

Dermatoglyphs in Patients with Beta-Thalassemia Major and Their Thalassemia Carrier Parents

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ABSTRACT

Dermatoglyphs are cutaneous ridges on the fingers, palms, and soles, formed during early intrauterine life. During this period, and only then, genetic and environmental factors can influence their formation. Beta-thalassemia major is an genetic disease. The aim of the present work was to analyze dermatoglyphs traits in subjects with beta-thalassemia major and their thalassemia carrier parents. The sample included 59 patients with beta-thalassemia major (39 males, 20 females). We also analyzed a total of 61 thalassemia carrier parents. There were 38 mothers and 23 fathers in the study. The control group comprised 120 healthy subjects (64 men, 56 women). All ten fingers and right hand palm prints of all participants were taken and statistically analyzed. The results of analyses showed that the frequency of loops on the fifth finger of left hand was significantly higher in female patients than that of carriers and controls ($p < 0.05$). It was also significantly higher in female patients than that male patients ($p = 0.03$). The increase in mean c-d ridge count was noticed in female patients only ($p = 0.04$) compared to carriers and controls. In addition to that there was also differences according to the mean c-d ridge count between female and male patients ($p = 0.01$). Our study showed that dermatoglyphs were helpful for the diagnosis of beta-thalassemia major. However, it does not help to detect thalassemia carriers. This study is the comprehensive dermatoglyphic research on thalassemia, and its dermatoglyphic data will be useful for future research in genetics and medicine.

Key words: Beta-thalassemia major, carriers, dermatoglyphs, genetics, diagnosis

Introduction

Dermatoglyphs are patterns made by epidermis on fingers, palms and soles. They are formed during early intauterine life, between the 7th and 21st week of gestation. The dermatoglyphic pattern of human palms and soles are individually unique and unchangeable during the life time¹.

Beta-thalassemia major (β -Thalassemia major) is an genetic disease characterized by profound anemia and severe hemosiderosis which may cause functional and physiological abnormalities in various organ systems. Relatively low cost of premarital screening and genetic counseling are very important in preventing the birth of patients with thalassemia². Providing laboratories for prenatal diagnosis is the most recent advancement in this field³.

Dermatoglyphs are used since they are very convenient and easily accessible tool in the study of genetically

influenced diseases. The diagnostic role of dermatoglyphic patterns is promising especially in chromosome abnormalities. In nearly all the chromosome disorders the dermatoglyphic patterns are unusual. It was hypothesized that thalassemic participants and their thalassemia carrier parents would exhibit higher rates of dermatoglyphic anomalies compared to control participants. If there were some differences between the groups we could use them as new diagnostic parameters for thalassemia in near future.

Methods

This study was approved by the Ethics committee of the Faculty of Medicine of the University of Mustafa Kemal. The sample included 59 patients with β -thalassemia major (39 males and 20 females) aged between 9

months and 37 years old (mean 11.08 ± 6.8 years). We also analyzed a total of 61 thalassemia carrier parents (either mother or father or both). There were 38 mothers and 23 fathers in the study. The control group comprised of 120 healthy subjects (64 men, 56 women). The group was fairly comparative to the thalassemia patients and thalassemia carriers in age, although this bears little importance because dermatoglyphic features do not change with age. The analysed and control groups were properly matched as regards ethnic origin. All ten fingers and right hand palm prints of all participants were taken and analyzed according to the Cummins and Midlo method⁴. The standard ink and roller method was employed to obtain the finger and palm prints.

Finger ridge counts were determined by counting the number of ridges that intersected a straight line connecting the triradial point (the point of ridge intersection) to the point of the core (the ridge in the center of the pattern). If more than one triradius was present on a finger, higher ridge count was taken. Total finger ridge count (TFRC) was computed by summing the ridge counts of all 10 fingers.

The frequency of pattern type on the fingers was also analyzed. Finger patterns were classified into three categories: (1) whorls (W), closed patterns characterized by having at least two triradii and a core(s); (2) loops (L), characterized by a triradius that opens towards a side of the hand, and a core; (3) and arches (A), which do not typically contain a triradius.

In the case of palms, palmar ridge counts (between the strait line connecting two digital triradial points located on the base of each finger, ie, »a«, »b«, »c«, and »d«) on the right hand a-b, b-c and c-d were also counted

Statistical Chi-square (χ^2) Test, Mann Whitney U, Kruskal-Wallis and multivariate analysis of variance (MANOVA) analyses were used to evaluate the findings.

