History of Disease Entity and Diagnosis Criteria

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Abstract

This chapter reviews the history of this mysterious moyamoya disease. Since the 1950s, this rare disease has been discovered in Japan and gradually gaining recognition around the world. It has been just 50 years since Suzuki and Takaku first published the term "moyamoya disease" in an English language journal in 1969. Looking back over the past 50 years, the author would like to review the state of affairs at the time when the causes and concepts of this disease were hotly debated. It may be helpful to us in the future when new diseases appear in front of us. It is no secret, however, that it is nearly impossible to write a history that will satisfy everyone. Therefore, the author appreciates it if the readers would kindly understand that this historical review is only from the author's point of view and they would forgive him.

In this chapter, the author describes the evolution of the diagnostic criteria for moyamoya disease. Since the diagnostic criteria for moyamoya disease were first established in 1979 by a Japanese research group, they have been revised approximately every 10 years in 1988, 1995, 2009, and 2018. The author would like to look back at that history and explore the background with the readers. Finally, the challenges of the diagnostic criteria and the prospects for the future will be discussed.

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S. Kuroda (ed.), *Moyamoya Disease: Current Knowledge and Future Perspectives*, https://doi.org/10.1007/978-981-33-6404-2_1

Keywords

Moyamoya disease · History · Diagnosis criteria

1.1 Introduction

Moyamoya disease is a very specific disease characterized by progressive stenosis of the terminal portion of the internal carotid artery and the formation of an abnormal network of dilated, fragile perforators around the basal ganglia, thalamus, and lateral ventricles. Moyamoya disease was first reported in Japan in the 1950s and has widely been recognized over the world. It occurs in children and adults, and presents with a variety of symptoms, including headache, TIA, ischemic stroke, intracranial bleeding, and involuntary movements. Initially, there was no effective treatment for the disease; however, since the late 1960s, surgical revascularization was found to be an effective treatment.

In this chapter, the author reviews the history of moyamoya disease and describes in detail the historical transition of the diagnosis criteria for moyamoya disease. The issues and future perspectives of the diagnosis criteria are also discussed.

1.2 History of Moyamoya Disease

The history of moyamoya disease has recently been described in the monographs written in English [1, 2]. In writing this chapter, however, the author tried to revisit the history of the moyamoya disease by reading the monographs, proceedings, and review articles from the 1960s. Based on previous review articles, Takeuchi and Shimizu reported a case of bilateral internal carotid arteries in 1955 and published it as a case report in 1957, which is believed as probably the first case of moyamoya disease on cerebral angiography that has appeared in the literature [3]. Since then, a significant number of similar cases were reported from Western countries as well as from Japan in the 1960s and 1970s. Interestingly, the first three cases reported from California were all Japanese Americans [4, 5].

At that time, various theories have been raised as to the cause of this peculiar cerebral angiography seen in moyamoya disease, including carotid artery hypoplasia [3], hypoplasia of the circle of Willis [6], cerebral juxta-basal telangiectasia [7], carotid artery occlusion [8], a network-like vascular anomaly of internal carotid artery [9], and angiomatous malformation [10, 11]. In addition to the papers cited here, many other reports were made in Japan at the time, which strongly suggests that Japanese neurologists and neurosurgeons were engaged in heated discussions about this novel, mysterious disease discovered in Japan in the 1960s and 1970s. However, many of these records are from the annual scientific meetings and their proceedings in Japan and are written in Japanese, and so the author must say that he only quotes original articles written in Japanese or English without citing them in

this book, which is likely to be read by readers in countries other than Japan. These valuable records are described in detail in a proceeding by Prof. Tatsuyuki Kudo from Keio University, Tokyo (1967) [12], a review article by Prof. Akira Nishimoto from Okayama University, Okayama (1979) [13] and a monograph by Prof. Jiro Suzuki from Tohoku University, Sendai (1983) [14]. So, the author would like to recommend to the readers who are interested in this topic to read them. Especially, the monograph edited by Prof. Suzuki has been translated and published in English, which will be of great help to the non-Japanese readers (Fig. 1.1).

In 1963, Suzuki et al. performed a detailed review of the findings on cerebral angiography in six cases of moyamoya disease, and concluded that these abnormal net-like vessels at the base of the brain are collateral blood vessels in response to the acquired and gradual narrowing of the terminal portion of the internal carotid artery at the 22nd Annual Meeting of the Japan Neurosurgical Society. At the same time, they also speculated that this group of cases belonged to a single, novel clinical entity. In fact, on the front page of the monograph mentioned above, he put his own words, "Moyamoya vessels are collateral pathways of the brain (1963)" along with the words by Dr. John Hunter in 1785, "Blood goes where it is needed." [14].

