

## PARENTAL DERMATOGLYPHIC ASPECTS IN TRISOMY 21

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**Abstract.** The paper deals with an analysis of digito-palmar dermatoglyphics with the parents of 54 children with Trisomy 21 (Down Syndrome) studied before [15]. The results obtained confirm the data found in the foreign specialised literature according to which many of the dermatoglyphic malformations present at children with Trisomy 21 can also be found to their apparently healthy parents but in a smaller number, because here they signal only the presence of the Trisomy 21 (partial Trisomy 21, Trisomy 21 by balanced translocation or mosaic Trisomy 21) while in the case of the affected children these abnormalities are also the graphic expression of the multiple malformations that accompany their total or unbalanced translocation Trisomy 21. Consequently, the author considers that the dermatoglyphic distortions illustrated in both cases (parents and their children), could be used as pathological indicators in the identifying of the carrier persons of the inapparent phenotypically Trisomy 21 but having the risk of giving birth to children with complete Trisomy 21 and specific multiple malformations.

**Key words:** Dermatoglyphics, Trisomy 21 (Down Syndrome), distortions (anomalies).

**Rezumat.** Lucrarea cuprinde o analiză a dermatoglifelor digito-palmare la părinții a 54 copii cu Trisomie 21 (Sindrom Down) din Moldova studiați de autor într-o lucrare anterioară [15]. Rezultatele obținute susțin datele din literatura străină de specialitate după care multe dintre stigmatele dermatoglifice malformative prezente la copiii cu Trisomie 21 se regăsesc și la părinții acestora aparent sănătoși cu o pondere însă mai redusă, dat fiindcă la aceștia, ele semnalizează doar simpla prezență a uneia din formele de Trisomie 21 inaparentă fenotipic (Trisomie parțială, Trisomie prin translocare balansată ori Trisomie în mozaic), în timp ce la copiii afectați aceste anomalii sunt și expresia grafică a multiplelor malformații care însoțesc Trisomia 21 totală sau prin translocare neechilibrată. Din aceste motive considerăm că distorsiunile dermatoglifice evidențiate de noi în ambele cazuri (părinți și copii) ar putea fi utilizate ca indicatori patologici în identificarea persoanelor purtătoare de Trisomie 21 inaparentă fenotipic dar care prezintă riscul de a da naștere la copii cu Trisomie 21 completă și cu multe malformații specifice acestei grave maladii.

**Cuvinte cheie:** Dermatoglifice, Trisomie 21 (Sindromul Down), distorsiuni (anomalii).

### INTRODUCTION

The mongolism or Down disease is the most frequently and well known malformative syndrome determined by a chromosomal abnormality which is characterized by a specific clinical aspects, mentally retard ranging to imbecility and Trisomy 21 (an extra G

group chromosome) [5]. In Romania, before 1989 the global frequency of the disease had reached an average rate of 1:650, while at the category of under 30 - year - old mothers the rate was 1:1500. It is considered [2] that the risk increases with the mother's age, reaching 1:300 at 35, 1:100 at 40

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and 1:50 at over 45 years old. But in spite of the mother's age 92-95% of the mongolians have complete or total Trisomy 21, while the remaining ones present only partial Trisomy 21, balanced or unbalanced translocation Trisomy 21 and mosaic Trisomy 21 (when triplication has not covered all cell lines and the patients can range from apparently normal people to classical mongolians depending on the number of affected cells). It has been proved although [6,9,10,16] that the parents of plurimalformed downian children, even if they are apparently normal phenotypically, they show important signs or malformations of the disease in their dermatoglyphic picture and present the risk of giving birth to children suffering from complete Trisomy 21, accompanied by multiple malformations specific to this disease. Since in our country the researches in this field are very few, the present paper completes the previous study on finger and palmar prints children with Down Syndrome from Moldavia (eastern Romania).

### MATERIAL AND METHODS

The parents of 54 children with Trisomy 21 (Down Syndrome) from Moldavia [15], have been investigated from a dermatoglyphic point of view, in a collaborative work with the Mental Health Laboratory (M.H.L) from Iassy. For all the dermatoglyphic pathology indicators noticed at the parents, the analysis was focused on their frequency compared to that of their affected children and both were related to normality, taking also into

account the sexual dimorphism and the bilateral differences.

The working methods are those used in pathological dermatoglyphic studies [5,6,9,10,16,17].

### RESULTS

It is worth underlining from the very beginning that, at the group level, the dermatoglyphic distortions with deep clinical significance found to both parents and their mongolian children are nothing else but deviations of the frequency of some digital and palmar dermatoglyphic features from their normal values, of the general population they come from [Personal thesis, 1995].

Thus, at the level of digital picture a first and serious abnormality with clinical implications found exclusively to the fathers not to the children (Table 1) supposes an excessive percentage for arches (A) up to 20.02% compared to only 5.05% of the normal population from Moldavia. Another serious distortion existing this time both in parents and in their children, although in a reduced percentage for the first ones, is **the growing incidence of the left and right hand monomorphism and of the individual one**. Each of the three types of monomorphism both for mothers (Table 1) and their girls are found with high frequencies comparatively to the normal population, taking into account that for the left monomorphism, the normal population has only 16.0%, for the right one 12.5% and for the individual one 6.1%.