Results

A statistically significant increase in ulnar loop pattern was observed in female patients (62%) when compared to that of male patients (44%) ($p < 0.05$). In contrast, the whorl pattern was decreased in female patients (38%), which was found to be statistically significant when compared to that of the male patients (56%) ($p < 0.05$).

The comparison of frequencies between patients, their parents and controls showed that the frequencies of various types are different but they were not statistically significant. The frequency of loops on the fifth finger of left hand was significantly higher in female patients than that of female thalassemia carrier parents and female controls ($p < 0.05$) (Table 1). It was also significantly higher in female patients than that of male patients ($p = 0.03$).

According to the dermatoglyphs differences by sexes, there were significant differences in 11 variables except a-b ridge count and c-d ridge count (Table 2). When evaluated based on groups finger ridge count of the left thumb (FRCL1) was statistically different ($p = 0.009$). The only difference was c-d ridge count variable when both sexes and groups were considered ($p = 0.01$). The cause of the difference was the presence of the female patients.

The mean TFRC was 143 ± 39.0 in female patients, which was significantly less than that of male patients (165 ± 38.1) ($p < 0.05$). There was no statistically significant difference in patient, their carrier parents and control groups according to the mean TFRC.

As shown in Table 3, the increase in mean c-d ridge count was noticed in female patients only ($p = 0.04$) compared to thalassemia carrier parents and controls. In addition to that there were also differences according to the

TABLE 1
FINGERPRINT PATTERNS ON EVERY FINGER FOR THALASSAEMIA PATIENTS, CARRIER PARENTS AND CONTROL GROUPS WITH FEMALE AND MALE

	Female			p*	Male			p*
	Patient	Carrier	Control		Patient	Carrier	Control	
Right hand								
F1 W/L	9/9	24/9	23/19	0.12	23/9	13/7	36/18	0.84
F2 W/L	7/11	22/11	27/15	0.85	20/11	12/8	29/24	0.67
F3 W/L	4/14	11/20	10/32	0.46	14/17	11/8	25/28	0.65
F4 W/L	11/7	22/9	27/15	0.74	26/5	12/7	36/17	0.18
F5 W/L	4/14	11/21	17/25	0.39	15/17	8/12	24/30	0.88
Left hand								
F1 W/L	9/9	20/12	29/13	0.37	16/16	11/8	28/26	0.85
F2 W/L	9/9	22/9	30/12	0.22	21/10	10/8	32/21	0.66
F3 W/L	4/14	11/20	16/26	0.48	10/21	9/9	24/29	0.38
F4 W/L	10/8	23/8	30/12	0.36	22/9	16/2	32/21	0.07
F5 W/L	1/17	8/24	17/25	0.02	12/20	5/14	22/32	0.53

* χ^2 test, F- finger, W- whorl, L- loop (ulnar or radial)

TABLE 2
MULTIVARIATE ANALYSIS OF VARIANCE (MANOVA) FOR 13 VARIABLES; FACTORIAL MODEL FOR SEX AND GROUPS
(PATIENT, CARRIER AND CONTROL)

	Corrected Model Mean square				Hypothesis p* values by source		
	Model (df=5)	Error (df=160)	F	p*	Sex (df=1)	Group (df=2)	Sex X Group (df=2)
a-b ridge count	78.7	48.1	1.6	0.15	0.06	0.21	0.71
b-c ridge count	52.0	37.6	1.3	0.23	0.03	0.26	0.88
c-d ridge count	114.3	50.0	2.2	0.04	0.47	0.51	0.01
FRCR1	51.7	17.9	2.8	0.01	<0.01	0.25	0.72
FRCR2	52.4	27.2	1.9	0.09	0.01	0.20	0.75
FRCR3	45.5	17.5	2.5	0.02	<0.01	0.72	0.50
FRCR4	41.9	17.8	2.3	0.04	<0.01	0.38	0.95
FRCR5	81.7	16.8	4.8	0.00	<0.01	0.11	0.81
FRCL1	74.2	23.5	3.1	0.01	<0.01	0.009	0.75
FRCL2	89.3	30.9	2.8	0.01	0.01	0.05	0.71
FRCL3	50.3	24.1	2.1	0.07	0.01	0.46	0.63
FRCL4	51.5	27.0	1.9	0.09	0.01	0.49	0.95
FRCL5	33.7	18.1	1.8	0.10	0.04	0.23	0.52

*MANOVA, FRCR – finger ridge count right, FRCL – finger ridge count left

TABLE 3
DERMATOGLYPHIC VARIABLES OF THE DIGITOPALMAR COMPLEX IN THALASSEMIA PATIENTS, CARRIER PARENTS
AND CONTROL GROUP WITH FEMALE AND MALE

