

World Journal of *Clinical Cases*

World J Clin Cases 2022 June 26; 10(18): 5934-6340



MINIREVIEWS

- 5934** Development of clustered regularly interspaced short palindromic repeats/CRISPR-associated technology for potential clinical applications
Huang YY, Zhang XY, Zhu P, Ji L
- 5946** Strategies and challenges in treatment of varicose veins and venous insufficiency
Gao RD, Qian SY, Wang HH, Liu YS, Ren SY
- 5957** Diabetes mellitus susceptibility with varied diseased phenotypes and its comparison with phenome interactome networks
Rout M, Kour B, Vuree S, Lulu SS, Medicherla KM, Suravajhala P

ORIGINAL ARTICLE

Clinical and Translational Research

- 5965** Identification of potential key molecules and signaling pathways for psoriasis based on weighted gene co-expression network analysis
Shu X, Chen XX, Kang XD, Ran M, Wang YL, Zhao ZK, Li CX
- 5984** Construction and validation of a novel prediction system for detection of overall survival in lung cancer patients
Zhong C, Liang Y, Wang Q, Tan HW, Liang Y

Case Control Study

- 6001** Effectiveness and postoperative rehabilitation of one-stage combined anterior-posterior surgery for severe thoracolumbar fractures with spinal cord injury
Zhang B, Wang JC, Jiang YZ, Song QP, An Y

Retrospective Study

- 6009** Prostate sclerosing adenopathy: A clinicopathological and immunohistochemical study of twelve patients
Feng RL, Tao YP, Tan ZY, Fu S, Wang HF
- 6021** Value of magnetic resonance diffusion combined with perfusion imaging techniques for diagnosing potentially malignant breast lesions
Zhang H, Zhang XY, Wang Y
- 6032** Scar-centered dilation in the treatment of large keloids
Wu M, Gu JY, Duan R, Wei BX, Xie F
- 6039** Application of a novel computer-assisted surgery system in percutaneous nephrolithotomy: A controlled study
Qin F, Sun YF, Wang XN, Li B, Zhang ZL, Zhang MX, Xie F, Liu SH, Wang ZJ, Cao YC, Jiao W

- 6050** Influences of etiology and endoscopic appearance on the long-term outcomes of gastric antral vascular ectasia

Kwon HJ, Lee SH, Cho JH

Randomized Controlled Trial

- 6060** Evaluation of the clinical efficacy and safety of TST33 mega hemorrhoidectomy for severe prolapsed hemorrhoids

Tao L, Wei J, Ding XF, Ji LJ

- 6069** Sequential chemotherapy and icotinib as first-line treatment for advanced epidermal growth factor receptor-mutated non-small cell lung cancer

Sun SJ, Han JD, Liu W, Wu ZY, Zhao X, Yan X, Jiao SC, Fang J

Randomized Clinical Trial

- 6082** Impact of preoperative carbohydrate loading on gastric volume in patients with type 2 diabetes

Lin XQ, Chen YR, Chen X, Cai YP, Lin JX, Xu DM, Zheng XC

META-ANALYSIS

- 6091** Efficacy and safety of adalimumab in comparison to infliximab for Crohn's disease: A systematic review and meta-analysis

Yang HH, Huang Y, Zhou XC, Wang RN

CASE REPORT

- 6105** Successful treatment of acute relapse of chronic eosinophilic pneumonia with benralizumab and without corticosteroids: A case report

Izhakian S, Pertzov B, Rosengarten D, Kramer MR

- 6110** Pembrolizumab-induced Stevens-Johnson syndrome in advanced squamous cell carcinoma of the lung: A case report and review of literature

Wu JY, Kang K, Yi J, Yang B

- 6119** Hepatic epithelioid hemangioendothelioma after thirteen years' follow-up: A case report and review of literature

Mo WF, Tong YL

- 6128** Effectiveness and safety of ultrasound-guided intramuscular lauromacrogol injection combined with hysteroscopy in cervical pregnancy treatment: A case report

Ye JP, Gao Y, Lu LW, Ye YJ

- 6136** Carcinoma located in a right-sided sigmoid colon: A case report

Lyu LJ, Yao WW

- 6141** Subcutaneous infection caused by *Mycobacterium abscessus* following cosmetic injections of botulinum toxin: A case report

