Utility of phenotypic dermal indices in the detection of down syndrome patients

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ABSTRACT

Down syndrome (DS) is the most common cause of mental retardation. The frequency of DS patients is about 1:800 and is mainly because of the presence of extra copy of chromosome number 21. Dermatoglyphic has been well established as a diagnostic aid in number of diseases having hereditary basis. Dermatoglyphic data was obtained by the use of ink and prints on a paper, from 15 cytogenetically confirmed patients of Down syndrome attending to the genetic clinic at BPKIHS. The data were correlated and compared with equal number of controls. Dermatoglyphic prints were used to evaluate the variation in the fingerprint patterns, the presence of simian crease and the difference in 'atd' 'dat' and 'adt' angles between the control and the DS patients. The results showed that both the 'atd' and 'adt' angles differed significantly from the control group. The dactylography study revealed higher incidence of loops and lower incidence of whorls in the DS patients as compared with the controls. This method is non-invasive and cost effective. The observed changes in the 'atd' and 'adt' angles plus the fingerprint patterns in the dermatoglyphic study proved that this simple technique could be a valuable tool for selecting patients of DS for cytogenetics analysis.

Keywords: Down syndrome, dermatolgyphy, simian crease, triradius angles, fingerprints.

INTRODUCTION

Down syndrome (DS) is one of the most common and best studied chromosomal disorder. It leads to mental retardation, with a frequency distribution of about 1:800 per live birth. J. L. H. Down, an English physician in 1866 first time described the clinical description of the condition which was subsequently given his name.[1] The terms "mongols", "mongoloid" and "mongolism" were widely adopted at that time because the condition reflected oriental cast of countenance produced by the characteristic epicanthal folds and upslanting palpebral fissures. The term is now considered inappropriate, abandoned and replaced by DS or Trisomy 21.

Dermatoglyphic is the study of epidermal ridges and their configuration on palms, fingers, soles and toes. The synonyms in use for epidermal ridges are papillary ridges, friction ridges and fingers prints. The dermatoglyphic (pattern of the ridged skin) is highly characteristic in Down syndrome patients. The dermatoglyphic features commonly noted in the hands of Down's syndrome patients include, increased incidence of ulnar loops, very high and L shaped ulnar loops, often have 10 ulna loops, reduced incidence of whorls and arches, decreased incidence of radial loops but increased incidence of radial loops on fingers other than Water finger (Fire/Air fingers normally virtually never have radial loops), displaced axial triradius to t2 position, large dermatoglyphic patterns in hypothenar area of the hand, interdigital Loop I3 on DS hands, simian crease commonly present, low a-b ridge count etc.[2]

Even before the awareness of chromosomal abnormality, Cummins had pointed out a set of characteristic dermatoglyphic features such as a single transverse crease, wide 'atd' angles in palm, increased occurrence of ulnar loops, hypothenar patterns in palms etc.[3,4] The dermal pattern in DS is as unique and as typical of the condition as the facial feature which is used in diagnosis.[5-7] Although dermatoglyphic features are known to be influenced by sex and age in normal but the dermatoglyphic features of DS patients are more or less irrespective of sex and even ethnic origin.[8]

An attempt has been made in this article to reiterate the already known fact that the presence of abnormal dermatoglyphic pattern may indicate the need for chromosomal analysis in suspected DS cases later followed by genetic counseling.

MATERIALS AND METHODS

Fifteen phenotypically diagnosed cases of Down syndrome were enrolled for the present study. These cases were selected from the patients attending the genetic clinic held every Wednesday in the Department of Pediatrics. In

order to study the dermatoglyphic patterns, palmar and fingerprints were obtained simply by the use of Indian ink and prints were taken on a paper. The diagnosis was confirmed by carrying out chromosomal analysis in the cytogenetic lab, Department of Anatomy, B. P. Koirala Institute of Health Sciences, Dharan, Nepal.

All the phenotypically diagnosed Down syndrome patients taken for this study was individually asked to wash his hand thoroughly and dry completely. He/she was then asked to put his palmer side of his hand on the stamp pad to completely smear with the ink and plant his palm on a clean white non-glazzed paper sheet. Care was taken to obtain complete and a clear identifiable print. The resultant pattern 'a'-'d' &'t' triradius were marked with the help of a magnifying glass. With the help of a scale and a protractor, the 'atd', 'dat' and 'adt' angles were measured. Similarly the presence of Simian crease and the fingerprints (loops, whorls and arches) were noted in details. The data were correlated with equal number of controls. For the classification of fingerprint patterns Henry- Galton system was followed.[9]

RESULTS

The presence of bilateral simian crease (Table-1) was observed in 60.0% of the total study cases. Only the complete transverse palmar crease (simian crease) was taken into account. In the control group the simian crease was absent.

