Author's response to reviews

Title: Co-existence of Phenylketonuria and Fabry Disease on a 3 year-old boy: Case report

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Author's response to reviews: see over
Dear Editor,

thank you for your help regarding our manuscript “Co-existence of Phenylketonuria and Fabry Disease on a 3 year-old boy: Case report”. We modified the paper according to the suggestions of the reviewers as follows:

REVIEWER 1

Comments

Page 1: We inserted “Giuseppe” Bonapace instead of “G.” Bonapace. We wrote all authors with full first name except “G. Bonapace” for error.

Page 2: We corrected “hypoidrosis” with “hypohidrosis/hyperhidrosis” because in FD may be present an abnormal pattern of sweating and added the suggested reference

We modified “symptoms appear...group” in “...symptoms may appear...”

We inserted the sentence “FD diagnosis is confirmed by enzyme/genetic analysis...”

Page 3: We replaced abbreviations “DHPR, AGA, EMA, tTG” with their full name “Dihydropterine reductase, antigliadin antibodies, antiendomisium antibodies, tissue transglutaminase”.

We included “At clinical examination no angiokeratomas were found” because during examination no cutaneous markers of FD were observed in our patient.

REVIEWER 2

We modified the genetic part of the work as follows:

Case presentation section:

Page 4, end of the first paragraph : we included, as recommended, a short family history and a Fig.1 which shows the family tree, and we wrote: “Family history was unremarkable for FD except for a maternal uncle deceased at the age of 53 years of cerebral stroke, and a grandmother suffers of systemic hypertension. A maternal niece has developed
severe renal failure at birth and underwent to renal transplantation at the age of 19 years for polycystic kidneys (Fig.1).”

Page 4 (6 line of the first paragraph): we corrected “p.[L48S]” with “p.L48S” and corrected the sentence as following “……homozygous for c.143 T>C mutation on exon 2 that changes in the protein Lys with Ser (p.L48S)”

Page 4 (3 line of the second paragraph): we corrected “p.[R112H]” with “p.R112H” and corrected the sentence as following “……mutation g.5234G>A 2 in the exon 2 that changes in the protein Arg with Hys (p.R112H)”

Page 4 (2 line of the second paragraph): we deleted the word “GLA”

Page 4 (end of paragraph 2): we inserted the sentences “…..and mutation analysis to brothers, sisters, nephews of the mother that resulted normal. The grandmother (II.2) and a brother (III.1) of the mother refused to undergo to molecular analysis (Fig.1)” to explain which members of the maternal side of the pedigree were tested for enzymatic activity.

We hope that the revised paper is now suitable for the publication as short report.

I look to hearing soon from you
Best wishes

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