Deng L, Luo YZ, Liu F, Yu XH

- 6148** Overlapping syndrome of recurrent anti-N-methyl-D-aspartate receptor encephalitis and anti-myelin oligodendrocyte glycoprotein demyelinating diseases: A case report
Yin XJ, Zhang LF, Bao LH, Feng ZC, Chen JH, Li BX, Zhang J
- 6156** Liver transplantation for late-onset ornithine transcarbamylase deficiency: A case report
Fu XH, Hu YH, Liao JX, Chen L, Hu ZQ, Wen JL, Chen SL
- 6163** Disseminated strongyloidiasis in a patient with rheumatoid arthritis: A case report
Zheng JH, Xue LY
- 6168** CYP27A1 mutation in a case of cerebrotendinous xanthomatosis: A case report
Li ZR, Zhou YL, Jin Q, Xie YY, Meng HM
- 6175** Postoperative multiple metastasis of clear cell sarcoma-like tumor of the gastrointestinal tract in adolescent: A case report
Huang WP, Li LM, Gao JB
- 6184** Toripalimab combined with targeted therapy and chemotherapy achieves pathologic complete response in gastric carcinoma: A case report
Liu R, Wang X, Ji Z, Deng T, Li HL, Zhang YH, Yang YC, Ge SH, Zhang L, Bai M, Ning T, Ba Y
- 6192** Presentation of Boerhaave's syndrome as an upper-esophageal perforation associated with a right-sided pleural effusion: A case report
Tan N, Luo YH, Li GC, Chen YL, Tan W, Xiang YH, Ge L, Yao D, Zhang MH
- 6198** Camrelizumab-induced anaphylactic shock in an esophageal squamous cell carcinoma patient: A case report and review of literature
Liu K, Bao JF, Wang T, Yang H, Xu BP
- 6205** Nontraumatic convexal subarachnoid hemorrhage: A case report
Chen HL, Li B, Chen C, Fan XX, Ma WB
- 6211** Growth hormone ameliorates hepatopulmonary syndrome and nonalcoholic steatohepatitis secondary to hypopituitarism in a child: A case report
Zhang XY, Yuan K, Fang YL, Wang CL
- 6218** Vancomycin dosing in an obese patient with acute renal failure: A case report and review of literature
Xu KY, Li D, Hu ZJ, Zhao CC, Bai J, Du WL
- 6227** Insulinoma after sleeve gastrectomy: A case report
Lobaton-Ginsberg M, Sotelo-González P, Ramirez-Renteria C, Juárez-Aguilar FG, Ferreira-Hermosillo A
- 6234** Primary intestinal lymphangiectasia presenting as limb convulsions: A case report
Cao Y, Feng XH, Ni HX
- 6241** Esophagogastric junctional neuroendocrine tumor with adenocarcinoma: A case report
Kong ZZ, Zhang L

- 6247** Foreign body granuloma in the tongue differentiated from tongue cancer: A case report
Jiang ZH, Xu R, Xia L
- 6254** Modified endoscopic ultrasound-guided selective N-butyl-2-cyanoacrylate injections for gastric variceal hemorrhage in left-sided portal hypertension: A case report
Yang J, Zeng Y, Zhang JW
- 6261** Management of type IIb dens invaginatus using a combination of root canal treatment, intentional replantation, and surgical therapy: A case report
Zhang J, Li N, Li WL, Zheng XY, Li S
- 6269** Clivus-involved immunoglobulin G4 related hypertrophic pachymeningitis mimicking meningioma: A case report
Yu Y, Lv L, Yin SL, Chen C, Jiang S, Zhou PZ
- 6277** De novo brain arteriovenous malformation formation and development: A case report
Huang H, Wang X, Guo AN, Li W, Duan RH, Fang JH, Yin B, Li DD
- 6283** Coinfection of *Streptococcus suis* and *Nocardia asiatica* in the human central nervous system: A case report
Chen YY, Xue XH
- 6289** Dilated left ventricle with multiple outpouchings – a severe congenital ventricular diverticulum or left-dominant arrhythmogenic cardiomyopathy: A case report
Zhang X, Ye RY, Chen XP
- 6298** Spontaneous healing of complicated crown-root fractures in children: Two case reports
Zhou ZL, Gao L, Sun SK, Li HS, Zhang CD, Kou WW, Xu Z, Wu LA
- 6307** Thyroid follicular renal cell carcinoma excluding thyroid metastases: A case report
Wu SC, Li XY, Liao BJ, Xie K, Chen WM
- 6314** Appendiceal bleeding: A case report
Zhou SY, Guo MD, Ye XH
- 6319** Spontaneous healing after conservative treatment of isolated grade IV pancreatic duct disruption caused by trauma: A case report
Mei MZ, Ren YF, Mou YP, Wang YY, Jin WW, Lu C, Zhu QC
- 6325** Pneumonia and seizures due to hypereosinophilic syndrome – organ damage and eosinophilia without synchronisation: A case report
Ishida T, Murayama T, Kobayashi S
- 6333** Creutzfeldt-Jakob disease presenting with bilateral hearing loss: A case report
Na S, Lee SA, Lee JD, Lee ES, Lee TK

