Toward a Noonan syndrome specific social cognitive training

Neurocognition, adaptive functioning, and psychopathology in Kleefstra syndrome

Karijn Vermeulen, Tijlkske Kleefstra, Wouter Staal, Hans Van Bokhoven, Jos Egger

Objective: The diagnostic yield for rare genetic causes for ID has increased tremendously over the last years. Studies that focus on sub-cohorts with known underlying genetic causes may enable to define more specific profiles that potentially could guide tailor made management. In our present study we aimed to examine if EHMT1 gene effects, which are also known as Kleefstra Syndrome (KS) in human, are associated with specific profiles for adaptive and maladaptive functioning.

Participants and Methods: In total we studied 58 subjects with ID (28 males, 30 females): 24 with Kleefstra Syndrome and 34 controls. They were examined with the Vineland Adaptive Behavior Scale, mini PAS-ADD interview, Autism Diagnostic Schedule, and the Cambridge Neuropsychological Test Automated Battery (CANTAB) to obtain measures of adaptive and maladaptive functioning. This study has an explorative nature and statistical analysis were used to contrast the results (Fisher’s exact test for prevalences, Mann-Whitney tests for subscale scores).

Results: KS-participants have low levels of adaptive functioning. Autism spectrum disorders are extremely prevalent (about 80%, p=0.001). There are also significantly high prevalences and symptom scores for depressive episodes (41.6%, p=0.043), obsessive compulsive disorders (33.3%, p=0.03) and psychotic symptoms (29.2%, p=0.005). The performance and results at the CANTAB are discussed in line with these. All together this results in a discriminating neuropsychiatric picture in KS patients.

Conclusions: KS patients are extremely vulnerable to develop neuropsychiatric disorders and should be carefully monitored for this.

Delineation of the cognitive phenotype of KBG syndrome

Linde Van Dongen, Tijlkske Kleefstra, Ellen Wingbermühle, Conny Stumpel, Jos Egger

Objective: KBG syndrome is caused by a mutation in the ANKR11 gene and characterized by a short stature and specific craniofacial and skeletal anomalies. The relatively limited amount of literature on phenoypical presentation, mentions delayed speech and motor development as well as mild to moderate intellectual disabilities. As to psychopathology, autism and ADHD are often described, but not yet substantiated in terms of neurocognitive variables. Aim of the current study was to investigate neurocognitive aspects of KBG syndrome, in particular attentional and social cognitive functioning.

Participants and Methods: Seventeen patients (aged 6-66 years; ten females) with an ANKR11 mutation were compared with two different groups of patients with genetic disorders and similar mental ages (n=14 and n=10). Neuropsychological assessment was performed focusing on the level of intellectual functioning and on attention, memory, executive functioning, and social cognition.

Results: Preliminary results showed mild to moderate intellectual disabilities (TIQ 45-84, M=63.5, SD=10.7). Mean mental age (M=6.4 years, SD=2.6 years) was lower than mean chronological age (M=11 years, SD=5.7 years). When compared to both control groups, results indicated a relatively strong processing speed and social cognitive functioning and a relatively weak performance on the direct recall of auditory memory tasks.

Conclusions: The cognitive profile of this group of 17 patients with KBG is characterized by mild intellectual disability and diminished sustained attention in verbal tasks that fits the ADHD symptoms described in the scarce literature on KBG. Implications for diagnostic procedures and clinical management the syndrome are discussed.

Toward a Noonan syndrome specific social cognitive training

Renée Roelofs, Ellen Wingbermühle, Ineke Van der Burgt, Roy Kessels, Jos Egger

Objective: The neuropsychological profile of patients with Noonan syndrome (NS) is characterised by lowered processing speed and impairments in social cognition (SC). Treatment of SC deficits has proven to be effective in other neuropsychiatric populations. The aim of this study is to perform a systematic review and to incorporate the results in a new, customised intervention protocol for the improvement of SC in patients with NS.

Participants and Methods: Controlled studies on SC interventions for adults with neuropsychiatric disorders, published between 1-1-2003 up to 1-1-2015, were identified through a systematic literature search in PubMed, Web of Knowledge, and PsycINFO databases.

Results: From the initial 4,565 hits, 101 full-text articles were assessed for eligibility, and 34 articles were included in the review. Studies predominantly focused on patients with schizophrenia (n=25), acquired brain injury (n=5), and autism (n=4). ‘Simple’ SC processes like emotion recognition were the main intervention targets. Treatment duration and intensity were highly variable (1-56 sessions; 1 week-2 years). Group size ranged from 1 to 12 patients, with an average of 5.

Conclusions: As patients with NS show deficits in the perception, interpretation, and expression of social-emotional information, a comprehensive approach seems most appropriate to improve SC in this group. Therefore, besides training of emotion recognition and theory of mind strategies, a specific SC treatment for adults with NS should also address problems in the identification and verbalization of (own) emotions. A training protocol comprising the aforementioned elements will be presented.

Symposium Session: Complexity of assessment for people in prolonged disorders of consciousness 15.30 - 16.30

Convenor: Anita Rose

Discussant: Agnes Shiel

Speakers: Samira Dhamapurkar, Olivia Gossers

Delayed recovery from the Vegetative and Minimally Conscious States

Samira Dhamapurkar, Barbara Wilson, Anita Rose, Gerhard Florschütz

Objective: Severity of brain injury is determined by the depth and duration of coma. Most patients who recover from coma open their eyes by four weeks post injury. They are then no longer in coma. They may have recovered full consciousness or they may still have reduced awareness/discrimination of consciousness (DOC). There is disagreement as to the percentage of patients who remain with a DOC after several months and then show some recovery. The aims of this study were to identify patients who made a delayed recovery 12 or more months after remaining with a DOC. Investigating if recovery was more likely for those who survived traumatic brain injury (TBI) and those who survived from other causes (mainly hypoxia).

Participants and Methods: All patients (N=33) with a disorder of consciousness, admitted to a rehabilitation centre over a five year period, were assessed with the Wessex Head Injury Matrix (WHIM) and the Disability Rating Scale (DRS) to determine if they had emerged from a DOC.

Results: Seven patients died. Twenty patients remained with a DOC for 12 or more months (11 in the Vegetative State and 9 in the Minimally Conscious State). Six patients (18%) emerged from a DOC (showed delayed recovery). Of these 5 had sustained a TBI and 1 had hypoxic damage.

Conclusion: We found that 18% of people who had a DOC for 12 or more month’s recovered consciousness and that survivors of a TBI were more likely to show delayed recovery than non TBI patients.

The use of neuroimaging technique in the diagnosis assessment of disorders of consciousness

Olivia Gossers, Steven Laureys

Objective: Evidences that some patients with disorders of consciousness (DOC) have higher level of cognition than behaviourally accessible are growing. Recently, cerebral positron emission tomography has shown its prognostic value in this population. The perturbational complexity index derived from