

### DERMATOGLYPHICS OF THE DIGITS AND INTER-DIGIT AREAS IN DOWN SYNDROME PATIENTS OF A SAMPLED NIGERIA POPULATION

#### ABSTRACT

This study was carried out to determine the possible differences between the dermatoglyphic characteristics of the digit and inter-digit areas of Down Syndrome (DS) patients and healthy Nigerians. The dermatoglyphic prints of the digit and inter-digit areas were obtained by using the Improvised digit method. Parameters evaluated were the digit patterns, inter-digit patterns and single flexion crease of the fifth digit. Comparison of dermal patterns in DS patients and normal ones was done using Chi-square test at a significant level of  $P = .05$ . The result of the digit patterns showed that the total mean percentage frequency for arch, radial loop, ulnar loop and whorl were 5.28%, 0.34%, 75.94% and 18.45% in DS patients and 9.55%, 1.11%, 60.11% and 29.22% in normal subjects respectively. The distribution of dermal ridge differ significantly in digits I, II and III of the right hand and digits II and III of the left hand in patients and normal subjects at  $P = .05$  level. Down syndrome patients had more of open fields in all the inter-digit areas except in inter-digit C. The difference in patterns between DS patients and normal subjects was significant in inter-digit C and D of the left hand only. One normal subject had single flexion crease compared to four in Down syndrome patients. The findings of the study could be used as supplementary diagnostic aid for Down syndrome.

Keywords: digit, inter-digit, dermatoglyphics, crease, down syndrome, Nigerians

#### 1. INTRODUCTION

Down syndrome is a developmental genetic anomaly caused by an extra copy of chromosome 21 [1,2,3,4]. It is the most commonly recognized genetic cause of mental retardation, and is characterized by a number of dysmorphic features such as epicanthic folds, furrowed tongue, short broad palms, transverse palmar crease, incurved fifth finger, altered dermatoglyphic patterns and congenital malformations like congenital heart diseases and leukaemia [5,6,7]

After the pioneering description of Down syndrome by a physician, Langdon J. Down in 1866, almost one century was needed to find the aetiology. Lejeune *et al.* [4] discovered an extra chromosome 21 in people with Down syndrome. The partial 21 trisomy was found later by Poissonnier *et al.* [8]. The general consensus in molecular research has established that the gene over dosage unbalances the biochemical and physiological pathways of development, resulting in the abnormal phenotype seen in Down syndrome [2,4,9,10].

Dermatoglyphics refers to unique pattern configurations of the ridges of the skin especially in the palm, sole and lip, which develop under genetic control and environmental influence from the 6th week of gestation to the 24th week [11,12, 13,14,15]]. From this stage onwards, they are unaffected by the environment and unchanging. This explains their unique role as an ideal marker for detection of altered development; the distinction between the normally and the abnormally developed individual being manifest not in the appearance of peculiar types of dermatoglyphics, but rather in alteration of the frequencies of the normal types [6,16]

Cummins [6] was the first man to describe the association between dermatoglyphic patterns and medical disorders through his study of Europeans, Americans and Jewish Down

syndrome patients. He was able to correctly detect 90% of the patients by means of the dermatoglyphic abnormalities typical of the syndrome. Extensive investigations on dermatoglyphics and Down syndrome correlations in several different populations revealed findings that were similar, but not identical, due to the pattern differences between the racial groups [17, 18, 19,20,21]. Sparse literature on the differences between the dermatoglyphics of digit and inter-digit areas of Nigerian patients and controls, and the need to compare these with findings from other races are the objectives of this study.

## **2. MATERIALS AND METHODS**

In this analytical study, a total of 31 patients (14 males and 17 females), between the age range of 4-28 years, all phenotypically and clinically diagnosed with Down syndrome were used. The patient sample was obtained as follows; 2 were from The OLG Foundation, Woji, Port Harcourt; 12 were from The Child Special School, Daughters of Charity, D/Line, Port Harcourt, both in Rivers state; 10 were from The Abuja School for the Handicapped, Kuje; 7 were from Hope Foundation, Maitama, both in the FCT, Abuja.

