

Dermatoglyphics and Karyotype Analysis in Primary Amenorrhoea

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ABSTRACT

Background: Dermatoglyphics is the scientific study of the skin ridge patterns on the fingers, toes, palms of the hands and soles of feet. Dermatoglyphics is in use as a supportive diagnostic tool in genetic or chromosomal disorders as well as in clinical conditions with genetic etiologies. Primary amenorrhoea and Dermatoglyphics, both have the suspected multifactorial (genetic and environmental) aetiologies.

Objective: In the present study the finger dermatoglyphic patterns were studied in primary amenorrhoea cases and age matched fertile control females and also attention was given to find out whether a specific dermatoglyphic trait exists in primary amenorrhoea cases and whether it was statistically significant.

Materials and Methods: To study the role of dermatoglyphics in primary amenorrhoea, a study was conducted on 30 subjects

with primary amenorrhoea (as cases) and compared it with equal number of age matched fertile females (as controls). We studied fingertip patterns in all the subjects enrolled. Simultaneously we have assessed the Karyotype of primary amenorrhoea cases.

Result and Conclusion: Two subjects in experimental group have shown abnormal Karyotypes. The most significant finding in present study was increased total finger ridge count (TFRC) in primary amenorrhoea cases which was statistically significant. We also found higher frequency of loops and arches in primary amenorrhoea with abnormal karyotypes. This type of study may be quite useful as a supportive investigation, in stating the predisposition of an individual to primary amenorrhoea and referral of an individual for karyotyping.

Keywords: Dermatoglyphics, Karyotype, Primary amenorrhoea

INTRODUCTION

The term Dermatoglyphics has its origin from Greek words, derma means skin and glyphic means carvings [1]. The term was coined by Dr. Harold Cummins- the Father of Dermatoglyphics, at the University of Oklahoma in 1926. Dermal ridge differentiation takes place early in fetal development. The resulting ridge configurations are genetically determined and influenced or modified by environmental factors [2]. They are stable throughout life, unique to the individual and significant as a means of identification. Moreover, characteristic dermatoglyphic abnormalities have been reported in genetic disorders like Down's syndrome [3], Klinefelter's syndrome [4], breast cancer [5] etc. In down's syndrome, mostly ulnar loop pattern was observed. Abnormal dermatoglyphic features such as simian crease, Sydney line, and patterns in the hypothenar and Interdigital areas have been observed more frequently in down's syndrome [3]. In Klinefelter's syndrome increased in whorls and decreased in loops was observed when study was conducted in six such patients [4]. A pattern of six or more digital whorls was identified more frequently in women with breast cancer than in those without the disease [5]. Dermatoglyphic study in children of oral cleft have showed increased frequency of ulnar loops [6]. When dermatoglyphic study was conducted on diabetic males, increased radial, ulnar loops and arches with decreased whorls were observed [7]. Finger tip study in students presented with extensive dental caries showed more whorls [8].

Dermatoglyphics is in use as a diagnostic tool in genetic or chromosomal disorders as well as in clinical conditions with genetic etiology. WHO Annual reports 1982 and 1985 have estimated that 15% of the human population is infertile and that amenorrhoea is the 6th major cause of female infertility [9]. So, the study was designed to see the dermatoglyphic pattern in primary amenorrhoea subjects and also to find out the dermatoglyphic pattern in primary amenorrhoea subjects who have abnormal karyotypes. Amenorrhoea and Dermatoglyphic patterns both have multifactorial determinants which can be broadly categorized as genetic and

environmental [9]. In a number of diseases having a strong genetic basis dermatoglyphics may be a supportive diagnostic aid. Some of these genetic disorders like Turner's syndrome and gonadal dysgenesis manifest with 'primary' amenorrhoea. Failure of onset of menstruation beyond the age of 16 y regardless of development of secondary sexual characters is called as primary amenorrhoea [10]. When there is absence of menstruation for six months or more following normal menstruation, it is called secondary amenorrhoea [11]. Our aim was to establish the relationship between primary amenorrhoea and Dermatoglyphics, due to their suspected genetic aetiology.

MATERIALS AND METHODS

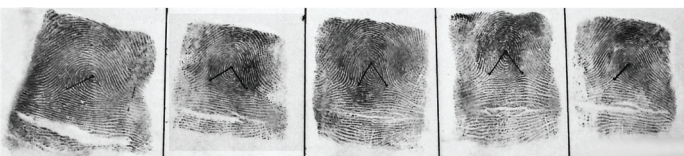
The present study was carried in the Department of Anatomy and Department of Obstetrics & Gynecology from January 2010 to August 2011. Thirty primary amenorrhoea subjects, from central Indian population with the age ranging from 17 to 25 y, were enrolled as 'cases' and thirty age matched fertile females as 'controls'. For all the subjects informed consent was taken in their local language along with detailed history before starting the procedure. All the records were maintained.

