

DERMAL PRINTS PATHOLOGY IN DOWN SYNDROME

Ana Țarcă

Romanian Academy, Iași Department of Anthropology

Abstract. A study of the palmar dermatoglyphics pathology, on a group of 54 mongoloid children (33 boys and 21 girls) in order to know the main malformative stigmata specific to Down syndrome have been set up. A large scale of palmar dermatoglyphic anomalies have been found, most of which were on both hands of patients; they were correlated with a high degree of their somatic, physiological, neuropsychical handicap. The new elements of dermatoglyphic pathology which are added to the other four used so far in the dermatoglyphic diagnosis of Trisomy 21: (L^U in Hp; the distal displacement of the triradius t'' , the increase of the *adt* angle and the transverse palmar sulcus), are able to ensure a greater diagnosis accuracy. The identification of the persons with forms of partial Trisomy 21, in mosaic or through balanced translocation, apparently in good health, but having the risk of transmission of the disease to their offsprings in its full or complete form (accompanied by malformations) could prevent this serious disease.

Key words: dermatoglyphics, Trisomy 21, anomalies

Rezumat. Lucrarea curpinde un studiu al patologiei dermatoglifelor palmare în lumina noilor descoperiri, pe un lot de 54 copii mongoloizi din Moldova (33 băieți și 21 fete) în scopul cunoașterii principalelor stigmat malformative tipice bolii Down. A fost evidențiată o gamă foarte largă de anomalii (distorsiuni) dermatoglice palmare, mare parte dintre ele prezente pe ambele palme ale pacienților, care se corelează cu gradul înalt de degenerare somatică, fiziologică și neuropsihică a acestora. Noile elemente de patologie dermatoglică care se adaugă celor patru utilizate până în prezent în diagnoza dermatoglică a Trisomiei 21 (L^u în Hp; distalizarea triradiusului t'' , creșterea mărimii unghiului *adt* și sulcusului palmar transvers), sunt în măsură să asigure o mai mare precizie în diagnoza acestei grave maladii dar și în prevenția și profilaxia ei, cel puțin în zona Moldova. Identificarea persoanelor cu forme de Trisomie 21 parțială, în mozaic sau prin translocație echilibrată, aparent sunt sănătoase somato-psihic dar prezentând riscul transmiterii bolii în descendență în forma ei totală sau completă (însoțită de malformații), ar putea preveni această boală gravă.

Cuvinte cheie: dermatoglife, Trisomia 21, anomalii

INTRODUCTION

Among the multiple malformations related with Trisomy 21 or Down syndrome, so-called abnormalities or distortions of dermatoglyphic picture, which are actually signals of somatic, physiological or neuropsychical degeneration of patients are used on a

large scale in the world, for diagnosis of this congenital disease (1,2,5,7,10). Dermatoglyphic studies in different countries (3,5,8,9,10), on patients suffering from Down syndrome demonstrated identical distortions in their dermatoglyphic picture, whatever the ethnic origin or race, except some small differences consisting in the

DERMAL PRINTS PATHOLOGY IN DOWN SYNDROME

frequency with which they manifest themselves. This, because at the group level these abnormalities are deviations of some dermatoglyphic features from the existing values of population they belong to, as well as deviations from their classical line of distribution depending on sex, laterality or fingers. In addition to that, these percentage differences also depend on the types of Trisomy 21 of the patients included in the study (total or complete trisomy 21, partial trisomy 21, translocation trisomy 21: balanced or unbalanced and mosaic trisomy 21), that imply different degrees of somatic, physiological or neuropsychical degeneration, reaching to apparently normal phenotypes (in the case of mosaic and balanced translocation trisomy 21).

In our country, the research on dermatoglyphics in Down syndrome are very few (3,12), and the results, although important from clinical implications of some abnormalities (12), are no longer updated to the latest discoveries in the field. The aim of this paper was to study the palmar dermatoglyphics pathology on a group of children suffering from Down syndrome in the idea of distinguishing the main distortions typical of subjects living in this area not well investigated until now.