	Female (mean ± SD)				Male (mean ± SD)			
	Patient	Carrier	Control	p*	Patient	Carrier	Control	p*
Right hand								
FRCR1	18.5 ± 3.4	19.2 ± 3.5	17.2 ± 4.8	0.15	19.7 ± 4.5	20.4 ± 4.6	20.7 ± 5.1	0.53
FRCR2	13.2 ± 4.4	14.5 ± 5.8	13.8 ± 5.6	0.28	14.8 ± 5.5	14.8 ± 6.2	15.5 ± 6.3	0.57
FRCR3	13.6 ± 5.2	13.3 ± 5.7	13.2 ± 5.1	0.16	15.6 ± 4.6	14.8 ± 4.6	14.8 ± 5.5	0.70
FRCR4	15.4 ± 5.1	16.1 ± 4.7	15.3 ± 6.0	0.78	17.6 ± 4.3	18.7 ± 5.3	17.5 ± 5.3	0.86
FRCR5	12.8 ± 3.2	13.8 ± 4.9	12.8 ± 5.4	0.41	16.4 ± 3.3	16.4 ± 4.7	16.0 ± 4.6	0.84
a-b ridge count	37.5 ± 8.9	39.2 ± 6.1	38.4 ± 6.1	0.61	38.6 ± 5.8	40.4 ± 8.7	41.1 ± 6.3	0.28
b-c ridge count	24.1 ± 4.1	26.1 ± 7.2	24.2 ± 6.3	0.34	26.7 ± 5.4	27.1 ± 5.7	25.6 ± 6.4	0.61
c-d ridge count	38.1 ± 5.2	33.6 ± 8.1	33.6 ± 7.4	0.04	33.7 ± 6.9	37.0 ± 5.9	35.7 ± 6.0	0.16
Left hand								
FRCL1	16.1 ± 4.3	18.2 ± 4.3	18.1 ± 5.4	0.18	18.7 ± 5.1	20.8 ± 5.6	20.3 ± 5.1	0.24
FRCL2	11.8 ± 6.7	14.3 ± 6.1	12.2 ± 6.1	0.23	15.1 ± 5.8	15.4 ± 5.5	14.9 ± 5.8	0.98
FRCL3	13.8 ± 4.6	13.9 ± 6.2	13.3 ± 5.2	0.76	16.7 ± 3.4	15.4 ± 5.4	15.7 ± 5.5	0.64
FRCL4	15.6 ± 5.3	16.2 ± 5.2	15.9 ± 5.2	0.88	18.2 ± 5.2	17.6 ± 6.9	18.1 ± 6.2	0.87
FRCL5	14.5 ± 4.7	14.7 ± 4.5	13.3 ± 5.0	0.41	15.3 ± 4.4	15.9 ± 4.5	16.4 ± 4.6	0.57

*Kruskall-Wallis test, FRCR – finger ridge count right, FRCL – finger ridge count left

mean b-c and the c-d ridge counts between female and male patients (Table 4). The mean b-c ridge count was increased in male patients, which was found to be statistically significant when compared to that of the female patients (p=0.04). However, the mean c-d ridge count was increased in female patients, which was found to be statistically significant when compared to that of the male patients (p=0.01).

Discussion

β-thalassemia major is an important health problem in Turkey because of its high carrier rate and the frequency of consanguineous marriages. The carrier detection procedures, genetic counselling, population screening and prenatal diagnosis of beta-thalassemias are the most important aspects of the disease. As part of this ef-

TABLE 4
THE MEAN PALMAR A-B, B-C AND C-D RIDGE COUNTS
IN PATIENTS GROUP ACCORDING TO SEXES

	Female (n=19) (mean ± SD)	Male (n=34) (mean ± SD)	P*
a-b ridge count	37.5 ± 8.9	38.6 ± 5.8	0.97
b-c ridge count	24.1 ± 4.1	26.7 ± 5.4	0.04
c-d ridge count	38.1 ± 5.4	33.7 ± 6.9	0.01

* Mann-Whitney U

fort, we initiated this study to investigate the presence of dermatoglyphs abnormalities in patients with β -thalassemia major and their thalassemia carrier parents. It is clear from our results that the possession of the gene for β -thalassemia has effect on fingerprint patterns especially for female patients.