Inclusion criteria

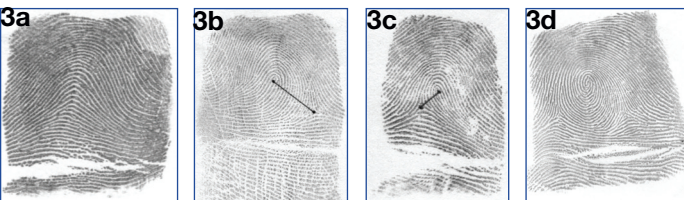
1. **For 'cases':** Female subjects of 17 to 25 y of age who presented themselves in Obstetrics/Gynecology OPD with the problem of failure to have onset of menses (primary amenorrhoea). All the subjects were referred from Obstetrics/ Gynecology Department to the division of human cytogenetic for karyotyping.
2. **For 'controls':** Married females up to the age of 25 y who had given birth to at least one live child without the history of miscarriages or genetic abnormalities. All the subjects were examined by specialist and found healthy. The subjects were also not suffering from any known genetic or clinical disorders and were not on medication.



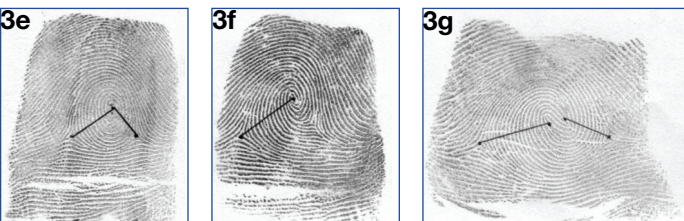
[Table/Fig-1]: Dermatoglyphic print of fingertips of control female
R1-W^uR2-L^u R3-L^u R4-L^uR5-L^u



[Table/Fig-2]: Dermatoglyphic print of fingertips of primary amenorrhoea female
R1-L^uR2-W^c R3-W^c R4-W^c R5-L^u



[Table/Fig-3a]: Arch, [Table/Fig-3b]: Ulnar Loop(L^u) Ridge counting done along the line, [Table/Fig-3c]: Radial Loop(L_r), [Table/Fig-3d]: Simple whorl (Spiral whorl-W^s)



[Table/Fig-3e]: Simple whorl (Concentric whorl-W^c) [Table/Fig-3f]: Composite whorl (Central pocket loop) [Table/Fig-3g]: Composite whorl (Lateral pocket loop)

10 Finger average	TL%	TW%	TA%
CF(Rt+Lt)/2	67.33	26.33	6.33
PAF(Rt+Lt)/2	63.33	33.99	2.67

[Table/Fig-4]: Distribution of finger tip patterns on both hands in two groups

Parameter	Controls (%)	Primary amenorrhoea subjects (%)	Chi Square	p-value
TUL	199 (66.33%)	186 (62%)	0.34	0.55 NS, p>0.05
TRL	3 (1%)	4 (1.33%)	0.00	1.00 NS, p>0.05
TL	202 (67.33%)	190 (63.33%)	0.35	0.55 NS, p>0.05

[Table/Fig-5]: Statistical analysis of loops

Cytogenetic analysis

All these 30 primary amenorrhoea subjects were also subjected for karyotyping in the division of human cytogenetic in the Department of Anatomy; Karyotypes were analysed by conventional Giemsa staining and G-banding by Trypsin and Giemsa (GTG) [12].

- **Method of dermatoglyphic recording:** Ink method was selected in our study for dermatoglyphic analysis [13]. Palmar and each fingertip dermatoglyphic prints of control females as well as primary amenorrhoea females were taken by rolling the finger from radial to ulnar side to include whole of the patterns [Table/Fig-1&2]. Detailed dermatoglyphic analyses of the prints were done with the help of magnifying lens.

