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PHENOTYPIC AND GENOTYPIC ANALYSIS IN A LARGE TURKISH BIPOLAR FAMILY

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Objective: To document the phenotypic and genotypic characteristics of a three generational large family loaded with affective disorders.
Method: Linkage analysis is done in a large three generational Turkish family loaded with affective disorders. 8 patients and 17 relatives are examined with SCID-I and the clinical characteristics of the patients are documented.. Blood is drawn from all family members to isolate DNA but only the patients DNA are included in the molecular analysis and these patients are screened for five susceptibility regions implicated for both schizophrenia and bipolar disorder (10p12, 13q32, 18p11.2, 22q11 ve 20 p12-q12).

Results: Clinical assesment of the patients revealed that, the depressive episodes of the patients are likely to be melancoholic and suicidal in nature and the manic episodes are usually associated with mood-congruent psychotic symptoms. No linkage could be detected for the screened regions in molecular analysis. Only a loss of heterozygosity could be detected in 18p11.2 region.

Conclusion: The allelic loss from the maternal allele in this region did not seem to segregate with the disorder and this maternal allele loss could not be explained by an imprinting mechanism, since there was no parental transmission of the disorder in this pedigree. This region must be screened with multiple markers and sitogenetic analysis should be done in order to be able to understand whether there is a real loss of in this part of the chromosome.

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