Cheiloscopy in individuals with Down syndrome and their nonsyndromic biological siblings

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ABSTRACT

The study aimed to carry out a comparative analysis between the lip print patterns in individuals with Down Syndrome and their nonsyndromic biological siblings. This was a cross-sectional blind study using an inductive approach and extensive direct observation procedures. A total of 68 cheiloscopic charts, named cheilograms, were divided into two groups (n=34), as follows: G1, including Down Syndrome individuals; and G2, including their nonsyndromic siblinggs. The convenience sample was selected in the city of João Pessoa, PB, Brazil. The following features were evaluated in eight labial regions called sub-quadrants: oral commissures (downturned, horizontal and upturned); lip thickness (thin, medium, thick and mixed); and labial grooves (I - complete vertical; I '- incomplete vertical; II – bifurcated; III – criss-cross; IV – reticular; or V - undefined). The data were analyzed by paired Student's t test and McNemar's Chi-square, with a 5% significance level. Most Down Syndrome individuals were found to have downturned oral commissures in 73.5% of cases, while their siblings showed a predominance of horizontal commissures in 73.5% of cases (p=0.009). There was no statistically significant difference for lip thickness between groups. In the analysis of labial groove patterns, Down Syndrome individuals (G1) showed a significant prevalence of the type I pattern (52.2%) as compared to their nonsyndromic siblings (30.1%) (p<0.001). Due to the tendency of having vertical labial groove patterns and downturned commissures, Down Syndrome individuals present cheiloscopic differences in relation to their nonsyndromic biological siblings, which suggests that syndromic genetics influences the development of these features. However, this may imply in a reduced potential of cheiloscopic identification due to the low divergence of labial phenotypes among Down Syndrome individuals.

INTRODUCTION

Down Syndrome (DS) was first described by John Langdon Down in 1866 as a chromosomal disorder resulting from the trisomy of the 21st pair. It is considered a natural and universal genetic event affecting 0.6-1/1,000 live births, and this proportion increases with maternal age. [1,2] DS is the most prevalent and studied congenital disease affecting the individual's intellectual capacity. [3] DS individuals may have changes in their physical, behavioral and cognitive development, [1,3-5] as well as a number of rare and/or anomalous patterns in their digital, palmar and plantar dermatoglyphics. These peculiar markers have been frequently used as an aid in diagnosis of this genetic alteration in 80% of cases. [6-8]

Cheiloscopy (from the Greek "*Cheilos*", lips; "Skopein", marks) refers to the registration and classification of the lip as to thickness, commissural arrangement and groove impressions, printed by an individual onto a given substrate. The study of cheiloscopy is based on the fact that the mucosal lip is covered with small grooves that reflect individual differences from a genetic basis and ethnic influence, thus providing specificity and variability to this method. [9-12]

Given their peculiar pattern – which is comparable to that of fingerprints – labial marks constitute a feasible identification technique applicable to forensic sciences. They possess the technical (classificability and practicability) and biological (uniqueness and immutability) requirements recommended by the literature and are hardly changeable, even in cases of trauma and inflammation. [11-16]

Due to their genetic background, DS individuals are more alike among themselves than when compared to their nonsyndromic biological siblings. As labial grooves have a genetic basis, it is still questionable whether such background influences the development of labial features and groove patterns in DS individuals. Thus, this study investigated the cheiloscopic features of DS individuals and their nonsyndromic siblings, in order to verify whether such chromosomal alterations may influence labial phenotypes and thus be used as a novel forensic tool for individual identification.

MATERIALS AND METHODS:

This study was carry out in accordance with the Resolution no. 466/12 of the National Health Council, Ministry of Health, which regulates research involving human beings. This study had prior approval of the Research Ethics Committee at the Center for Health Sciences, Federal University of Paraíba (CAAE no. 02955912.0.0000.5188).

This was a cross-sectional blind study using an inductive approach and extensive direct observation procedures.[17] A pilot study including 20 cheiloscopic records was previously performed to train the examiners. For the numerical variable (lip thickness), the intraclass correlation coefficient ranged from 0.922 to 0.984, while the Kappa statistics (labial grooves and commissures) showed agreement values between 0.767 and 1. Both analyses indicated satisfactory agreement between examiners with regard to the criteria used in our cheiloscopic study.

The sample was composed of 68 individuals divided into two groups (n=34): G1, consisting of DS individuals; and G2, consisting of their nonsyndromic biological siblings. The convenience sample was selected from a reference center for disabled individuals in the city of João Pessoa, PB, Brazil.

