

# Dermatoglyphic Assessment in Down and Klinefelter Syndromes

M. Hassanzadeh Nazarabadi,  
R. Raoofian, R. Abutorabi<sup>1</sup>,  
H.B. Hosseini<sup>1</sup>

## Abstract

**Background:** Dermatoglyphics are the dermal ridge configurations on the digits, palms and soles. Dermatoglyphic polymorphism results from the co-operation of genetic and environmental factors. The Dermatoglyphic analysis is a valuable completion of initial diagnosis of some syndromes genetically determined. Our objective was to assess dermatoglyphics study results against standard chromosomal analysis in Down and Klinefelter syndromes.

**Methods:** In this study we applied clear plastic tape and graphite powder for finger and palm prints of 90 persons. Cytogenetic study was also performed for patients with Down (n=29) and Klinefelter (n=22) syndromes and 39 normal individuals who served as the control group.

**Results:** Dermatoglyphic investigations indicated that in Down syndrome, simian line, ulnar loops, whorl, t", t'" and t' were significant, whereas arch and interdigital III pattern were more indicative for Klinefelter syndrome.

**Conclusion:** Dermatoglyphic can be used both as an initial diagnostic step and for screening purposes.

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**Keywords** • Dermatoglyphics • Down syndrome • Klinefelter syndrome

## Introduction

**D**ermatoglyphics are the dermal ridge configurations on the digits, palms and soles.<sup>1-6</sup> The scientific study of papillary ridges of the hands and feet was first begun in 1823 by Evangelista Purkinje—a Czech physiologist and biologist. He was the first who systematically categorized fingerprint patterns. In 1892, Sir Francis Galton published his classic treatise on fingerprints. He studied the hereditary aspects of fingerprints, compared siblings, twins and genetically unrelated individuals and was the first who reported concordance of papillary ridge patterns among relatives.<sup>2,7,8</sup>

Then, many studies have been emerged which increasingly shown how dermatoglyphics can predict a range of conditions and diseases.<sup>3,4,9-13</sup> The dermatoglyphic analysis is now a valuable companion to other methods used for diagnosis of some genetic diseases (e.g., phenylketonuria) and syndromes genetically determined (e.g., Down, Turner or Klinefelter syndromes).<sup>14-16</sup>

Dermatoglyphic polymorphism results from the interplay of

Department of Medical Genetics,  
<sup>1</sup>Endocrinology,  
Ghaem Hospital,  
Mashad University of Medical Sciences,  
Mashad, Iran.

## Correspondence:

Mohammad Hassanzadeh  
Nazarabadi PhD,  
Head of Medical Genetics Dept.,  
Medical Faculty,  
Mashad University of Medical Sciences,  
Mashad, Iran.  
**Tel:** +98 511 8828575  
**Fax:** +98 511 8419796  
**Email:** nazarabadi@hotmail.com

genetic and environmental factors during the early stages of ontogenesis.<sup>15</sup> Dermal ridge configurations begin to develop about the 13<sup>th</sup> week of gestation as the fetal mounds on the digit tips, and interdigital, thenar and hypothenar areas of the hand. The pattern formation is complete by the 19<sup>th</sup> week.<sup>6,17-19</sup> These findings on ridge development, have been recently confirmed by Blechschmidt, Penrose and O'Hara, Okijima, and Babler.<sup>7,20,21</sup>

There are three basic fingerprint patterns: arches, loops and whorls or mixtures of them. The loops may be ulnar or radial.

A triradius is a three-way fork—a confluence of three-ridge systems. Arches have no triradii; a loop has one and the whorl has two or more triradii. A triradius is seen at the base of the palm—the axial triradius. This may be displaced distally in certain conditions. The palmar creases are usually made up of proximal and distal transverse creases and a thenar crease. A single transverse (crosswise) palmar flexion crease in the hand is called “simian crease” which is most often associated with chromosomal abnormalities such as trisomy 21, trisomy 18, trisomy 13, etc.<sup>22</sup>

Dermal and palmar ridges are highly useful tools in medical studies including autosomal and sex chromosomal anomalies.<sup>1,5,9,11,14</sup> Their notably variable characteristics are not duplicated in other people, even in monozygotic twins or in the same person, from location to location.<sup>2</sup>

In this study, we assessed the dermatoglyphic study results against standard chromosomal analysis on two groups of patients with Down or Klinefelter syndrome and normal individuals.