Around this time, Prof. Suzuki's concept began to be recognized by many neurologists and neurosurgeons, but the names of the disease still varied widely among them, including spontaneous occlusion of the circle of Willis [15, 16], cerebral arterial rete [17], Nishimoto disease, and Nishimoto-Takeuchi-Kudo disease [18–22]. In 1966, Suzuki and co-workers first proposed the term "moyamoya" disease in a Japanese written medical journal [23]. He described how he arrived at this naming in his monograph as follows: *"Moyamoya" was thought to be appropriate since the abnormal net-like vessels at the base of the brain, as seen in angiograms, are hazy in appearance – like a puff of cigarette smoke drifting in the air, which is commonly described as moyamoya by native speakers of Japanese. As described below,*

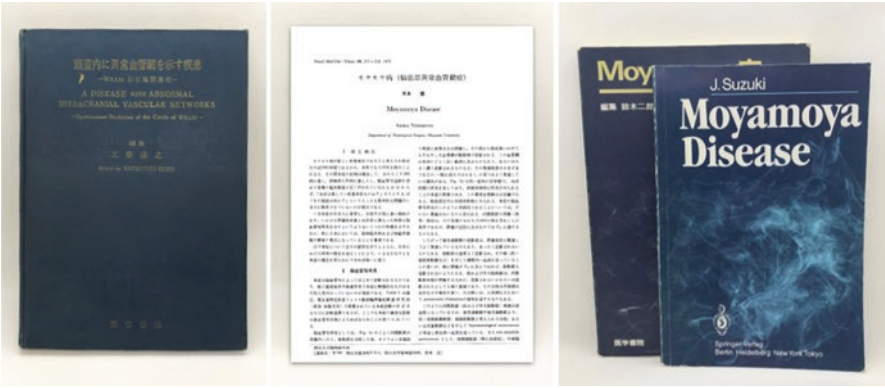

Fig. 1.1 History of moyamoya disease. (*Left*) a proceeding by Prof. Tatsuyuki Kudo from Keio University, Tokyo (1967) [12], (*Middle*) a review article by Prof. Akira Nishimoto from Okayama University, Okayama (1979) [13], and (*Right*) Japanese- and English-written monographs by Prof. Jiro Suzuki from Tohoku University, Sendai (1983) [14]

moreover, in follow-up angiograms of this disease in young patients, the sequential changes – from an initially thick "cloud", to a gradually thinning and shrinking "fog", to the eventual disappearance of the net-like vessels around the internal carotid artery – are also reminiscent of the misty "moyamoya" changes in cigarette smoke. He further stated as follow: *Certainly, moyamoya is an unusual word to enter into medical terminology, but it is appropriate for a number of reasons. It expresses the fact that its etiology is as hazy as the pattern of the abnormal vessels itself. Moreover, the disease was first reported in Japan and continues to show a high incidence there. Usage of a Japanese word emphasize these facts and has thus far been well received in the West as well as in Japan* [14]. Subsequently, Suzuki and Takaku published the first English written article entitled, "Cerebrovascular moyamoya disease, Disease showing abnormal net-like vessels in base of brain," using their new term "moyamoya." Since then, the term "moyamoya" is rapidly recognized worldwide because of its ease of use and euphony [24]. One of the authors, Prof. Akira Takaku was appointed to the University of Toyama as a professor and moved from Sendai to Toyama in 1980. Much later on, the author also moved to the same University of Toyama as a professor in 2012, and had the chance to ask Prof. Takaku about the backgrounds of the naming of moyamoya disease in the 1960s. The following is the narrative by Prof. Takaku: At that time, Prof. Suzuki was concerned that the term "moyamoya" was a Japanese term that was somehow familiar but not international and inappropriate as an academic term. However, he thought that since the disease was discovered in Japan, it would be appropriate to describe it in Japanese. He also thought that if he could properly explain why he named it as moyamoya disease, people overseas might understand it. He also considered that the rhythmic rhyming word "moyamoya" would be easy for foreigners to pronounce. Anyway, Prof. Suzuki was wondering if and how the four syllable words "moyamoya" would be received by foreigners. When Prof. Suzuki and Prof. Takaku submitted their paper to the *Archives of Neurology* journal, they initially chose the main title of their article as "Diseases showing abnormal net like vessels in base of brain," and added the subtitle "Cerebrovascular Moyamoya disease" with diffidence. However, Prof. Merritt, who was a chief editor of the journal at the time, insisted that "Cerebrovascular Moyamoya disease" should be the main title of the article. Prof. Takaku still believed that Prof. Merritt's wise decision was a major turning point in making the name "moyamoya disease" widely known throughout the world (cited from Annual Report 2013 of Department of Neurosurgery, University of Toyama). Very regretfully, Prof. Takaku passed away in 2015, so the author would like to write down Prof. Takaku's narrative here by translating it into English.

