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Thesis Abstracts

Use of molecular markers in the identification of species of the genus *Cassia* (Leguminosae)

(Uso de marcadores moleculares na identificação de espécies do gênero *Cassia* (Leguminosae))

*Goran Kuhar Jezovsek**

Cassia (Leguminosae) species were studied using molecular markers. Two identification methodologies were used to study two known species, *C. obtusifolia* and *C. occidentalis*, and a population similar to *C. obtusifolia*, though with some morphological differences. Nine different systems were analyzed by using isoenzymatic protein markers. Eleven primers were used to generate RAPDs using polymerase chain reaction (PCR). A high degree of polymorphism was found among the three samples. Analysis of genetic similarity with the Jaccard's coefficient showed that there is a possibility that the third sample is actually a third species and not a subpopulation of *C. obtusifolia*. The congruent results indicate that the combined use of the two techniques may be a good tool for the identification of species.

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*1995. Departamento de Tecnologia, Faculdade de Ciências Agrárias e Veterinárias de Jaboticabal, UNESP, Jaboticabal, SP, Brasil. Master's thesis. Orienting Professor: Dr. Eliana G. de Machado Lemos.

Genetic and neurologic aspects of autism; proposal of an interdisciplinary approach in the diagnostic evaluation of autism and related disorders

(Aspectos genéticos e neurológicos do autismo; proposta de abordagem interdisciplinar na avaliação diagnóstica do autismo e distúrbios correlatos)

*Carlos Eduardo Steiner**

Autism is a neurodevelopmental disorder, clinically characterized by the presence of abnormalities in three main areas: social interaction, language and communication, and interest and activities. The onset is before the age of three years. Additional findings include mental retardation, epilepsy, hyperactivity, hearing loss, minor dysmorphic features and other neurological signs.

No single cause for autism has been identified. Genetic and nongenetic factors have been described, including the association with genetic disorders, especially the fragile X syndrome and tuberous sclerosis. Familial recurrence and a high rate of concordance in monozygotic twins are evidence of a genetic basis. Most probably autism occurs as a multifactorial disorder.

Several laboratory findings have been described in association with autism, including biochemical, chromosomal, neuroanatomical and neurophysiological abnormalities, but none of them helps to confirm this disorder. Thus, in the absence of a biological marker, its diagnosis continues to be exclusively clinical. Furthermore, there is no standardization of the exams that should be requested to individuals with the initial diagnosis of autism.

On the basis of a literature review concerning clinical, laboratory and neuroimaging in autism, a protocol for the evaluation of individuals with this hypothesis is proposed. It comprises diagnosis based on well-established neuropsychological criteria, clinical genetic and neurological evaluation, screening for inborn errors of metabolism, chromosomal analysis, molecular study of the FRAXA, magnetic resonance and SPECT imaging.

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Apparently balanced chromosomal translocation associated to mental retardation and/or congenital malformations. Clinical-cytogenetic considerations and proposed related mechanisms to abnormal phenotype in six cases

(Translocação cromossômica aparentemente balanceada associada ao retardo mental e/ou malformações congênitas. Considerações clínico-citogenéticas e mecanismos propostos para o fenótipo anormal em seis casos)

*Elenice Ferreira Bastos**

Structural chromosomal abnormalities have been used as an important tool in research towards genetic diseases, specially in the construction of the Morbid Anatomy Map of the Human Genome (OMIM, 1998). In this context, balanced chromosomal translocations associated to abnormal phenotypes may be relevant to identification of specific chromosomal sites associated to a particular disease. This investigation comprises the study of six cases of apparently balanced chromosomal translocations associated to abnormal phenotypes through several cytogenetic techniques, including molecular cytogenetic (FISH). Referral for cytogenetic studies were: congenital malformations (cases I, II and V); mental retardation (cases

III and IV) and primary amenorrhea (case VI). The six karyotypes were characterized according to the ISCN (1995) as:

- Case I = 46,XX, t(3;8)(p23;q22.1);
- Case II = 46,XX, t(4;16)(q21;q22);
- Case III = 46,XY, t(2;15)(q23;q22);
- Case IV = 46,XY, t(2;17)(q11.2;q25.3);
- Case V = 46,XX, t(11;13)(p13;p33);
- Case VI = 46,XX, t(6;18)(p23;q22).

A phenotype-karyotype correlation was attempted based on the smallest overlapping region and delineated in cases I, IV and V as BPES syndrome and region 3p23, 2p deletion syndrome and region 2p11.2 and isolated aniridia and region 11p13, respectively. In cases II (Binder-like syndrome), III (unespecific mental retardation) and VI (primary amenorrhea) both chromosomal breakpoints involved in the translocation could be candidate regions. The parental origin of the anomalous chromosomes in two out of four informative *de novo* cases was paternal. Concerning the etiological mechanisms attributed to the abnormal phenotype, haploinsufficiency was considered in cases I, II and V, genomic imprinting, position effect, and microdeletion syndromes were considered in the remanescent cases.

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