

Bulbar dysfunction (BD) presents as dysphagia and dysarthria, which may lead to weight loss, impaired communication, orofacial pain and aspiration pneumonia. Bulbar dysfunction in MND is a negative prognostic indicator of survival, requires careful monitoring and necessitates well-timed multidisciplinary interventions; referral to speech-language pathology (SLP), neuropsychology, feeding tube placement, augmentative communication devices etc.

Pseudobulbar affect (PBA) manifests in disproportionate emotional responses to minimal or non-contextual stimulus. BD and PBA develop in a range of neurological conditions but are pathognomonic of motor neuron diseases (MNDs) with considerable diagnostic, management and quality of life (QoL) implications.

As reported by patients and their families, BD and PBA constitute one of the most burdensome clinical symptoms, with considerable QoL and social ramifications. Feedback from patient advocacy groups, our shared clinical experience and research highlighted the high incidence of misdiagnoses, delay in interventions and limited awareness for both BD and PBA. There is a striking lack of consensus in definitions, validated screening tools, diagnostic criteria, and research into the social and QoL implications. Differences in terminology, screening practices, access to multidisciplinary interventions, cultural factors, sex, and differences in health care systems in different geographical regions obstacle a development of uniform operational strategies.

The aim of the study is to evaluate biological underpinnings and QoL impact of BD and PBA in moderate to severe stage of ALS in patients and their caregivers.

Methods: The presence and severity of speech and swallowing dysfunction will be evaluated using a validated algorithm based on acoustic and mechanic parameters. A purpose-designed neuroimaging protocol will be implemented to evaluate the central and peripheral motor neuron components of bulbar function. Using patient reported outcome measures (PROMS), patient and caregiver perspectives on the impact of BD & PBA will be monitored. A comprehensive battery of neuropsychological tests, mood, affect, QoL questionnaires will be administered to map the incidence, clinical associations, longitudinal evolution and social sequelae of bulbar and pseudobulbar impairment. The interactions between PBA and frontotemporal dementia will be studied in detail.

Planned results: A transnational comparison approach will allow document cultural, ethical, and healthcare system differences in management of BD and PBA. It will also improve understanding the aetiology and raise public awareness of the clinical, social and QoL impact of BD & PBA. The applicants are not only passionate about clinical MND care, but they also have complementary research expertise in neuropsychology, speech pathology, social science and neuroimaging. They believe that achieving the objectives of this project is directly relevant to precision individualized patient care and meaningful caregiver support.