Journal of Rare Cardiovascular Diseases
ISSN: 2299-3711 (Print) | e-ISSN: 2300-5505 (Online)
www.jrcd.eu

CASE REPORT

Primary Hypokalemic Periodic Paralysis in a 13-year-old: Importance of Early Genetic Diagnosis

Dr. Ashwini Ambalavanan, Dr. Shalini Priya Nandagopal, Dr. Joshua Rajan X and Dr. Syed Mohammed H
Saveetha Institute of Medical and Technical Sciences

*Corresponding Author
Dr. Syed Mohammed H

Article History
Received: 08.08.2025
Revised: 15.09.2025
Accepted: 24.10.2025
Published: 05.11.2025

Abstract: ***Introduction***: Hypokalemic periodic paralysis is a rare form of genetic channelopathy characterized by sudden onset of weakness. This is one form of periodic paralysis, a rare group of disorder. Other forms of periodic paralysis include thyrotoxic periodic paralysis, hyperkalemic periodic paralysis and Anderson syndrome. We are reporting a case of young male who presented to the emergency room with sudden paralysis to highlight on such a diagnosis. ***Case Report:*** A case of 13year old male is discussed here. The patient presented with sudden onset of weakness of both upper and lower extremities since the day of admission. Physical examination showed paresis in both upper and lower limbs, with lower limb being more affected than upper limbs. No sensory deficit was observed during examination. Baseline investigations revealed low potassium levels. The patient's paralysis resolved upon repletion of his low potassium and he was discharged with no neurological deficit. ***Conclusion***: Periodic paralysis should be considered in patients presenting with sudden onset of weakness or and treatment are crucial as untreated cases can be life-threatening. Though this is a rare disease, its symptoms can have a significant impact on the quality of life. Addressing the underlying cause is essential to prevent paralysis from persisting or recurring.

Keywords: weakness; hypokalemia; periodic paralysis; potassium; neuromuscular disorder.

INTRODUCTION

Periodic paralysis (PP) is a rare neuromuscular disorder related to a defect in ion channel, characterized by episodes of painless muscle weakness, which may be precipitated by heavy exercise, fasting, or high-carbohydrate meals. There are several types of periodic paralysis associated with electrolyte and metabolic abnormalities [1].

Of the various forms of periodic paralysis, Hypokalemic Periodic Paralysis (HPP) is the most common with a prevalence of 1 in 100,000 [2]. HPP, also known as Westphal's syndrome, is an autosomal dominant channelopathy characterized by sudden muscle weakness or paralysis [2]. While rare, this condition can pose a potential threat to life. Nevertheless, prompt and accurate diagnosis and treatment can lead to complete reversal of its effects [3]. This case report explores the clinical progression, diagnostic complexities, and treatment approaches in a patient with hypokalemic periodic paralysis, with the aim to enhance the understanding and optimize management of this condition.

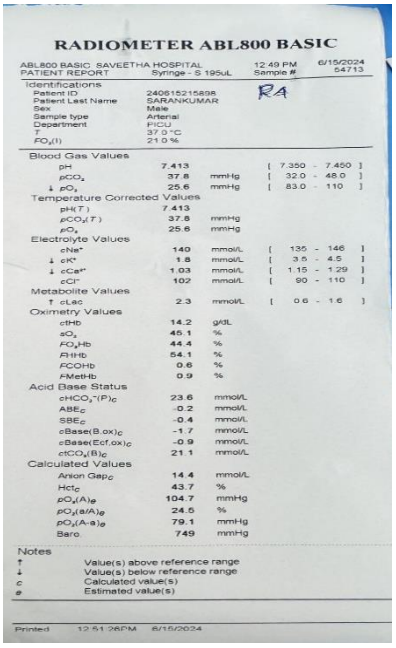
CASE PRESENTATION:

A 13year old male child with no significant past medical history presented to the emergency room with sudden onset of weakness of both upper and lower limbs since morning. The previous night, he went to bed without any complaints but was unable to move his upper and lower extremities the next morning. He did not experience any respiratory or swallowing difficulties and could move his neck and facial muscles. He reported no pain or paraesthesia.

