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**THE USE OF MOTION ANALYSIS SYSTEM TO EVALUATE THE EFFECT OF SINEMET IN IMPROVING THE MOTOR FUNCTION OF AN ADOLESCENT WITH CTNNB1 SYNDROME**

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**Introduction**

CTNNB1 syndrome is a newly discovered rare genetic condition caused by loss-of-function mutations in the CTNNB1 (beta-catenin) gene. CTNNB1 gene is important in brain development and maturation. In addition to developmental problems, CTNNB1 mutations are associated with intellectual disability, speech impairment, microcephaly and motor problems including truncal hypotonia, limbs spasticity and dystonia. Some patients reported improvement in dystonia after using Sinemet (carbidopa - levodopa). However, no objective evaluation has been used to document the improvement.

**Objectives**

In this case study, we used the motion analysis system to objectively measure the effect of drug on movement in a patient diagnosed with CTNNB1 syndrome.

**Methodology**

A single case study with an adolescent diagnosed with CTNNB1 syndrome was referred for pre-drug assessment on gait and movement analysis. Hypertonia Assessment tool (HAT) confirmed patient's movement disorder to be mainly caused by dystonia. Instrumental gait analysis, Oxygen consumption test (O<sub>2</sub> cost), Dyskinesia Impairment Scale (DIS) were done at baseline, 1, 3 and 6-month post-sinemet usage. Standardized video recording protocol was adopted in lying, sitting and standing positions. Motion analysis for eight regions of body parts (eye, mouth, neck, trunk, proximal arm, distal arm, proximal leg, distal leg) were captured and analyzed. Movement analysis through video viewing and scoring with the DIS scoring sheet were performed simultaneously by three independent paediatric physiotherapists.

## **Result**

In gait analysis, the patient showed 23% improvement in walking speed at 3-month post-sinemet. Oxygen consumption test demonstrated 20% reduction of O<sub>2</sub> cost at 6-month post-sinemet. For DIS, the patient showed significant improvement in Action-Dystonia Score at 1-month post-sinemet (120/192 to 79/192) (the lower score the better). Further improvement in this score was shown at 3-month (62/192) and 6-month (60/192) post-sinemet. Similar results were observed in the Rest-Dystonia Score, pre (26/96), 1-month (17/96), 3-month (7/96) and 6-month (6/96). The scores on Action Choreo-athetosis were pre (52/192), 1-month (26/192), 3-month (29/192) and 6-month (29/192), while the scores on Rest Choreo-athetosis were pre (6/96), 1-month (2/96), 3-month (4/96) and 6-month (2/96). Moreover, the movement of upper limbs also demonstrated improvement in drawing of circular pattern during the DIS test. Patient also reported subjective improvement on movement stability.

### **Conclusions:**

The utilization of motion analysis together with DIS test provided objective congruent evidence demonstrating favorable therapeutic effect of Sinemet in our patient. When innovative treatment is being contemplated with only scanty anecdotal reports available, proper usage of objective assessment tools rather than subjective reporting enable the clinicians to conclude on the therapeutic efficacy and from there generate further research hypothesis for this group of newly discovered conditions.