LETTER TO THE EDITOR

- 6338** Stem cells as an option for the treatment of COVID-19
Cuevas-González MV, Cuevas-González JC

ABOUT COVER

Editorial Board Member of *World Journal of Clinical Cases*, Cristina Tudoran, PhD, Assistant Professor, Department VII, Internal Medicine II, Discipline of Cardiology, "Victor Babes" University of Medicine and Pharmacy Timisoara, Timisoara 300041, Timis, Romania. cristina13.tudoran@gmail.com

AIMS AND SCOPE

The primary aim of *World Journal of Clinical Cases* (WJCC, *World J Clin Cases*) is to provide scholars and readers from various fields of clinical medicine with a platform to publish high-quality clinical research articles and communicate their research findings online.

WJCC mainly publishes articles reporting research results and findings obtained in the field of clinical medicine and covering a wide range of topics, including case control studies, retrospective cohort studies, retrospective studies, clinical trials studies, observational studies, prospective studies, randomized controlled trials, randomized clinical trials, systematic reviews, meta-analysis, and case reports.

INDEXING/ABSTRACTING

The WJCC is now indexed in Science Citation Index Expanded (also known as SciSearch®), Journal Citation Reports/Science Edition, Scopus, PubMed, and PubMed Central. The 2021 Edition of Journal Citation Reports® cites the 2020 impact factor (IF) for WJCC as 1.337; IF without journal self cites: 1.301; 5-year IF: 1.742; Journal Citation Indicator: 0.33; Ranking: 119 among 169 journals in medicine, general and internal; and Quartile category: Q3. The WJCC's CiteScore for 2020 is 0.8 and Scopus CiteScore rank 2020: General Medicine is 493/793.

RESPONSIBLE EDITORS FOR THIS ISSUE

Production Editor: *Ying-Yi Yuan*, Production Department Director: *Xu Guo*, Editorial Office Director: *Jin-Lei Wang*.

NAME OF JOURNAL

World Journal of Clinical Cases

ISSN

ISSN 2307-8960 (online)

LAUNCH DATE

April 16, 2013

FREQUENCY

Thrice Monthly

EDITORS-IN-CHIEF

Bao-Gan Peng, Jerzy Tadeusz Chudek, George Kontogeorgos, Maurizio Serati, Ja Hyeon Ku

EDITORIAL BOARD MEMBERS

<https://www.wjgnet.com/2307-8960/editorialboard.htm>

PUBLICATION DATE

June 26, 2022

COPYRIGHT

© 2022 Baishideng Publishing Group Inc

INSTRUCTIONS TO AUTHORS

<https://www.wjgnet.com/bpg/gerinfo/204>

GUIDELINES FOR ETHICS DOCUMENTS

<https://www.wjgnet.com/bpg/GerInfo/287>

GUIDELINES FOR NON-NATIVE SPEAKERS OF ENGLISH

<https://www.wjgnet.com/bpg/gerinfo/240>

PUBLICATION ETHICS

<https://www.wjgnet.com/bpg/GerInfo/288>

PUBLICATION MISCONDUCT

<https://www.wjgnet.com/bpg/gerinfo/208>

ARTICLE PROCESSING CHARGE

<https://www.wjgnet.com/bpg/gerinfo/242>

STEPS FOR SUBMITTING MANUSCRIPTS

<https://www.wjgnet.com/bpg/GerInfo/239>

ONLINE SUBMISSION

<https://www.f6publishing.com>



De novo brain arteriovenous malformation formation and development: A case report

Huan Huang, Xue Wang, An-Na Guo, Wei Li, Ren-Hua Duan, Jun-Hao Fang, Bo Yin, Dan-Dong Li

Specialty type: Clinical neurology

Provenance and peer review:

Unsolicited article; Externally peer reviewed.

