

Chapter 14

Case Study:

Communication Abilities of a Child With Leukoencephalopathy (LBSL)

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ABSTRACT

The present study aims to implement an intervention program on a 2.5-year-old boy with a rare metabolic syndrome, Leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation (LBSL). Although the recorded cases of the syndrome are unknown, there are approximately 100 patients worldwide that undergo treatment. This specific syndrome refers to a genetic biological disorder in expression. An attempt was made to investigate the outcome of an intervention program involving projects based on play therapy for the development of deficit communication skills. The ESCS was used as an approved psychometric scale of communication skills. The results showed that there was a quantitative and qualitative increase in the early communication skills of the child, and also the emergence of a more complex type of play.

INTRODUCTION

LBSL syndrome is a genetic metabolic syndrome that falls into the category of Leukodystrophy, in the spectrum of Leukoencephalopathies. Leukodystrophies are rare, metabolic, evolving and mainly inherited diseases, which can be inherited either by being autosomal, in a residual or predominant way or in a sex-linked way. However, leukodystrophies may also be due to random mutations. They were so named from the “white” substance-myelin, which insulates protecting structures of the nervous system, ensuring the high speed of the propagation of nerve signals. Leukodystrophies disrupt the growth or maintenance of myelin, which is due to a multitude of mutations in various genes. Depending on the gene and its’ mutation, there are many categories of leukodystrophies.

LBSL is caused by an abnormal variant in the DARS2 gene (Van Berge et al., 2014; Shimojima et al., 2017; Rathore et al., 2017). The people who are affected are affected in the white matter of the brain (leukoencephalopathy). Without the existence of myelin, signals between nerve cells cannot be properly transmitted, resulting in the development of basic neurological deficits and also having a negative effect on overall nerve development (Stevens & Iowe, 1993). Due to the fact that there have been only 100 reported or recognized cases of the syndrome worldwide there is a lack of the clinical trials that could be made. Having in mind that many combinations of mutation types can cause this syndrome make it difficult to develop a complete picture of symptoms and prognosis (A cure for Ellie, 2023).

LBSL syndrome can cause symptoms that develop either before birth with serious complications throughout infancy or early in childhood which disappear within the first two years of life. However, some people may not show obvious symptoms until school age or adulthood and these symptoms may remain mild for many years. The onset, progression, and severity of symptoms vary. The most common symptoms are spasticity or stiffness of the muscles and cerebellar ataxia, which is the difficulty of coordinating walking and performing fine motor skills (Uziel et al., 2011).

There is no cure for LBSL, nor has any standard treatment protocols or guidelines for affected individuals and treatment efforts focus on addressing each individual’s specific symptoms been found. Individuals who are affected may benefit from physical therapy and rehabilitation, as well as the use of assistive devices (e.g. orthotics and wheelchairs), speech therapy, and additional medical, social and in some cases occupational services may be needed, including special education (mdahellas.gr).

Regarding the correlation between playing and the neurodevelopmental level of the child, there has been evidence that there is a positive effect on the child’s nervous development when playing (Cozolino, 2010; Levy, 2011; Stewart et al., 2016). More specifically, during the game there is an increase in the levels of the hormone oxytocin (Stewart et al., 2016), as well as activation of mirror neurons (Levy, 2011). These neurodevelopmental challenges have behavioral manifestations in the individual, such as: stimulating the feeling of trust, reducing fear, benefits of the recognition of the feelings for the person involved in the play (Stewart et al., 2016), understanding and response of the therapist to the child’s feelings during play time all with the help of the biological process of mirror neurons (Levy, 2011).

The psychoanalytic view of the action of playtime is a theoretical pillar used for the present research. More specifically, the classical psychoanalyst Winnicott (1971) defines a developmental path of psychographic stages for the child’s play. Based on the child’s interaction with the play object and the presence of the nurturer to provide the child the opportunity for reality. Subsequently, the object exists in the next stage between acceptance and denial as “objectively perceived”. And the nurturer acts by participating in the return of what is rejected (Winnicott, 1971). In the present study, the participant is at this stage, in terms of play, and based on this, his attitude to the play was determined by the educator-researcher.

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