Name of Journal: World Journal of Clinical Cases

Manuscript NO: 88346

Manuscript Type: CASE REPORT

Reviewer #1:

Scientific Quality: Grade B (Very good)

Language Quality: Grade A (Priority publishing)

Conclusion: Minor revision

Specific Comments to Authors: The article you've provided outlines a case study on Paramyotonia Congenita (PMC), a rare genetic disorder affecting skeletal muscle function. Here's a breakdown and review of the key points of the article: 1. **Background on PMC**: PMC is identified as a sodium channelopathy, initially identified by Eulenburg. It affects skeletal muscle and is typically diagnosed through electromyography (EMG). 2. **Case Summary**: The study reports on a 3-year-old female child diagnosed with PMC. The child's symptoms include laryngeal stridor, muffled speech, and myotonia present from birth. The symptoms worsen with cold, exposure to cool water, crying, and physical activity, but improve in warmth. Notably, the child's myotonia did not normalize even with warmth and remained unchanged after consuming potassium-rich food. This observation helps differentiate PMC from hyperkalemic periodic paralysis. 3. **Diagnostic Observations**: The child's needle EMG showed two unique types of myotonic discharges not previously documented in PMC studies: giant-amplitude potentials and irregular wave trains. These are significant findings as they expand the understanding of EMG features in PMC. 4. **Genetic Testing and Treatment**: Genetic testing revealed a heterozygous mutation in the SCN4A gene. After a six-month treatment with mexiletine, the child's symptoms showed improvement. 5. **Conclusion and Clinical Implications**: The case is significant for its unique EMG findings, which could assist clinicians in distinguishing PMC from neurological forms of myotonia. It broadens the known characteristics of EMG in PMC, potentially aiding in more accurate diagnosis.

LANGUAGE POLISHING REQUIREMENTS FOR REVISED MANUSCRIPTS SUBMITTED BY AUTHORS WHO ARE NON-NATIVE SPEAKERS OF ENGLISH

As the revision process results in changes to the content of the manuscript, language problems may exist in the revised manuscript. Thus, it is necessary to perform further language polishing that will ensure all grammatical, syntactical, formatting and other related errors be resolved, so that the revised manuscript will meet the publication requirement (Grade A).

Authors are requested to send their revised manuscript to a professional English language editing company or a native English-speaking expert to polish the manuscript further. When the authors submit the subsequent polished manuscript to us, they must provide a new language certificate along with the manuscript.

Once this step is completed, the manuscript will be quickly accepted and published online. Please visit the following website for the professional English language editing companies we recommend: https://www.wjgnet.com/bpg/gerinfo/240.

ABBREVIATIONS

In general, do not use non-standard abbreviations, unless they appear at least two times in the text preceding the first usage/definition. Certain commonly used abbreviations, such as DNA, RNA, HIV, LD50, PCR, HBV, ECG, WBC, RBC, CT, ESR, CSF, IgG, ELISA, PBS, ATP, EDTA, and mAb, do not need to be defined and can be used directly.

The basic rules on abbreviations are provided here:

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(2) Running title: Abbreviations are permitted. Also, please shorten the running title to no more than 6 words.

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(4) Key Words: Abbreviations must be defined upon first appearance in the Key Words.

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(8) Figures: Abbreviations are not allowed in the Figure title. For the Figure Legend text, abbreviations are allowed but must be defined upon first appearance in the text. Example 1: A: Hepatocellular carcinoma (HCC) biopsy sample; B: HCC-adjacent tissue sample. For any abbreviation that appears in the Figure itself but is not included in the Figure Legend textual description, it will be defined (separated by semicolons) at the end of the figure legend. Example 2: BMI: Body mass index; US: Ultrasound.

(9) Tables: Abbreviations are not allowed in the Table title. For the Table itself, please verify all abbreviations used in tables are defined (separated by semicolons) directly underneath the table. Example 1: BMI: Body mass index; US: Ultrasound.