Parameter	Controls	Primary amenorrhoea subjects (%)	Chi Square	p Value
TSW	52 (17.33%)	58 (19.33%)	0.13	0.71, NS, p>0.05
TW ^c _{amp}	27 (9%)	44 (14.67%)	1.70	0.19, NS, p>0.05
TW	79 (26.33%)	102 (33.99%)	1.52	0.21, NS, p>0.05

[Table/Fig-6]: Statistical analysis of whorls

Parameter	Controls	Primary amenorrhoea subjects (%)	Chi Square	p Value
TA ^p	18 (6%)	8 (2.67%)	1.04	0.30, NS, p>0.05
TA ⁱ	1 (0.33%)	0	1.00	0.31, NS, p>0.05
TA	19 (6.33%)	8 (2.67%)	1.04	0.30, NS, p>0.05

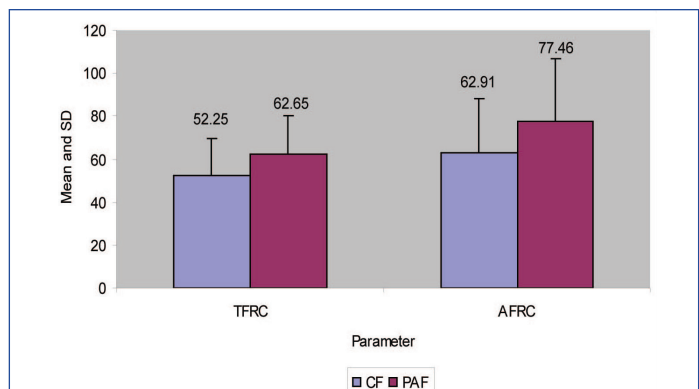
[Table/Fig-7]: Statistical analysis of arches

Parameter (out of 300 fingertips)	Controls (%)	Cases (%)	Chi Square value	p Value
TL	202 (67.33%)	190 (63.33%)	0.35	0.55, NS
TW	79 (26.33%)	102 (33.99%)	1.52	0.21, NS
TA	19 (6.33%)	8 (2.67%)	1.04	0.30, NS

[Table/Fig-8]: Statistical analysis of fingertip pattern

Parameter	Controls	Cases	z Value	p-value
TFRC	52.25	62.65	2.39	0.017, S
AFRC	62.91	77.46	1.82	0.068, NS

[Table/Fig-9]: Statistical analysis of finger ridge count (TFRC and AFRC)



[Table/Fig-10]: Total finger ridge count and absolute finger ridge count in primary amenorrhoea and control females

Following parameters were studied:

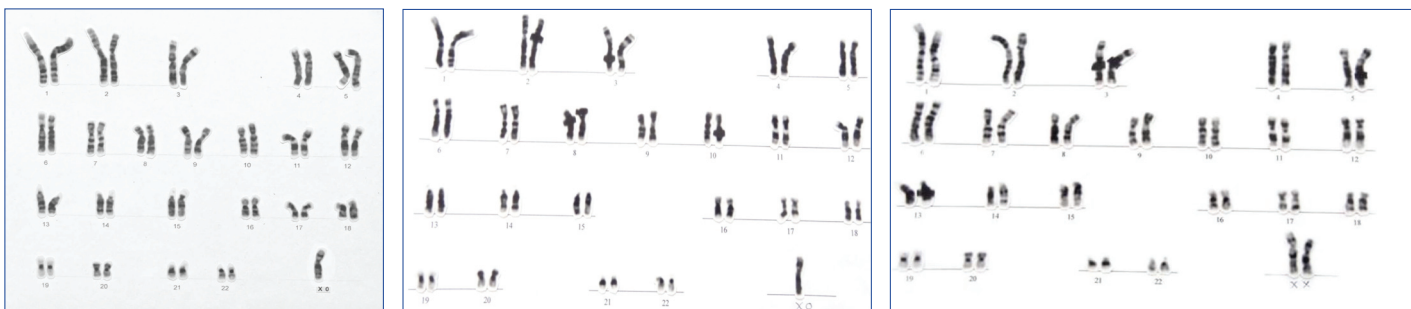
- 1) Qualitative analysis of finger tip patterns (FTP).
 - Study of arches (arch plain/ arch tented) [Table/Fig-3a].
 - Study of loops (ulnar loop/ radial loop) [Table/Fig-3b&c].
 - Study of whorls (simple whorl / whorl composite) [Table/Fig-3d-g].
- 2) Quantitative analysis of finger tip patterns by doing finger ridge count (Total finger ridge count (TFRC) and Absolute finger ridge count (AFRC)).

Total Finger Ridge Count (TFRC): It includes the sum of ridge counts of all ten digits, where only the larger count is used on those digits with more than one ridge count. It expresses size of pattern.

Absolute finger Ridge Count (AFRC): The additions of the ridge counts from all the separate triradii on all ten digits are included in AFRC. It reflects the pattern size and pattern type also.

STATISTICAL ANALYSIS

statistical analysis was done by descriptive statistics and inferential statistics using Mann-Whitney U-Test and Chi-square test using statistical software SPSS Version 17.00 and Graphpad Prism four.



