Author’s response to reviews

Title: SCL20A2 mutation presenting with acute ischemic stroke: a case report

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Responses to reviewers

Reviewer 1

1. Page 5, line 82: the sentence starts with ?And?, and sounds grammatically not clear.

Reply: (line 83) He presented with agitation when he was awaken.

2. Page 7, line 141: the sentence starts with ?And?, and sounds grammatically not clear.

Reply: (line 157-158) A small number of these arteries even exhibited obstruction of the lumen because of severe calcification.

3. Page 9, line 177: ?may attribute to? could be changed in ?may be attributed to?  

Reply: (line 191-193) Furthermore, PFBC patient can present with cognitive impairment which may be attributed to structural and functional network changes of frontotemporal lobar.

Reviewer 2

1. Term "neuropsychiatric" disease is not appropriate, it would be better to use "disease with neurological and psychiatric symptoms".

Reply: (line 29, 50, 125) “a rare neuropsychiatric disorder” has been changed as “a rare disorder”.
2. Please explain the whole diagnostic workup, including diagnostic workup for (cardio)embolic etiology of stroke.

Reply: (line 107-110) Sinus rhythm was showed on electrocardiogram. Mural thrombus was not detected in ultrasound cardiogram. We hypothesized that ischemic stroke may be related with hypoperfusion.

3. Please explain the reason for mRS of 2 with NIHSS of 1

Reply: (line 118-121) The score of NIHSS was one point (correctly answered one question when he was asked his age and the name of the current month) and modified Rankin Scale score was two points (unable to carry out all pre-stroke activities, such as working as a farmer).

4. The theoretic explanation of stenotic changes of intracranial arteries might not applicable for this case. CTangiography did not show any significant stenotic changes, and the bilateral localisation suggest other ethiology rather than thrombotic, probably haemodinamic (hypoperfusion?). Please explain all possible mechanisms of developement of stroke in this case.

Reply: (line 43-44) Ischemic stroke can occur in PFBC patients, which may be associated with hypoperfusion and calcification of arteries.

(line 169-173) In our case, hypovolemia caused by gastrointestinal bleeding may result in hypoperfusion of brain and cause acute ischemic stroke in bilateral basal ganglia and periventricular regions. We also excluded other etiologies which may cause bilateral multiple ischemic stroke such as embolism, cerebral amyloid angiopathy and immunologically-mediated small vessel diseases.

5. In discussion, please include other differential diagnostic possibilities for the clinical picture of the patient, as neuroimaging (which you have included in the paper) can also be interpreted differently.

Reply: (line 144-150) In clinical practice, other causes of intracranial calcification should be distinguished from PFBC, including physiological calcification, disorders of calcium metabolism (hypoparathyroidism, pseudo-hypoparathyroidism, pseudo-pseudo-hypoparathyroidism and hyperparathyroidism), mitochondrial diseases, severe hypomagnesemia, HIV infection and other toxic conditions. Detailed medical histories and laboratory tests are helpful in determining the causes of intracranial calcification.

Reviewer 3

1. Authors didn't give any information about patient's parents: Are they living? with or without any neurological problem? If they are dead, what is the cause of death?
Is there any other family member (uncle? aunt? grandmother/father et cet) with neurological problems, and/or stroke?

One of his daughters has brain calcification without any clinical symptom. How old is she?

Reply: (line 97-100) Family history revealed that one of his three daughters (34 years old) had also suffered from brain calcification, but without neurological problems (Fig. 3). His parents and two brothers also have no neurological problems. His father died of cardiac disease 10 years ago and his mother died of cancer 13 years ago.