The volunteers presenting inflammation, trauma, malformation or other condition in the oral or perioral regions, were excluded from the sample. The data were collected based on three analyses, as follows: [9]

1. Lip thickness: After making sure that lips were free from cosmetics and/or other impurities, we used a needle point to measure the upper and lower lip thickness individually, taking as anatomical references the upper edge of the lip cord to the oral rhyme (for the upper lip); and the oral rhyme to the lower edge of the lip cord (for the lower lip). All measurements were made at the level of the median sagittal line. The record of the numerical value corresponding to the measurements was made with the use of a millimeter ruler (Hyzer-Krauss scale, adopted by the American Board of Forensic Odontology, ABFO, no. 2). The lips were analyzed and classified as: [18] thin (<8 mm), medium (8-10 mm), thick or very thick (>10 mm) and mixed (lips which denoted different categories for the upper and lower components).

2. Oral commissures: A high-resolution digital camera (DSLR Nikon D3200 24.2 Mega Pixels, São Paulo, SP, Brazil) with no flash was used to obtain two photographs (one more general and another with a close-up) for the analysis of the disposition of the oral commissures. The participant was positioned with their Frankfurt plane parallel to the ground and lips at rest. The oral commissures were categorized [9] as downturned, horizontal and upturned based on their disposition in relation to a virtual line perpendicular to the labial median line (which was tangent to the labial tubercle).

3. Labial groove pattern: an individual sample containing 0.8 g of lipstick (Avon Renew Renovador Intensivo Batom Cor Intensa® - red color, long-wearing, São Paulo, SP, Brazil) was applied onto the lips of the subjects, which should be dry, static, closed and relaxed. Then the lips were pressed and slightly scrolled from left to right against a white cardstock supported on a glass plate. The lip print was secured with an adhesive tape (3M®, transparent color, 45 mm, Sumaré, SP, Brazil). For the analysis of labial grooves, the collected lip print was divided into eight sub-quadrants (four upper and four lower sub-quadrants) (Fig. 1), which were numbered clockwise starting in the upper right sub-quadrant. The identification of groove types was carried out with the aid of a magnifying glass, in which the predominant type in each sulcular sub-quadrant was noted in cheilograms according to the previously described method: [19] Type I - complete vertical line; Type I' – incomplete vertical line; Type II - bifurcated lines; Type III – criss-crossed lines; Type IV - reticular lines; and Type V – undefined pattern not applicable to the previous classification (Fig. 2).

Figure 1: Schematic division of the lips into eight sub-quadrants, as follows: 1 and 2 – upper right sub-quadrants; 3 and 4 – upper left sub-quadrants; 5 and 6 – lower left sub-quadrants; and 7 and 8 – lower right sub-quadrants.