[Table/Fig-11]: G-Banded Karyotype of a Subject with Primary Amenorrhoea Karyotype: 45, XO (Turner Syndrome)

[Table/Fig-12a]: G-Banded Karyotype of a Subject with Primary Amenorrhoea-Mosaic Turner, Karyotype: 45, XO

[Table/Fig-12b]: G-Banded Karyotype of a Subject with Primary Amenorrhoea-Mosaic Turner, Karyotype: 46, XX

	Control F(n=30)	PAF(n=30)	PAF with normal karyotype (n=28)	PAF with abnormal karyotype (n=2)	Chi square	P value
Arch	19(6.33%)	8(2.67%)	7(2.5%)	1(5%)	0.52	0.47, NS,p>0.05
Loop	202(67.33%)	190(63.33%)	174(62.14%)	16(80%)	7.86	0.005, S,p<0.05
Whorl	79(26.33%)	102(33.99%)	99(35.35%)	3(15%)	10.67	0.001, S,p<0.05

[Table/Fig-13]: Finger tip pattern in primary amenorrhoea subjects along with Karyotype correlation

RESULTS

The dermatoglyphic observations of finger tips of 30 control females and 30 primary amenorrhoea female subjects were analysed and recorded as per following details:

Basic finger tip patterns:

Control group

- Loops:** Loops were the most frequent pattern in controls. We found 202 loops out of 300 fingertips and the frequency was 67.33% [Table/Fig-4]. The proportion of ulnar loops to radial loops in control females was 199:3 [Table/Fig-5]. Thus there was increased percentage of total ulnar loops (66.33%) as compared to total radial loops (1%) in controls.
- Whorls:** In all controls the whorls were found to be the second in the descending order of frequency amongst digital patterns. 79 out of 300 fingertips of control females had whorls. The frequency was 26.33% in control females [Table/Fig-4]. The proportion of total simple whorl to total whorl composite in control females was 52:27 [Table/Fig-6]. Thus there was increased percentage of total simple whorl (17.33%) as compared to total whorl composite (9%) in controls.
- Arches:** In all control females arches were the least frequent pattern. 19 out of 300 fingertips of control females had arches. The frequency was 6.33% [Table/Fig-4]. The proportion of total arch plain to total arch tented in control females was 18:1 [Table/Fig-7]. Thus there was increased percentage of total arch plain (6%) as compared to total arch tented (0.33%) in controls.

Primary amenorrhoea subjects:

- Loops:** Loops were the most frequent pattern in primary amenorrhoea subjects (190/300). The frequency was 63.33% [Table/Fig-4]. The proportion of ulnar loops to radial loops in primary amenorrhoea females was 186:4 [Table/Fig-5]. Thus, there was increased percentage of total ulnar loops (62%) as compared to total radial loops (1.33%) in cases.
- Whorls:** 102 out of 300 fingertips of 30 primary amenorrhoea subjects had whorls. The frequency was 33.99% [Table/Fig-4]. The proportion of total simple whorl to total whorl composite

in primary amenorrhoea females was 58:44 [Table/Fig-6]. Thus there was increased percentage of total simple whorl (19.33%) as compared to total whorl composite (14.67%)

- Arches:** 8 out of 300 fingertips of 30 primary amenorrhoea subjects had arches. The frequency was 2.67% [Table/Fig-4]. The proportion of total arch plain to total arch tented in primary amenorrhoea females was 8:0 [Table/Fig-7]. Thus there was increased percentage of total arch plain (2.67%) as compared to total arch tented (0).

We observed decrease in total loops and total arches and increase in total whorls in primary amenorrhoea cases as compared to controls but the difference was found to be statistically insignificant [Table/Fig-8].

Comparative analysis of fingertip pattern between control females and primary amenorrhoea subjects:

- Loops:** Considering overall distribution of fingertip patterns, there was decreased percentage of total loops (TL- 63.33%) as well as total ulnar loops (TUL- 62 %) in primary amenorrhoea subjects as compared to that of control females (TL- 67.33% and TUL- 66.33%)[Table/Fig-4,5]. Whereas percentage of total radial loops (TRL-1.33 %) was increased in primary amenorrhoea subjects as compared to that of control females (TRL- 1%).
- Whorls:** Regarding overall distribution of whorls, it was observed that the percentage of total simple whorl (19.33%), total whorl composite (14.67%) and total whorls (33.99%) in primary amenorrhoea subjects were increased as compared to total simple whorl (17.33%), total whorl composite (9%) and total whorl (26.33%) in control females [Table/Fig-6].
- Arches:** When overall digital distribution of arches was analysed on both hands, we observed that total arch plain (2.67%), total arch tented (0) and total arches (2.67%) were decreased in primary amenorrhoea subjects as compared to total arch plain (6%), total arch tented (0.33%) and total arches (6.33%) of control females [Table/Fig-7].