Prior to this episode, the patient had a history of low-grade, intermittent fever associated with headaches five days earlier. Three days ago, he experienced one day of loose stools, with three watery, non-bloody, and non-foul-smelling episodes. He did not take any medications and denied using alcohol or drugs, as well as any significant changes in diet or activity levels. His parents and brother had no history of similar episodes or other significant illnesses.

On examination, child was conscious, oriented. Vitals were stable. On physical examination, child was awake and alert and of thin built. Cardiac examination revealed regular rhythm and no murmurs. Both the respiratory and abdominal examination were unremarkable. There were no deformities or oedema of the extremities and distal pulses were present and equal bilaterally. Neurologic exam revealed power of all 4 limbs reduced, with upper limbs power being 2/5 and lower limbs being 0/5 and absent reflexes. Sensation was intact, and Cranial nerves examination was normal. Blood gas revealed normal pH & gas, Electrolytes revealed Potassium of 1.8. Child was started on IVF DNS with 20mEq on peripheral Live.

Routine investigations, including liver enzymes and complete blood count were normal except for potassium level of 2.1, CPK – 340. Thyroid function was normal. Nerve conduction study of all four limbs showed pure motor axonal polyneuropathy involving upper and lower limbs (LL>UL). Electrocardiogram showed prolonged QT interval



RADIOMETER ABL800 BASIC

ABL800 BASIC; SAVEETHA HOSPITAL — 12:49 PM — 6/15/2024
PATIENT REPORT — Syringe - S 195uL — Sample # — 54713

Identifications

Patient ID	2406152158SR
Patient Last Name	SARANKUMAR
Sex	Male
Sample type	Arterial
Department	PICU
T	37.0 °C
$FO_2(I)$	21.0 %

RA

Blood Gas Values

pH	7.413		[7.350 - 7.450]
pCO_2	37.8	mmHg	[32.0 - 48.0]
↓ pO_2	25.6	mmHg	[83.0 - 110]

Temperature Corrected Values

pH(T)	7.413	
pCO_2(T)	37.8	mmHg
pO_2	25.6	mmHg

Electrolyte Values

cNa^+	140	mmol/L	[135 - 146]
↓ cK^+	1.8	mmol/L	[3.5 - 4.5]
↓ cCa^{2+}	1.03	mmol/L	[1.15 - 1.29]
cCl^-	102	mmol/L	[90 - 110]

Metabolite Values

↑ cLac	2.3	mmol/L	[0.6 - 1.6]

Oximetry Values

ctHb	14.2	g/dL
sO_2	45.1	%
FO_2Hb	44.4	%
FHHb	54.1	%
FCOHb	0.6	%
FMetHb	0.9	%

Acid Base Status

$cHCO_3^-(P)_c$	23.6	mmol/L
ABE_c	-0.2	mmol/L
SBE_c	-0.4	mmol/L
$cBase(B,ox)_c$	-1.7	mmol/L
$cBase(Ecf,ox)_c$	-0.9	mmol/L
$ctCO_2(B)_c$	21.1	mmol/L

Calculated Values

Anion Gap_c	14.4	mmol/L
Hct_c	43.7	%
$pO_2(A)_e$	104.7	mmHg
$pO_2(a/A)_e$	24.5	%
$pO_2(A-a)_e$	79.1	mmHg
Baro	749	mmHg