MATERIAL AND METHODS

54 children (33 boys and 21 girls) with Down syndrome registered in Mental Health Centre – Iași have been investigated. Family inquires shown 72.7% of affected girls and 66.7% of boys were last born child. Among physical stigmas and intellectual deficit

specific of mongoloid children, the most frequent are the serious eye disabilities, the congenital heart malformations and severe mental disorder, so 90% of these patients were in the first degree disability.

Sexual dymorphism and the bilateral differences have been taken into account and the results were compared with those found in Moldavian population.

The working methods are the ones frequently used in the dermal prints pathology studies (1,2,5,8).

RESULTS

The analysis of the main palmar dermatoglyphic characteristics with trisomy 21 from Moldavia revealed a large range of abnormalities with serious clinical significance, similar with those found in other ethnical groups of mongoloids (1,2,4,7,10,13).

The important modifications of total frequency of pattern from palm's compartments, mean an unexpected increase of this frequency in Hypothenar (Hp) and the III^d interdigital space (I₃) and also a sensible decrease in the IVth interdigital space (I₄) in the II^d one (I₂) and in the Thenar/I (Th/I).

Consequently, classical sequence of patterns distribution in these compartments for the first three positions in the scheme is overturned meaning: **Hp > III > IV > Th/I > II** instead of **IV > III > Hp > Th/I > II** (Table 1). Out of the 57.4% Hp patterns, the great majority (47.6% in girls and 42.4% in boys) are ulnar loops (L^u), patterns very uncommon among the Moldavian population (Table 2), and even among the Romanian and European population

(1,7,9,13). Mongoloid patients of both sexes show these patterns predominantly on the right hands.

Table 1. The total frequency of the patterns in the palmar area found in children with Trisomy 21, compared to the normal population

Area	Trisomy 21		Normal		Bilateral differences			
	Boys	Girls	Men	Women	Trisomy 21		Normal	
					Boys	Girls	Men	Women
Hp	57.6	57.1	35.2	35.1	R > L	R > L	R > L	R > L
Th/I	-	9.5	11.0	8.0	L = R	L = R	L > R	L > R
II	3.0	-	4.0	2.1	R > L	R = L	R > L	R > L
III	36.4	42.9	39.2	37.9	R > L	R > L	R > L	R > L
IV	13.6	14.3	42.7	45.7	L ≥ R	L = R	L > R	L > R

Another pathological sign was *the distal and solitary shifting of the axial triradius t in t'' and t''' positions found with a very high frequency* compared with the population they come from (Table 2), which appeared with a higher percentage on the left hands instead of the right ones, as in normal people.

The spectacular increase of cases where in the palm Hp there are 2, 3, or 4 triradii of which one at least in a distal position (t', t'', or t'''), is of equal importance as clinical implications. This distortion is determined by the presence in this compartment of L^u, either alone or in a combination with other patterns. As in other diseases (5,6,9), or in normal populations (1,12), these combinations of more triradii in the same palm will be encountered more often on the right palms in both sexes (Table 2).

The large increase of mongoloid children cases having distal and

solitary positions t'' and t''' as well as combined ones, led to *other serious distortion that consist of a marked reduction of the basal and solitary position t up to 10.2% compared to 63.5% representing the percentage for the Moldavian normal population.* In addition to that, while for the latter this position is predominantly on the left hands at both sexes, in our groups, it was found on the right ones in most of the cases (Table 2).

The very high position of the triradius t, on the palm either single or in a combination with other positions, had as a result the *increase of adt angle, over passing 57° and reaching even 95° and more.* The large and short palms of mongoloid patients could be responsible for this angle increase. Besides the high frequency of the cases with an increased *adt* angle, namely 89.4% for boys and 78.5% for girls, we found that at 84.4% of the male patients and 77.8% of the female

DERMAL PRINTS PATHOLOGY IN DOWN SYNDROME

ones this angle was on both palms, fact which is supposed to amplify its malformative effect.