6 EDITORIAL OFFICE'S COMMENTS

Authors must revise the manuscript according to the Editorial Office's comments and suggestions, which are listed below:

(1) Science editor:

1 Conflict of interest statement: Academic Editor has no conflict of interest. 2 Manuscript's theme: The topic is within the scope of the journal. 3 Academic misconduct: No academic misconduct was found. 4 Scientific quality and comments: (1) The authors report a case of paramyotonia congenita. The abundant myotonic discharges are detected in muscles by needle EMG. (2) Please supplement the contents of each part according to the WJCC publication requirements, including History of present illness, History of past illness, Personal and family history, Physical examination, Laboratory examinations, Imaging examinations, multidisciplinary expert consultation, Final diagnosis, and Treatment, etc. (3) Please add PMID and DOI numbers to your references. They are available at: http://doi.crossref.org/simpleTextQuery. If there are no PMID or DOI numbers, please provide the website. 5 Language evaluation: Grade A (Priority publishing). 6 Recommendation: Conditional acceptance.

(2) Company editor-in-chief:

I have reviewed the Peer-Review Report, full text of the manuscript, and the relevant ethics documents, all of which have met the basic publishing requirements of the World Journal of Clinical Cases, and the manuscript is conditionally accepted. I have sent the manuscript to the author(s) for its revision according to the Peer-Review Report, Editorial Office's comments and the Criteria for Manuscript Revision by Authors. Before final acceptance, uniform presentation should be used for figures showing the same or similar contents; for example, "Figure 1 Pathological changes of atrophic gastritis after treatment. A: ...; B: ...; C: ...; D: ...; E: ...; F: ...; G: ...". Please provide the original figure documents. Please prepare and arrange the figures using PowerPoint to ensure that all graphs or arrows or text portions can be reprocessed by the editor. In order to respect and protect the author's intellectual property rights and prevent others from misappropriating figures without the author's authorization or abusing figures without indicating the source, we will indicate the

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Thank you very much for your valuable comments and suggestions on our manuscript. Following the reviewer' and editors' comments, we have modified and improved our manuscript to your kind advices and referee's detailed suggestions. Our response is given in normal font and changes/additions to the manuscript are given highlighted the revised/added contents with yellow color in the revised manuscript. Below we address the reviewer's comments point-by-point. At the end of the letter is the revised manuscript.

1. Reviewer #1:

Scientific Quality: Grade B (Very good)

Language Quality: Grade A (Priority publishing)

Conclusion: Minor revision

Specific Comments to Authors: The article you've provided outlines a case study on Paramyotonia Congenita (PMC), a rare genetic disorder affecting skeletal muscle function. Here's a breakdown and review of the key points of the article: 1. **Background on PMC**: PMC is identified as a sodium channelopathy, initially identified by Eulenburg. It affects skeletal muscle and is typically diagnosed through electromyography (EMG). 2. **Case Summary**: The study reports on a 3-year-old female child diagnosed with PMC. The child's symptoms include laryngeal stridor, muffled speech, and myotonia present from birth. The symptoms worsen with cold, exposure to cool water, crying, and physical activity, but improve in warmth. Notably, the child's myotonia did not normalize even with warmth and remained unchanged after consuming potassium-rich food. This observation helps differentiate PMC from hyperkalemic periodic paralysis. 3. **Diagnostic Observations**: The child's needle EMG showed two unique types of myotonic discharges not previously documented in PMC studies: giant-amplitude potentials and irregular wave trains. These are significant findings as they expand the understanding of EMG features in PMC. 4. **Genetic Testing and Treatment**: Genetic testing revealed a heterozygous mutation in the SCN4A gene. After a six-month treatment with mexiletine, the child's symptoms showed improvement. 5. **Conclusion and Clinical Implications**: The case is significant for its unique EMG findings, which could assist clinicians in distinguishing PMC from neurological forms of myotonia. It broadens the known characteristics of EMG in PMC, potentially aiding in more accurate diagnosis.

Response: We thank the reviewer for raising the concern. In this manuscript, we aim at illustrate the special electromyographic features in a child with paramyotonia congenita. The reviewer only commented on the main points of the article and did not raise any other issues with our manuscript that needed to be revised. We revised the manuscript based on the editor's comments.

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Response: We revised manuscript to a professional English language editing company to polish the manuscript further. And we will provide a new language certificate along with the manuscript. Here the new language certificate.