**Table 1. Dermatoglyphic anomalies of parents and their children with Down Syndrome (%)**

Dermatoglyphic anomalies		Parents		Children with Down Syndrome	
		Fathers	Mothers	Boys	Girls
Arches on fingers	A	20.0	7.3	2.7	-
Monomorphism	Left	25.0	32.4	43.4	52.4
	Right	18.7	27.0	42.4	61.9
	Individual	6.2	13.5	21.2	42.8
Ulnar loop ( $L^U$ ) in palmar Hp		12.5	10.8	42.4	47.6
Dense and very dense network in Th/I		9.4	18.9	37.9	57.1
The distance a-b very much reduced compared to the case of normal people		34.4	17.5	81.8	54.8
The supression of C line	Cx	34.3	31.1	30.3	40.5
	Co	3.1	10.8	24.2	4.8
tt', t't'', tt't'', t't''t''' etc.		21.9	29.7	56.0	42.9
Average M.L.I.		9.6	11.6	10.9	10.4
Palmar transversal sulcus		6.2	9.5	46.9	23.8

A wider range of dermatoglyphic distortions is offered by **the palmar picture**, for some of them, as we see, the parents covering even greater frequencies than their ill children. Thus, **the ulnar loop ( $L^U$ ) from the palmar Hypothenar (Hp)**, that in normal people does not exceed 5%, reach a percentage 2.5 times higher for parents and 9 times higher for offspring. In the first case it is more frequent in fathers and on the left palms and in the second case at girls and on their right palms.

**In the Thenar/I (Th/I) compartment we found a very high incidence for the disposition of the palmar ridges under the form of a dense and very dense network**, a particularity that has already been highlighted in the case of the blind children and also in the case of those suffering from other severe

ocular diseases [13], and which can be only exceptionally seen at the normal people. These severe anomaly is more often at mothers found as well as girls and on the right hands of the subjects of both sexes rather than on the left ones (Table 1).

**The reduction of the distance between the a and b triradii (which delimitate the 2<sup>nd</sup> interdigital space) much behind the admitted average threshold of 21 mm for the normal female people and of 24 mm for the male**, is one of the palmar pathological indicators which has already been signaled in the case of the Moldavian subjects with congenital heart malformations [14], but which has also been reported in many of the anomalies of the sexual chromosomes [6,10,17], though in a smaller proportion. This severe

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distortion reaches an unusually high frequency, especially with fathers and boys (Table 1). Nevertheless, while in the case of the parents it is predominant on the left palms at their affected children of both sexes it is more frequent on the right ones.

**The partial or total suppression of the course of the C line (Cx and respectively Co)** ranges between 10 and 12% for Cx and only between 2% and 3% for Co, in the case of the normal Moldavian population. In Table 1 one can see that for Cx fathers exceed their sons, and for Co mothers exceed their daughters, although on the whole, the children with Trisomy 21 are the ones with higher percentages when compared with their parents. Also, while parents maintain the classical line of sexual dimorphism which implies a greater incidence for Cx, in the case of fathers and for Co in the case of mothers, in their offsprings Cx is more often present at girls and Co at boys. Cx and Co prevail on the left hands both for the parents and children with Trisomy 21, but in terms of disposition in the carriers, they appear in the first case more frequently on the left hands (44.4% for fathers and 41.6% for mothers) as it is the case of normal people as well, and to the latter on both palms of the carriers (50% for boys and 46.1% for girls).

**The presence in the same palm of 2, 3 or 4 triradii among which one in distal position**, is a malformative mark that reaches an average of 22.2% in Moldova population, and on the group of downian children 56% in boys and 42.9% in girls. Between the parents only mothers reach frequencies

exceeding the normal people by around 10%. In both groups (parents and children) this dermatoglyphic distortion, that had been reported in many other diseases, though in more reduced percentages [4,7,8,10,16,17), appears more frequently on the right hands of the subjects of both sexes as well as in the case of the normal population.

**A pathological indicator that is presents both parents and children are the tendency for an emphasized transversal line of the dermal ridges, in the superior region of the palm.** This was estimated by us through a M.L.I (Main Line Index), imagined by H. Cummins [2], and suggestively expressed in the both cases through average high values (Table 1) compared to those found in the Moldavian population of only 8.00. Most obviously this transversality of palmar ridges appeared on the right hands of the subjects in both cases.

**A last abnormality with serious clinical implications refers to the palmar transversal sulcus or Simian Line** that appear in a proportion of 9.5% in mothers and 6.2% in fathers compared to the 3.3% in the normal population of Moldova. In the case of ill children, palmar transversal sulcus reaches a very high percentages (23.8% at girls and 46.9% at boys) and have a preferential disposition on the right hands and not on the left ones as it appears to parents or to normal peoples.

The tendency for higher frequencies of the sulcus in women, as it appears in parents has been found in the people with congenital heart malformations [14] as well as for parents of the other

plurimalformed children from Moldavia [12]. The preferential disposition on the right hands of the ill children of both sexes has been reported also for children with congenital and hereditary deafness living in the same area [11].

#### CONCLUSIONS

- Apparently healthy parents but carrying as their offsprings one of the forms of Trisomy 21 with no phenotypical manifestation, have the same distortions. The occurrence of these distortions with severe medical implications was more limited in parents where they signal the simple presence of atypical Trisomy 21.
- The mothers had a small raised frequencies dermatoglyphic anomalies than the fathers supporting the hypothesis that mothers are more responsible in the spreading of this disease.
- For fathers, stigmata have found that were absent from their children (the excessive occurrence of arches on the fingers).
- Dermatoglyphic distortions were present on both hands of the carriers (in double dose) of downian children compared to their parents.
- Dermatoglyphic anomalies according to sex and laterality of the parents maintained the classical line of sexual dymorphism and of bilateral differences as it appears to normal people while in the case of ill children this line was different.

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