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ANOCTAMINOPATHY WITH PERSISTENT MILD SYMPTOMATIC HYPERCKEMIA AS INITIAL PRESENTATION.

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INTRODUCTION

Anoctaminopathies are a group of AR skeletal muscle disorders with male preponderance 2:1, caused by ANO5 gene mutations. clinical phenotypes range from asymptomatic hyperCKemia, exercise induced myalgia to proximal and/or distal muscle weakness. ANO5 mutations are highly prevalent in European countries; however it is not common in patients of Asian origin.

AIMS / OBJECTIVES

MATERIALS / METHODS

CASE REPORT:
A 37 year old man, born of non consanguineous marriage, presented with myalgias, excercise intolerance for 8yrs, proximal weakness of both upper limbs, proximal and distal weakness of both lower limbs since 3yrs.
O/E: He has wasting of pectorals, biceps>triceps, scapular winging diving position,thigh adductor wasting, ankle contracture with toe walking. power: proximal upper 4/5,above head abduction weak,proximal lower-hip addiction -4/5, ankle DF -4/5.His plantars are flexor with absent ankle jerk.

RESULTS & DISCUSSION

RESULTS: Blood routines, thyroid profile were normal, s. CPK levels elevated moderately, Ncs all 4 limbs -normal, EMG showed myopathic potentials in trapezius, biceps. 2D ECHO was normal. Whole exome seq- revealed homozygous mutation in ANO5 gene. and heterozygous mutation in DYSF gene.

DISCUSSION: isolated asymptomatic hyperCKemia, pseudo metabolic phenotypes frequently misdiagnosed as metabolic myopathies are also manifestations of hereditary Anoctaminopathy (pentilla etal). Regular follow up needed to look for onset of muscle weakness. Genetic analysis is the only way for definite diagnosis of these sporadic cases.

CONCLUSION

Awareness of anoctaminopathy as the cause of nonspecific muscular complaints or of isolated hyperCKemia is essential to correctly diagnose affected patients.