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ZiboYimore Translation CO. LTD

To whom it may concern:The purpose of this letter is to verify that Zibo Yimore Translation CO. LTD provided the English proofreading services for the following thesis : Special electromyographic features in a child with paramyotonia congenita：a case report

Author: Hui Yi, Chenxiang Liu , Shuxin Ye, Yvlin Liu

The basic language editing including the correction of grammar, punctuation and syntax was performed normatively with our best efforts. And the edited document was returned to the writer on 04/12/2023. We are unaware of any changes or additions made to the manuscript after that time. We have kept the specimen of the received original file in case of any legal dispute.

Sincerely,

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(9) Tables: Abbreviations are not allowed in the Table title. For the Table itself, please verify all abbreviations used in tables are defined (separated by semicolons) directly underneath the table. Example 1: BMI: Body mass index; US: Ultrasound.

Response: We checked all of the abbreviations and changed any that are inappropriate in the text to meet your requirements. This includes Title, Abstract, Key Words, Core Tip, Main Text, Figures. Detailed revision is as below. In the revised manuscript:

Keywords: Paramyotonia congenita; channelopathy; Electromyography; Child; Case report

Electromyography

Low-frequency (2Hz, 3Hz, 5Hz) and high-frequency (10Hz, 20Hz, 30Hz) repetitive frequency electrical stimulation (RNS) exhibited normal outcomes. EMG indicated challenging motor unit action discharge (MUP) differentiation, with abundant myotonic discharges detected in muscles examined. Facial nerve motor conduction and blink reflex testing were unremarkable.

The measurements for her sensory conduction velocity (SCV), sensory nerve action potential (SNAP), motor nerve conduction velocity (MCV), and compound muscle actionpotential (CMAP) yielded normal results, illustrated in Figures 4 and 5.

Figure Legends

Figure 4. motor nerve conduction velocity (MCV) and compound muscle actionpotential (CMAP) of the right tibial nerve

Figure 5. sensory conduction velocity (SCV) and sensory nerve action potential(SNAP) of the left median nerve

Figure 8-1. compound muscle actionpotential (CMAP) and motor nerve conduction velocity(MCV) of the facial nerve

Figure 8-3. repetitive frequency electrical stimulation (RNS) results of the left abductor hallucis digitorum (Low-frequency [2Hz, 3Hz, 5Hz] and high-frequency [10Hz, 20Hz] were normal.)

EDITORIAL OFFICE'S COMMENTS

Authors must revise the manuscript according to the Editorial Office's comments and suggestions, which are listed below:

(1) Science editor:

1 Conflict of interest statement: Academic Editor has no conflict of interest. 2 Manuscript's theme: The topic is within the scope of the journal. 3 Academic misconduct: No academic misconduct was found. 4 Scientific quality and comments: (1) The authors report a case of paramyotonia congenita. The abundant myotonic discharges are detected in muscles by needle EMG. (2) Please supplement the contents of each part according to the WJCC publication requirements, including History of present illness, History of past illness, Personal and family history, Physical examination, Laboratory examinations, Imaging examinations, multidisciplinary expert consultation, Final diagnosis, and Treatment, etc. (3) Please add PMID and DOI numbers to your references. They are available at: http://doi.crossref.org/simpleTextQuery. If there are no PMID or DOI numbers, please provide the website. 5 Language evaluation: Grade A (Priority publishing). 6 Recommendation: Conditional acceptance.

Response: We thank the reviewer for raising the concern. We added the Background, Case summary, Conclusion, Core tip, Chief complaints, History of present illness, History of past illness, Personal and family history, Physical examination, Laboratory examinations, Imaging examinations, Exome-wide genetic testing, Final diagnosis, Treatment, and Outcome and follow-up. We have also added PMID and DOI numbers for most references, except for a few whose PMID or DOI numbers were not found on http://doi.crossref.org/simpleTextQuery.Detailed revision is as below. In the revised manuscript:

BACKGROUND

Paramyotonia congenita (PMC) stands as a rare sodium channelopaty of skeletal muscle, initially identified by Eulenburg. The identification of PMC often relies on electromyography (EMG), a diagnostic technique. The child's needle EMG unveiled trains of myotonic discharges with notably giant amplitudes, alongside irregular wave trains of

myotonic discharges. This distinctive observation had not surfaced in earlier studies.

