EVENT ABSTRACT

Back to Event

Duchenne Muscular Dystrophy in monozygotic twins: first follow up study worldwide

D N. Wijekoon[1], P Ratnayake[2], C R. Karunarathne[1], K G. Kumari[1], B L. Gonawala[1], R Angalena[3], A Dalal[3], J Nazarian[4, 5], E Hoffman[4, 5] and K. Ranil De Silva[1*]

[1] University of Sri Jayewardenepura. , Genetic Diagnostic & Research Laboratory, Faculty of Medical Sciences, Sri Lanka
[2] Lady Ridgeway Hospital, Lady Ridgeway Hospital, Sri Lanka
[3] *Center for DNA Fingerprinting and Diagnostics, Diagnostics Division, India*
[4] Children's National Medical Center, Research Center for Genetic Medicine, United States
[5] George Washington University School of Medicine and Health Sciences, Department of Integrative Systems Biology, United States

Duchenne muscular dystrophy (DMD), is a rare incidence in monozygotic twins. We report, to the best of our knowledge the first follow up study worldwide on a genetically confirmed sporadic case of monozygotic twins with DMD along with career detection in two generations. Socio-demographic characteristics and clinical data were recorded through a standard questionnaire, and clinical assessment scales [Barthel Index (BI), Hammersmith motor ability score, North Star Ambulatory Assessment (NSAA), Vignos and Brook scales]. A follow-up was performed after 15 months. Mutation detection protocols included multiplex PCR (20 primers) followed by Multiple Ligation dependent Probe Amplification (MLPA) using SALSA MLPA Kit P034/P035. Zygosity confirmation was performed with 13 STR markers. Multiplex PCR revealed no deletion in the hotspot regions of the dystrophin gene. MLPA analysis confirmed dystrophin gene deletions in exons 61, 62 with no duplications. Carrier status of the mother and the sister of the twins were confirmed for exon 61, 62. Clinical scores at initial visit (Age 7yrs): Twin 1 [BI-55/100, Hammersmith-28/40, NSAA-8/34, Vignos scale-5/10, Brook scale-1/6] and Twin 2 [BI-60/100, Hammersmith-28/40, NSAA-7/34, Vignos-5/10, Brook scale-1/6]. Clinical scores at follow up (Age 8yrs): Twin 1 [BI-10/100, Hammersmith-4/40, NSAA-0/34, Vignos scale-9/10, Brook scale-5/6] and Twin 2 [BI-10/100, Hammersmith-5/40, NSAA-0/34, Vignos-9/10, Brook scale-5/6]. The higher rate of disease progression with different manifestations of the disease in the monozygotic twins may be due to environmental, genetic & epigenetic factors which shed a light on utilizing this unique sample to identify new disease modifying genes and protein biomarkers.

Keywords: Mutation, Twins, epigenetic, Duchenne muscular dystrophy, Monozygotic

Conference: 14th Meeting of the Asian-Pacific Society for Neurochemistry, Kuala Lumpur, Malaysia, 27 Aug - 30 Aug, 2016.

Presentation Type: Poster Presentation Session **Topic:** 14th Meeting of the Asian-Pacific Society for Neurochemistry

Citation: Wijekoon DN, Ratnayake P, Karunarathne CR, Kumari KG, Gonawala BL, Angalena R, Dalal A, Nazarian J, Hoffman E and De Silva K (2016). Duchenne Muscular Dystrophy in monozygotic twins: first follow up study worldwide. *Front. Cell. Neurosci. Conference Abstract: 14th Meeting of the Asian-Pacific Society for Neurochemistry.* doi: 10.3389/conf.fncel.2016.36.00135

Received: 04 Aug 2016; **Published Online:** 11 Aug 2016.

* **Correspondence:** Prof. K. Ranil De Silva, University of Sri Jayewardenepura., Genetic Diagnostic & Research Laboratory, Faculty of Medical Sciences, Gangodawila, Nugegoda, Sri Lanka, ranil@sjp.ac.lk