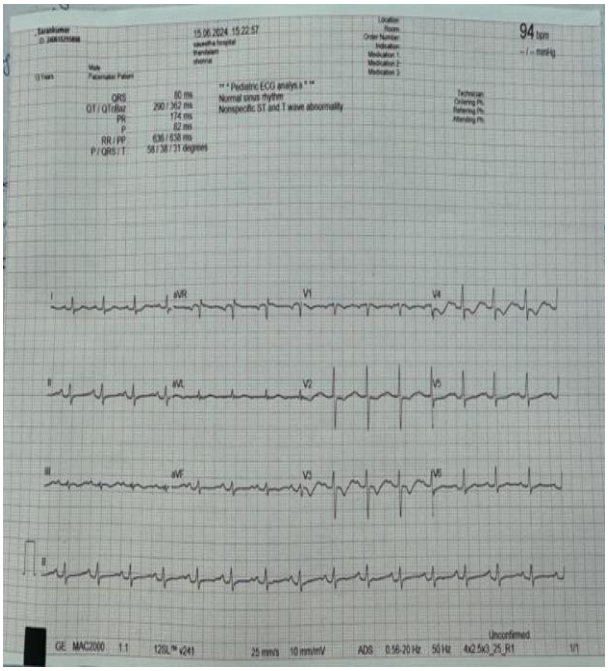
Notes

↑ Value(s) above reference range
↓ Value(s) below reference range
c Calculated value(s)
e Estimated value(s)

Printed 12:51:26PM 6/15/2024

BLOOD GAS OF THE CHILD IN ROOM AIR: showed potassium 1.8

Three hours after initiation of intravenous potassium replacement, the patient's neurologic symptoms had completely resolved. Repeat electrocardiogram revealed a normal sinus rhythm and rate.



Follow up studies were performed to determine the aetiology of the patient's hypokalaemia and genetic work up was done.

GENETIC ANALYSIS:

Whole exome sequencing was done. Reports detected Gene CACNA1S for Congenital Myopathy due to Dihydropyridine receptor defect OMIM#620246

RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS DETECTED

SNV(s)/INDELS

Gene# (Transcript)	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification‡
CACNA1S **(-)** (ENST00000362061.4)	Exon 30	c.3716G>A **(p.Arg1239His)**	Heterozygous**	Congenital myopathy 18 due to dihydropyridine receptor defect (OMIM#620246)	Autosomal dominant; Autosomal recessive**	Pathogenic (PS3,PM2,PM5, PP3,PP5)

Parental testing is recommended, and classification of the variant(s) may change based on segregation analysis.

**This is an autosomal recessive disorder caused by bi-allelic (homozygous or compound heterozygous) pathogenic/likely pathogenic variants in the *CACNA1S* gene. The assay has detected a single heterozygous Pathogenic variant in *CACNA1S* gene mentioned in the table above. No other clinically relevant variant is detected in the coding region and exon-intron boundaries of these genes. Kindly correlate clinically.

DISCUSSION:

Hypokalemic periodic paralysis (HPP) is a rare autosomal dominant channelopathy characterized by recurrent episodes of flaccid muscle weakness associated with low serum potassium levels. It primarily affects skeletal muscle excitability and is most commonly linked to mutations in the CACNA1S gene, which encodes the α1 subunit of the L-type calcium channel, and less frequently to mutations in the SCN4A gene, encoding the skeletal muscle sodium channel (4,5).

Our patient, a 13-year-old male with his first episode of acute flaccid paralysis and genetically confirmed CACNA1S mutation, represents the classical presentation of HPP. The typical age of onset is between 10 and 20 years, with attacks often triggered by rest after vigorous exercise, high-carbohydrate meals, stress, or cold exposure (6,7). The absence of sensory, cranial, or sphincter involvement and rapid improvement after potassium supplementation are characteristic clinical features that help distinguish HPP from other causes of acute flaccid paralysis such as Guillain–Barré syndrome, myasthenia gravis, or toxic neuropathies (8).

The pathophysiology of HPP involves an abnormal shift of potassium from the extracellular to the intracellular compartment, leading to depolarization of muscle cell membranes and reduced muscle excitability. Mutations in CACNA1S or SCN4A alter ion channel function and predispose muscle fibres to sustained depolarization in the setting of elevated insulin or catecholamine levels after carbohydrate-rich meals or rest following exercise (9,10). Genetic testing plays a critical role in confirming the diagnosis, differentiating between primary (familial) and secondary causes of hypokalaemic paralysis, and enabling family screening and counselling (11).

The management of acute HPP focuses on prompt potassium replacement. Oral potassium is preferred if the patient can swallow and has no respiratory compromise, while intravenous supplementation is indicated in severe weakness or dysphagia. Continuous cardiac monitoring is essential during therapy to prevent rebound hyperkalaemia or arrhythmias(12). Our patient responded rapidly to intravenous potassium, with complete resolution of weakness within hours, consistent with classical HPP behaviour.

