Channelopathy chronicles - A case series.

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INTRODUCTION

- Membrane excitability, which is critical for the function of muscle and nerve is regulated by voltage gated ion channels. It is therefore not surprising that ion channels are involved in the pathogenesis of diseases of these tissues. In muscle these diseases are characterized by either transient, membrane hyperexcitability (i.e, myotonia) or hypoexciatbility (i.e paralysis)or both. This simple way of distinguishing the main symptoms corresponds quite well to the types of ion channels involved and allows one to discern.
- 1. Classic congenital myotonia without paralysis (chloride channel)
- 2. Varying combination of myotonia and paralysis (sodium channel)
- 3. Paralysis without myotonia. (various cation channels)

OBJECTIVE

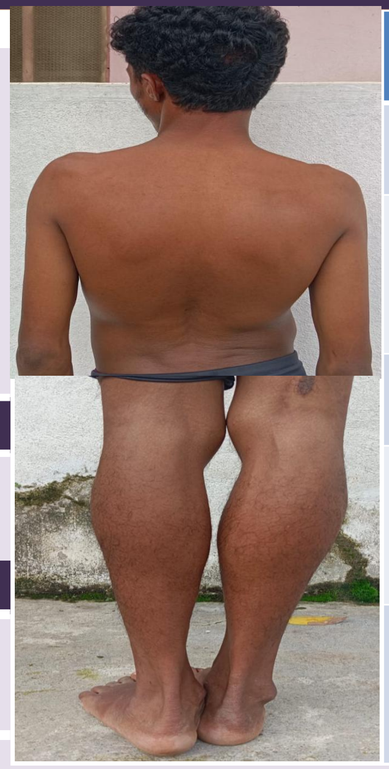
The objective of the study is to characterize the clinical presentations and genetic profiles of patients presented with periodic paralysis through a comprehensive case series analysis.

MATERIALS AND METHODS

- Here, we present a case series detailing five patients presenting with periodic paralysis and myotonia. Clinical features, family history, electrodiagnostic data, genetic test results, and treatment outcomes were analyzed.

OBSERVATION

In this case series 5 patients were analyzed. Their ages ranged from 20 to 37 years, with three of them having a history of consanguineous parentage. All of them were male. Two patients presented with muscle stiffness which would worsen with initial movements after a state of inactivity. The stiffness would get better when they moved around. Three patients presented with episodic weakness which would worsen with strenuous exercise and carbohydrate intake. one patient had a severe attack with neck flexor weakness requiring intensive care.



PHENOTYPE	GENE
Myotonia	CLCN 1
Myotonia and periodic paralysis	CLCN 1
Periodic paralysis	CACN1AS
Hypokalemic periodic paralysis	SCN4A
Hyperkalemic periodic paralysis	SCN4A

Their clinical features, family history and electrodiagnostic tests were analysed. Whole exome sequencing revealed chloride channel CLCN1 mutation in two patients, sodium channel SCN4A mutation in two patients and, calcium channel CACN1AS mutation in one patient

CONCLUSION

The clinical and genetic spectrum of muscle channelopathies is highlighted in this case series. Two patients with myotonia who had typical pathogenic mutations of the CLCN 1 gene were at one extreme of the spectrum. On the opposite end of the scale, we found three individuals whose primary complaint was periodic paralysis. One patient had a mutation in CACN1AS, while two patients had mutations in SCN4A.