According to a review article by Nishimoto [13], however, there were other groups in Japan that called the disease as "chiri-chiri" disease rather than "moyamoya" disease at the same time. Nishimoto described that Maki and Nakada reported a 1-year-old girl and named it "chiri-chiri" disease in 1967 [25]. The Japanese term "chiri-chiri" means "curly" or "wrinkled," e.g., curly hair is called as "chiri-chiri hair" in Japanese; the people may have imagined that the blood vessels within the abnormal vascular networks were comparable to hair, rather than to cigarette smoke. So, in the late 1960s, there was a moyamoya—chiri-chiri controversy

in Japan. In fact, when the author started his residency program at the Department of Neurosurgery, Hokkaido University Hospital, Sapporo in 1986, he heard the same story from senior colleagues many times.

1.3 Diagnosis Criteria of Moyamoya Disease

In the 1970s, the Japanese Ministry of Health and Welfare organized a research group to investigate the causes of moyamoya disease and to establish treatment and prevention methods for the disease. First, the Research Committee on Vascular Abnormality in the Brain and Spinal Cord (headed by Prof. Katsutoshi Kitamura) was organized in 1974, and 189 cases of moyamoya disease were registered from all over Japan during the 2 years from 1974 to 1975. Then, in 1977, the research committee was reorganized and given a new name. Thus, the Research Committee on Spontaneous Occlusion of the Circle of Willis ("Moyamoya Disease"), headed by Prof. Fumio Gotoh, was newly established for the same purpose [13]. This research group first established the guideline for the diagnosis of spontaneous occlusion of the circle of Willis (moyamoya disease) in 1978 and published it in their annual report in 1979 [26]. This guideline consisted of the following structure: (1) a general description of moyamoya disease, (2) indispensable angiographic findings, (3) unknown cause and no special underlying diseases, and (4) pathological findings for diagnostic reference. The guidelines clearly stated that cerebral angiography is mandatory for diagnosis and that at least the findings shown in Table 1.1 must be present on cerebral angiography (Table 1.1). In this guideline, ***definite case*** must fulfill all three findings on cerebral angiography, plus other special underlying diseases must not be presented. On the other hand, ***probable case*** should fulfill two findings on cerebral angiography (2-a and 2-b), plus other special underlying diseases must not be presented. In other words, a definite case was defined as having bilateral lesions and a probable case was defined as having unilateral lesions. Since then, moyamoya disease has been diagnosed on the basis of morphological findings of the lumen of the affected arteries on cerebral angiography, which has been maintained to date.

This guideline was revised by the Research Committee, headed by Prof. Hajime Handa, in 1988 [27]. In the revised version, the possibility of familial occurrence of moyamoya disease was first noted in the general description. Specific examples of

Table 1.1 Cerebral angiographic findings to be presented for diagnosis moyamoya disease (An excerpt from the Guideline 1978) Ref. [13]. *English translation by the author*

Cerebral angiography is mandatory for diagnosis and at least the following findings must be present on cerebral angiography
a. Stenosis or occlusion at the terminal portion of the internal carotid artery and at the proximal portion of the anterior and the middle cerebral arteries.
b. Abnormal vascular networks in the vicinity of the occlusive or stenotic lesion on the arterial phase.
c. These findings should present bilaterally.

causative comorbidities that should be excluded when diagnosing moyamoya disease were presented. They included atherosclerosis, meningitis, brain tumor, Down syndrome, von Recklinghausen's disease, head trauma, and irradiation. In addition, pediatric cases with typical angiographic findings on one side and an apparent stenosis of the terminal portion of the contralateral internal carotid artery were classified into ***definite cases***, because these patients usually develop moyamoya vessels thereafter [28].

