

## BIOMECHANICAL EVALUATION OF FEMALE'S LOWER LIMB JOINTS WITH RETT SYNDROME

Chong-Ku Hah, Uk-Hyeon Seo\*, Jae-Hun Yi\*, and Hyun-Min Jun\*

Institute for Natural Science, Seoul Women's University, Seoul, Korea

\* Biomechanics Laboratory, Korea National Sport University, Seoul, Korea

**KEY WORDS:** Rett syndrome, moment, MECP2, mutation

**INTRODUCTION:** Rett syndrome (symbolized RTT) is caused by sporadic mutations in the gene MECP2 located on the X chromosome. It almost exclusively affects girls -- male fetuses with the disorder rarely survive to term. Development is typically normal until 6-18 months, when language and motor milestones regress, purposeful hand use is lost and acquired deceleration in the rate of head growth (resulting in microcephaly in some) is seen. Rett syndrome is usually caused (95% or more) by a de novo mutation in the child (so it is inherited from a genotypically normal mother, i.e. without a MECP2 mutation). The purpose of this paper is to evaluate moments(flexion/extension) of girl's lower limbs with Rett Syndrome and compare them with a normal girl's moment.

**METHOD:** One subject(11yrs, 29.7 kg, 128.0 cm) was a girl with RS and the other subject (10yrs, 31.3 kg, 138.1 cm) was a normal girl. Their gaits were captured with seven infrared cameras(MCU-240, Qualisys Inc., Sweden) and recorded with two force plate(type9286A, Kistler Com., Switzerland) at the same time. Data were analyzed with a visual 3D-software(C-motion Inc., USA)

### RESULTS:

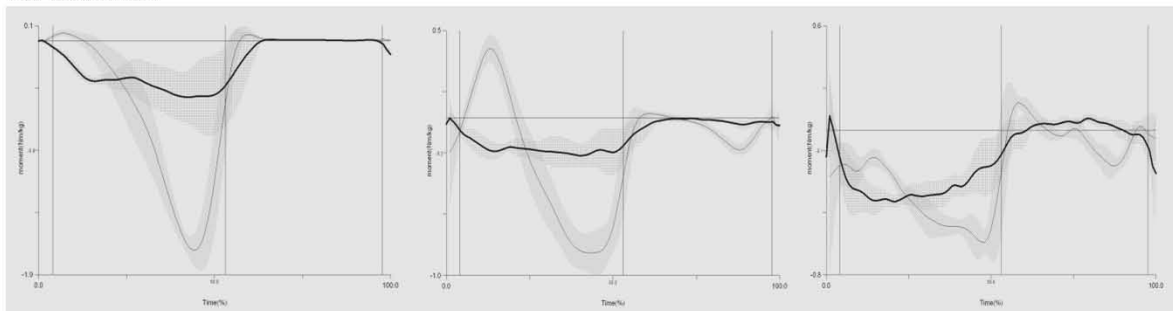


Figure 1. Right Ankle, Knee, Hip Joint Moment (unit: y-axis:Nm/kg, x-axis: time%) (RS: bold, Normal: solid)

**DISCUSSION:** As follows results, it is similar with the angle displacement pattern about flexion/extension of each lower extremity's joint during gait but different with the moment pattern. Especially the flexion/extension moment of ankle joint and knee joint of RTT's gait wasn't similar with the normal gait from heel contact to toe off. These results wasn't seemed to be smoothness between contraction and relaxation of lower leg's muscle group during gait and resulted from a genetic disorder with an apraxia which is one of RTT's characteristic.

**CONCLUSION:** The gait patterns of them were very different. A moments of girl's gait with RS were far less than normal gait(except hip moment), and the variations of moments with RS was smaller than normal gait.

### REFERENCES:

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