The control group comprised of 62 male and 28 female healthy normal individuals within the age range of 4-28, majority were drawn from the University of Port Harcourt Demonstration Secondary School and Friendship Centres in Port Harcourt, Rivers state. Both the patients and the controls were unrelated Nigerians, hailing from the 5 geographical zones of the country; North, South-South, South-East, South-West, and Middle belt. Consent was granted by both the institutions and parents for the research. The age, sex and state of origin of each subject were taken as indicated in the identification and bio-data form. Digital prints of the finger and palmar area were obtained using the improvised digital method according to Oghenemavwe & Osaat [22].

The digit patterns measured were whorl (this could be central whorl, double loop whorl, central pocket loop whorl and accidental whorl), ulnar loop (UL), radial loop (RL), arch (this could be plain arch or tented arch). The inter-digit patterns evaluated were loop (L), radial loop (RL), vestige (V), loop + vestige (L+ V), radial loops + vestige (RL + V) (as seen in the bee vestige), carpal loops + vestige (CL + V) (as seen in the bee vestige) and open field (OF). Open field are no pattern or "patternless". Single flexion crease on the 5th digit was also evaluated. The variables obtained were analysed using Excel statistical tool pack version 2010. Results are presented in percentage and mean frequencies. Chi-square test was used to determine statistical differences in patterns occurrence between Down syndrome patients and normal subjects.

## **3. RESULTS**

### **3.1 Digit Patterns**

Tables 1 and 2 show the percentage frequency of patterns seen in each digit/finger of the right and left hands of all the subjects used in the study. Nigeria Down Syndrome patients have more of ulnar loops in all fingers than the control subjects except in the fourth digit of the left hand and the fifth digits of both hands. In digit I and II the ratio ulnar loop in patient and

control is approximately 1:2. The occurrence of whorl is more in the normal subjects. The total mean percentage frequency for arch, radial loop, ulnar loop and whorl were 5.28%, 0.34%, 75.94% and 18.45% in patients and 9.55%, 1.11%, 60.11% and 29.22% in normal subjects respectively. Radial loop is absent in all digits of the right hand of patients. The distribution of dermal ridge differ significantly in digits I,II and III of the right hand and digits II and III of the left hand in patients and normal subjects at P = .05 level.

**Table 1: Percentage Frequency Distribution of Digital Patterns in the Right Hand**

	I*	I*	II**	II**	III**	III**	IV	IV	V	V	MEAN I-V	MEAN I-V
	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT
	N	DS	N	DS	N	DS	N	DS	N	DS	N	DS
<b>A</b>	12.23	13.79	16.67	6.67	10.00	0.00	2.22	0.00	2.22	0.00	8.67	4.09
<b>RL</b>	0.00	0.00	2.22	0.00	1.11	0.00	1.11	0.00	0.00	0.00	0.89	0.00
<b>UL</b>	34.44	62.07	38.89	86.67	71.11	96.55	53.33	66.67	92.22	78.57	58.00	78.11
<b>W</b>	53.33	24.14	42.22	6.67	17.78	3.45	43.33	33.33	5.56	21.43	32.44	17.80

*N= control, DS = Down syndrome, I = thumb, II = index finger, III= middle finger, IV = ring finger, V = little finger*

**Table 2: Percentage Frequency Distribution of Digital Patterns in the Left Hand**

	I	I	II**	II**	III*	III*	IV	IV	V	V	MEAN I-V	MEAN I-V	TOTAL MEAN % (LEFT + RIGHT)
	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	N DS
	N	DS	N	DS	N	DS	N	DS	N	DS	N	DS	N DS
<b>A</b>	13.34	22.58	17.78	3.23	16.67	3.23	2.22	3.33	2.22	0.00	10.44	6.47	9.55 5.28
<b>RL</b>	0.00	0.00	4.44	0.00	1.11	0.00	1.11	3.33	0.00	0.00	1.33	0.67	1.11 0.34
<b>UL</b>	43.33	54.84	46.67	90.32	67.78	90.32	60.00	53.33	93.33	80.00	62.22	73.76	60.11 75.94
<b>W</b>	43.33	22.58	31.11	6.45	14.44	6.45	36.67	40.00	4.44	20.00	26.00	19.10	29.22 18.45

*N= control, DS = Down syndrome, I = thumb, II = index finger, III= middle finger, IV = ring finger, V = little finger*

### 3.2 Inter-digit Patterns

Compare to normal subjects, Down syndrome patients had more of open fields in all the inter-digit areas except in inter-digit C. The difference in patterns between patients and normal subjects was significant in inter-digit C and D of the left hand only (see tables 3 and 4).