The guidelines for the diagnosis and treatment of spontaneous occlusion of the circle of Willis ("Moyamoya" disease) have been extensively revised in 1995 [29], and was published in English in 1997 by the Research Committee headed by Prof. Masashi Fukui [30]. The guideline consisted of (1) guidelines for the diagnosis, (2) general information, (3) treatment, and (4) guidelines for making a diagnosis using MRI and MRA. The most significant change in the diagnosis of moyamoya disease at that time was the emergence of MR angiography. Although cerebral angiography had been considered essential for the diagnosis of moyamoya disease until then, noninvasive MR angiography has been reported to be useful for the diagnosis of moyamoya disease since the early 1990s [31–33]. Yamada et al. (1992) reported that MR angiography can accurately detect occlusive lesions at the terminal portion of the internal carotid artery and associated moyamoya vessels, although some occlusive lesions were overestimated because of a flow-related artifact on MR [33]. Houkin et al. (1994) also analyzed the findings on MR angiography in 39 patients with moyamoya disease and found good correlations between cerebral angiography and MR angiography when detecting the stenotic lesions at the carotid fork and moyamoya vessels [32]. Numerous studies to date have recognized that MR angiography is an extremely useful modality not only for the diagnosis of moyamoya disease, but also for determining the stage of the disease and for observing the progression of the disease and collateral formation after surgical revascularization due to its noninvasive nature [34–39]. As a result, the 1997 revision of the guidelines for the diagnosis of moyamoya disease stated for the first time that the diagnosis of moyamoya disease can be made without cerebral angiography only if the findings on MR angiography meet certain criteria (Table 1.2). At the same time, the guidelines for making a diagnosis using MRI and MRA were also presented (Table 1.3) [30]. In addition, autoimmune disease was added as a comorbidity that should be ruled out when diagnosing moyamoya disease of unknown etiology [30].

Subsequently, the guidelines for diagnosis and treatment of moyamoya disease (spontaneous occlusion of the circle of Willis) were revised by the Research Committee on the Pathology and Treatment of Spontaneous Occlusion of the Circle of Willis, headed by Prof. Nobuo Hashimoto, in 2009 [40] and its English version was published in 2012 [41]. From this revised guideline, the world's preferred name of the disease, moyamoya disease, has been used as the official name of the disease, whereas the previous guideline used spontaneous occlusion of the circle of Willis as the official name of the disease, with moyamoya disease written in parentheses. The guideline consisted of eight chapters, including (1) concepts of the disease, (2) epidemiology, (3) pathology/etiology, (4) symptoms, (5) similar conditions, (6) diagnosis, (7) treatment, and (8) prognosis. Although the diagnostic criteria were not

Table 1.2 Guideline 1995 for the diagnosis of moyamoya disease (Fukui et al. 1997) Ref. [30]

1.1. Diagnosis criteria

(A) Cerebral angiography is indispensable for the diagnosis, and should present at least the following findings:

1. Stenosis or occlusion at the terminal portion of the internal carotid artery and/or at the proximal portion of the anterior and/or the middle cerebral arteries.
2. Abnormal vascular networks in the vicinity of the occlusive or stenotic lesions in the arterial phase.
3. These findings should present bilaterally.

(B) When magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) clearly demonstrate all the below-described findings, conventional cerebral angiography is not mandatory.

1. Stenosis or occlusion at the terminal portion of the internal carotid artery and at the proximal portion of the anterior and middle cerebral arteries on MRA.
2. An abnormal vascular network in the basal ganglia on MRA.

Note: An abnormal vascular network can be diagnosed when more than two apparent flow voids are seen in one side of the basal ganglia on MRI.

3. (1) and (2) are seen bilaterally (refer to the image diagnostic guideline by MRI and MRA).

(C) Because the etiology of this disease is unknown, cerebrovascular disease with the following basic diseases or conditions should thus be eliminated:

1. Atherosclerosis
2. Autoimmune disease
3. Meningitis
4. Brain neoplasm
5. Down syndrome
6. Recklinghausen's disease
7. Head trauma
8. Irradiation to the head
9. Others

(D) Instructive pathological findings:

1. Intimal thickening and the resulting stenosis or occlusion of the lumen are observed in and around the terminal portion of the internal carotid artery usually on both sides. Lipid deposits are occasionally seen in the proliferating intima.
2. Arteries constituting the circle of Willis such as the anterior and the middle cerebral and the posterior communicating arteries often show stenosis of various degrees of occlusion associated with fibrocellular thickening of the intima, a waving of the internal elastic lamina, and an attenuation of the media.
3. Numerous small vascular channels (perforators and anastomotic branches) are observed around the circle of Willis.
4. Reticular conglomerates of small vessels are often seen in the pia mater.