At the Areal Thenar level (Th/I), there were a *lot of cases with the dermal ridges disposed in a dense and very dense net*, a feature not existent in the normal population, but which we already found, although in much smaller percentage, at the blind people from Moldavia, as other authors in the sexual chromosomes diseases (7,9,13). This palmar abnormality, in our cases was more frequent at the girls and especially on the right palms for both sexes (Table 2).

In the interdigital space III (I₃), from the 42.9% patterns present at girls and 36.4% at boys, most were loops (L) formed by the bending of C line in this compartment, a characteristic feature for this serious disease (2,4,7,9). In this compartment the mongoloid children of both sexes have also *an important distortion that implies the partial or total suppression of the C line (Cx and respectively Co)*. It exists in a quite high proportion in comparison with the population from Moldavia (Table 2), and especially on the left palms where a substantial reduction of pattern frequency have been observed. Table 2 shows that while at normal people Cx is predominant for boys and Co for girls, at patients with Trisomy 21 appeared a reversal of the classical line of the sexual dimorphism from this point of view. The fact which come in for attention was the very high percentage of cases where these two rare abnormalities (Cx and Co) were

found on both palms (50.0% at boys and 46.1% at girls) the remaining being disposed either exclusively on the left hands or only on the right ones.

In the interdigital space II (I₂) a serious distortion by its medical implications (2,4,5,7,9), is the *substantial decrease (much less normality - 21 mm for women and 24 mm for men), of the distance between the triradius a and b that mark this compartment*. We found in 86.4% of boys and 54.8% of girls, comparatively to only few cases in the normal population (Table 2), and more on the left palms at boys and on the right ones at girls. In 83.8% of the cases with this distortion for boys and 64.3% for girls, it appears on both palms at the same time which implies the doubling of its clinical implications.

The intense transversality of the dermal ridges in the distal region of the palm is another pathological indicators characteristic for the patients with Down syndrome, evaluated by main line index (M.L.I.), suggested by H. Cummins (1). This transversal alignment of the ridges in our group is suggestively illustrated by the average values which are high for M.L.I. - namely 10.9 for the affected boys and 10.4 for the girls compared to those for the Moldavian population where they reach only 8.9, suggesting an intermediary slope of ridges. Like as for normal people, the transversality of the palmar ridges of our patients was always pronounced in the right palms at both sexes (Table 2).

Table 2. The frequency of the palmar dermatoglyphic anomalies found in children with Trisomy 21 compared to the normal local population

Palmar anomalies	Trisomy 21		Normal		Bilateral differences			
	Boys	Girls	Men	Women	Trisomy 21		Normal	
(%)					Boys	Girls	Men	Women
L ^u in Hp	42.4	47.6	6.2	5.4	R=L	R>L	L>R	L>R
t ^u , t ^{uu}	25.7	38.1	2.0	3.1	L>R	L>R	R>L	R>L
tt ^u , t ^u t ^u , tt ^u t ^u , etc.	57.0	42.1	20.6	18.1	R>L	R>L	R>L	R>L
basal "t"	10.6	9.5	65.8	61.3	R>L	R>L	L>R	L>R
adt angle over passing 57°	89.4	78.5	3-5%	2-4%	L>R	R>L	L>R	L>R
Dense and very dense net in Th.I	37.9	57.1	-	-	R=L	R=L	-	-
a-b distance under 21 mm for women and 24 mm for men								
	86.4	54.8	3.9	1.8	L>R	R>L	L>R	L>R
Cx	30.3	40.5	19.7	18.7	L>R	L>R	L>R	L>R
Co	24.2	4.8	7.4	8.7	L=R	L=R	L>R	L>R
M.L.I. (mean)	10.9	10.4	8.9	8.9	R>L	R>L	R>L	R>L
Transversal palmar sulcus	46.9	23.8	8.1	5.4	R>L	R>L	L>R	L>R