CASE SUMMARY

We report the case of a 3-year-old female child with PMC, who exhibited laryngeal stridor, muffled speech, myotonia from birth. Cold, exposure to cool water, crying, and physical activity exacerbated the myotonia, which was relieved in warmth, yet never normalized. Percussion myotonia was observable in bilateral biceps. Myotonia symptoms remained unchanged after potassium-rich food consumption like bananas. Hyperkalemic periodic paralysis was excluded. Cranial magnetic resonance imaging (MRI) yielded normal results. Blood potassium remained within normal range, while creatine kinase showed slight elevation. Exome-wide genetic testing pinpointed a heterozygous mutation on chromosome SCN4A: c.3917G>A (p.G1306E).After a six-month mexiletine regimen, symptoms alleviated.

CONCLUSION

In this case revealed the two types of myotonic discharges, and had not been documented in other studies. We underscore two distinctive features: giant-amplitude potentials and irregular waves.

Core Tip:

Paramyotonia congenita (PMC) is a rare sodium channelopathy of skeletal muscle, first identified by Eulenburg. The distinguishing feature of PMC is paradoxical myotonia, where myotonia worsens with cold and exercise. In instances where genetic testing is unavailable, electromyographic (EMG) is a swift, cost-effective diagnostic and differential diagnostic method for PMC. This article elaborates on the EMG characteristics of a recently diagnosed PMC case at our hospital. In this particular case revealed the two types of myotonic discharges, and had not been documented in other studies. We underscore two distinctive features: giant-amplitude potentials and irregular waves.

Chief complaints

A 3-year-old child presented to the rehabilitation department, Qilu Children's Hopital of Shandong University with a complaint of inflexible physical activity for 3 years on July 16, 2022.

History of present illness

A 3-year-old child exhibited laryngeal stridor, muffled speech, and sporadic stridor from birth, a condition unaffected by positioning. This manifestation occurred after feeding and crying, leading to issues with water consumption, mild breathlessness, and occasional breath-holding. Severe cases brought about breathlessness, apnea, lip and facial cyanosis, and myotonia. Cold, exposure to cool water, crying, and physical activity exacerbated the myotonia, which was relieved in warmth, yet never normalized. At 6 months, restricted upper limb movement was observed. The child managed independent standing and walking by 1 year and 3 months, but upper limb inflexibility persisted. At 2 years and 8 months, calf spasms, walking stiffness, knee-flexed lower limb posture abnormalities, and occasional limb rigidity after trips were noted. Instances of ptosis, difficulty in eye opening, and eyeball movement restriction following face washing were present. Myotonia symptoms remained unchanged after potassium-rich food consumption like bananas.

History of past illness

The patient underwent inguinal hernia repair(bilateral) in 2021 at Qilu Children's Hopital of Shandong University.

Personal and family history

Non-consanguineous parents and no family history of similar neurological disorders.

Physical examination

Physical traits encompassed a short neck, hunched back, limited bilateral eye abduction, motor skill delays, and firm muscles with evident muscle bulges. Percussion myotonia was observable in bilateral biceps.

Laboratory examinations

Blood potassium remained within normal range (4.1-4.8 mmol/L), while creatine kinase showed slight elevation (279-771 U/L). Liver function, renal function, thyroid function and inorganic ions were normal.

Imaging examinations

Cranial magnetic resonance imaging (MRI) yielded normal results.

Exome-wide genetic testing

Exome-wide genetic testing pinpointed a heterozygous mutation on chromosome SCN4A: c.3917G>A (p.G1306E).

FINAL DIAGNOSIS

The diagnosis aligned with paramyotonia congenita, grounded in clinical features, lab tests, neurophysiology, and genetic evaluations[3, 11-14, 16]. Neither parent carried mutations in the gene, and familial history was absent, suggesting sporadic occurrence.

TREATMENT

The child received mexiletine at a initial dose of 50 mg three times a day. The dose was increased to 75 mg three times a day after 2 weeks, and to 100 mg three times a day after a month. In additon, the patients also participated in physical exercises for rehabilitation.