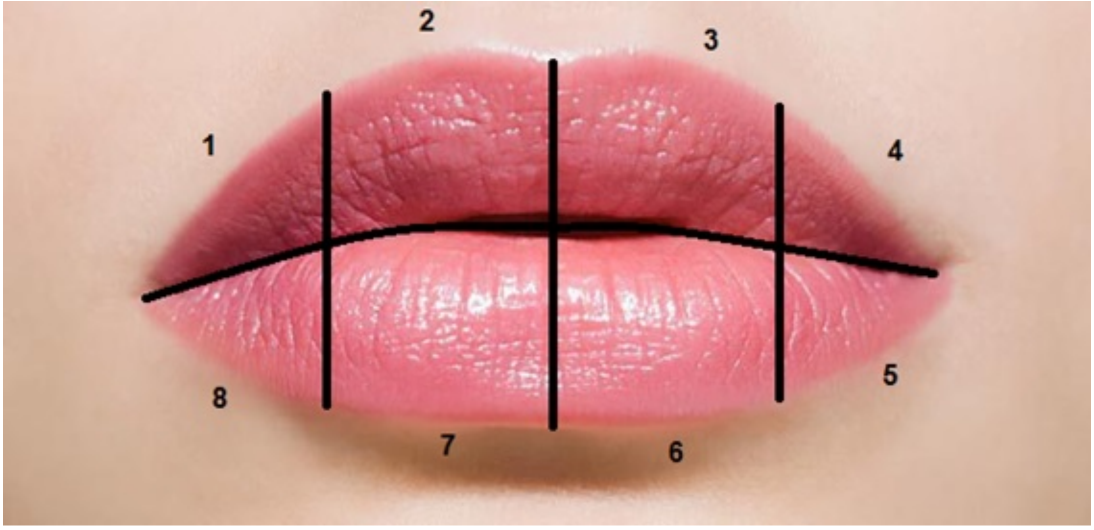
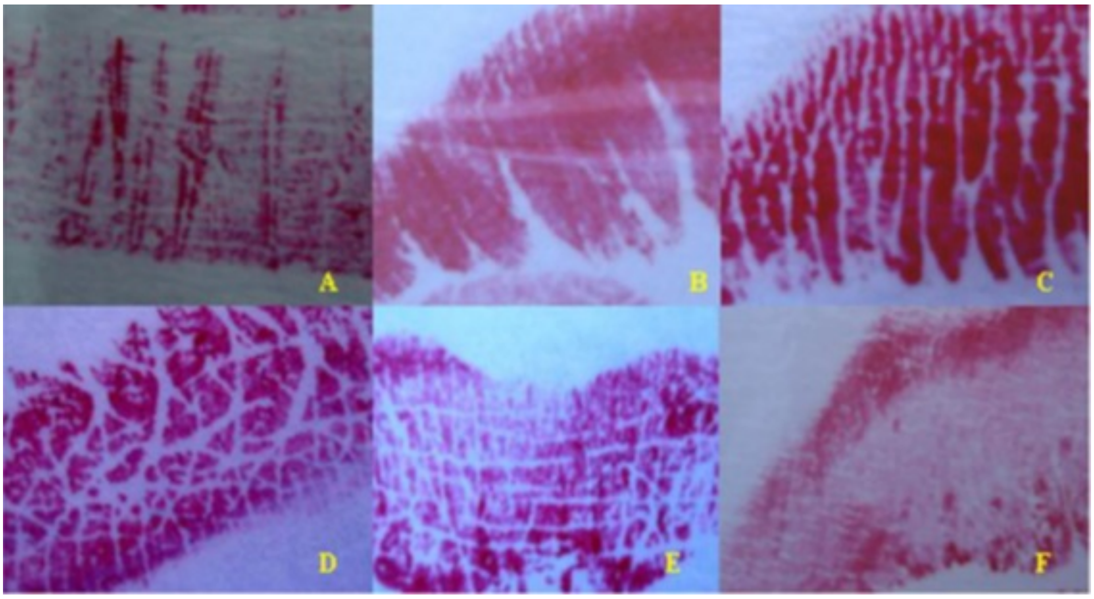


Figure 2: Labial groove types: Type I (A); I' (B); II (C); III (D); IV (E) and V (F). Source: Oliveira; Rabello; Fernandes (2012).



The data were processed on the Statistical Package for Social Sciences version 20.0 (SPSS Inc., Chicago, IL, USA). Descriptive and inferential statistical techniques were used. Student's *t* test was used for pair-wise comparisons between numerical variables, and McNemar's Chi-square test was used to compare categorical variables, in addition to Chi-square test of equal proportions in a single group. A 5% significance level ($p \leq 0.05$) was adopted.

RESULTS

Of the total sample, 63.24% were females. Individually, the groups had similar proportions of subjects regarding sex: G1 (70.58% females) and G2 (63.24% females). The age of DS individuals in G1 ranged from 3 to 42 years, with a mean age of 13.88 years, while their nonsyndromic siblings (G2) were aged between 3 and 39 years, with a mean age of 17.35 years (p=0.004).

As for lip thickness (Table 1), G1 individuals showed mean thickness of 8.88 mm and 9.62 mm for the upper and lower lips, respectively. G2 individuals

showed mean lip thickness of 9.84 mm and 11.07 mm, respectively, with a statistically significant difference between groups for both the upper (p=0.002) and lower (p=0.001) lips. Furthermore, we observed that individuals in G1 had a higher percentage of medium lips (50.0%), followed by mixed (35.3%), thick/very thick (11.8%), and thin lips (2.9%). Among the individuals with mixed lips, their upper lips showed predominance of the medium thickness (23.5%), in contrast with their lower lips showing thick or very thick patterns (20.6%). Individuals in G2 were found to be the ones with the highest prevalence of thick or very thick lips (44.1%), followed by medium (38.2%) and thin (17.6%) types, with no mixed lips identified in the sample.

The analysis of oral commissures (Table 1) revealed that DS individuals tend to have downturned commissures (73.5%). No upturned commissure was identified in DS individuals (G1). Nevertheless, their siblings (G2) showed a high percentage of the horizontal pattern (73.5%), followed by the downturned (20.6%) and upturned (5.9%) patterns (p=0.009).

