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Case Report

COL4A1 with Cerebral Small Vessel Disease and Multiple Anomalous cerebral vascular origins: A case report

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Abstract

Cerebral small vessel disease (cSVD) is a main cause of stroke, cognitive impairment, and vascular dementia. COL4A1 mutations have been identified as a cause of hereditary cerebral small vessel disease. COL4A1 mutation is associated with nephropathy, aneurysms, and muscle cramps, which we call the HANAC syndrome. Mutations in *COL4A1* have recently been identified in both a mouse model and families, characterized by cystic brain cavities and cerebral white-matter lesions. But it has not been reported with vascular malformations. We here first report a case of COL4A1 with cerebral small vessel disease and multiple anomalous cerebral vascular origins.

Key words: COL4A1; cerebral small vessel disease (cSVD); stroke ; vascular origins.

Introduction:

The case was a 43-year-old man who had stroke in 2014 when he was 36 years old. The case had no special past medical history. The initial presentation was in 2014.The MRI shown acute lacunar infarction in the pons. His farther dead when 40 more years old because of cerebral infarction. His brother got infarction when 35years old. He got difficult in language in September 2021 and was referred to our hospital for further therapy. High-throughput sequencing was performed for the case by JINAN KINGMED CENTER FOR CLINICAL LABORATORY(Jinan,China). Brain magnetic resonance image(MRI) showed cSVD. The CT angiography of head and neck shown multiple anomalous cerebral vascular origins.

Genetic Analysis

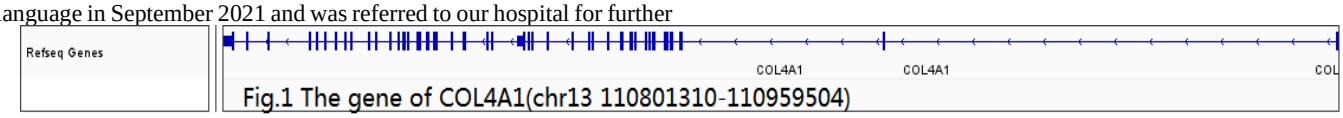

Figure 1. *High-throughput sequencing showed duplication and recombination occurred in the whole COL4A1.*

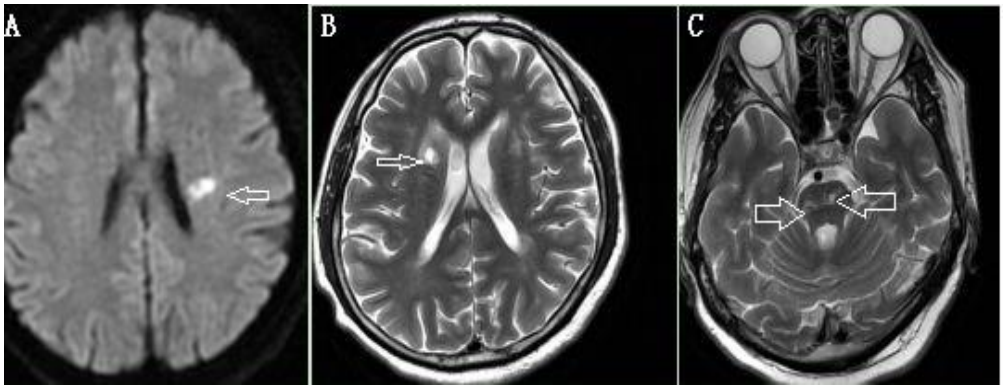

Figure 2. *The DWI findings acute infarct in the left corona radiata(A,DWI).The MRI T2 also showed multiple lacunar infarcts in the corona radiata and corona pans(B and C, T2-weighted).*

