

# **Reply to the editor's comments**

Reviewer Number 06400042	Original comments of the editor	Reply by the author(s)	Changes done on page number and line number
1	In line 103, fever in his right knee should be changed to hotness of his right knee.	We have completed the modification.	Page 5, Line 102
2	In line 112, There was no muscle amyotrophy should be corrected to There was no muscle atrophy.	We have completed the modification.	Page 5, Line 123
3	In line 112, fibrillation should be corrected to fasciculation, clawfoot to claw-feet.	We have completed the modification.	The "fasciculation" was deleted. claw-feet (Page 7, Lin 163)
4	The following sentence (There was no muscle amyotrophy, muscle tract fibrillation, and abnormal involuntary movement. He was intellectually normal and had no ataxia, cognitive impairment, peripheral neuropathy, bladder dysfunction, clawfoot, and scoliosis) have English a grammar problem regarding the use of negative sentence?	We have completed the modification. We made a new edit of this paragraph.	Page 7, Line162- Line 165
5	In line 135 the following sentence (weakness and spasticity of both lower limbs in his 40 years and had required a cane to walk in his 60 years) should be corrected to (weakness and spasticity of both lower limbs at age 40 years and at age 60 years he start using cane to walk)	We have completed the modification. We made a new edit of this paragraph.	Page 6-7, Line151-Line 156
6	In line 255 what you mean by (avoiding	We have completed the	Page 10, Line 258-Line 263

	neuro-disease-inducing substances)? it should be changed to appropriate pharmacological term.	modification. We made a new edit of this paragraph.	
7	The examination of his nervous system showed increased muscle tension? here what you mean by muscle tension, did you mean tone?	We have completed the modification. We made a new edit of this paragraph.	Page 5, Line121-Line 123
8	in line 110, you should explain what type of muscle weakness does the patient had? is it proximal, distal or pyramidal? please you have to re-write the physical examination using appropriate neurological terms with detailed neurological examination.	We have completed the modification. We made a new edit of this paragraph.	Page 5 - 6, Line 117-Line 114
9	Can you please upload the lab. result for the studied gene with manuscript files.	I uploaded reports from all those performing genetic testing.	See the Supplementary Material for the related reports.

Reviewer Number 004010431	Original comments of the editor	Reply by the author(s)	
1	The SPRS scale evaluation should be included in the text and summarized in the Table. Moreover, bioinformatic methods should be reported in details (Mutation Taster, Polyphen-2, SIFT, Provean). See Carosi et al,	1 、 The evaluation of the SPRS scale has been summarized in the table. 2、 Mutation Taster assessed the novel mutation to be pathogenic	1、 Table is in Word document And explained. 2、 P7, Line 163 - 165.

	JNNP 2015 (see Suppl. File).		
2	The English language needs an improvement.	We have obtained the English language editing certificate and have uploaded.	

Company editor-in-chief	Original comments of the editor	Reply by the author(s)	
1	However, the quality of the English language of the manuscript does not meet the requirements of the journal. Before final acceptance, the author(s) must provide the English Language Certificate issued by a professional English language editing company.	We have obtained the English language editing certificate and have uploaded.	
2	Before its final acceptance, the author(s) must provide the Signed Consent for Treatment Form(s) or Document(s)	We have completed this work	
3	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.	We have completed this work. Figure, tables and text are in PowerPoint.	
4	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	We determined as the raw data.	

	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).		
5	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.	Copyright information has been added	
6	Authors are required to provide standard three-line tables, that is, only the top line, bottom line, and column line are displayed, while other table lines are hidden. The contents of each cell in the table should conform to the editing specifications, and the lines of each row or column of the table should be aligned. Do not use carriage returns or spaces to replace lines or vertical lines and do not segment cell content.	Three-line tables have been made and provided as required	

7	Please upload the approved grant application form(s) or funding agency copy of any approval document(s).	We are able to provide it and have uploaded it.	
---	--	---	--