OUTCOME AND FOLLOW-UP

The frequency and severity after a eight-month mexiletine regimen, which affirmed the therapeutic effect.

CONCLUSION

The special case revealed two types of myotonic discharges, which had never been documented until now. We underscore two distinctive features: giant-amplitude potentials and irregular waves. These features are linked to the synchronous activation of multiple muscle fibers and the impairment of sodium channel inactivation. By sharing this case, our intent is to assist clinicians in distinguishing this type of myotonic discharge from neurological myotonia and widens the known the special feature of EMG in PMC.

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(2) Company editor-in-chief:

I have reviewed the Peer-Review Report, full text of the manuscript, and the relevant ethics documents, all of which have met the basic publishing requirements of the World Journal of Clinical Cases, and the manuscript is conditionally accepted. I have sent the manuscript to the author(s) for its revision according to the Peer-Review Report, Editorial Office's comments and the Criteria for Manuscript Revision by Authors. Before final acceptance, uniform presentation should be used for figures showing the same or similar contents; for example, "Figure 1 Pathological changes of atrophic gastritis after treatment. A: ...; B: ...; C: ...; D: ...; E: ...; F: ...; G: ...". Please provide the original figure documents. Please prepare and arrange the figures using PowerPoint to ensure that all graphs or arrows or text portions can be reprocessed by the editor. In order to respect and protect the author's intellectual property rights and prevent others from misappropriating figures without the author's authorization or abusing figures without indicating the source, we will indicate the author's copyright for figures originally generated by the author, and if the author has used a figure published elsewhere or that is copyrighted, the author needs to be authorized by the previous publisher or the copyright holder and/or indicate the reference source and copyrights. Please check and confirm whether the figures are original (i.e. generated de novo by the author(s) for this paper). If the picture is 'original', the author needs to add the following copyright information to the bottom right-hand side of the picture in PowerPoint (PPT): Copyright ©The Author(s) 2023. When revising the manuscript, it is recommended that the author supplement and improve the highlights of the latest cutting-edge research results, thereby further improving the content of the manuscript. To this end, authors are advised to apply PubMed, or a new tool, the RCA, of which data source is PubMed. RCA is a unique artificial intelligence system for citation index evaluation of medical science and life science literature. In it, upon obtaining search results from the keywords entered by the author, "Impact Index Per Article" under "Ranked by" should be selected to find the latest

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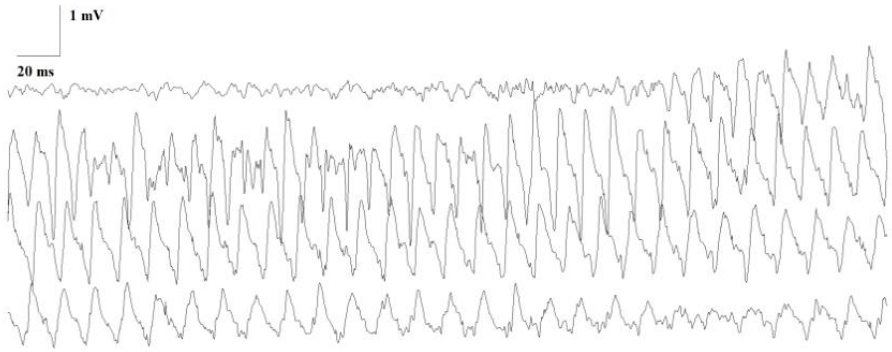

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Figure 1-1. Right rectus femoris (horizontal bar 20ms/D, vertical bar 1mV/D). The train of positive waves in myotonic discharges, displaying fluctuations in frequency and amplitude, with a frequency range of 70-150Hz and an amplitude range of 0.5-3mV, is depicted.

