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the Frontal Systems Behavior Scale- FrSBE) and psychological measures (e.g. mood, purpose in life, illness perceptions, spirituality) were obtained. We followed patients up at 3-monthly intervals to monitor whether they had accepted / refused NIV or gastrostomy. After intervention decisions, further interviews repeated baseline measures and assessed further intervention-specific beliefs.

**Results:** We observed higher premorbid IQ and educational level and an active approach towards illness in those making a decision to refuse/accept an intervention. Aspects of executive dysfunction related to the FrSBE at baseline and post decision appeared related to intervention refusal. Also relevant were patients' own concerns, understanding of and perceived necessity for an intervention, as well as aspects of spirituality.

**Conclusions:** Since other studies have demonstrated the relevance of executive dysfunction for treatment compliance and prognosis in ALS, current findings raise the possibility that cognitive impairment in people with ALS may be also be influential in how interventions are made available or information is presented to patients. Greater awareness needs to be raised of these non-illness related factors that may influence decision-making and therefore survival.

#### **Understanding the goal-setting process in cognitive rehabilitation for people with early-stage dementia** Aleksandra Kudlicka, Suzannah Evans, Linda Clare

**Objective:** There is promising evidence that people with early-stage dementia (PwD) can benefit from individualised cognitive rehabilitation interventions, and can successfully engage in the process of eliciting therapeutic goals. The Bangor Goal Setting Interview schedule was developed as a means of structuring the process of identifying therapy goals. In this study we examined the goal-setting process and the nature of the goals identified.

**Participants and methods:** The Bangor Goal Setting Interview was used to elicit therapy goals for people with early stage AD, vascular, or mixed dementia as part of the baseline assessment in the ongoing GREAT trial. Researchers and PwD worked together to identify two or three specific, measurable, achievable, personally relevant and time-bound therapy objectives related to everyday functioning. Thematic analysis was used to reveal common themes in the identified goals.

**Results:** We analysed a total of 591 therapy goals identified by 209 PwD. Goals were associated with nine domains: socialising, exercising, engaging in meaningful activities, using new technology, carrying out activities of daily living, remembering names, locating lost items, managing medication and improving orientation. Participants were motivated to work on these goals for a range of reasons, including reducing dependence and improving enjoyment of life.

**Conclusions:** Significant numbers of people with early stage dementia are able to identify meaningful therapy objectives. These personalised rehabilitation goals provide information about the areas where support would be most welcomed by people with dementia, and can be used to inform the development of genuinely person-centred rehabilitation interventions.

#### **Developing a cognitive rehabilitation approach for people with Parkinson's Disease Dementia and Dementia with Lewy Bodies: the CORD-PD trial**

Tamlyn Watermeyer, Julie Roberts, Linda Clare, John Hindle

**Objective:** Approximately 30% of people with Parkinson's disease (PD) experience a dementia that shares a similar neuropsychological profile to that of Dementia with Lewy Bodies (DLB). Pharmacological treatments are available, but due to possible side-effects, may not be suitable for all patients. Non-pharmacological interventions may offer an alternative to support people with early Parkinson's disease dementia (PDD) and DLB. Cognition-focused interventions, mostly cognitive training, have been explored in people with PD who have mild cognitive impairment, but no studies to date have assessed such interventions in PDD or DLB. The efficacy of cognitive rehabilitation (CR) in Alzheimer's disease and other dementias is currently being explored in the ongoing GREAT trial. However, since the application of CR in PDD and DLB may be complicated by the distinct features of these disorders, notably their parkinsonian symptoms, these patients were not included in GREAT. Our objective was to adapt the CR approach for people with PDD and DLB.

**Participants and methods:** CORD-PD is an ongoing pilot RCT that aims to assess the feasibility and potential effectiveness of CR for people with early PDD and DLB.

**Results:** We will make use of case studies from our work so far to illustrate the types of goals identified. Issues surrounding goal-setting and implementing CR with PDD and DLB patients will be discussed.

**Conclusions:** We will consider the expected results and possible clinical implications of this research. Finally, we will reflect upon the lessons learned from this pilot study for the development of a future fully powered RCT.

#### **Symposium Session: The clinical utility of neuropsychological genetics: Treatment follows cognitive phenotyping 15.30 - 17.00** Convenor: Jos Egger

Discussant: Tjitske Kleefstra

Speakers: Tjitske Kleefstra, Jos Egger, Karlijn Vermeulen, Linde Van Dongen, Renée Roelofs

#### **The scientific and clinical relevance of studying rare genetic/neurodevelopmental disease**

Tjitske Kleefstra, Rolf Pfundt, Han Brunner, Jos Egger

**Objective:** Intellectual disability (ID) with or without autism spectrum disorders (ASD), is one of the main reasons for referral to a clinical geneticist. ID has a major impact on affected individuals, their families and society. The recent advances in genetic technologies have enabled to identify disease causing variants throughout the whole human exome, even at the single base-pair level of the DNA. This significant increase in diagnostic potential is of high value for proper genetic counseling and paves the way for studying fundamental aspects of brain functioning on one hand and the more personalized approach of syndrome specific management of ID disorders at the clinical site on the other hand.

**Participants and Methods:** So far, over 2000 ID patients have been investigated in our genome diagnostic center by whole exome sequencing which revealed a diagnostic yield of around 30%. Though most genes are very rarely affected, by collaborative efforts with other (inter)national genetic departments, substantial cohorts enable the definition of numerous such novel rare genetic syndromes.

**Results:** Examples of novel syndromes that we have recently defined are GATAD2B, POGZ, KBG, USP9X and DDX3X. The increase in novel syndromes needs multidisciplinary expertise and care including neuropsychiatric involvement.

**Conclusions:** The potential of novel genetic techniques will be discussed and examples of novel syndromes will be given. Syndrome specific management and how to centralize expert knowledge will be highlighted by our experience through our expert center and the formation of European Reference Networks.

#### **Phelan-McDermid syndrome: Neuropsychological phenotype, cerebellar functioning and treatment selection**

Jos Egger, Willem Verhoeven, Renée Zwanenburg, Conny Van Ravenswaaij, Clara Bonaglia, Tjitske Kleefstra

**Objective:** The 22q13.3 deletion syndrome or Phelan-McDermid syndrome is characterized by a variable degree of intellectual disability, impaired speech and language as well as social communicative skills, and mild dysmorphic features. The SHANK3 gene is thought to be a major contributor to the phenotype. Apart from the syndrome associated autistic features, symptoms from the bipolar spectrum can be discerned, in particular behaviour instability and fluctuating mood culminating in a (hypo)manic state. In case of coincident major somatic events, a deteriorating course may occur.

**Participants and Methods:** The present study comprises seven adult patients (four females, three males; aged 21-44 years) with genetically proven Phelan-McDermid syndrome. Data from medical records were collected and extensive assessment of neuropsychological variables was performed to identify cognitive characteristics and their relation with psychopathology and treatment.

**Results:** All patients showed profound communication deficits and their developmental functioning ranged from 1;0 to 6;3 years. In addition, they had slow speed of information processing, impairment of attentional and executive functions, and cognitive alexithymia. As to psychopathology, features from the affective and anxiety domains were prominent findings in these seven patients suggesting the