

SPEECH IMPAIRMENT IN GLUT1DS – De Giorgis Valentina

GLUT1 Deficiency Syndrome (GLUT1DS) most frequent symptoms are epilepsy, movement disorders and intellectual disability.

As a rare disease, few studies in the literature deeply describe the neuropsychological characteristics of patients, in particular language and communication disorders that though have a major impact on the social and family sphere.

An assessment protocol for speech and language impairments in GLUT1 patients will be presented along with the result of a perspective study that highlighted the most impaired abilities found.

We characterized the dysarthria of GLUT1DS patients in an ataxic form. This finding appears to be stable or slightly improving over time, although patients perceived them more disabling, probably due to the high environmental/social/work demands.

In our study population, dysarthria is more disabling in patients with a more pronounced movement disorder and who started ketogenic diet later.

Finally, some diagnostic and rehabilitation suggestions aimed at the speech of patients with GLUT1DS will be presented.