**Table 3: Percentage Frequency Distribution of Inter-digit Patterns in the Right Hand of All Subjects**

	A	A	B	B	C	C	D	D	Mean A-D	Mean A-D
	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	RIGHT	LEFT
	N	DS	N	DS	N	DS	N	DS	N	DS

CL+V	2.22	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.56	0.00
L	0.00	0.00	6.67	0.00	28.89	51.61	67.78	35.48	25.83	21.77
L+V	0.00	0.00	0.00	0.00	0.00	0.00	1.11	0.00	0.28	0.00
OF	91.11	100.00	85.56	93.55	55.56	41.94	23.33	54.84	63.89	72.58
RL	1.11	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.28	0.00
RL +V	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.00
V	5.56	0.00	7.78	6.45	15.56	6.45	7.78	9.68	9.17	5.65

**Table 4: Percentage Frequency Distribution of Inter-digit Patterns in the Left Hand of All Subjects**

	A	A	B	B	C**	C**	D*	D*	Mean A-D	Mean A-D
	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	LEFT	RIGHT	LEFT
	N	DS	N	DS	N	DS	N	DS	N	DS
CL+V	2.22	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.56	0.00
L	0.00	0.00	1.11	6.45	24.44	29.03	71.11	41.94	24.17	19.35
L+V	0.00	0.00	0.00	0.00	0.00	0.00	4.44	0.00	1.11	0.00
OF	73.33	96.77	98.89	87.10	72.22	41.94	16.67	38.71	65.28	66.13
RL	5.56	0.00	0.00	0.00	0.00	0.00	0.00	0.00	1.39	0.00
RL +V	2.22	0.00	0.00	0.00	0.00	0.00	0.00	0.00	0.56	0.00
V	16.67	3.23	0.00	6.45	3.33	29.03	7.78	19.35	6.94	14.52

CL+V - Carpal loop and vestige; L - Loop; L+V - Loop and vestige; OF - Open field; RL - Radial loop; RL+V - Radial loop and vestige; V - Vestige

### 3.3 Single Flexion Crease on 5th Digit

The result of the occurrence of single flexion crease is presented on Table 5. Only one normal subject had single flexion crease compared to four in Down syndrome patients.

**Table 5: Percent Frequency and Test for Differences in Single Flexion Crease in the 5th Digit**

Hand	Subject	Double	Single	%	Calculated Chi value	Critical Chi value	Inference
Right	N	89	1	1.11	8.84	3.84	Differences in pattern frequency is significant (P = 0.00)
	DS	27	4	12.90			
Left	N	90	0	0.00	2.66	3.84	Differences in pattern frequency not significant (p = 0.13)
	DS	27	4	12.90			

## 4 DISCUSSIONS

This study has evaluated the digit and inter-digit patterns, and single flexion crease of the 5th digit in a sample of Down syndrome patients of Nigeria extraction. The high occurrence of loop pattern in the digit of Nigerian Down syndrome patients are similar to those reported for Europeans, Jews and Czechs [6, 7]. Cummins [6] in his study of Europeans and Jews, reported the overall frequencies for the digital patterns in the Down syndrome patients were: whorls, 19.8%; ulnar loops, 75.4%; radial loops, 2.2%; and arches, 2.6%. A study among Canadians reported that total ulnar loop, total whorl, total arches and total radial loop in Down syndrome patients were 75.1%, 20.1%, 2.3% and 2.5% compared to 61.2%, 28.4%,

5.6% and 4.9% in normal subjects respectively [18]. The digit patterns reported by Anderman [23] in a study of 70 Down syndrome patients in Canada, Borbolla *et al.*[24] in a study of Cuban patients and Barbosa *et al.* [2] in Brazilian patients were similar to the findings of Walker.