1.2. Diagnosis

In reference to 1.1 mentioned above, the diagnostic criteria are classified as follows: Autopsy case not undergoing cerebral angiography should be investigated separately while referring to (D).

1. ***Definite case***:

One which fulfills either (A) or (B) and (C). In children, however, a case which fulfills (A) (1) and (2) or (B) (1) and (2) on one side and with remarkable stenosis at the terminal portion of the internal carotid artery on the opposite side is also included.

2. ***Probable case***:

One which fulfills either (A) (1) and (2) or (B) (1) and (2) and (C) (unilateral involvement).

Table 1.3 Guideline 1995 for the usage of MRI and MRA to make a diagnosis of moyamoya disease (Fukui et al. 1997) Ref. [30]

Guidelines for making a diagnosis using MRI and MRA
(A) When magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) clearly demonstrate all the findings described below, conventional cerebral angiography: 1. Stenosis or occlusion at the terminal portion of the internal carotid artery and at the proximal portion of the anterior and middle cerebral arteries on MRA. 2. An abnormal vascular network in the basal ganglia. 3. (1) and (2) are seen bilaterally.
(B) Imaging methods and judgment 1. More than a 1.0-tesla magnetic field strength is recommended. 2. There are no restrictions regarding the MRA imaging method. 3. The imaging parameters, such as the magnetic field strength, the imaging methods and the use of contrast medium, should be clearly documented. 4. An abnormal vascular network can be diagnosed when more than two apparent flow voids are seen on one side of the basal ganglia on MRI. 5. Either an over- or underestimation of the lesion could be made according to the imaging conditions. To avoid a false-positive diagnosis, only definite cases should thus be diagnosed based on the MRI and MRA findings.
(C) Because similar vascular lesions secondary to other disorders are sometimes indistinguishable from this disease in adults, a diagnosis of MRI and MRA without conventional angiography is thus only recommended in childhood cases.

Table 1.4 Suzuki's angiographical stage Ref. [24]

Stage	Cerebral angiographic findings
I	Narrowing of the carotid fork
II	Initiation of the moyamoya (dilated major cerebral artery and a slight moyamoya vessel network)
III	Intensification of the moyamoya (disappearance of the middle and anterior cerebral arteries, and thick and distinct moyamoya vessels)
IV	Minimization of the moyamoya (disappearance of the posterior cerebral artery, and narrowing of individual moyamoya vessels)
V	Reduction of the moyamoya (disappearance of all the main cerebral arteries arising from the internal carotid artery system, further minimization of the moyamoya vessels, and an increase in the collateral pathways from the external carotid artery system)
VI	Disappearance of the moyamoya (disappearance of the moyamoya vessels, with cerebral blood flow derived only from the external carotid artery and the vertebrobasilar artery systems)

revised, Suzuki's angiographic stage (Table 1.4) [24] and MRA-based disease stage (Table 1.5) [36] were described for the first time in Chap. 6 "Diagnosis." In Chap. 7 "Treatment," the recommendation grade and evidence level are first described for each treatment in accordance with the development and spread of evidence-based medicine (EBM) in the medical community over the world, although the author will skip it here [41].

The most recent guideline was established in 2015 by The Research Committee on Spontaneous Occlusion of the Circle of Willis ("Moyamoya Disease"), headed by Prof. Kiyohiro Houkin and was published in Japanese in 2018 [42]. An English

Table 1.5 Houkin's MRA stage Ref. [36]

MRA findings	Score
(1) Internal carotid artery	
Normal	0
Stenosis of C1	1
Discontinuity of the C1 signal	2
Invisible	3
(2) Middle cerebral artery	
Normal	0
Stenosis of M1	1
Discontinuity of the M1 signal	2
Invisible	3
(3) Anterior cerebral artery	
Normal A2 and its distal	0
A2 and its distal signal decrease	1
Invisible	2
(4) Posterior cerebral artery	
Normal P2 and its distal	0
P2 and its distal signal decrease	1
Invisible	2
MRA total score	MRA stage
0–1	1
2–4	2
5–7	3
8–10	4