Dermatoglyphs abnormalities have been associated with Down's syndrome, mental retardation, primary open angle glaucoma, multiple sclerosis, Alzheimer's disease, cervical and endometrial carcinoma, Wilson's disease, psoriasis, vitiligo, brachial plexus palsy, congenital heart disease, borderline personality disorder, schizophrenia, and autism⁵⁻¹⁵. Chok et al found that the schizotypic patients had a significantly lower TFRC, and number of whorls than the control group, and significantly more loops. The groups was not differ on either of the palmar measures¹⁶. Investigation on dermatoglyphs in patients with mitral valve prolapsus revealed that there was no dermatoglyphic differences between patients and control group¹⁷. Rodewald et al showed that carriers of balanced 15;21 translocation also had dermatoglyphs abnormalities¹⁸. With regards to thalassemia disease, there have been a few previous dermatoglyphic studies of thalassemia from different countries dating from the 1970s¹⁹⁻²¹. Gualdi Russo et al suggest that digital patterns are the most efficient dermatoglyphic characteristics that differentiate between the patients with thalassemia and normal controls. They found that female patients had an increased frequency of loops in comparison to the controls¹⁹. Similarly, the current study revealed

that female patients' frequency of loops on the fifth finger of left hand was significantly higher in comparison to the male patients, their female carrier parents and female controls. In contrast to our study, Saha et al found that the mean TFRC was higher in female patients in comparison to the male patients (154.2 and 127.81 respectively)²⁰. Mutalimova et al found that thalassemia patients have more whorls compared to other finger patterns²¹.

Consistent with primary hypothesis of the study, the finding of such characteristics would be helpful for the diagnosis of β -thalassemia major and therefore it can be used to aid genetic counselling. However, since we could not find any dermatoglyphs differences in thalassemia carriers, we suggest that unfortunately dermatoglyphs study will not help detect thalassemia carriers.

In most anthropological studies, comparison between the male and female sexes is one of the goals. So, in the present work, we compared all findings according to sexes in thalassemia patients, thalassemia carriers and controls. Indeed, we found that female patients' frequencies of fingertip patterns and palmar ridge counts were different than that of male patients. Arrieta et al. investigated genetic component of variables a-b, b-c and c-d ridge counts in healthy persons and concluded that in healthy men, concerning the c-d ridge count findings, there is a stronger influence of environment while in healthy women all variables a-b, b-c and c-d ridge counts have a strong genetic component that affect their phenotypic expression²². Our data seem to indicate that the factors determining the c-d ridge count are canalized in females and males in different way.

The limitation of this study is that the only right hand palm prints of all study subjects were examined for the investigation of dermatoglyphs traits in patients with β -thalassemia major and their thalassemia carrier parents.

We conclude that in agreement with previous studies, dermatoglyphs can be used as a genetic tool for the diagnosis of β -thalassemia disease. A further studies with larger number of β -thalassemia patients and thalassemia carriers by different regions are essential to learn the real significance of the dermatoglyphs features.

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ANALIZA DERMATOGLIFA KOD PACJENATA SA BETA TALESEMIJOM I NOSITELJIMA OVE BOLESTI

S A Ž E T A K

Dermatoglifi su kožni nabori a prstima i dlanovima formirani tijekom ranog intrauterinog života. Samo tijekom tog perioda genetski i okolišni čimbenici mogu utjecati na njihovo formiranje. Beta talesemija je genetska recesivna autosomalna bolest. Cilj ovog rada bio je analizirati dermatoglifska svojstva kod ispitanika s beta talesemijom te ustanoviti koji su roditelji nositelji tog gena. Uzorak je sadržavao 59 pacijenata (39 muških i 20 ženskih). U studiji je također analiziran 61 roditelj, 38 majki i 23 očeva. Kontrolna grupa sadržavala je 120 zdravih ispitanika (64 muškaraca i 56 žena). Uzeti su otisci sa svih 10 prstiju ruke kao i dlanova koji su zatim statistički obrađeni. Analize su pokazale kako su petlje na petom prstu lijeve ruke statistički značajno veće kod oboljelih u odnosu na kontrolnu grupu ($p < 0,05$). Također, utvrđene su značajno veće petlje kod ženskih pacijenata nego kod muških ($p = 0,03$). Povećanje vrijednosti c-d brazde primijećeno je kod ženskih pacijenata samo ($p = 0,04$) uspoređujući sa kontrolnim skupinama. Nadalje, također su pronađene razlike u vrijednostima c-d brazde između muških i ženskih pacijenata ($p = 0,01$). Ova studija pokazala je kako je proučavanje dermatoglifa uspješno za dijagnosticiranje beta talesemije, iako ne može detektirati nositelje ove nasljedne bolesti. Ova studija trebala bi koristiti u budućim genetskim i medicinskim istraživanjima o talesemiji.