Finger ridge count

It was observed that the mean total finger ridge count (TFRC= 62.65) and absolute finger ridge count (AFRC= 77.46) in primary amenorrhoea subjects were increased when compared with the mean of TFRC (52.25) and AFRC (62.91) in control females. Increase in TFRC was found to be statistically significant [Table/Fig-9,10].

Karyotype analysis (performed in 'cases' only)

In the present study, out of 30 primary amenorrhoea subjects there were two subjects with abnormal karyotype- one subject with Turner syndrome, karyotype 45, XO [Table/Fig-11] and second of Mosaic Turner (Karyotype 45, XO/46, XX) [Table/Fig-12a&b]. When overall digital distribution of fingertip pattern (FTP) of these subjects was analysed on both hands, we observed that loops seemed to be of higher frequency in primary amenorrhoea females with abnormal karyotype (80%) as compared to primary amenorrhoea females

with normal karyotype (62.14%). Arches seemed to be of higher frequency in primary amenorrhoea with abnormal karyotype (5%) as compared to primary amenorrhoea with normal karyotype (2.5%). But the frequency of whorls in primary amenorrhoea females with abnormal karyotype (15%) was decreased as compared to primary amenorrhoea with normal karyotype (35.35%) [Table/Fig-13].

DISCUSSION

Dermatoglyphics has been a useful tool in understanding basic biological questions in medicine, genetics and evolution since the past 150 y, apart from its wide application for personal identification. Dermatoglyphic analysis is now beginning to prove itself as an extremely useful tool for preliminary investigations into conditions with a suspected genetic basis [14].

In the present study we found reduction in total loops (TL) and total arches (TA) in 'cases' of primary amenorrhoea group. Some authors found increased ulnar loops (6 of 10 cases) and increased whorls (3 of 10 cases with Turner's syndrome) when dermatoglyphic study was carried out in ten female patients with sex chromatin abnormalities and genital tract anomalies complaining of primary amenorrhoea [15]. We also found increased total whorls (TW). Another author reported increased percentage of total loops and decreased percentage of arches in primary amenorrhoea subjects as compared to controls [16]. When dermatoglyphic analysis of 29 patients of classical gonadal dysgenesis was done it was observed that there was increased incidence of ulnar loops on the first digit of both hands [17]. Some authors found increased frequency of ulnar loops on first finger of right hand when study was carried out in 52 women with the Ullrich-Turner syndrome [18]. Dermatoglyphic study of the females having children with Trisomy 21 showed more arches and less whorls [19]. Increased ulnar loop was observed in schizophrenics with Down's syndrome [20].

In our study frequency of loops and arches was highest in primary amenorrhoea subjects with abnormal karyotype (2 cases) when compared with primary amenorrhoea subjects with normal karyotype. Some workers observed an increase in ulnar loops and whorls but decrease in radial loops and arches in a study carried on Swedish cases of Turner's syndrome showing the symptoms of ovarian dysgenesis [21]. Another author found higher frequency of loops in both controls and primary amenorrhoea cases frequency was especially higher in subjects with abnormal karyotype [9]. Some authors studied total finger ridge count (TFRC) in primary amenorrhoea cases. They found increased total finger ridge count in cases of primary amenorrhoea which coincides with our study [15,21].

CONCLUSION

Dermatoglyphics has been considered as a growing science with an immense practical value and it has been emphasized as one of the diagnostic tool which aids the cytogeneticist to look for specific abnormalities and identification of abnormal chromosomes.

In the present study, we found statistically significant increase in mean total finger ridge count in primary amenorrhoea cases. We also observed increased mean absolute finger ridge count, increased total radial loops and total whorls which were statistically insignificant. While total ulnar loops, total arches were decreased which were also statistically insignificant.

Thus the utility of dermatoglyphics is not to diagnosis, but to prognosis of disease and also the identification of people with the genetic predisposition to develop certain disease. Dermatoglyphics may be a useful screening method to identify the population at risk and referral of such individuals for karyotyping so that a watch may be kept for the early onset of symptoms.

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FINANCIAL OR OTHER COMPETING INTERESTS: None.

Date of Submission: **Jun 12, 2014**
Date of Peer Review: **Sep 29, 2014**
Date of Acceptance: **Oct 06, 2014**
Date of Publishing: **Dec 05, 2014**