Proximal to each finger an apex a triradius is formed by the dermal ridges by which exact center of each mount under the finger can be located. These triradii located proximal to the fingers are known as 'a', 'b', 'c' and 'd' triradius starting from the thumb to the little finger respectively. The triradius on the proximal palm in the center is known as 't'. The line joining the 'a', 'd' and 't' triradius forms angles 'atd', 'adt' and 'dat'. In normal being, the 'atd' angle ranges between 40º -45º while the normal 'dat' angle ranges between 56º-57º.

In the comparison of the 'atd' angle (Table-2) of the right and left hand respectively between the study cases and of the control group was observed. The 'atd' angle of the right hand of DS (48.07±2.3) was significantly ($p<0.0001$) higher than that of the controls (40.20±3.1). Similarly 'atd' angle of the left hand of DS (48.67±3.08) was significantly ($p<0.001$) higher in comparison to the controls (38.93±2.6). The 'adt' angle (Table-2) of the right hand of study group (72.67±5.8) was found to be significantly lower ($p<0.0001$) than that of the right hand of the control (80.27±4.1). Significant difference ($p<0.0001$) was also noted in the 'adt' angle of the left hand between the study group (73.80 ± 6.1) and the controls (81.13 ± 2.7). No significant difference was observed in the comparison of the 'dat' angle (Table-2) between the DS patients and the control of both the right and the left hand respectively.

Observation of the total no of fingerprints of both hand (Table-3) showed that the total number of loops was significantly higher ($p<0.0001$) in patients with DS (79.3%) as compared to the control group (47.3%). Significantly ($p<0.005$) fewer whorls were observed in the DS patients (17.3%) when compared with the controls (41.3%). No significant differences were seen in the number of arches in the two groups.

In right hand (Table 4) the total number of loops was significantly higher ($p<0.0001$) in patients with DS (77.3%) as compared to the control group (45.3%). A significant decrease ($p<0.0005$) was noted when comparing the whorls between the DS groups (17.3%) and the control (46.6%). No significant differences were found in the number of arches between the two groups. Comparison of the fingerprints in the left hand (Table-4) elucidated that total number of different types of fingerprints in the left hand between the DS group and the control illustrated significant difference ($p<0.00001$) in the pattern of loops between the DS (81.3%) and the control (49.3%). Number of whorls were also significantly ($p<0.01$) higher in controls (36.0%) than in the DS (17.3%). No significant difference was observed in the number of arches in the DS and controls. Observation of the individual fingerprints patterns showed that all the digits in DS patients showed significant increase in the percentage of loops, in right index finger ($p<0.01$) and right ring finger ($p<0.001$) and left index finger ($p<0.001$) and middle finger ($p<0.05$). Ulnar loop on all the ten fingers was observed in 3 (20.0%) study cases (Table-5).

All the digits showed lower percentage of whorls in DS patients as compared to control group. The difference between two groups was statistically significant in right index finger ($p<0.05$) and ring finger (0.05) and left index finger ($p<0.01$) (Table-6).

DISCUSSION

Transverse palmar crease (simian crease) (Fig. 1.) is the best known visible features of DS. It occurs much more frequently in DS than in controls and is supposed to be in about 50.0% of DS cases. It has been reported either as a complete or as an incomplete type.[10] In the present study the simian crease was observed in 60.0% of the total cases and was totally absent in the controls. Rajangam et al. noted the simian crease in 50.0% as complete and 91.7% as an incomplete type in the DS patients.[11] In our study only the complete type of simian crease was taken into account. If the latter is also included, the frequency would be higher.

The normal 'atd' angle is around 40-45º (Fig. 2). In the present study the atd angle was observed to be around 48º while the control was around 38-40º. Rajangam *et. al.* have reported wide 'atd' angle over 57º in 60.2% of patients.[5] Observation of the present study displayed 33.3% cases with the 'atd' angle over 50º.[12] The most peculiar dermatoglyphic feature of DS was the distal position of axial triradius. The pattern in the hypothenar area might be the cause for the presence of distal triradius in the palms of DS patients, resulting in the wide 'atd' angles. Most of the previous investigators have not observed 'adt' and 'dat' angles. However in a study conducted by Mandasescu

S, the normal 'dat' angle is reported to be around 57.7º.[13] Rignell and Davee also reported similar findings.[14,15] In the present study the 'dat' angle was observed to be around 60º in DS patients and 60-61º in controls. Similarly 'adt' angle was observed to be around 72-73º in DS patients and 80-81º in controls.

