Strategies to improve PCS management times are critical to decrease these disparities, including faster pre-hospital recognition and in-hospital workflows.

A.4

Automating gait analysis in children with Cerebral Palsy using an artificial intelligence-augmented pipeline

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Background: Cerebral palsy (CP) is a neuromotor disorder whereby gait abnormalities are predominant. Motion analysis is instrumental in management. While 3D kinematic labs exist, they are costly to operate, and the expertise required to interpret limits their availability to only a handful of facilities. In response, we have developed an Automated Intelligence (AI) driven pipeline to automate gait evaluation using 2-dimensional video. We assess the performance of this tool in comparison to traditional evaluation using visual assessment by trained human expert. Methods: A dataset of 109 patients with CP (6–37 years) (GMFCS I – II) was processed using our tool. The Edinburgh Visual Gait Score (EVGS) was derived using videos capturing sagittal and coronal views. Algorithm performance was determined by comparing automated EVGS scores against clinical expert scoring. Results: The AI pipeline successfully analysed 105/109 patient videos. For most EVGS parameters (14/17), the algorithm demonstrated moderate to high accuracy (70-94%), while 3 parameters (hindfoot valgus/varus, maximum lateral trunk shift, pelvic rotation at midstance) demonstrated lower accuracy (58-62%). Conclusions: This study validates the feasibility of an AI-augmented pipeline for automating EVGS-based gait assessments. With ongoing development, this technology has potential to improve accessibility to gait analysis and allows deployment outside of traditional settlings.

A.6

The clinical validation of a comprehensive neural autoantibodies testing

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Background: New neural antibodies are being identified each year and determining how to measure them and how to interpret test results is complex. In addition, screening with two methods is recommended for most antibodies, particularly paraneoplastic antibodies. We report the clinical validation and profile of a series of neural autoantibodies detected with a comprehensive testing algorithm. Methods: This is an ongoing study in which we are asking for the clinical correlation and final diagnosis of patients whose serum and/or CSF samples were tested at the BC Neuroimmunology Lab, Vancouver for neural autoantibodies. We performed immunofluorescence screening assay/IHC in rat brain sections in combination with confirmatory fixed or live Cell-Based assays and/or immunoblots. Results: We obtained clinical information from 219 samples (22 positive), Upon clinical inquiry, we obtained clinical information on 12 cases (five positive and seven negative). One Ttitin positive case was associated with anti-acetylcholine receptor antibody myasthenia and one Zic4 antibody was detected as a false positive by immunoblot but was negative by Rat Brain IHC. Conclusions: We have identified 10 percent seropositivity on 219 samples testing for Mosai-6 and full paraneoplastic testing. Further clinical validation studies are ongoing to evaluate the accuracy of our serological testing for neural antibodies.

A.7

A novel Xdystonia-parkinsonism gene variant responsive to bilateral GPi DBS: a video case study

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Background: X-linked dystonia-parkinsonism (XDP) is a rare movement disorder primarily affecting males of Filipino descent characterized by dystonia and parkinsonism. This case illustrates a patient with a novel gene variant responsive to deep brain stimulation (DBS). Methods: Case study of Filipino male with XDP followed for 15 years. Results: A 32-year-old Filipino male presented with oromandibular and cervical dystonia which later generalized. He went on to develop parkinsonism with significant gait impairment, incomprehensible speech, and required PEG tube placement. His symptoms were refractory to pharmacologic therapy. At age 43, he underwent bilateral globus pallidus internus (GPi) DBS placement with significant improvement of his symptoms as illustrated by videos accompanying this report. He had marked improvement of gait, speech, and pharyngeal dystonia resulting in removal of his PEG tube with return to full oral intake. He continues to benefit 3 years after DBS placement. Genetic testing identified a missense hemizygous non-coding transcript exon variant TAF1 n.5776C>T which is a novel gene variant of XDP not previously reported in the literature. Conclusions: This case illustrates a patient with a novel TAF1 gene variant associated with XDP not previously reported in the literature. This variant was responsive to bilateral GPi DBS placement.