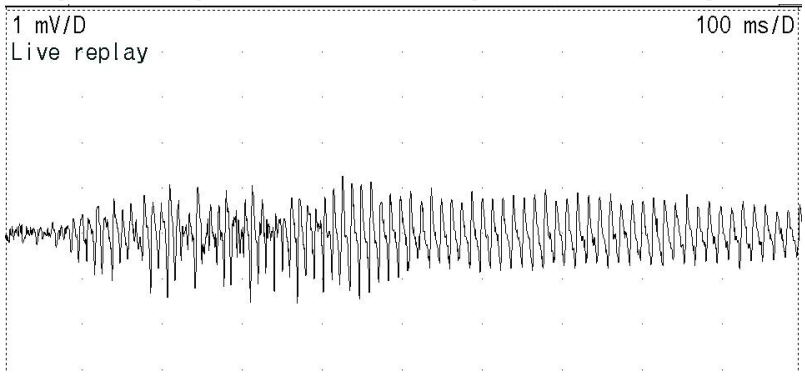

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Figure 1-2. Right rectus femoris (horizontal bar 100ms/D, vertical bar 1mV/D). A section of the train of positive waves in myotonic discharges showcases an oscillating pattern in amplitude and frequency, characterized by a rapid surge followed by a gradual decline.

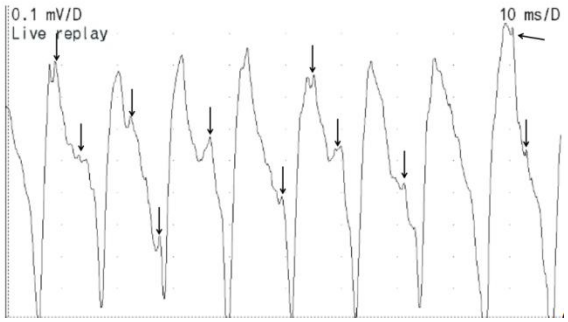

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Figure 1-3. Right rectus femoris (horizontal bar 10 ms/D, vertical bar 0.1mV/D). This represents a small section of the train of positive waves within myotonic discharge depicted in Figure 1-2. A complete positive wave is visible, and the arrows denote the presence of negative phase waves, each exhibiting varying frequencies and shapes.

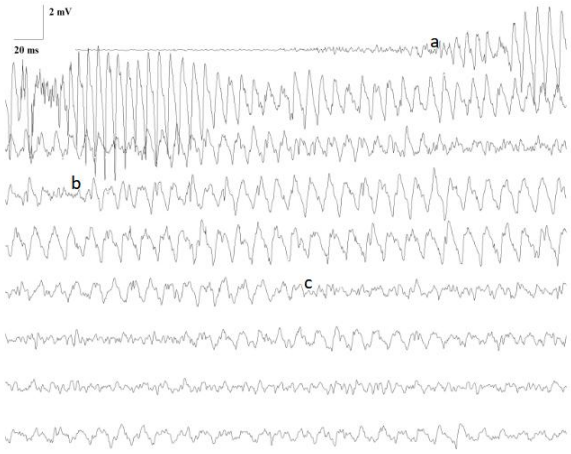

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Figure 2-1. Right rectus femoris (horizontal bar 20ms/D, vertical bar 2mV/D). a: Train of giant-amplitude of myotonic discharge displaying waxing-waning patterns in frequency (100-150Hz) and amplitude (3-15mV); b: Train of positive waves within myotonic discharge, with frequency (100-150Hz) and amplitude (0.5-3mV); c: Train of irregular waves within myotonic discharge, characterized by irregular frequency and waveform, with amplitude (0.2-2mV), and challenging frequency estimation.

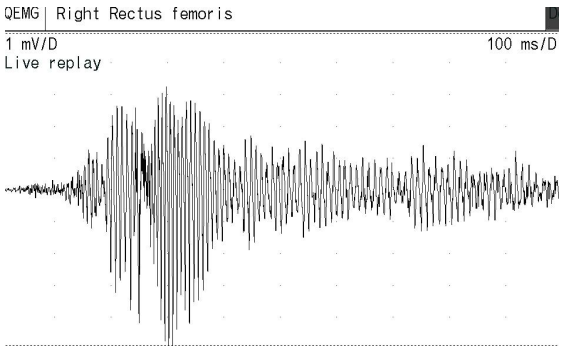

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Figure 2-2. Right rectus femoris (horizontal bar 100ms/D, vertical bar 1mV/D). Sequence of giant-amplitude myotonic discharge with sudden initiation and cessation.