Peer-review model: Single blind

Peer-review report's scientific quality classification

Grade A (Excellent): A

Grade B (Very good): 0

Grade C (Good): C

Grade D (Fair): 0

Grade E (Poor): 0

P-Reviewer: Almzeogi MA, Serbia;
Medina SM, Bolivia

Received: January 13, 2022

Peer-review started: January 13, 2022

First decision: March 23, 2022

Revised: April 1, 2022

Accepted: April 27, 2022

Article in press: April 27, 2022

Published online: June 26, 2022



Huan Huang, Xue Wang, An-Na Guo, Department of Radiology, The Second Affiliated Hospital and Yuying Children's Hospital of Wenzhou Medical University, Wenzhou 325000, Zhejiang Province, China

Wei Li, Ren-Hua Duan, Jun-Hao Fang, Bo Yin, Dan-Dong Li, Department of Neurosurgery, The Second Affiliated Hospital and Yuying Children's Hospital of Wenzhou Medical University, Wenzhou 325000, Zhejiang Province, China

Corresponding author: Dan-Dong Li, MD, Neurosurgeon, Department of Neurosurgery, The Second Affiliated Hospital and Yuying Children's Hospital of Wenzhou Medical University, No. 108 Xueyuan West Road, Wenzhou 325000, Zhejiang Province, China.

andonglmn@163.com

Abstract

BACKGROUND

Brain arteriovenous malformation (AVM), an aberrant vascular development during the intrauterine period, is traditionally considered a congenital disease. Sporadic reports of cases of *de novo* AVM formation in children and adults have challenged the traditional view of its congenital origin.

CASE SUMMARY

In this report, we have presented the case of a child with a *de novo* brain AVM. Magnetic resonance imaging and magnetic resonance angiography of the brain showed no AVM at the age of 5 years and 2 mo. Brain AVM was first detected in this child at the age of 7 years and 4 mo. The brain AVM was significantly advanced, and hemorrhage was seen for the first time at the age of 12 years and 8 mo. There was further progression in the AVM, and hemorrhage occurred again at the age of 13 years and 5 mo. Genetic analysis of this patient revealed a mutation in the *NOTCH2* (p.Asp473Val) gene.

CONCLUSION

In short, our case has once again confirmed the view that brain AVM is an acquired disease and is the result of the interaction of genes, environment, and molecules.

Key Words: *De novo* arteriovenous malformation; Angiogenesis; Hemorrhage; *NOTCH2*; Case report

Core Tip: Brain arteriovenous malformation (AVM) is one of the main causes of spontaneous cerebral hemorrhage in children. At present, the mechanism of the occurrence and development of AVM is not clear, and there have been very few case reports that have documented the progression of a *de novo* brain AVM. In this report, we present the case of a child with a *de novo* brain AVM. Brain AVM was first detected in this child at the age of 7 years and 4 mo and was significantly advanced and hemorrhaging at the age of 12 years and 8 mo. Genetic analysis of this patient revealed a mutation in the *NOTCH2* (p.Asp473Val) gene.

Citation: Huang H, Wang X, Guo AN, Li W, Duan RH, Fang JH, Yin B, Li DD. *De novo* brain arteriovenous malformation formation and development: A case report. *World J Clin Cases* 2022; 10(18): 6277-6282

URL: <https://www.wjgnet.com/2307-8960/full/v10/i18/6277.htm>

DOI: <https://dx.doi.org/10.12998/wjcc.v10.i18.6277>

INTRODUCTION

Brain arteriovenous malformation (AVM) is one of the main causes of spontaneous cerebral hemorrhage in children[1] and accounts for 30%-50% of spontaneous cerebral hemorrhage in children[1,2]. It is generally believed that AVM is a kind of congenital vascular disease[3]. The abnormality is formed in the third to fourth week of embryonic development, when the length is about 40-80 mm, during the process of cerebrovascular development and manifests as abnormal coiled and interlaced vascular clusters[4]. Arteries and veins are directly connected by malformed vascular clusters. At present, the mechanism of the occurrence and development of AVM is not clear, and there have been very few case reports that have documented the progression of a *de novo* brain AVM[5-7]. Here, we have documented for the first time a case of a child with a *de novo* brain AVM that progressed and ruptured twice within 6 years. We have also reported for the first time the potential role of heterozygous mutations in the *NOTCH2* gene in the pathogenesis of AVM.

CASE PRESENTATION

Chief complaints

A 15-year-old boy was admitted to the hospital in November 2019 with complaints of an acute headache for 3 h.

History of present illness

The child presented with a sudden severe headache for 3 h with no inducing factors. He was conscious and had no history of nausea, dizziness, or fever.

