control.	108a
Table (18) Percent distribution of modal types of A, C and D main lines	
in XX patients and male control.	108b
Table (19) Bilateral percent distribution of modal types of C palmar line	
in XX patients and female control.	108c
Table (20) Bilateral percent distribution of modal types of C palmar line	
in XX patients and male control.	108c
Table (21) Bilateral percent distribution of modal types of D palmar line	
in XX patients and female control.	108d
Table (22) Bilateral percent distribution of modal types of D palmar line	
in XX patients and male control.	108d
Table (23) Bilateral percent distribution of modal types of A palmar line	
in XX patients and female control.	108e
Table (24) Bilateral percent distribution of modal types of A palmar line	
in XX patients and male control.	108e
Table (25) Bilateral percent distribution of Simian crease in XX patients	
and female control.	108f
Table (26) Bilateral percent distribution of Simian crease in XX patients	
and male control.	108f
Table (27) Bilateral percent distribution of Sydney crease in XX patients	
and female control.	108g

Table (28) Bilateral percent distribution of Sydney crease in XX patier	.40
and male control.	
Table (29) Bilateral percent distribution of ATD angle in XX patients a	108g
female control.	
Table (30) Bilateral percent distribution of ATD angle in XX patients a	109a
male control.	nd
	109a
Table (31) Percent incidence of T, T', T" in XX patients and female con Table (32) Percent incidence of T, T', T" in XX patients and male control.	trol. 109b
Table (33) Number and percent distributions and male control	ol. 109b
Table (33) Number and percent distribution of ABO blood group and Rhesus factor in VV periods.	
Rhesus factor in XX patients and female control. Table (34) Number and page 4 V. 17	110a
Table (34) Number and percent distribution of ABO blood group and	
Rhesus factor in XX patients and male control.	110a
Table (35) Percent distribution of the digital pattern in XY patients and	
female control.	112a
Table (36) Percent distribution of digital patterns in XY patients and	
male control.	112a
Table (37) Bilateral percent distribution of digital patterns in XY patients	
and female control.	112b
Table (38) Bilateral percent distribution of digital patterns in XY patients	-120
and male control.	112d
Table (39) Mean, standard deviation and standard arror of the digital ridge	1120
counts in XY patients and female control.	
Table (40) Mean, standard deviation and standard arror of the digital ridge	112f
counts in XY patients and male control.	
Table (41) Percent distribution of Hypothenar and Thenar/I patterns	112 f
in XY patients and female control.	
Table (42) Percent distribution of Hypothenar and Thenar/I patterns	114a
in XY patients and male control.	
will conduit.	114b

Table (43) Percent distribution of patterns of the interdigital areas	
in XY patients and female control.	114c
Table (44) Percent distribution of patterns of the interdigital areas	1140
in XY patients and male control.	114d
Table (45) Bilateral percent distribution of palmar patterns	2170
in XY patients and female control.	114e
Table (46) Bilateral percent distribution of palmar pattern	1110
in XY patients and male control.	114f
Table (47) Percent distribution of modal types of A, C and D main lines	1111
in XY patients and female control.	115a
Table (48) Percent distribution of modal types of A, C and D main lines	115 u
in XY patients and male control.	115b
Table (49) Bilateral percent distribution of modal types of C palmar line	1100
in XY patients and female control.	115c
Table (50) Bilateral percent distribution of modal types of C palmar line	1150
in XY patients and male control.	115c
Table (51) Bilateral percent distribution of modal types of D palmar line	1130
in XY patients and female control.	115d
Table (52) Bilateral percent distribution of modal types of D palmar line	1130
in XY patients and male control.	115d
Table (53) Bilateral percent distribution of modal types of A palmar line	1150
in XY patients and female control.	115e
Table (54) Bilateral percent distribution of modal types of A palmar line	1130
in XY patients and male control.	115f
Table (55) Bilateral percent distribution of Simian crease	1151
in XY patients and female control.	116a
Table (56) Bilateral percent distribution of Simian crease	1104
in XY patients and male control.	116a

Table (57) Bilateral percent distribution of Sydney crease	
in XY patients and female control.	116b
Table (58) Bilateral percent distribution of Sydney crease	
in XY patients and male control.	116b
Table (59) Bilateral percent distribution of ATD angle	
in XY patients and female control.	117a
Table (60) Bilateral percent distribution of ATD angle	
in XY patients and male control.	117a
Table (61) Percent incidence of T, T', T" in XY patients and female control	
Table (62) Percent incidence of T, T', T" in XY patients and male control.	117b
Table (63) Number and percent distribution of ABO blood group	
and Rhesus factor in XY patients and female control.	118a
Table (64) Number and percent distribution of ABO blood group	
and Rhesus factor in XY patients and male control.	118a

Introduction and Aim of the Work

Sex chromosomes have been found to influence a large number of developmental systems in humans (Barlow et al. 1973)

Throughout this work we are concerned with two aspects of human development that are known to be influenced by sex chromosomes.

The first is dermatoglyphics or dermal ridge pattern configurations which recently have gained much interest as a prenatal marker and a valuable diagnostic tool (Schaumann and Kimura 1991).

The second is the development of external genitalia whose abnormalities compose one of the much complicated problems in diagnosis and management (Pagon 1987).

Dermal ridge patterns begin to develop very early in prenatal life and are not completely developed until the 6th month of pregnancy. During this period, many different genetic and environmental intrauterine events may bring their effect on the final outcome of dermal ridge pattern differentiation that is, at birth will be permanent for life (Schaumann and Alter 1976).

Sex chromosomes effect on dermatoglyphics have been proved on both the qualitative and quantitative aspects of palmar ridges (Alter 1965, Penrose 1968 Loesh and Huggins 1992)

Trying to illucidate the mechanism by which Sex chromosomes influence dermatoglyphyics, Polani and Polani (1979) found that certain dermatoglyphic traits are influenced by fetal androgens. Janz and Hunt (1986) suggested that sex chromosomes actually control the tissue sensitivity to sex steroids and that digital dermatoglyphic traits are then affected by hormones and not chromosomes.