The ulnar loop was the most frequent type of finger pattern observed (77.3%), followed by whorls (17.3%) and arches (5.3%).The percentage frequency of the fingerprint pattern showed that the ulnar loop on all ten fingers was found to be occurring in 20.0% of the DS subjects. It is stated that one of the diagnostic indices in DS is the high frequency of ulnar loops in all the ten fingers. Rajangam *et al*. (1995) reported the presence of ulnar loop on all the ten fingers on 21.3% of the DS subjects and also noted higher frequency of ulnar loops (7.6%) which was comparable to our observation.[5] Excess of ulnar loops in the present study was observed on IInd (90.0%), Ist and IIIrd (83.3%) and Vth (80.0%) finger respectively. This corroborates with the previous findings of Borbolla *et al*. (1980) and Loesch who reported excess of ulnar loops on IInd and IIIrd fingers and Rajangam *et al*. (1995) who also observed excess of ulnar loops on IInd and IIIrd fingers. [11,16,17]

Hence the determination of dermatoglyphic pattern is a good indicator for diagnosis of DS patients which can then be further confirmed by chromosomal analysis. This method is non-invasive and cost effective. Thus the observed changes in the 'atd' and 'adt' angles plus the fingerprint patterns in the dermatoglyphic study proved that this simple technique could be a valuable tool for selecting patients of DS for cytogenetics analysis.

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Table-1: Showing the percentage frequency of simian crease in DS subjects and control

Simian crease	Down syndrome	Control
Right hand	60.0%	Absent
Left hand	60.0%	Absent

Table-2: Showing the comparision of 'atd', 'adt' and 'dat' angles between study group and control

	Right hand			Left hand		
	DS	**Control**	**p-value**	**DS**	**Control**	**p-value**
'atd' angle	48.07 ± 2.3	40.20 ± 3.1	0.0001	48.67 ± 3.0	38.93 ± 2.6	0.0001
'adt' angle	72.67 ± 5.8	80.27 ± 4.1	0.0001	73.80 ± 6.1	81.13 ± 2.7	0.0001
'dat' angle	61.07 ± 4.1	60.67 ± 4.1	NS	59.40 ± 4.5	61.33 ± 4.1	NS

n= 15 in each group.

Table-3: Showing the comparison of percentage of total no of fingerprints between study group and control

Type	**DS**	**Control**	**p-value**
Loops	79.33	47.33	0.0001
Whorls	17.33	41.33	0.005
Arches	3.33	6.66	NS

n= 15 in each group.

Table-4: Showing the comparison of percentage of total no of fingerprints in right and left hand separately between study group and control

	Right hand			Left hand		
	Loops	**Whorls**	**Arches**	**Loops**	**Whorls**	**Arches**
DS	77.3	17.3	5.3	81.3	17.3	1.3
Control	45.3	46.6	4.0	49.3	36.0	8.0
p-value	0.0001	0.0005	NS	0.00001	0.01	NS

Table-5: Showing the comparison of percentage of Loops in each digit in two groups

Loops	**Right hand**					**Left hand**				
Group	**I**	**II**	**III**	**IV**	**V**	**I**	**II**	**III**	**IV**	**V**
DS	80	86.70	80	66.70	80	86.70	93.33	86.70	53.33	80
Control	46.70	40	60	20	60	66.70	33.33	46.70	33.33	66.70
p-value	NS	<0.01	NS	<0.001	NS	NS	<0.001	<0.05	NS	NS

Table-6: Showing the comparison of percentage of Whorls in each digit in two groups:

Whorls	**Right hand**					**Left hand**				
Group	**I**	**II**	**III**	**IV**	**V**	**I**	**II**	**III**	**IV**	**V**
DS	20	6.70	6.70	33.33	20	13.33	0	13.33	46.70	20
Control	53.33	46.70	33.33	73.33	26.70	26.70	46.70	26.70	60	20
p-value	NS	<0.05	NS	<0.05	NS	NS	<0.01	NS	NS	NS

NS= Not Significant

Right hand

Left hand

Fig 1. Showing the simian crease

Fig. 2. Showing the three triradius (a,d & t) and the ‘atd’ angle