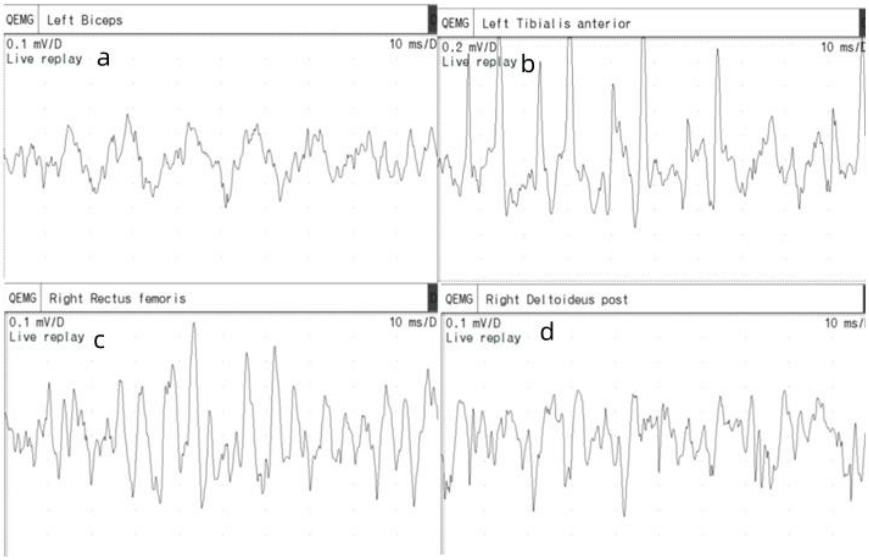

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Figure 3. Trains of irregular waves within myotonic discharge across different muscles. a: left biceps, b: Left tibialis anterior, c: Right rectus femoris, d:right deltoideus post.

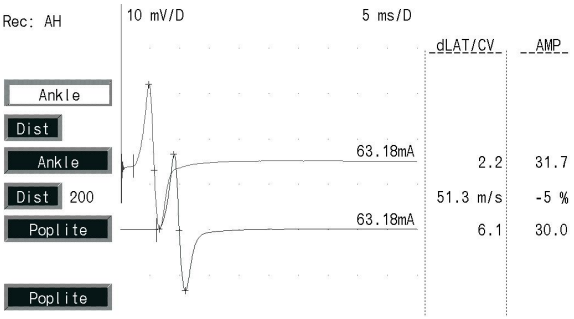

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Figure 4. motor nerve conduction velocity (MCV) and compound muscle actionpotential (CMAP) of the right tibial nerve

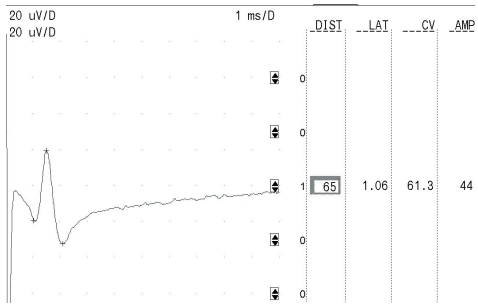

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Figure 5. sensory conduction velocity (SCV) and sensory nerve action potential(SNAP) of the left median nerve

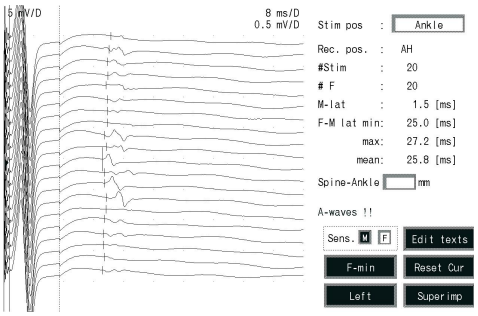

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Figure 6. F-wave of the right tibial nerve

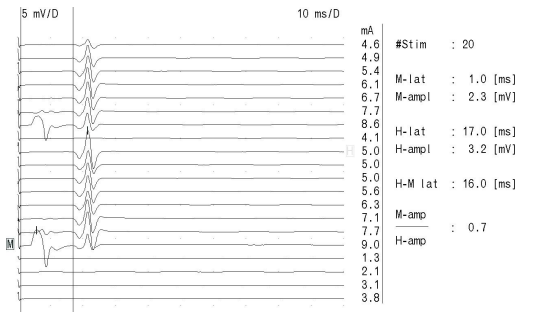

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Figure 7. H -reflex of the right tibial nerve

