

Space-Occupying Intraventricular Vascular Lesion in Tuberous Sclerosis Complex

To the Editor:

We read your *Neurologist* case report “Hemorrhage into a subependymal giant cell astrocytoma in an adult with tuberous sclerosis” published in July 2021,¹ and found it very interesting. In the article, Barbiero and colleagues presented a case of intratumoral hemorrhage of a subependymal giant cell astrocytoma (SEGA), which is a rare phenomenon, in

an adult diagnosed with tuberous sclerosis complex (TSC). In fact, there is a similar case of TSC of our own whereby we are glad to discuss the disease further in this short letter.

TSC is a rare condition that can affect virtually any organ in the body,² first reported in 1862 by von Recklinghausen,³ tentative diagnostic criteria had been made and improved now and then based on the

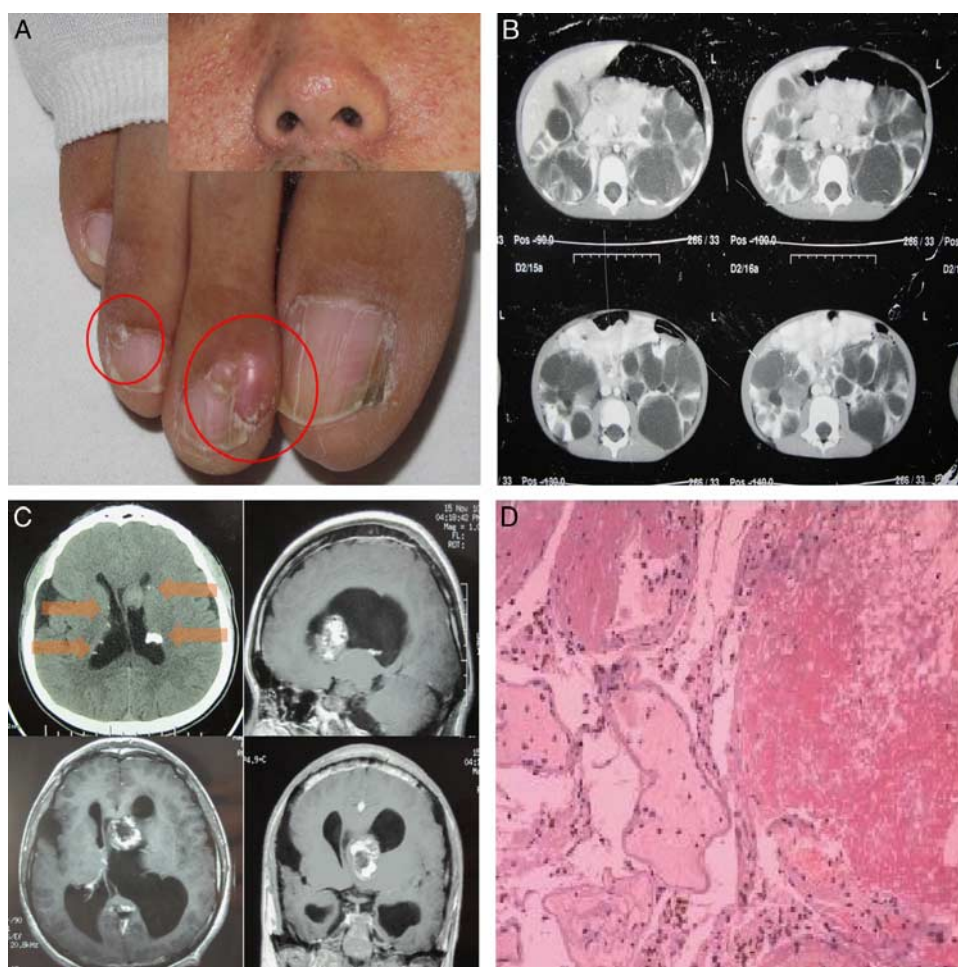


FIGURE 1. Clinical findings, radiology, and pathologic results of the patient. (A) Multiple facial angiofibromas and ungual fibromas (red circles). (B) Bilateral renal cysts, which support the diagnosis of tuberous sclerosis complex. (C) Subependymal nodules (orange arrows) in computed tomography and a space-occupying lesion inside the ventricle causing obstructive hydrocephalus with heterogenous enhancement on magnetic resonance imaging. (D) Pathologic test showed (intraventricular) vascular lesion with hemorrhage and clots, partially organized, in which hemosiderosis was observed. With dispersive distribution of lymphocytes, plasmocytes, and phagocytes, and absence of tumor cells, a final diagnosis of intraventricular vascular lesion with fresh and remote hemorrhage was determined. This is a very uncommon nature for a tuberous sclerosis complex–related lesion. Vimentin(+), glial fibrillary acidic protein(–), NF(–), NeuN(–), SYN(–), factor VIII, and CD34(+), Ki-67(–).