As shown in Table 2, the most prevalent type of labial groove was type I in all sub-quadrants analyzed in G1 individuals (1- 44.1%; 2- 64.7%; 3- 41.2%; 4- 32.4%; 5-50.0%; 6- 70.6%; 7- 70.6%; 8- 44.1%) (Fig. 3). In G2, type I was most prevalent pattern in sub-quadrants 2 (32.4%), 3 (26.5%), 6 (55.9%) and 7 (52.9%). There was a predominance of type III in sub-quadrants 1 (29.4%) and 4 (32.4%), and of type II in sub-quadrants 5 (44.1%) and 8 (32.4%)" (Fig. 4)".

Type I labial groove was the most prevalent pattern in all sub-quadrants of G1 individuals (52.2%), followed by types I' and II (both 16.9%). Likewise, type I was the most prevalent pattern (30.1%) in G2 as well, followed by types II (23.9%) and III (19.9%), although with a significant difference between groups G1 and G2 ($p<0.001$) (Table 3).

DISCUSSION

Down syndrome is currently the most prevalent human trisomy worldwide, which is responsible for intellectual, physical and cognitive changes. [1-5] Since ancient times it has been known that singularities present in an individual may facilitate the task of their forensic identification. [20-23]

DS individuals have highly typical dermal ridge patterns, making it possible to diagnose this syndrome just by dermatoglyphic examination. Examples of irregularity in the palmprint pattern of DS individuals include the presence of a single palmar transverse crease or "simian crease"; prevalence of ulnar loop; and of a t-like axial palmar triradius displaced distally (which is present in about 70% individuals), usually associated with some patterns in the hypothenar area. [6-8]

This cheiloscopic study revealed common features shared by DS individuals (G1), such as the prevalence of downturned commissures and complete vertical lines (type I) in all labial sub-quadrants analyzed. These findings disagree with those found for their nonsyndromic biological siblings (G2), thus indicating that the trisomy of the chromosome 21 influences the lip phenotype. As for the grooves, such a finding is confirmed mainly in the central sub-quadrants of the lips. Studies have shown that marks on the lip corners are not clearly captured in the printing process, even those of great quality, which makes the central portion of the lip a decisive area for human identification procedures. [22,23]

Labial phenotypes are transmitted on the basis of heredity precepts, e.g., thin and thick lips are more typically found in white and black individuals, respectively. The results observed in the present study are in disagreement with those

Table 1: Description of the variables lip thickness and oral commissure according to the study group.

Variable	DS (G1) n	DS (G1) %	Nonsyndromic sibling (G2) n	Nonsyndromic sibling (G2) %	p-value
TOTAL	34	100,0	34	100,0	
Maximum thickness of the upper lip (average)	8.88 mm		9.84 mm		$p^{(1)} = 0.002$*
Maximum thickness of the lower lip (average)	9.62 mm		11.07 mm		$p^{(1)} < 0.001$*
Lip thickness					
Thin	1	2.9	6	17.6	
Medium	17	50.0	13	38.2	**
Thick/Very thick	4	11.8	15	44.1	
Mixed	12	35.3	-	-	
Mixed upper lips					
Thin	4	11.8	1	2.9	
Medium	8	23.5	13	38.2	**
Thick/Very thick	-	-	1	2.9	
Not mixed	22	64.7	19	55.9	
Mixed lower lips					
Thin	1	2.9	-	-	
Medium	4	11.8	2	5.9	**
Thick/Very thick	7	20.6	13	38.2	
Not mixed	22	64.7	19	55.9	
Oral commissure					
Horizontal	9	26.5	25	73.5	
Upturned	-	-	2	5.9	**
Downturned	25	73.5	7	20.6	

(*): Non-significant difference at a 5.0% level.
(**): It was not determined due to the difference in the number of categories.
(1): Paired Student's *t* test.

Table 2: Description of the variable type of labial groove according to the study group and the sub-quadrant.