Studies of digit patterns in Asia population are similar to those of this study except that in the Chinese patients with significant amount of radial loop [19,25], a high occurrence of whorl (52.5%) pattern was seen in the digits Iranian Down syndrome (52.5%)[26]. Early study of Down syndrome patients in Nigeria recorded 94% occurrence of ulnar loops in the patients as against 62.5% in the control, and occurrence of whorls was more in the controls (31.1%) than in the Down syndrome patients (4%). No arch pattern was found in the digits of Down syndrome patients [27]. In all, the digit patterns of Down syndrome patients in the present study are generally similar to previous studies with the predominating similarities being; marked elevation in the frequency of ulnar loops especially on the first, second and third digits and consequently reduced occurrence of whorls, radial loops and arches. The peculiarity in Down syndrome patients of Nigeria extraction is that digit IV and V had no radial loop in both hands and the occurrence of arches are comparatively higher when compared to other populations

Some researches had established that the factors affecting digit pattern types include symmetry and shape of volar pad, timing of volar pad regression and timing of the onset of primary ridge formation. Thus a symmetrical, round-shaped volar pad results in a symmetrical pattern such as a whorl while an asymmetrical, leaning volar pad results in an asymmetrical pattern such as a loop. Early regression of volar pads (leading to a volar pad that was in a more regressed state (flatter size) at the time of the onset of primary ridge formation) results in a low-ridge-count pattern such as an arch [11,13,15, 27]. It has also been established that the size of the volar pad with respect to the finger is mainly under genetic regulation of growth rate, timing of volar pad regression and timing of the onset of primary ridge formation [5]. Given also, is that gene over dosage as seen in Down syndrome is associated with dermatoglyphic aberrations [2, 9,10, 29]. The result of digit pattern as seen in this study supports the inference that gene over dosage as seen in Down syndrome, could cause:

- (1) an abnormally increased occurrence of asymmetrical, leaning volar pads, reflected in the higher occurrence of loops, especially of the ulnar type in patients.
- (2) a complementary abnormally decreased occurrence of symmetrical, round-shaped volar pads, reflected in the lower occurrence of whorls in patients.
- (3) an abnormally persistent volar pad caused by late regression of the pad which is reflected in the lower occurrence of arches in the patients.

The dermal configurations of the inter-digit areas could be described as “patternless” (open field) or “pattern”. The patterns seen in our samples were Carpal loop and vestige (CL+V); Loop (L), Loop and vestige (L+V), Radial loop (RL); Radial loop and vestige (RL+V), Vestige (V). The significant difference found in inter-digit C of the left hand was probably due to the presence of more open field in control (72.22%) compared to patients. Similarly,

the significant difference in dermal ridge configuration found in the inter-digit D of patients and normal subjects are due to variation and distributions of the dermal ridge patterns. The study of Cummins's [6] report that there are more patterns in the second and third inter-digit areas of the Down syndrome patients agrees partially with our study as more patterns are found in the second inter-digit area. A study among Canadians showed that 82% of the translocation patients had loop patterns in the third inter-digit space compared to 86% of the trisomic patient. Another study showed that 50.7% of their patients had true patterns in the third inter-digit space on the left hand, and 76.0% on the right compared to 25.7% and 49.5% in controls.

There are reports of inter-digital patterns in some Asian populations. Bryant *et al.*[19] found high occurrence of patterns in the third inter-digit area of Chinese children with Down syndrome compared with the normal. Among Iranian population high occurrence of patterns in the third inter-digit space are more indicative of Klinefelter syndrome than Down syndrome [26]. The Nigerian patients in the present study displayed inter-digit pattern similar to Down syndrome patients of European and North American extraction when compared with Iranian population

The occurrence of single flexion crease in the 5th digit was significantly different in Down syndrome patients and control in this study. Single flexion crease in the fifth digit was reported to occur more in Down syndrome patients than in normal individuals [30].The study by Soltan & Clearwater [3] revealed that single flexion crease in the fifth digit occurred in 22% of the translocation patients and 27% of the trisomic patients. Anderman [23] reported similar pattern for Canadians. There was no occurrence of single crease in fifth digit in the normal Chinese children; however there was a high frequency of its occurrence amongst Chinese patients.

## **5. CONCLUSION**

A dermatoglyphic study of a group of 31 Down syndrome patients and 90 controls of Nigeria origin has been presented, and results compared with studies on European, North American, South American, Asian groups. Nigeria patients had results similar, but not identical, to findings in the other racial groups. The diversity in the results, are mostly due to diverse racial traits. The occurrence frequencies of ulnar loop type of digit pattern, third inter-digit patterns and single flexion crease on 5th digit showed strong diagnostic value in the Nigeria patients. Considering the findings in the present study, dermatoglyphic criteria can be used as a supplementary diagnostic aid for Down syndrome.

### **Consent**

All volunteers gave written informed consent before they participated in the study

### **Ethical Approval**

Ethical approval was given by the Ethical Committee College of Health Sciences University of Port Harcourt.

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