

Pilot Study for Subgroup Classification for Autism Spectrum Disorder Based on Dysmorphology and Physical Measurements in Chinese Children

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Background: Previous autism spectrum disorder (ASD) researches indicate that early identification combined with a targeted treatment plan involving behavioural interventions and multidisciplinary therapies can provide substantial improvement for ASD patients. Currently there is no cure for ASD, and the clinical variability and uncertainty of the disorder still remains. Hence, the search to unravel heterogeneity within ASD by subgroup classification may provide clinicians with a better understanding of ASD and to work towards a more definitive course of action.

Methods: In this study, a norm of physical measurements including height, weight, head circumference, ear length, outer and inner canthi, interpupillary distance, philtrum, hand and foot length was collected from 658 typical developing (TD) Chinese children aged 1 to 7 years (mean, 4.19 years). The norm collected was compared against 80 ASD Chinese children aged 1 to 12 years (mean, 4.36 years). We then further attempted to find subgroups within ASD based on identifying physical abnormalities; individuals were classified as (non)dysmorphic with the autism dysmorphology measure (ADM) from physical examinations of 12 body regions.

Results: Our results show that there were significant differences between ASD and TD children for measurements in: head circumference ($P=0.009$), outer ($P=0.021$) and inner ($P=0.021$) canthus, philtrum length ($P=0.003$), right ($P=0.023$) and left ($P=0.20$) foot length. Within the 80 ASD patients, 37 (46%) were classified as dysmorphic ($P=0.00$).

Conclusion: This study attempts to identify subgroups within ASD based on physical measurements and dysmorphology examinations. The information from this study seeks to benefit ASD community by identifying possible subtypes of ASD in Chinese population, and seeks for a more definitive diagnosis, referral and treatment plan.

Klein Levin Syndrome is a Steroid-responsive, Non-N-methyl-D-aspartate Receptor-mediated Encephalitis

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Background: Klein Levin syndrome is a rare neuropsychiatric disorder with periodic hypersomnia, cognitive and behavioural disturbance. It is postulated to be triggered by a viral illness or is a post-infectious immune-mediated encephalitis. With an increasing awareness of immune-mediated encephalitis in the past 10 years, especially N-methyl-D-aspartate receptor (NMDAR)-mediated encephalitis, testing for the presence of NMDAR antibodies in patients with Klein Levin syndrome might give us more understanding about the relationship between these two disease entities. Immunotherapy has not been reported to be a treatment option. Current evidence suggested the use of lithium to improve the abnormal behaviour with recovery of symptoms in patients with Klein Levin syndrome, including a decrease in duration of each attack and frequency of relapses.

Methods: A 15-year-old boy with Klein Levin syndrome presented with episodic attacks of repetitive excessive masturbation, hypersomnia, short-term memory loss, compulsive water drinking, and fluctuation in blood pressure was being investigated. Clinical course and treatment was given to the patient.

Results: Intravenous pulse methylprednisolone was given for 3 days, followed by a 4-week course of oral prednisolone. The boy went into complete remission few days after treatment. However, the neuropsychiatric symptoms recurred twice upon tapering of the oral steroid. Again, complete remission could be achieved after optimising the dose of oral prednisolone back to 1 mg/kg/day. Currently, his condition is well controlled with lithium while oral prednisolone was successfully tailed off.

Conclusion: This is the first reported case on the successful treatment of Klein Levin syndrome with steroid. We would like to support that Klein Levin syndrome, a neuropsychiatric disorder, is a steroid-responsive immune-mediated encephalitis. However the underlying mechanism is not NMDAR antibody mediated. Further study on a larger group of patients would help to elucidate the underlying aetiology, treatment and the prognosis of this disease.