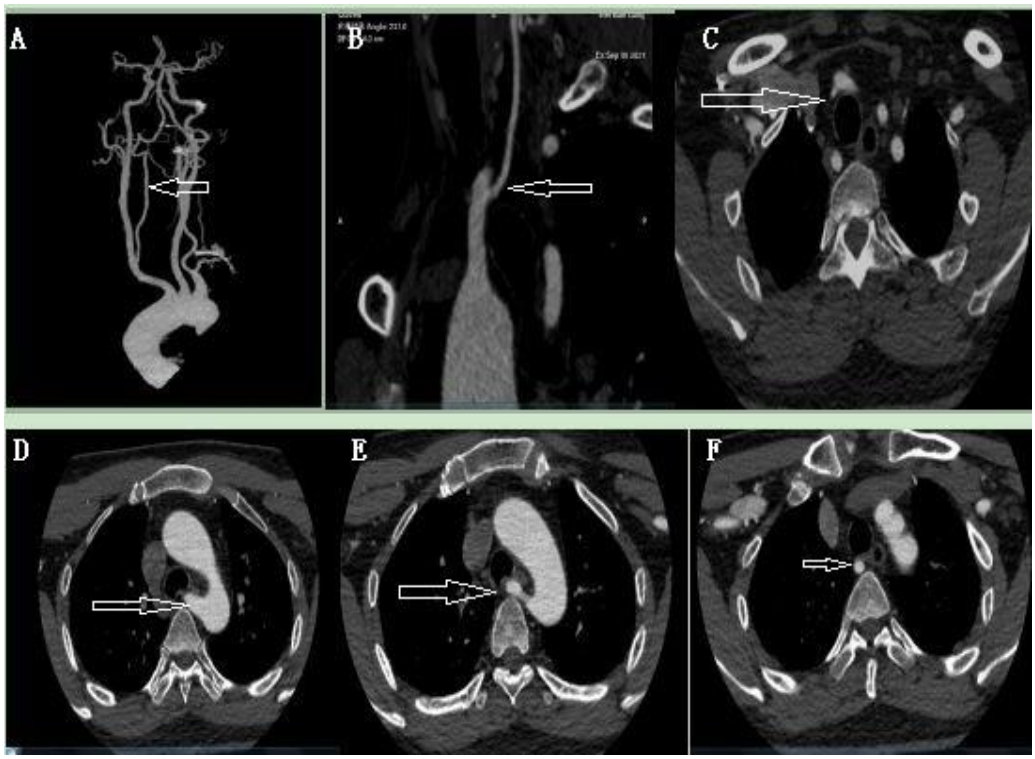

Figure 3 *The CT angiography of the head and neck findings the right vetebral artery arise dierctly form the right common carotid artery with finding no right subclavian artery(A and B CTA). The axial CTA findings the origin of the right vertebaral artery(C). The axial CTA findings the course of the right subclavian artery arosing directely from the thoracic aorta which also called aberrant right subclavian artery(ARSA) (D,E and F).*

Discussion

To our knowledge, the COL4A1 mutations can cause HANAC syndrome, which may attributed to the procollagen type IV α1 [1,2] The widespread expression of the α1.α1.α2(IV) network suggests that *COL4A1* mutations may lead to a systemic phenotype [3].

Studies have shown that the COL4A1 mutations are associated with cerebral small vessel disease (cSVD), defined as lacunar infarcts, deep intracerebral hemorrhages (ICH), and leukoaraiosis. MRI brain showed lacunar infarcts in the deep white matter and pons in our patient.

There is some reported that it related to fetal vascular origins of schizencephaly[4].

ARSA is an uncommon congenital anomaly of the aortic arch with incidence of 0.02 to 1.7%.5 It is rare that the right vertebral artery arise directly from the right common carotid artery. It also reported that the ARSA associated with Genetic Abnormalities [6]. It has not been reported of the coexistence of the two deformities. We hypothesized that the two deformities may not be accidence but rather result from the COL4A1 mutations.

Conclusion: We describe a patient with cSVD thought to be related to a novel COL4A2 variant. CTA finding multiple anomalous cerebral vascular origins. This has not been reported and it may deserves further study.

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