Type	Sub-quadrant 1				Sub-quadrant 2				Sub-quadrant 3				Sub-quadrant 4			
	G1		G2		G1		G2		G1		G2		G1		G2	
	N	%	N	%	N	%	N	%	N	%	N	%	N	%	N	%
I	15	44.1	7	20.6	22	64.7	11	32.4	14	41.2	9	26.5	11	32.4	4	11.8
I'	8	23.5	4	11.8	6	17.6	5	14.7	8	23.5	5	14.7	9	26.5	8	23.5
II	4	11.8	5	14.7	5	14.7	6	17.6	6	17.6	8	23.5	7	20.6	5	14.7
III	1	2.9	10	29.4	-	-	5	14.7	1	2.9	5	14.7	1	2.9	11	32.4
IV	-	-	6	17.6	-	-	-	-	-	-	7	20.6	-	-	6	17.6
V	6	17.6	2	5.9	1	2.9	7	20.6	5	14.7	-	-	6	17.6	-	-
Type	Sub-quadrant 5				Sub-quadrant 6				Sub-quadrant 7				Sub-quadrant 8			
	G1		G2		G1		G2		G1		G2		G1		G2	
	N	%	N	%	N	%	N	%	N	%	N	%	N	%	N	%
I	17	50.0	7	20.6	24	70.6	19	55.9	24	70.6	18	52.9	15	44.1	7	20.6
I'	3	8.8	3	8.8	4	11.8	2	5.9	3	8.8	3	8.8	5	14.7	4	11.8
II	7	20.6	15	44.1	4	11.8	8	23.5	4	11.8	7	20.6	9	26.5	11	32.4
III	3	8.8	7	20.6	1	2.9	3	8.8	-	-	3	8.8	1	2.9	10	29.4
IV	-	-	1	2.9	1	2.9	1	2.9	1	2.9	2	5.9	1	2.9	-	-
V	4	11.8	1	2.9	-	-	1	2.9	2	5.9	1	2.9	3	8.8	2	5.9

Table 3: Description of the variable *type of prevailing labial groove* according to the study group.

Variable	DS (G1) n	DS (G1) %	Nonsyndromic sibling (G2) n	Nonsyndromic sibling (G2) %	p-value
TOTAL	272	100.0	272	100.0	
Total of sub-quadrants					
I	142	52.2	82	30.1	
I'	46	16.9	34	12.5	
II	46	16.9	65	23.9	$p^{(1)} < 0.001$*
III	8	2.9	54	19.9	
IV	3	1.1	32	11.8	
V	27	9.9	5	1.8	

(*): Significant difference at a 5.0% level.
(1): According to McNemmar's test.

Figure 3: Distribution of the most prevalent type of labial groove in G1 individuals.

