



# **129<sup>th</sup> Anniversary International Medical Congress**

**Sri Lanka Medical Association**

*“Moving from Millennium Development Goals to  
Sustainable Development Goals”*

**Inauguration - 24<sup>th</sup> July 2016**

**Cinnamon Grand, Colombo**

**Main Congress - 25<sup>th</sup> to 27<sup>th</sup> July 2016**

**Galadari Hotel, Colombo**

management were assessed using an interviewer administered questionnaire at the patients' homes. Caregivers' skill level on home management was assessed by a practical session. Data were analyzed using SPSS 21 Version.

### Results

Mothers were the caregivers of the majority of patients (n=21, 67.7%). 58.06% (n=18) of the caregivers had some form of higher education. The mean general knowledge score about DMD was 87.90 (SD ± 10.94). Only 6.4% were having poor general knowledge regarding DMD. The mean knowledge score about home management was 41.94 (SD ± 21.82). 67.7% (n=21) had poor perceptions on home management. The mean home management skill level score of the caregivers was 25.52% (SD ± 22.52). Majority of the caregivers (80.6%, n=25) had poor home management skill levels. The caregivers' home management skill levels differed significantly by gender of the caregiver (P<0.01).

### Conclusions

Though caregivers had good general knowledge regarding DMD, their knowledge and skills level regarding home management were poor. Therefore home based rehabilitation seems to be less effective for these patients. More programmes should be conducted to improve caregivers' knowledge regarding home management.

#### PP111

### Demographic, clinical and biochemical characteristics of patients with homocystinuria in Sri Lanka

Dilanthi HW<sup>1</sup>, Indika NLR<sup>1</sup>, Kularatnam GAM<sup>1</sup>, Chandrasiri NDPD<sup>1</sup>, Jayasena KLSPKM<sup>1</sup>, Jasinge E<sup>1</sup>

<sup>1</sup>Department of Chemical Pathology, Lady Ridgeway Hospital for Children, Colombo

### Introduction and objectives

Homocystinuria is a group of disorders caused by defects in the remethylation or transsulfuration of Homocysteine. In this study, we assessed the demographic, clinical

and biochemical characteristics of patients with homocystinuria in Sri Lanka.

### Method

The study included the patients who were referred to the Department of Chemical Pathology, Lady Ridgeway Hospital, Sri Lanka, with suspected homocystinuria from 2014 to 2016 and their asymptomatic siblings with positive silver nitroprusside test. Each patient with clinical features of homocystinuria and positive silver nitroprusside test were further investigated with plasma methionine, plasma homocysteine, serum vitamin B12 levels, acylcarnitine profile, urine organic acid analysis, complete blood count and blood picture to arrive at a specific diagnosis.

### Results

Out of the 74 patients screened, 11 (14.8%) patients had a positive screening test (silver nitroprusside test). Three patients (27.2%) had probable cystathionine β synthase deficiency, 2 (18.2%) had probable cobalamin defect and one (10.1%) had combined methylmalonic aciduria and homocystinuria. The two commonest presentations were impaired vision and learning difficulties. Parental consanguinity was seen in 54.5% patients. Average gap between the onset of symptoms and the diagnosis was two years.

### Conclusions

A significant number of patients were detected with homocystinuria. Hence it is important to improve the awareness of the disease among clinicians in Sri Lanka about the disease. Improvement of laboratory facilities to detect the disease early to prevent complications is important.

#### PP112

### Parental perceptions on nutritional status and health related quality of life of their primary school children in Colombo educational zone

Amarathunga AADS<sup>1</sup>, Wickramasinghe VP<sup>2</sup>, Siriwardana HVYD<sup>3</sup>

<sup>1</sup>Allied Health Sciences Unit, Faculty of Medicine, University of Colombo