ABSTRACT

| TITLE OF THE ABSTRACT | : The manifestations of Wilson's disease with respect to renal tubular dysfunction, bone health and body composition. |
|-----------------------|---|
| DEPARTMENT | : Endocrinology, Diabetes and Metabolism, Christian Medical College, Vellore. |
| NAME OF THE CANDIDATE | : Nitin Kapoor |
| DEGREE AND SUBJECT | : D.M (Endocrinology) |
| NAME OF THE GUIDE | : Professor Thomas Paul |
| NAME OF CO-GUIDE | : Professor Nihal Thomas |
| | |

KEY WORDS:

Wilson's Disease Renal Tubular Acidosis Bone Mineral parameters Body composition

AIM / OBJECTIVES:

To study the prevalence of Renal Tubular Acidosis (RTA) among patients with Wilson's Disease (WD). To also study the bone health, body composition and factors that affect bone mineral density in these individuals.

MATERIAL AND METHODS:

All patients aged > 12 years attending the Hepatology and Neurology outpatient and inpatient departments at CMC, Hospital over a period of 1 year who were diagnosed to have Wilson's disease and who met the inclusion/exclusion criteria were included in this cross sectional study after a prior written informed consent. After a detailed history and examination, they underwent biochemical testing to assess the renal tubular dysfunction including ammonium chloride loading test. Bone mineral density (BMD) and body composition analysis were analysed using Dual energy X-Ray absorptiometry(DXA) machine.

RESULTS:

A total of 25 subjects were recruited in this study. Fifty six percent of patients with Wilson's Disease had Renal Tubular Acidosis. Distal RTA was found in 24% patients. RTA was more common in patients with Hepatic WD and who had a prolonged duration of illness. Patients with WD had significantly lower BMD as compared to control subjects. Low BMI (p=0.04), low IGF -1(p=0.002) and a shorter duration of therapy(p=0.001) were the key determinants of low bone mass. Patients with WD had significantly more body fat(p=0.01) and lower lean mass(p=0.03) when compared to controls.

CONCLUSIONS:

Renal tubular acidosis was common in patients with Wilson's disease. These patients often have lower bone mineral density and probable higher risk of development of fractures. Hence all patients of WD should be screened and treated for these parameters to improve their quality of life. Further prospective studies are needed to validate these findings.