

DERMATOGLYPHIC STUDIES IN RETT SYNDROME

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ABSTRACT

Palmar and digital dermatoglyphics from 12 patients with Rett syndrome and from 144 healthy women were compared. Significant differences were found for the mean atd angle (higher in Rett syndrome patients) and for the frequencies of arches and whorls in the 4th digit, the first higher and the latter lower in Rett syndrome patients.

INTRODUCTION

Rett syndrome is an encephalopathy expressed by mental deficiency, stereotyped movements and autistic behaviour. Considering that this syndrome is restricted to the female sex and that no exogenous agent has been so far identified, some researchers, including us, are investigating a possible genetic origin for Rett syndrome. The dermatoglyphic studies here presented constitute one of the points we have studied in patients affected by this syndrome.

MATERIALS AND METHODS

Palmar and digital dermatoglyphics were collected from 12 patients with the Rett syndrome; they were compared to the ones observed in a sample of 144 healthy women (Dal Colletto, unpublished results).

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In all patients the diagnosis of Rett syndrome was made following a standard rigorous protocol, so that the chances of clinical error were negligible. The normal women sample was assembled among staff and students from the University of São Paulo.

Dermatoglyphic analysis was performed on standard prints obtained with ink. For the description of most dermatoglyphical features, we followed carefully the procedures indicated by Penrose (1968).

Statistical analysis made use of different tests, which are mentioned in the respective results.

RESULTS

Tables I and II present the result of the Rett syndrome patient's dermatoglyphic analysis, compared with the normal sample. We have found significant differences between patients and normal controls only for the mean atd angle ($t = 2.21$; 154 d.f.; $P < 0.05$), higher in Rett syndrome patients than in normal controls. Table III presents the frequencies of digital patterns of Rett syndrome patients compared to normal women. Chi-squared tests on 3×2 contingency tables gave significant figures only for the fourth digits on both hands. Fisher's exact tests performed on 2×2 partitions of the original 3×2 table showed significant differences between the frequencies of arches in the 4th digit of the right hand – higher in Rett syndrome patients ($P = 0.03$) – and of whorls in the 4th digit of the left hand – lower in Rett syndrome patients ($P = 0.03$).

DISCUSSION

Rett syndrome patients seem to have slight dermatoglyphic abnormalities that could suggest, as proposed by Opitz (1987), a first trimester of pregnancy onset of the pathogenesis.

However, since ten different chi-squared tests were performed on 3×2 contingency tables using the data from Table III, it is possible that some of these differences were due to an increase in type II error, so we should treat them cautiously.

Table I - Results of atd angle, TRC, A'-d and a-b counts (mean and standard deviation) of the Rett syndrome patients, compared with the results of a normal sample (t test).

			atd	TRC	A'-d	a-b
Rett syndrome		\bar{x}	96.58	119.75	106.75	81.58
Patients	R + L					
N = 12		sd	8.38	44.82	18.16	9.68
Normal Women		\bar{x}	88.18	133.84	95.51	83.23
Sample	R + L					
N = 144		sd	12.93	46.80	20.88	10.01
t test			2.21	1.01	1.81	0.55
(d.f. = 154)						

Table II - Frequency (%) of palmar patterns in Rett syndrome patients compared with the results of a normal sample (Fisher's exact test):

		H	T	I1	I2	I3	I4
Rett	R	41.7	16.7	8.4	0.0	25.0	41.7
syndrome							
Patients	L	25.0	16.7	0.0	0.0	16.7	58.4
N = 12							
Normal	R	44.4	7.6	3.5	4.2	36.8	50.0
Women							
Sample	L	33.3	12.5	9.7	0.7	23.6	54.2
N = 144							
Fisher's	R	P = 1.0000	P = 0.2625	P = 0.3864	P = 1.0000	P = 0.5392	P = 0.7655
test	L	P = 0.7521	P = 0.6535	P = 0.6029	P = 1.0000	P = 0.7343	P = 1.0000

H = hypothenar area; T = thenar area; I1, I2, I3 and I4 = first, second, third and fourth interdigital areas.

Table III - Frequency (%) of digital patterns in Rett syndrome patients, compared with the results of a normal sample.

		Digits					
		I	II	III	IV	V	
Rett syndrome Patients (N = 12)	R	L ^u	66.7	75.0	50.0	41.7	91.7
		L ^r	0.0	0.0	8.4	0.0	0.0
		W	33.4	16.7	25.0	41.7	8.4
		A	0.0	8.4	16.7	16.7	0.0
	L	L ^u	50.0	75.0	58.4	83.4	83.4
		L ^r	0.0	8.4	0.0	0.0	0.0
		W	50.0	8.4	25.0	8.4	8.4
		A	0.0	8.4	16.7	8.4	8.4
Normal Women Sample (N = 144)	R	L ^u	55.6	43.1	81.3	55.6	87.6
		L ^r	0.0	11.8	0.7	0.0	0.0
		W	41.7	38.2	12.5	43.1	11.1
		A	2.8	6.9	5.6	1.4	1.4
	L	L ^u	53.5	38.9	77.1	53.5	81.3
		L ^r	0.7	13.2	2.1	0.7	0.0
		W	42.4	35.4	13.2	43.1	17.4
		A	3.5	12.5	7.6	2.8	1.4

R = right hand; L = left hand; Lu = ulnar loops; L^r = radial loops; W = whorls; A = arches.

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RESUMO

Foram analisadas as impressões dermatoglíficas digitais e palmares de 12 pacientes com síndrome de Rett. Os resultados obtidos foram comparados com os de uma amostra de 144 mulheres normais.

Diferenças significativas foram encontradas nas medidas do ângulo atd médio (mais alto nas pacientes) e nas frequências de arcos e verticilos no 4^o dedo: as primeiras mais altas e as segundas mais baixas nas pacientes.

REFERENCES

- Opitz, J.M. (1987). Rett syndrome – A Review and Discussion of Syndrome Delineation and Syndrome Definition. *Brain & Develop.* 9: 445-450.
- Penrose, L.S. (1968). Memorandum on dermatoglyphic nomenclature. *Birth. Def. Orig. Art. Ser.* 4.

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