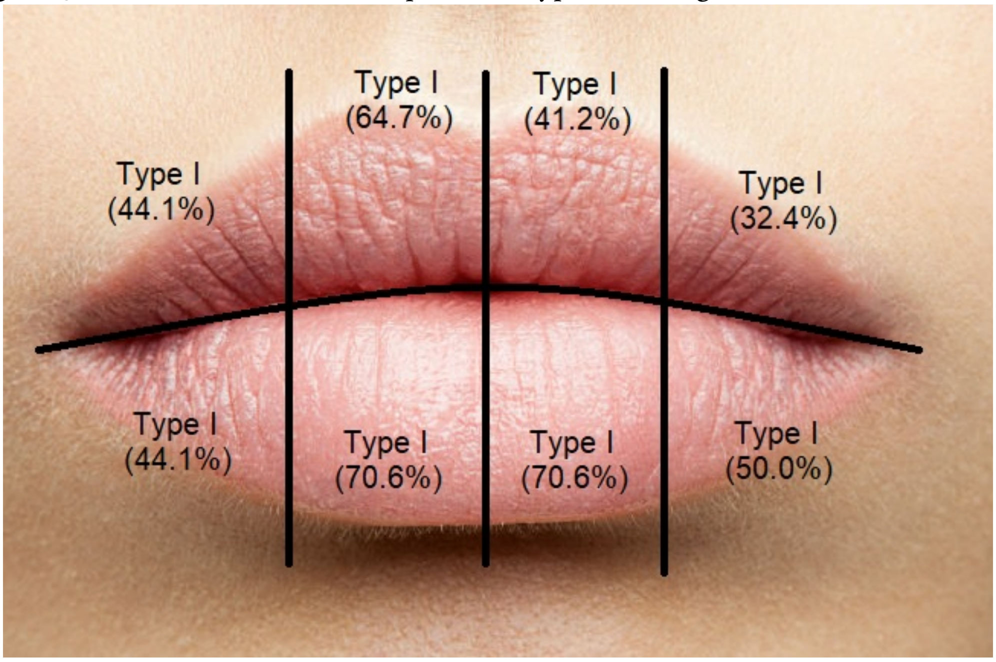
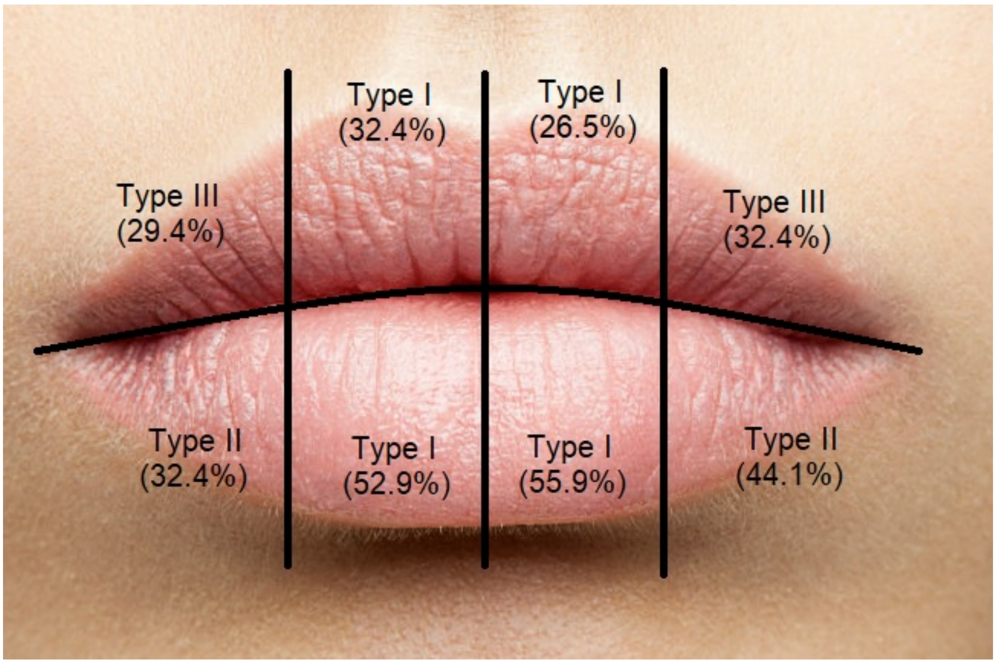


Figure 4: Distribution of the most prevalent type of labial groove in G2 individuals.



found in a previous similar study[9] including individuals from northeastern Brazil. The authors pointed out that the upper lips appeared more often with the thin type and that, contrary to this trend, the lower lips predominated as the thick type. The variation of thickness patterns in both groups can be attributed to ethnical miscegenation as a result of the historical and heterogenic Brazilian colonization, especially in northeast where africans, Indians and Europeans settled down. [9,16,24]

Other studies [9,16] have shown the predominance of horizontal commissures in their sample subjects, followed by downturned and upturned commissures. These results corroborate our findings concerning the group of nonsyndromic siblings, but diverge from those of the DS group, which showed a high percentage of the downturned commissural standard. This could be explained by the position and arrangement of the muscle fibers of perioral muscles in this group of individuals.

Despite lip thickness and commissural disposition are easily obtainable, such variables are considered essential for the initial acceptance or refusal of printings that are compared in a pairwise manner. [16,25] Only the detailed analysis of labial grooves by experienced examiners confronting the lip prints questioned with those of a possible suspect, can confirm or not the identification of the given evidence.

A study investigating lip prints in a sample from Lebanon [26] showed a predominance of groove types I, II and III, in that order, which was also observed herein in the group of nonsyndromic siblings (G2).

The cheiloscopy is considered a new method of human identification – including monozygotic individuals. The biological properties (uniqueness, permanence and immutability) of labial grooves are comparable to that of fingerprinting methods. [27,28] The latter are adopted worldwide as important resources for human identification, because although there is a resemblance between parents' and their children's features, their lines and groove patterns are not identical among them. [29-32] However the queiloscopy still needs of more accurate methods, since the labial anatomy can be altered according to the age, sex and ethnicity of the individual. [12,28,33]

CONCLUSION

It may be concluded that DS individuals present cheiloscopic differences in relation to their nonsyndromic biological siblings as the former have vertical groove patterns and downturned commissures. The findings reported herein suggest that chromosomal trisomy influences the development of such labial phenotypes. This fact minimizes the potential of cheiloscopic identification, since its technical foundations of classificability and practicability would be compromised due to the low divergence of labial phenotypes among DS individuals.

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