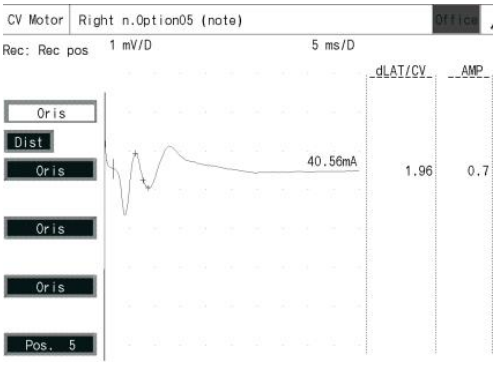

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Figure 8-1. compound muscle actionpotential (CMAP) and motor nerve conduction velocity(MCV) of the facial nerve

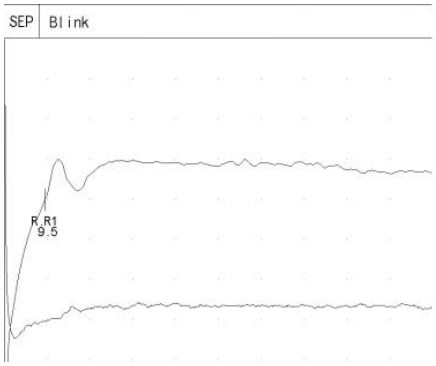

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Figure 8-2. Blink reflex(at sedation)

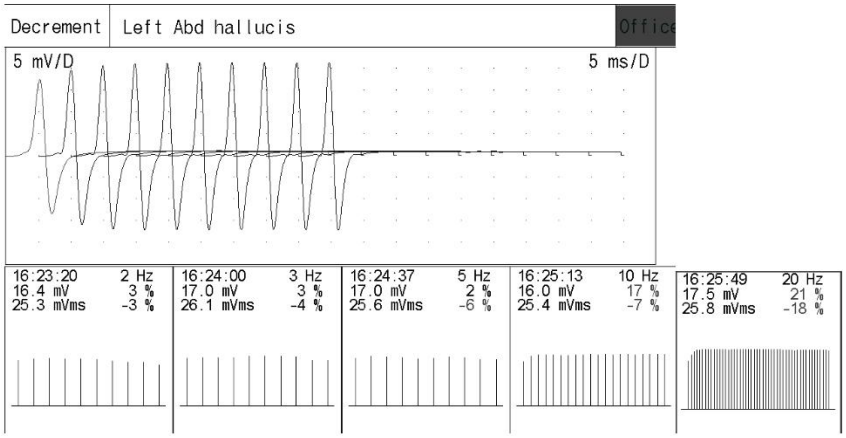

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Figure 8-3. repetitive frequency electrical stimulation (RNS) results of the left abductor hallucis digitorum (Low-frequency [2Hz, 3Hz, 5Hz] and high-frequency [10Hz, 20Hz] were normal.)

PMC is a sodium channelopathy of skeletal muscle caused by mutations in the SCN4A gene. EMG can confirm the presence, severity and distribution of myotonic discharge, which can support the diagnosis of PMC, and determine whether or not a patient has myopathy. Paradoxical myotonia is the typical feature of PMC. Nearly all patients' EMG showing myotonic potentials in previous studies[26-28]. CMAP decreased following exposure to cold or cold water tests in some patients[29-31]. Additionally, some people may also show a decrease in CMAP after a short period of exercise[27]. According to a report in China, one patient with PMC had delayed nerve conduction velocity and low F-wave appearance in both lower limbs[32]. If patients with PMC has not yet experienced clinical symptoms, their EMG were normal[33]. According to the studies by S Zhang and J Song[32, 34], EMG in some patients with PMC exhibited both myotonic and myopathic potentials. In presenting

this case of PMC with unique potentials identified via needle EMG, we underscore two distinctive features: high-amplitude potentials and irregular waves. These features are linked to the synchronous activation of multiple muscle fibers and the impairment of sodium channel inactivation. By sharing this case, our intent is to assist clinicians in distinguishing this type of myotonic discharge from neurological myotonia.