version has not been published officially. Its structure remains the same as the 2009 version, but there have been major revisions to the diagnosis criteria. First, the site of an occlusion or stenotic lesion was previously defined as "the terminal portion of the internal carotid artery and the proximal portion of the anterior and middle cerebral arteries," but the 2015 revision changed it to "*the arteries centered on the terminal portion of the intracranial internal carotid artery*." It is because there have been several reports of cases with occlusion or severe stenosis of the proximal portion of the middle cerebral artery associated with typical moyamoya vessels, but without obvious stenotic lesions at the terminal portion of the internal carotid artery. Previously, such cases were not diagnosed as moyamoya disease because they were not considered to meet the diagnostic criteria for moyamoya disease. However, Kim et al. (2016) collected 81 cases with an isolated middle cerebral artery stenosis or occlusion. All of them were younger than 60 years. According to the findings on high-resolution MRI (see Chap. 14), they were classified into atherosclerosis group (n = 45) and non-atherosclerosis group (n = 36). The latter was characterized by younger age, smaller number of vascular risk factors, thinner intima-media thickness, and more frequent mutation at *RNF213*, a susceptibility gene for moyamoya disease. More interestingly, Choi et al. (2012) reported a 25-year-old female with chronic headache due to an isolated middle cerebral artery stenosis on the right side. She was conservatively followed up and repeated cerebral angiography revealed the

isolated middle cerebral artery stenosis progressed to typical moyamoya disease [43]. These observations strongly suggest that stenotic lesions may start to develop at the proximal portion of the middle cerebral artery, but not at the terminal portion of the internal carotid artery in moyamoya disease. Based on these considerations, the pathogenesis of these cases is now thought very similar to that of moyamoya disease, but a significant number of such cases have not been diagnosed as moyamoya disease according to previous diagnostic criteria. Therefore, the Research Committee decided to revise the diagnosis criteria as written above in 2015.

Second, the 2015 edition eliminated the distinction between definite and probable cases. In the past, cases with bilateral stenotic lesions at the terminal part of the internal carotid artery and its branches were classified as ***definite*** cases and cases with unilateral stenotic lesions were classified as ***probable*** cases, but all cases are simply defined as moyamoya disease without distinction between definite cases and probable cases since 2015 [42]. It is based on increasing evidence that a significant number of unilateral moyamoya disease progress to bilateral moyamoya disease, especially in pediatric patients. They also share a similar genetic background (see Chap. 3) [44–46]. Therefore, the pathophysiology and genetics are recognized as very similar between bilateral and unilateral moyamoya disease, and the distinction between bilateral and unilateral cases has gradually become less significant.

Although different from the diagnostic criteria, this 2015 version of the diagnostic and treatment guidelines for moyamoya disease is notable for incorporating new findings throughout, including the revision to the treatment guidelines for hemorrhagic-type adult moyamoya disease in view of the recent series of studies of the Japan Adult Moyamoya (JAM) Trial. However, this 2018 edition has been published in Japanese only [42], while the English written version has not yet been officially released. Therefore, there is an urgent need to publish an English written version that should serve as the basis for discussions on the future development of a unified international moyamoya disease concept and diagnostic criteria over the world.

Finally, the author will also discuss future issues. As pointed out earlier, moyamoya disease is diagnosed based on the findings on cerebral angiography or MR angiography, i.e., the diagnosis completely depends on morphological information about the lumen of the affected arteries. Therefore, when encountering a patient with stenosis or occlusion of an intracranial artery due to other causes, it is not infrequently difficult to distinguish them from moyamoya disease. Particularly in patients older than 50 years, it is not uncommon to meet with difficulty in accurately diagnosing whether the stenotic lesion is caused by moyamoya disease or by atherosclerosis. This obscures the debate over whether *RNF213* mutants are involved in intracranial artery stenosis lesions other than moyamoya disease or not. Therefore, we need to further improve the accuracy of the diagnosis of moyamoya disease in the next revision.

Acknowledgment This work was partly supported by a grant from the Research Committee on Moyamoya Disease, the Japanese Ministry of Health, Labour and Welfare.