History of past illness

The boy had normal growth and development and no history of other special diseases. In August 2011, the child presented at another hospital with complaints of a headache for more than 1 wk. At that time, magnetic resonance imaging (MRI), magnetic resonance angiography (MRA), and magnetic resonance venography (MRV) were performed. The scans revealed that there was no vascular abnormality, abnormal signal intensity, or any other findings suggestive of an AVM (Figure 1A-C). At that time, the doctor did not administer any treatment, and the headache was ameliorated by itself. In October 2013, the child again presented at another hospital with headache and nausea for 2 d. His MRA scan revealed a small right temporal AVM, the size of which was 6.2 mm × 1.9 mm (Figure 1C). Digital subtraction angiography revealed the presence of a suspicious lesion in the right temporal lobe (Figure 1D). The boy was treated for headaches and was not examined further. In February 2019, the boy presented at our hospital complaining of a sudden headache and vomiting for 2 h. He was conscious when he came to our hospital. His Glasgow coma scale score was 14, and National Institutes of Health Stroke Scale score was 1. Computed tomography of the head demonstrated acute intracerebral hemorrhage in the right temporal lobe. The MRA revealed a small AVM in the right temporal lobe (Figure 1E). Symptomatic treatment was given to the child during hospitalization, and gamma knife radiosurgery was suggested. Unfortunately, the child's parents rejected radiotherapy.

