Case Report

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Spontaneous Subdural Hemorrhage in a Patient with Marfan Syndrome: Case Report and Literature Review

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Background: Marfan syndrome is autosomal dominant with ocular, cardiovascular, and musculoskeletal manifestations. It is caused by mutation that encodes the fibrin gene on chromosome 15.
Case Presentation: A 54-year-old man presented to our center with a sudden onset of severe headache followed by a gradual loss of consciousness. His medical history was significant for Marfan syndrome. We found a spontaneous subdural hematoma in the left frontoparietotemporal area. The patient underwent emergency surgical intervention consisting of excision of the acute lesion and decompressive craniectomy in the left frontoparietal-temporal area. A postoperative brain computed tomography scan was conducted, which revealed a reduced midline shift and a subdural hematoma drainage. Laboratory investigations and intensive treatment were continued; however, the patient's clinical condition deteriorated progressively.
Conclusion: This case underscores the rare occurrence of spontaneous acute subdural hematoma in the setting of Marfan syndrome, which calls for prompt recognition and surgical intervention.

Keywords: *Marfan Syndrome; Acute Subdural Hematoma; Neurological Complications; Surgical Intervention; Multidisciplinary Approach*

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INTRODUCTION

Marfan syndrome is an autosomal dominant connective tissue disorder caused by mutations in the FBN1 gene located on chromosome 15. These mutations result in abnormalities in fibrillin-1, a crucial component of the elastic fibers that support the structural integrity of connective tissue. The musculoskeletal, cardiovascular, and ocular systems are the primary targets of the disorder, but its cerebrovascular consequences are increasingly recognized. Fibrillin-1 dysfunction leads to aneurysms, arterial dissections, and other vascular abnormalities, which can lead to ischemia or hemorrhage due to its role in preserving vascular elasticity [1].

Subdural hematomas (SDHs) can spontaneously happen on their own. However, it has also been documented in people with vascular anomalies. For instance, one study found 129 cases of Marfan syndrome and 826 patients associated with symptomatic intracranial aneurysms [2]. The first reported case was that of Gabrielle, a 5-year-old girl who displayed the classic symptoms of Marfan syndrome. Antoine Bernard-Jean Marfan (1858–1942) first defined the condition [3]. A study found, in the databases, that the SNP gene (rs112287730), indicated as a genetic variant, has an estimated frequency of 0.1 per 20,000 in Russia [4]. Marfan syndrome was also found to have a prevalence of 6.5 per 100,000 in 2014 and an annual incidence of 0.19 per 100,000. An annual increase of 1.03% (95% CI: 1.02–1.04; $p < 0.001$) was observed

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[5]. In this case study, the aim of this rare manifestation of spontaneous SDH in a patient with Marfan syndrome is examined, along with the diagnostic and therapeutic implications.

CASE REPORT

A 54-year-old man was brought to the emergency department by his wife after experiencing a sudden, severe headache and gradual loss of consciousness. His medical history included an aortic root aneurysm, aortic regurgitation (grade 2–3), mitral regurgitation (grade 3), a thoracic aortic aneurysm, prosthetic aortic and mitral valves, hypertension (stage 3, controlled), atrial fibrillation, a left renal cyst, and gallbladder polyposis. Urgent imaging was recommended, and owing to his history and Marfanoid features, a cerebral infarction was suspected. He was therefore admitted to our center, Moscow City Clinical Hospital No. 68, Demikhova V.P. The magnetic resonance imaging showed cerebral compression due to a hypertensive subdural hematoma, with no trauma to the left hemisphere. Computed tomography (CT) revealed parenchymal hemorrhage in the dorsal aspect of the left hippocampus, bordering the occipital lobe, with mild perifocal edema. We diagnosed the patient with Marfan syndrome and a spontaneous subdural hematoma and performed a decompressive craniotomy in the left frontoparietotemporal area. After excision of the acute lesions, a 120 cm³ subdural hematoma was recovered in the left hemisphere