### Patients and Methods

To obtain the prints, we used 3/4"-wide clear plastic tape for fingerprints and 4"-wide tape for palm prints in 29 patients with Down syndrome, 22 patients with Klinefelter syndrome and 39 unrelated normal individuals who served as the control group. Graphite powder was rubbed over the area to be printed and the tape was gently pressed against the surface. When the tape was peeled off, the image of the print which was transferred to the tape pressed onto a sheet of paper.<sup>2,8,16</sup> Cytogenetic study was performed based on the G-banding technique at high resolution.<sup>23</sup>

### Results

Simian crease was observed significantly ( $p < 0.001$ ) more frequently in those with Down

syndrome ( $n=13$ , 45%) than in those with Klinefelter (0%) and controls ( $n=1$ , 3%).

The whorls (53%) and ulnar loops (66%) were more frequent in Down syndrome, whereas arches (18%) were more frequent in those with Klinefelter syndrome. On the other hand, radial loops were less frequent in both Down and Klinefelter syndromes (0%, and 2%, respectively) as compared with the control group (6%).

The arch patterns were more prevalent ( $p=0.033$ ) in Klinefelter syndrome (18%) than in Down syndrome (3%) and controls (6%). The frequency of ulnar loops and whorls were essentially the same in Klinefelter patients and controls.

Figures 1 and 2 summarize the results total ridge count (TRC) and mean ridge breadth (MRB) in Down and Klinefelter syndromes. No significant associations was found between the incidence of mean TRC in Down (93.4) and Klinefelter (97.5) syndromes, in contrast with the control group (136.2). The highest number of TRC in Down and Klinefelter syndromes were 113 and 114, respectively; the lowest number in controls was 115 (fig 3). No significant differences was found in MRB between the two syndromes and the control group (51.7, 57, 61.2, respectively).

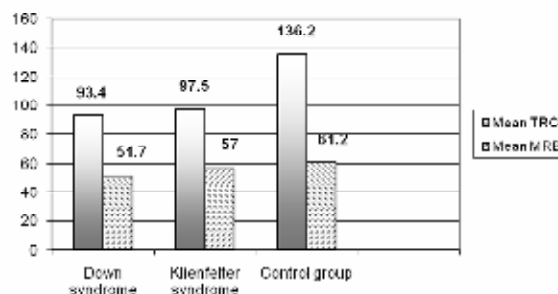


Fig 1: TRC and MRB in the three study groups.