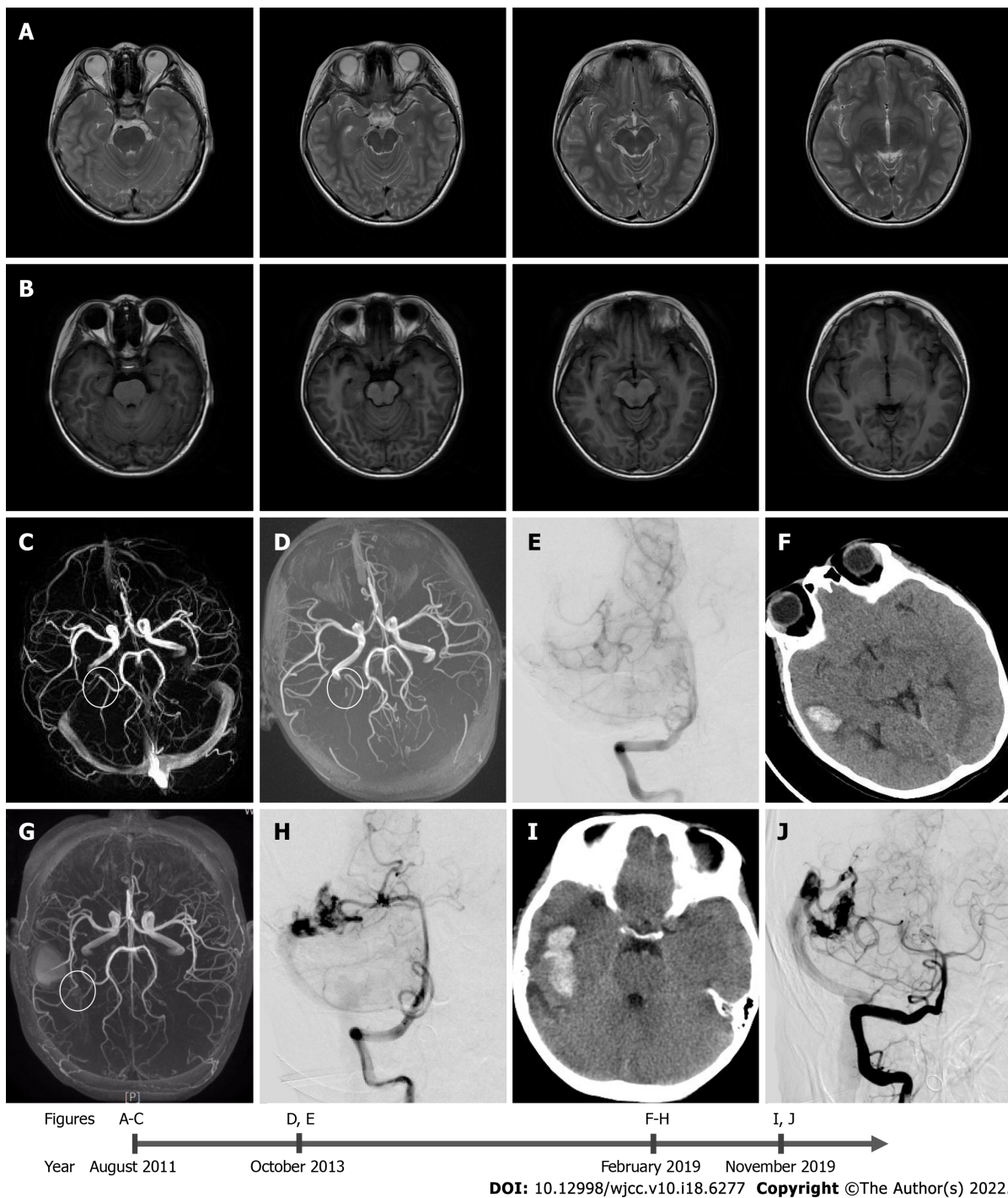


Figure 1 Diagnostic images from August 2011 to November 2019. A-C: T1 (A) and T2 (B) weighted images of magnetic resonance imaging, and (C) magnetic resonance angiography (MRA) done in August 2011, at the age of 5 years and 2 mo. There is no evidence of vascular lesion; D and E: The results of (D) MRA and (E) the first anteroposterior digital subtraction angiography (DSA) performed in October 2013, indicating *de novo* arteriovenous malformation; F-H: The results of (F) the head computed tomography (CT) scan, (G) MRA, and (H) the second DSA, which were performed in February 2019; I and J: Results from (I) the post-hemorrhage non-contrast head CT scan depicting blood in the right temporal lobe and (J) the third DSA, which were performed in November 2019.

Personal and family history

The patient did not have any personal or family history of brain AVM.

Physical examination

The patient was conscious when examined at our hospital in November 2019. His Glasgow coma scale score was 15 and his National Institutes of Health Stroke Scale score was 1.

Laboratory examinations

Laboratory examinations showed no significant abnormality.

Imaging examinations

A small AVM within the right temporal lobe was visualized in the MRA (Figure 1G), and cerebral angiogram revealed the presence of a 21.6 mm × 21.2 mm right temporal AVM, which was supplied by the inferior temporal branches of the right posterior cerebral artery. Venous drainage occurred through superficial cerebellar veins into the right sigmoid sinus. It was classified as Spetzler-Martin grade 1 (Figure 1H).

In November 2019, computed tomography of the head demonstrated the presence of an acute intracerebral hemorrhage in the right temporal lobe (Figure 1I). Digital subtraction angiography displayed abnormal vascular clusters, which were slightly larger than those seen 10 mo ago. The size of these clusters was 24.3 mm × 22.8 mm. The AVM was classified as a Spetzler-Martin grade 1 (Figure 1J).

Further diagnostic work-up

After receiving consent from the child's parents, we conducted genetic analysis for the child and his parents. We discovered a mutation that might be related to the incidence of AVM, *NOTCH2* heterozygous mutation [NM_024408.3; c.1418A>T; p.Asp473Val]. The mutation was only found in the child.

FINAL DIAGNOSIS

Brain AVM, Spetzler-Martin grade 1.

TREATMENT

The parents refused to conduct further interventional treatment. Supportive treatment was administered during the hospitalization period, and the boy was discharged after 10 d in good clinical condition.

OUTCOME AND FOLLOW-UP

The clinical condition of the child was good during the final follow-up, which was performed on January 2021. The child was in good general condition at the time. After January 2021, he was lost to follow-up.

DISCUSSION

The patient underwent MRI, MRA, and MRV scans in August 2011, at the age of 5 years and 2 mo for nonspecific headaches. No abnormal vascular lesions or secondary signs of brain AVM were observed. A brain AVM was first identified in October 2013. Bleeding occurred for the first time in February 2019 and occurred again in November 2019. The malformed vascular clusters progressed significantly during this period. No risk factors were associated with AVM in this case. Literature review revealed that most of the reported AVM cases had underlying pathology, and only a few of them had no cause of *de novo* AVM[5,8]. Our case report is the first to define the progression and eventual rupture within 6 years following its primary discovery. We also report for the first time that heterozygous mutations in the *NOTCH2* gene might be playing a role in the pathogenesis of AVM.

Currently, the pathogenesis of brain AVM is not clearly understood. The imbalance of some signal molecules during embryonic development is believed to be the most likely reason for no capillary formation between the arteries and veins[9]. This balance is dependent on the strict regulation of various angiogenic factors, and any interference for these automatic regulation factors may lead to the formation of AVM[10,11]. There was no high-risk factor for AVM in this case, and the only high-risk factor that may have a relation in the development of AVM was the heterozygous mutation of the *NOTCH2* gene. The *NOTCH2* protein is the receptor of the Notch signaling pathway, and its activity can directly affect Notch signaling[12]. However, the Notch signaling pathway has complex and context-dependent effects on angiogenesis[13,14]. Studies have demonstrated that abnormal activation of Notch signaling in human brain AVM was associated with AVM bleeding[15]. The inhibition and activation of Notch signaling were both associated with AVM formation. The mutation in this case (p.Asp473Val) was first described by Gilbert *et al*[16]. The mutation is located in exon 8, and exon 8 encodes the epidermal growth factor-12 domain of *NOTCH2*[16,17]. Unfortunately, there has been no research on the effect of this mutation on the function of the *NOTCH2* protein.

