



Thesis Abstract

**Psychogenetics of Ullrich-Turner syndrome
(an investigation of 28 subjects and respective
controls through the Bender test and Piagetian
scales)
(Psicogenética da síndrome de Ullrich-Turner
(investigação de 28 sujeitos com respectivos
controles através do teste de Bender e das
escalas de Piaget))**

Fátima do Carmo Fonseca Ricardi

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Orienting Professor: Pedro Henrique Saldanha

Piagetian scales (PS) and Bender visual motor gestalt test (BT), as well as laterality and Ishihara tests, were applied to 28 subjects with universal 45,X Ullrich-Turner syndrome (UTS), and their respective controls, in order to investigate their cognitive performance. Dermatoglyphics were also analyzed to obtain “clues” of embryological changes that may have appeared during nervous system development and that could be associated with the cognitive performance of the UTS individuals. Statistical analysis was performed by parametric (descriptive statistics, *t*-test, *t*-test on paired data, and stepwise multiple regression analysis) and non-parametric tests (Fisher’s exact test, Spearman and Mann-Whitney). Dermatoglyphic pattern distribution was similar to that reported in previous studies of UTS individuals: ulnar loops (L^u) in the digital patterns and finger ridge, a-b, and A’-d counts were more frequent, while arch and whorl patterns were less frequent compared to controls. However, we did not find higher frequencies of hypothenar pattern, maximum atd angle, and ulnarity index in our UTS subjects, unlike other investigations. Furthermore, we found significant differences between UTS and control TI values.

BT scores were also lower in the probands (as has been previously reported), revealing a neurocognitive deficit of visual motor perception in UTS individuals, which could be due to an absence of or deficiency in cerebral hemispheric lateralization. However, UTS subjects seemed to improve their performance on BT with age. Cognitive performance of the UTS subjects was not significantly different from that of controls, confirming previous study in which UTS perfor-

mance was found to be similar to that of the normal Brazilian population. Nevertheless, there was a significant correlation between the BT scores and the PS values, suggesting that the development of the visual motor perception and the construction of the reflective abstraction and the representation space are somehow linked. There were significant correlations between the BT scores and the PS levels with dermatoglyphic parameters. This association could be explained by changes in the common ectodermal origin of the epidermis and the central nervous system. UTS subjects seem to succeed in compensating their spatial impairments to adapt their cognitive and social contacts. Consequently, genetic counseling should consider cognitive and psychosocial difficulties presented by UTS subjects, providing appropriate treatment and orientation for them and their families.

Key words: Turner syndrome, Bender test, Piagetian scales, Laterality test, Dermatoglyphics, Cognition

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