Long-term management aims to prevent recurrent attacks. Lifestyle modification—such as avoiding known triggers like excessive carbohydrate intake, prolonged rest after exertion, and high-salt foods—is the mainstay. In patients with frequent episodes, carbonic anhydrase inhibitors (e.g., acetazolamide or dichlorphenamide) can reduce episode frequency by inducing a mild metabolic acidosis and stabilizing muscle membrane potential (13). However, response to these agents varies depending on the underlying genetic mutation, and patients with SCN4A mutations may not benefit or may even worsen (14).

Given that this was the first episode in our patient, the long-term prognosis is favourable. Nevertheless, periodic follow-up and education of the patient and family about recognizing early symptoms and avoiding triggers are crucial to prevent recurrences and complications such as fixed myopathy that can occur in long-standing or poorly controlled cases (14).

This case underscores the importance of considering HPP in any child or adolescent presenting with acute, reversible muscle weakness and hypokalaemia. Early recognition, appropriate potassium replacement, and genetic confirmation are essential for optimal management and prevention of future episodes.

REFERENCES:

1. Soule BR, Simone NL. Hypokalemic Periodic Paralysis: a case report and review of the literature. Cases J. 2008 Oct 21;1(1):256. doi: 10.1186/1757-1626-1-256. PMID: 18939979; PMCID: PMC2584072.
2. Fontaine B, Vale-Santos J, Jurkat-Rott K, Reboul J, Plassart E, Rime CS, Elbaz A, Heine R, Guimaraes J, Weissenbach J, et al. Mapping of the hypokalaemic periodic paralysis (HypoPP) locus to chromosome 1q31-32 in three European families. Nat Genet. 1994;6:267–272. doi: 10.1038/ng0394-267.
3. Abdelmasih R, Abdelmaseih R, Hasan M, et al. (May 25, 2021) A Case of Hypokalemic Periodic Paralysis in a Young Athlete. Cureus 13(5): e15236. doi:10.7759/cureus.15236
4. Jurkat-Rott K, Lehmann-Horn F. Muscle channelopathies and critical points in functional and genetic studies. J Clin Invest. 2005;115(8):2000–2009.
5. Statland JM, Fontaine B, Hanna MG, Johnson NE, Kissel JT, Sansone VA, et al. Review of the diagnosis and treatment of periodic paralysis. Muscle Nerve. 2018;57(4):522–530.
6. Sansone VA, Griggs RC, Meola G. Hypokalemic periodic paralysis: Clinical and genetic aspects. Rev Neurol (Paris). 2008;164(7):547–556.
7. Miller TM, Dias da Silva MR, Miller HA, Kwiecinski H, Mendell JR, Tawil R, et al. Correlating phenotype and genotype in the periodic paralyses. Neurology. 2004;63(9):1647–1655.
8. Lin SH. Thyrotoxic periodic paralysis. Mayo Clin Proc. 2005;80(1):99–105.
9. Cannon SC. Channelopathies of skeletal muscle excitability. Compr Physiol. 2015;5(2):761–790.
10. Venance SL, Cannon SC, Fialho D, Fontaine B, Hanna MG, Ptácek LJ, et al. The primary periodic paralyses: Diagnosis, pathogenesis and treatment. Brain. 2006;129(1):8–17.
11. Jurkat-Rott K, Holzherr B, Fauler M, Lehmann-Horn F. Sodium channelopathies of skeletal muscle: The periodic paralyses. Pflugers Arch. 2010;460(2):239–248.
12. Tawil R, Griggs RC. Periodic paralysis. Muscle Nerve. 2002;25(3):395–411.
13. Bendahhou S, Cummins TR, Kula RW, Fu YH, Ptácek LJ. Impairment of slow inactivation as a common mechanism for periodic paralysis in sodium channel mutations. Neurology. 2002;58(8):1266–1272.
14. Matthews E, Hanna MG. Muscle channelopathies: Does the predicted channel gating pore offer new treatment insights for hypokalemic periodic paralysis? Curr Opin Neurol. 2010;23(5):466–471