CONCLUSION

We report for the first time a case of *de novo* AVM formation in a child, which progressed and eventually ruptured within 6 years. Our case contests the traditional view that brain AVM is congenital, and our case once again confirms the view that brain AVM is an acquired disease that is the result of an interaction of genes, environment, and molecules.

ACKNOWLEDGEMENTS

We thank the Department of Neurosurgery and the Department of Radiology of The Second Affiliated Hospital and Yuying Children's Hospital of Wenzhou Medical University for the diagnosis and treatment of the patient.

FOOTNOTES

Author contributions: Li DD was in charge of case review and preparation of the manuscript; Huang H, Wang X, Guo AN, and Li W collected clinical opinions regarding this case and drafted the manuscript; Duan RH, Fang JH, and Yin B participated in the coordinating the manuscript; Huang H and Li DD revised the manuscript; All authors read and approved the final manuscript.

Supported by the Science and Technology Program of Wenzhou, China, No. Y20190145 to Huan Huang; and the Beijing New Health Industry Development Foundation, No. XM2020-02-002 to Bo Yin.

Informed consent statement: Written informed consent was obtained from the patient's parents.

Conflict-of-interest statement: The authors declare there are no conflicts of interest.

CARE Checklist (2016) statement: The authors have read the CARE Checklist (2016), and the manuscript was prepared and revised according to the CARE Checklist (2016).

Open-Access: This article is an open-access article that was selected by an in-house editor and fully peer-reviewed by external reviewers. It is distributed in accordance with the Creative Commons Attribution NonCommercial (CC BY-NC 4.0) license, which permits others to distribute, remix, adapt, build upon this work non-commercially, and license their derivative works on different terms, provided the original work is properly cited and the use is non-commercial. See: <https://creativecommons.org/licenses/by-nc/4.0/>

Country/Territory of origin: China

ORCID number: Huan Huang 0000-0001-8842-2424; Xue Wang 0000-0001-6178-6316; An-na Guo 0000-0003-3631-1401; Wei Li 0000-0002-3401-0077; Ren-Hua Duan 0000-0003-2776-7060; Jun-Hao Fang 0000-0002-1395-0756; Bo Yin 0000-0002-5487-0197; Dan-Dong Li 0000-0003-4973-8818.

S-Editor: Gong ZM

L-Editor: Filipodia

P-Editor: Gong ZM

REFERENCES

- 1 Sison V, Stackhouse T, Breeze R, Hall T, McKenzie P, Tartaglia N. Arteriovenous Malformation in a Youth with Atypical Autism Symptoms. *J Child Dev Disord* 2017; **3** [PMID: 28989994 DOI: 10.4172/2472-1786.100042]
- 2 Jimenez JE, Gersey ZC, Wagner J, Snelling B, Ambekar S, Peterson EC. Role of follow-up imaging after resection of brain arteriovenous malformations in pediatric patients: a systematic review of the literature. *J Neurosurg Pediatr* 2017; **19**: 149-156 [PMID: 27911246 DOI: 10.3171/2016.9.PEDS16235]
- 3 Leblanc GG, Golanov E, Awad IA, Young WL; Biology of Vascular Malformations of the Brain NINDS Workshop Collaborators. Biology of vascular malformations of the brain. *Stroke* 2009; **40**: e694-e702 [PMID: 19834013 DOI: 10.1161/STROKEAHA.109.563692]
- 4 Mullan S, Mojtahedi S, Johnson DL, Macdonald RL. Embryological basis of some aspects of cerebral vascular fistulas and malformations. *J Neurosurg* 1996; **85**: 1-8 [PMID: 8683257 DOI: 10.3171/jns.1996.85.1.0001]
- 5 Santos R, Aguilar-Salinas P, Entwistle JJ, Aldana PR, Beier AD, Hanel RA. De Novo Arteriovenous Malformation in a Pediatric Patient: Case Report and Review of the Literature. *World Neurosurg* 2018; **111**: 341-345 [PMID: 29294397 DOI: 10.1016/j.wneu.2017.12.145]
- 6 Chen W, Choi EJ, McDougall CM, Su H. Brain arteriovenous malformation modeling, pathogenesis, and novel therapeutic targets. *Transl Stroke Res* 2014; **5**: 316-329 [PMID: 24723256 DOI: 10.1007/s12975-014-0343-0]