References

1. Tominaga T. Overview. Moyamoya disease update. Tokyo: Springer; 2010.
2. Yoshimoto T, Numagami Y, Shirane R. History and dedicated researchers. Moyamoya disease. Rolling Meadows: American Association of Neurological Surgeons; 2001.
3. Takeuchi K, Shimizu K. Hypoplasia of the bilateral internal carotid arteries. No To Shinkei. 1957;9:37–43.
4. Leeds NE, Abbott KH. Collateral circulation in cerebrovascular disease in childhood via rete mirabile and perforating branches of anterior choroidal and posterior cerebral arteries. Radiology. 1965;85:628–34.
5. Weidner W, Hanafeewmarkham CH. Intracranial collateral circulation via leptomeningeal and rete mirabile anastomoses. Neurology. 1965;15:39–48.
6. Kudo T, Takayama R, Mikawakuchi K, Ishimori S, Otake S, KNakagawa K. Occlusion of internal carotid artery. Brain Nerve (Tokyo). 1957;9:757.
7. Sano K. Cerebral juxta-basal telangiectasia. Brain Nerve (Tokyo). 1965;17:748–50.
8. Takeuchi K, Kobayashi S. Arterial occlusion at the base of brain in children. No To Shinkei 1965;17:779–80.
9. Moriyasu N, Nishio S. FIve cases of network like vascular anomaly of internal carotid artery. Brain Nerve (Tokyo). 1965;17:761–3.
10. Maki Y, Nakada Y. Autopsy cases showing angiomatous anomaly of carotid artery at the base of the brain. Brain Nerve (Tokyo). 1965;17:764–6.
11. Nishimoto A, Sugiu R, Takeuchi S. Hemangiomatous malformation of bilateral internal carotid artery at the base of brain. Brain Nerve (Tokyo). 1966;17:750–2.
12. Kudo T. Historical overview. In: Kudo T, editor. 25th congress of Japan neurosurgical society, vol. 1966. Tokyo: Igaku Shoin; 1967. p. 12–22.
13. Nishimoto A. Moyamoya disease(author's transl). Neurol Med Chir (Tokyo). 1979;19:221–8.
14. Suzuki J. Moyamoya disease. Berlin Heidelberg: Springer; 1983.
15. Kudo T. Occlusion of the circle of Willis with special reference to its developmental process. No To Shinkei. 1966;18:889–96.
16. Kudo T. Spontaneous occlusion of the circle of Willis. A disease apparently confined to Japanese. Neurology. 1968;18:485–96.
17. Handa H, Tani E, Kajikawa H, Sato K, Yamashita J. Autopsy case of the so-called cerebral arterial rete in an adult. No To Shinkei. 1969;21:181–91.
18. Klaus E, Farkova H, Urbanek K. Case of "Moyamoya disease" (Nishimoto-Takeuchi-Kudo syndrome). Fortschr Geb Rontgenstr Nuklearmed. 1970;113:603–8.
19. Nishimoto A, Takeuchi S. Abnormal cerebrovascular network related to the internal cartoid arteries. J Neurosurg. 1968;29:255–60.
20. Simon J, Sabouraud O, Guy G, Turpin J. A case of Nishimoto's disease. Apropos of a rare and bilateral disease of the internal carotid. Rev Neurol (Paris). 1968;119:376–83.
21. Urbanek K, Farkova H, Klaus E. Nishimoto-Takeuchi-Kudo disease: case report. J Neurol Neurosurg Psychiatry. 1970;33:671–3.
22. Vuia O, Alexianu M, Gabor S. Hypoplasia and obstruction of the circle of Willis in a case of atypical cerebral hemorrhage and its relationship to Nishimoto's disease. Neurology. 1970;20:361–7.
23. Suzuki J, Takaku A, Asahi M. Evaluation of a group of disorders showing an abnormal vascular network at the base of the brain with a high incidence among the Japanese. 2. Follow-up studies by cerebral angiography. No To Shinkei. 1966;18:897–908.
24. Suzuki J, Takaku A. Cerebrovascular "moyamoya" disease. Disease showing abnormal net-like vessels in base of brain. Arch Neurol. 1969;20:288–99.
25. Maki Y, Nakada Y. So-called "Chiri-Chiri" disease. J Chiba Medical Soc. 1976;43:667.
26. The Research Committee on Spontaneous Occlusion of the Circle of Willis ("Moyamoya Disease") (1979) The diagnostic guide of the "spontaneous occlusion of the circle of Willis".