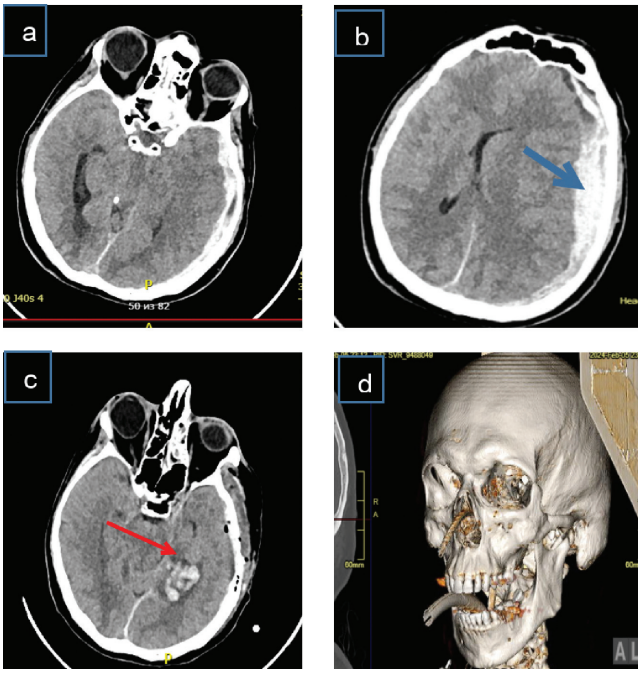

Figure 1 Identification of a spontaneous subdural hematoma in the left frontoparietotemporal area. (**a**) Preoperatory image of a cerebral herniation with signs of parenchymal hemorrhage and vascular edema. (**b**) The blue arrow points to a significant acute subdural hematoma. (**c**) The red arrow indicates an area of cerebral ischemia. (**d**) A three-dimensional (3D) reconstruction of the patient's skull.

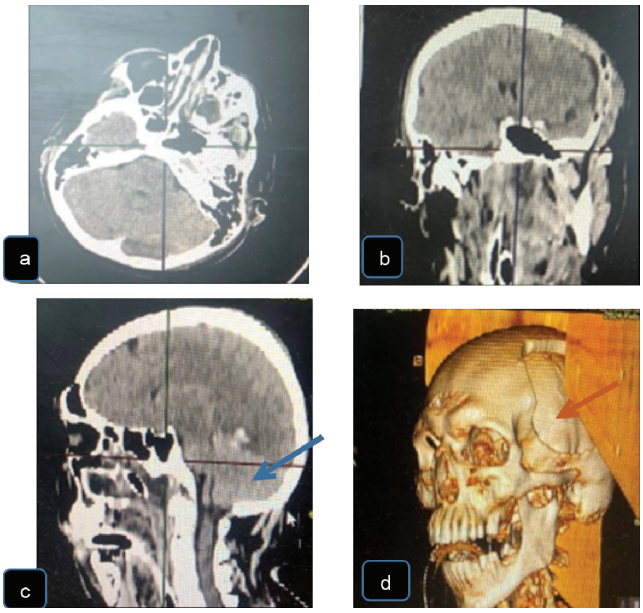

Figure 2 Postoperative images of the patients skull. (**a**) Postoperative, axial CT scan showing remnants of acute subdural hematoma and mild narrowing of the left hemisphere. (**b**) Coronal image with decreased transverse cerebral dislocation. (**c**) The blue arrow points towards the formation of a parenchymal hematoma in the left posterior occipital region. (**d**) A 3D reconstructive image with the red arrow pointing towards a postoperative left temporal decompressive craniotomy.

(Figures 1 and 2). Postoperative follow-up in the intensive care unit (ICU), the outcomes, and therapeutic intervention were provided, such as cephalosporin that was changed to vancomycin after sepsis. The patient remained in a critical condition and required continuous respiratory support from the eighth day in the ICU; he presented with moderate edema and coma; therefore intensive treatment was continued. The appointment sheet was modified based on clinical and laboratory findings. A transfusion protocol was initiated, and anemia monitoring was started, with ferric iron exchange and pathogenesis treatment. Because of this patient's prosthetic valve and subarachnoid hemorrhage, the chosen heparin dose was verified by activated partial thromboplastin time every six hours to maintain continuous anticoagulant therapy. However, no improvement was observed. We recommended active antibiotic therapy after the patient presented with fever during follow-up, which confirmed sepsis. No improvement was observed. The patient moved from a moderate to a severe coma with a Glasgow Coma Score of 5 at the time. Despite all concomitant efforts by our team, there was no improvement, and the patient died from respiratory arrest (Figure 3).

Ethical Approval and Informed Consent

An ethical approval was granted at the Moscow City Clinical Hospital No. 68, Demikhova V.P. in Moscow, Russia (Ref. number 1857, dated 02.07.1992, No.

