Brachmann - de Lange syndrome:
Behavioural problems and quality of life of patient and family
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Brachmann - de Lange (B-dL) is a rare genetic syndrome with variant expression. Its typical clinical presentation includes growth retardation, multiple congenital anomalies, mental retardation, and behavioral disturbances. Patients present with a limited capacity for social contact and communication, stereotypical behavior, adjustment difficulties and outbursts of anger with aggressive and self-destructive behavior. The severity and the manifestation type of these features present themselves within a wide clinical spectrum, so that the diagnosis of the syndrome is often delayed. In the present study the clinical course of a patient with B-dL is described. She was admitted to our clinic because of severe behavioral disturbances, within the context of deep mental retardation. The diagnosis of B-dL syndrome was made during her hospitalization, based on the characteristic cognitive phenotype she presented and on her cognition and behavioral profile. The pharmaceutical response (a combination of small doses of alopéridol and gabapentin) and the structure of a steady schedule helped in the amelioration of her family's burn out feelings. The clinical improvement of the patient and the psychoeducation of the family facilitated the interaction between the patient and her caretakers and lead to the adoption of more effective strategies for the achievement of psychosocial support. Therefore, a steady improvement of the quality of life of the patient and of her family was noted, an improvement that is maintained until today, two years after her hospitalization.

Key words: Brachmann - de Lange syndrome, behavioural disturbances, quality of life, treatment.

ΒΙΒΛΙΟΓΡΑΦΙΑ