L = left

R = right

Finally, *a last serious abnormality* illustrated by us for the mongoloid children palm *is the presence of the palmar transversal sulcus in very high percentages considering the normal people (table 2) and with a preferential distribution on the right palms and not on the left ones as it appears to normal people or in the case of other serious diseases* (9,11,13). As far as the sulcus is concerned it was found on both palms at the same time in 55% of the male series and in only 25% of the female one, but for the latter we noticed its exclusive presence on the right hands in most of the cases (62.5%). This preferential disposition on the right hands of the palmar sulcus we have found at children with congenital and hereditary deafness.

CONCLUSIONS

1. The study of the palmar dermatoglyphics in cases of Trisomy 21 in Moldavia revealed a large scale of anomalies with clinical implications. 15-20 years ago the identification of the Down syndrome from a dermatoglyphic perspective was based on only four pathological indicators (L^u from the Hp of the palm, the distal displacement of the t triradius, the maximal *adt* angle and the presence of the transversal palmar sulcus on at least one of the hands).
2. Our research brings some new elements of pathology, able of assure more accurate diagnosis of this serious disease.
3. Furthermore, the new dermatoglyphic nomogram could be used for a screening of the disease at the population level, by the identification

DERMAL PRINTS PATHOLOGY IN DOWN SYNDROME

of the persons suffering from the partial or mosaic Trisomy 21. Although phenotypically normal, they bear the malformative stigmata which are specific to the disease and face the risk of giving birth to children with complete or total Trisomy 21 (accompanied by well-known multiple malformations).

REFERENCES

1. Cummins H., Midlo Ch. - *Finger Prints Palms and Soles*, Dower Publications, INC, New York, 1961, 279–281.
2. Digamber S., Borgaonkar Ph. D. - *Dermatoglyphic Studies and Their Usefulness in Clinical Diagnosis by the Method Predictive Discrimination*, Birth Defects, Original Article Series, 1979, V, 6, 621–625.
3. Geormăneanu C., Geormăneanu M. - *Introducere în Genetica pediatrică*, Ed. Medicală, București, 1986, vol. I, 95–116.
4. Holt B., Rignell A. - *High Variability in Mongoloid Dermatoglyphics*, Clin. Genet., 1976, 9, 606–608.
5. Holt Sarah B. - *The Genetics of Dermal Ridges*, Ch. C. Thomas – Publisher, Springfield–Illinois–USA, 1968, 112–145.
6. Penrose L.S. - *The Distal Triradius “t” on Hands of Parents of Mongol Imbeciles*, Ann. Human. Genet., 1954, 19, 10–38.
7. Reed T. E., Borgaonkar D. S., Christian J. C. - *Dermatoglyphic Nomogram for the Diagnosis of Down’s Syndrome*, J. Pediatr., 1970, 77, 1024–1032.
8. Rignell A. - *Significant Variations of Fingertype Dermatoglyphics in Trisomy 21 or Down’s Syndrome*, Ann. J. Physical Anthrop., 1987, 72, 277–286.
9. Schauman Blanka, Milton A. - *Dermatoglyphics in Medical Disorders*, Springer–Verlag, New York – Heidelberg –Berlin, 1976, 146–162.
10. Rajangam S., Janakiram S. - *Dermatoglyphics in Down’s Syndrome*, J. Indian – Med– Assoc., 1995, 93 (1), 10–30.
11. Schauman Blanka, Kimura S. - *Palmar, Plantar and Digital Flexion Crease: Morphologic and Clinical Considerations*, Birth Defects, 1991, 27 (2), 229–252.
12. Țurai C., Leonida C. I. - *Amprente papilare*, Ed. Medicală, București, 1979, 231–236.
13. Walker Norma F. - *The Use of Dermal Configurations in the Diagnosis of Mongolism*, Pediatr. Clin. North. Ann., 1958, 531–543.