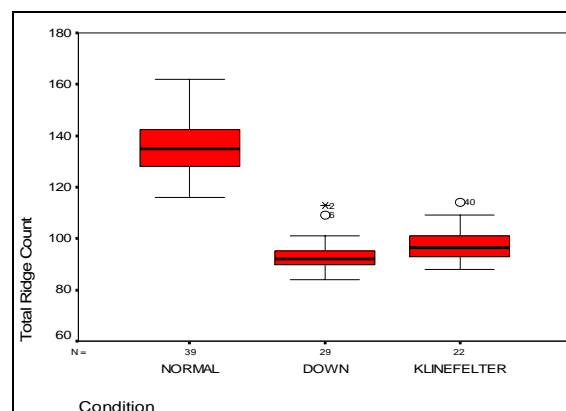


Fig 2: Box plot of TRC in the three study groups. \* , 0 : Outliers

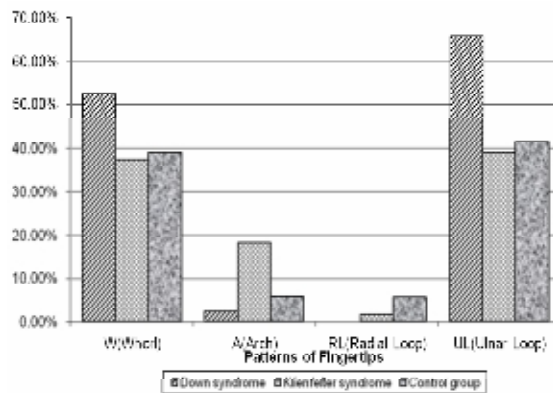


Fig 3: Prevalence of fingertip patterns

The data concerning additional triradii of the palm are shown in fig 4. The t' (28%), t'' (30%) and especially t''' (42%) were significantly more frequent in Down than Klinefelter syndrome (20%, 0%, 9%, respectively)(p<0.01 for t'' and t''' patterns). The prevalence of interdigital II and III patterns were higher in Klinefelter (8%, 8%, respectively) than in Down (5%, 0%, respectively) syndrome; the difference for interdigital III pattern was statistically significant (p=0.042). The prevalence of interdigital IV pattern was also high in Down (16%) and Klinefelter (17%) syndrome as compared with the controls (8%) (fig 4 table 1).

Fig 4: Prevalence of Triradii in palms.

Using a logistic regression analysis, the discriminant equation:

$$y = 4.5 \times \text{Simian line} + 3.5 \times t' + 2.7 \times \text{Ulnar loop} - 3.2$$

yielded a sensitivity of 72% and a specificity of 93.3% for diagnosing Down syndrome.

The arch pattern can be used as a screening test for diagnosis of Klinefelter syndrome with a sensitivity of 80% and a specificity of 79%.

Discussion

The reported frequencies in the three groups were compatible with reports of McGovern, Fogle and others which had reported that the prevalence of simian line was 40%–50% in Down syndrome. Therefore, simian line could be a valuable aid when the clinical diagnosis is uncertain.<sup>24-30</sup>

These frequencies of fingertip patterns in our study were also comparable with other reports which revealed that increase of ulnar loops was more frequent in Down syndrome. However, results of some previous studies, which reported excess of radial loops in the 4<sup>th</sup> and 5<sup>th</sup> fingers, were inconsistent with the our findings.<sup>3,24,29,31,32</sup> Such a discrepancy can be explained by ethnohistoric and geographic variations between different population.<sup>7,33,34</sup>

The arch patterns in our study groups resembled the results of Shiono, *et al*, who found high frequencies of fingertip arches in patients with Klinefelter.<sup>35</sup> Komatz and Yoshida, reported that in Klinefelter syndrome, arch patterns of digit I had a significantly higher incidence, but in digit II, the ulnar loops were significantly more frequent and whorls were significantly less frequent.<sup>36</sup> However, in our study, the frequency of ulnar loops and whorls were essentially the same in Klinefelter patients and controls. The diverse results outlined in these studies, can be explained by racial traits and environmental factors in different countries.<sup>33,37,38</sup>

Our results were in keeping with other reports which stated that TRC were significantly lower in both Down and Klinefelter syndromes than in normal people.<sup>1,9,31,35,39</sup>

Considering our findings and other reports there are clear associations between dermatoglyphics and some chromosomal disorders. Therefore dermatoglyphics may be treated as a valuable initial diagnostic step for suspicious cases.<sup>4,5,14,15,25</sup>

Table1: Summary of analyzed parameters

	W	A	RL	UL	M TRC	M MRB	t'	t''	t'''	ID II	ID III	ID IV	S
N	39.2%	5.8%	5.7%	41.6%	136.2	61.2	8%	0%	0%	5%	0%	8%	2.5%
D	52.5%	2.5%	0%	66%	93.4	51.7	28%	42%	30%	5%	0%	16%	45%
K	37.6%	18.3%	1.6%	39%	97.5	57	20%	9%	0%	8%	8%	17%	0%

N: Normal , D: Down , K: Klinefelter , W: Whorl , A: Arch ,RL: Radial loop ,UL:Ulnar loop , ID: Interdigital , S: Simian

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