Annual Report 1978 of The Research Committee on Spontaneous Occlusion of the Circle of Willis (“Moyamoya Disease”) of the Ministry of Health and Welfare, Japan.
27. Kitamura K (1988) Revision of the diagnostic guide of the spontaneous occlusion of the circle of Willis. Annual Report 1987 of the Research Committee on the Pathology and Treatment of Spontaneous Occlusion of the Circle of Willis (“Moyamoya Disease”) of the Ministry of Health and Welfare, Japan.
28. Ikezaki K, Fukui M. Definition (guidelines). In: Moyamoya disease. Rolling Meadows: American Association of Neurological Surgeons; 2001.
29. The Research Committee on Spontaneous Occlusion of the Circle of Willis (“Moyamoya Disease”) (1996) The diagnostic guideline of the “spontaneous occlusion of the circle of Willis” (new diagnostic criteria). Annual Report 1995 of the Research Committee on Spontaneous Occlusion of the Circle of Willis (“Moyamoya Disease”) of the Ministry of Health and Welfare, Japan.
30. Fukui M. Guidelines for the diagnosis and treatment of spontaneous occlusion of the circle of Willis (‘moyamoya’ disease). Research Committee on Spontaneous Occlusion of the Circle of Willis (Moyamoya disease) of the Ministry of Health and Welfare, Japan. Clin Neurol Neurosurg. 1997;99(Suppl 2):S238–40.
31. Hasuo K, Yasumori K, Yoshida K, Hirakata R, Kuroiwa T, Mizushima A, Matsushima T, Fukui M, Masuda K. Magnetic resonance imaging compared with computed tomography and angiography in moyamoya disease. Acta Radiol. 1990;31:191–5.
32. Houkin K, Aoki T, Takahashi A, Abe H. Diagnosis of moyamoya disease with magnetic resonance angiography. Stroke. 1994;25:2159–64.
33. Yamada I, Matsushima Y, Suzuki S. Moyamoya disease: diagnosis with three-dimensional time-of-flight MR angiography. Radiology. 1992;184:773–8.
34. Houkin K, Kuroda S, Ishikawa T, Abe H. Neovascularization (angiogenesis) after revascularization in moyamoya disease. Which technique is most useful for moyamoya disease? Acta Neurochir. 2000;142:269–76.
35. Houkin K, Nakayama N, Kuroda S, Ishikawa T, Nonaka T. How does angiogenesis develop in pediatric moyamoya disease after surgery? A prospective study with MR angiography. Childs Nerv Syst. 2004;20:734–41.
36. Houkin K, Nakayama N, Kuroda S, Nonaka T, Shonai T, Yoshimoto T. Novel magnetic resonance angiography stage grading for moyamoya disease. Cerebrovasc Dis. 2005;20:347–54.
37. Kuroda S, Ishikawa T, Houkin K, Nanba R, Hokari M, Iwasaki Y. Incidence and clinical features of disease progression in adult moyamoya disease. Stroke. 2005;36:2148–53.
38. Smith ER, Scott RM. Progression of disease in unilateral moyamoya syndrome. Neurosurg Focus. 2008;24:E17.
39. Yoon HK, Shin HJ, Lee M, Byun HS, Na DG, Han BK. MR angiography of moyamoya disease before and after encephaloduroarteriosynangiosis. AJR Am J Roentgenol. 2000;174:195–200.
40. Research Committee on intractable diseases of the Ministry of Health LaWJ. Recommendations for the management og moyamoya disease. A statement from Research Committee on Spontaneous Occlusion of the Circle of Willis (Moyamoya disease) surgery for cerebral. Stroke. 2009;37:321–37.
41. The Research Committee on the Pathology and Treatment of Spontaneous Occlusion of the Circle of Willis. Guidelines for diagnosis and treatment of moyamoya disease (spontaneous occlusion of the circle of Willis). Neurol Med Chir (Tokyo). 2012;52:245–66.
42. Tominaga T, Suzuki N, Miyamoto S, Koizumi A, Kuroda S, Takahashi JC, Fujimura M, Houkin K. Recommendations for the management of moyamoya disease: a statement from Research Committee on Spontaneous Occlusion of the Circle of Willis (Moyamoya disease) [2nd edition]. Surg Cerebr Stroke. 2018;46:1–24.
43. Choi HY, Lee JE, Jung YH, Cho HJ, Kim DJ, Heo JH. Progression of isolated middle cerebral artery stenosis into moyamoya disease. Neurology. 2007;68:954.

44. Inoue T, Murakami N, Sakadume S, Kido Y, Kikuchi A, Ichinoi N, Suzuki K, Kure S, Sakuta R. Differing phenotypes of Moyamoya disease in a familial case involving heterozygous c.14429G > A variant in RNF213. Pediatr Int. 2015;57:798–801.
45. Mineharu Y, Takagi Y, Takahashi JC, Hashikata H, Liu W, Hitomi T, Kobayashi H, Koizumi A, Miyamoto S. Rapid progression of unilateral moyamoya disease in a patient with a family history and an RNF213 risk variant. Cerebrovasc Dis. 2013;36:155–7.
46. Miyawaki S, Imai H, Shimizu M, Yagi S, Ono H, Mukasa A, Nakatomi H, Shimizu T, Saito N. Genetic variant RNF213 c.14576G>A in various phenotypes of intracranial major artery stenosis/occlusion. Stroke. 2013;44:2894–7.