The authors declare no conflict of interest.

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ISSN: 2331-2637/22/2704-0222

DOI: 10.1097/NRL.0000000000000415

progression of our understanding about this peculiar disease. The *Updated Diagnostic Criteria for TSC 2012* is the most recent and generally accepted program for TSC diagnosis to date.² Subependymal nodules (SENs) and SEGA are considered to be the major characteristics of intracranial lesions of TSC and were both explicitly made diagnostic on the rare genetic disease in this criteria.² Taking no account of the correlation between SEN and SEGA for now, however, an interesting non-SEGA but SEGA-like intraventricular vascular lesion, assumed vascular malformation, has never been reported and it is controversial with current understanding about TSC.

We would like to share an unique case of a tumor-like intraventricular vascular lesion, instead of SEGA, in a TSC patient. In 2010, a 19-year-old boy desperately sought surgical treatment of a “brain tumor” for intermittent attacks of seizures over the past 11 years, which were marginally controlled by multiple medications yet remained repeatedly recurrent. Visually he had facial angiofibromas and unguinal fibromas (Fig. 1A) that were fairly recognizable, meanwhile, computed tomography and magnetic resonance imaging revealed multiple renal cysts of both sides (Fig. 1B) as well as SENs in his head (Fig. 1C). These findings were amply in compliance with the 2012 Criteria with presence of at least 3 major (facial angiofibromas, unguinal fibromas, and SENs) and 1 minor clinical features (renal cysts). However, a space-occupying lesion near left foramen of Monro led us to a diagnosis of SEGA which seemed the only rational reflection. Fresh, preoperative magnetic resonance imaging at that time indicated heterogeneous enhancement within the “tumor” as well as prominent growth compared to 7 years before then (Fig. 1C). A frontal transcortical tumorectomy was scheduled several days later, which went well with a complete resection of the lesion. No signs of epileptic relapse were found postoperatively. However, unfortunately, the patient suffered from an infection of the central nervous system which gave us no choice for betterment but to engage a

sustained lumbar cistern drainage, which proved worked subsequently.

Surprisingly, a pathologic test showed unexpected results that the majority of the specimen consisted of vasculopathy but tumoral cells (Fig. 1D). Clotted blood and abnormal vascular plexus were found at the site, pathologic diagnosis was determined as an intraventricular vascular lesion with hemorrhage in the end and the patient was discharged before long, with optimistic expectancy of fine recovery. The patient is well till publishing, without any signs for recurrence of the intraventricular vascular lesion, after a follow-up over 10 years.

We regret a genetic screening was not conducted regarding *TSC1/TSC2* mutations to aid in confirmation of TSC but the clinical findings were just sufficient for the diagnosis: This is a TSC patient. But the question goes, is the mass in his head a SEGA?

In this case, we believe the pathologists have ultimate priority to say the nature of this “tumor”, and when the fact goes “wrong,” it is time we must challenge the traditional ideas. Both SENs and SEGAs were extensively reported and frequently detected prenatally or at birth,² but usually before the age of 20 years.⁴ Up to 80% TSC patients had SENs while SEGAs are less common (5% to 15%) and it is believed that SEGA derives from SENs in many cases.⁴ It is widely accepted that SEGAs typically arise from SENs, especially near the foramen of Monro.² Our patient had SENs but it does not mean the mass “has to” be a SEGA, not necessarily. Thus, a non-SEGA intraventricular lesion in a TSC patient, with hemorrhage, should add to diversification of central nervous system features of TSC as we consider. To say the least, hemorrhage within a SEGA is still exceedingly rare.¹

The report of our case is a rare phenomenon in a rare, complicated disease. It argued that intracranial lesions of TSC may be diversified, not limited to SEN or SEGA, with mechanisms awaiting exploring. More concerns shall be

arisen for this matter. As neurosurgeons and neurologists, we might as well give some reinspection to the essence of TSC’s central nervous system manifestations and apprehending its natural history, conceivably.

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ACKNOWLEDGMENTS

The surgical procedure mentioned in this case was performed on November 26, 2010, by Dr Yazhuo Zhang, Dr Chuzhong Li, and Dr Hongtao Lv at Beijing Tiantan Hospital, where Dr Hongtao Lv received his resident training and finished his PhD program. After achieving his doctoral degree, Dr Hongtao Lv had been working at The First Affiliated Hospital of Dalian Medical University since 2011. Professor Rongyao Liu, Director of Neurosurgery, The First Affiliated Hospital of Dalian Medical University, provided general support during the preparation of this manuscript.

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