- 7 **Florian IA**, Beni L, Moisoiu V, Timis TL, Florian IS, Balaşa A, Berindan-Neagoe I. 'De Novo' Brain AVMs-Hypotheses for Development and a Systematic Review of Reported Cases. *Medicina (Kaunas)* 2021; **57** [PMID: [33652628](#) DOI: [10.3390/medicina57030201](#)]
- 8 **Yeo JJ**, Low SY, Seow WT, Low DC. Pediatric de novo cerebral AVM: report of two cases and review of literature. *Childs Nerv Syst* 2015; **31**: 609-614 [PMID: [25537110](#) DOI: [10.1007/s00381-014-2609-y](#)]
- 9 **Storkebaum E**, Quaegebeur A, Vikkula M, Carmeliet P. Cerebrovascular disorders: molecular insights and therapeutic opportunities. *Nat Neurosci* 2011; **14**: 1390-1397 [PMID: [22030550](#) DOI: [10.1038/nn.2947](#)]
- 10 **Achrol AS**, Guzman R, Varga M, Adler JR, Steinberg GK, Chang SD. Pathogenesis and radiobiology of brain arteriovenous malformations: implications for risk stratification in natural history and posttreatment course. *Neurosurg Focus* 2009; **26**: E9 [PMID: [19409010](#) DOI: [10.3171/2009.2.FOCUS0926](#)]
- 11 **Ma L**, Guo Y, Zhao YL, Su H. The Role of Macrophage in the Pathogenesis of Brain Arteriovenous Malformation. *Int J Hematol Res* 2015; **1**: 52-56 [PMID: [26495437](#) DOI: [10.17554/j.issn.2409-3548.2015.01.12](#)]
- 12 **Wang MM**. Notch signaling and Notch signaling modifiers. *Int J Biochem Cell Biol* 2011; **43**: 1550-1562 [PMID: [21854867](#) DOI: [10.1016/j.biocel.2011.08.005](#)]
- 13 **Gridley T**. Notch signaling in vascular development and physiology. *Development* 2007; **134**: 2709-2718 [PMID: [17611219](#) DOI: [10.1242/dev.004184](#)]
- 14 **Murphy PA**, Lu G, Shiah S, Bollen AW, Wang RA. Endothelial Notch signaling is upregulated in human brain arteriovenous malformations and a mouse model of the disease. *Lab Invest* 2009; **89**: 971-982 [PMID: [19546852](#) DOI: [10.1038/labinvest.2009.62](#)]
- 15 **Wang LJ**, Xue Y, Huo R, Yan Z, Xu H, Li H, Wang J, Zhang Q, Cao Y, Zhao JZ. N6-methyladenosine methyltransferase METTL3 affects the phenotype of cerebral arteriovenous malformation via modulating Notch signaling pathway. *J Biomed Sci* 2020; **27**: 62 [PMID: [32384926](#) DOI: [10.1186/s12929-020-00655-w](#)]
- 16 **Gilbert MA**, Bauer RC, Rajagopalan R, Grochowski CM, Chao G, McEldrew D, Nassur JA, Rand EB, Krock BL, Kamath BM, Krantz ID, Piccoli DA, Loomes KM, Spinner NB. Alagille syndrome mutation update: Comprehensive overview of JAG1 and NOTCH2 mutation frequencies and insight into missense variant classification. *Hum Mutat* 2019; **40**: 2197-2220 [PMID: [31343788](#) DOI: [10.1002/humu.23879](#)]
- 17 **Duan Z**, Li FQ, Wechsler J, Meade-White K, Williams K, Benson KF, Horwitz M. A novel notch protein, N2N, targeted by neutrophil elastase and implicated in hereditary neutropenia. *Mol Cell Biol* 2004; **24**: 58-70 [PMID: [14673143](#) DOI: [10.1128/MCB.24.1.58-70.2004](#)]



Published by **Baishideng Publishing Group Inc**
7041 Koll Center Parkway, Suite 160, Pleasanton, CA 94566, USA

Telephone: +1-925-3991568

E-mail: bpgoffice@wjgnet.com

Help Desk: <https://www.f6publishing.com/helpdesk>

<https://www.wjgnet.com>