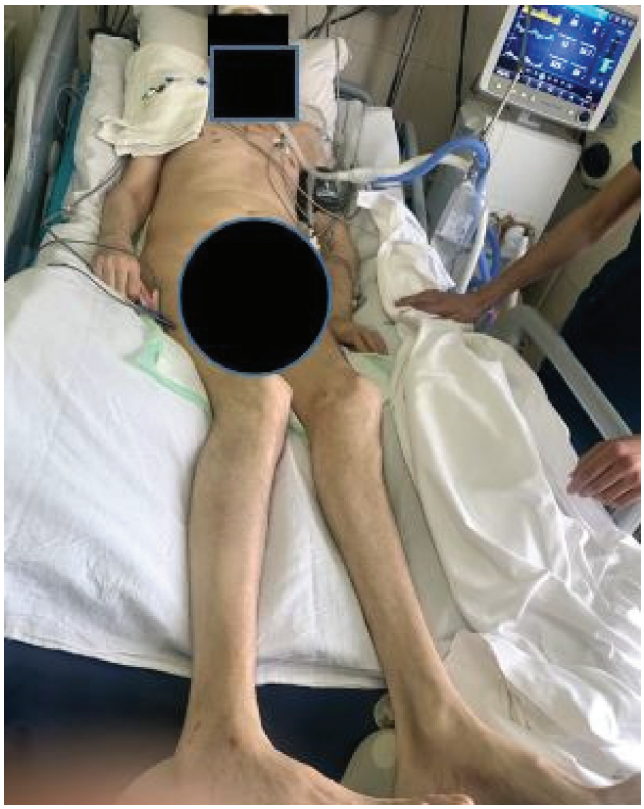

Figure 3 Postoperative image of the patient in the ICU where he was under mechanical ventilation and periodic monitoring.

2300-1; amended on 06.11.2021). The ethical guidelines outlined in Good Clinical Practice in the Declaration of Helsinki of the World Medical Association were strictly followed, ensuring that each patient provided informed consent before participating in the research.

DISCUSSION

Marfan syndrome is an autosomal dominant genetic disorder characterized by systemic manifestations that affect various organ systems, including the ocular, cardiovascular, and musculoskeletal systems. The condition arises from mutations on the FBN1 gene on chromosome 15, which leads to structural abnormalities in connective tissues, especially elastic fibers. Consequently, individuals with Marfan syndrome commonly present with clinical features such as tall stature, arachnodactyly (elongated fingers), chest deformities, joint hypermobility, and ocular abnormalities, including lens subluxation (Figure 4). Acute SDH is a neurosurgical emergency involving the accumulation of blood between the dura mater and the arachnoid matter. Although typically caused by traumatic brain injury, it may also occur spontaneously, particularly in individuals with vascular abnormalities or coagulopathies. Spontaneous SDH is often characterized by severe headache, altered levels of consciousness, focal neurological deficits, or signs of increased intracranial pressure [1,5–7].

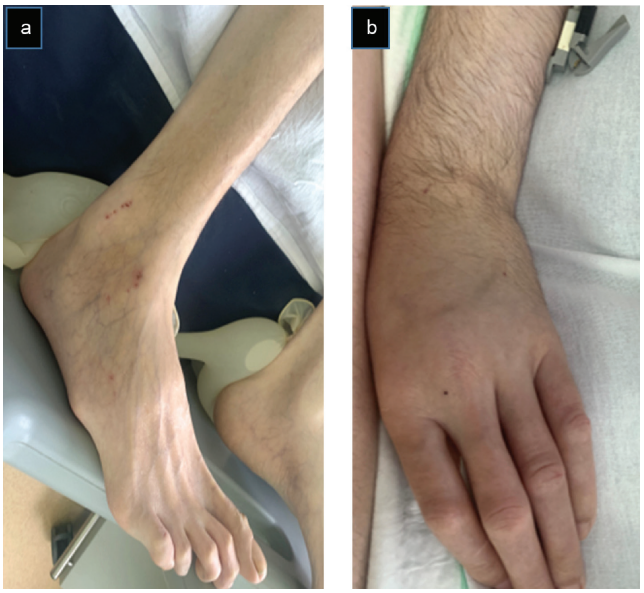

Figure 4 Characteristic features of a patient with Marfan Syndrome. Arachnodactyly or (**a**) lengthy toes and (**b**) fingers of the patient.

A retrospective case series study from 1992 in the Dominican Republic examined five patients diagnosed with Marfan syndrome. Only individuals aged 10 to 27 years were officially reported on the island at the Pediatric Medical Genetics Unit of the Robert Reid Hospital in Santo Domingo City. The patients exhibited classic Marfanoid characteristics, such as musculoskeletal abnormalities, lens subluxation, aortic root dilation, and dilatation of vessels for the fibrillin gene, holosystolic mitral valve prolapse, left ventricular hypertrophy in two instances, and systolic clicks in two instances [6].

Hemodynamic factors are critical in the management of aortic disease in Marfan syndrome. Aortic diameters exceeding 5.0 cm are strongly associated with a heightened risk of dissection or rupture. The use of β-adrenergic blockers has been advocated since 1971 to mitigate hemodynamic stress on the aorta, particularly in cases of malignant hypertension [8,9].

Spontaneous SDH associated with severe hypertension is considered an indicator of end-organ damage in hypertensive emergencies. In such cases, treatment protocols include administering up to four doses of 20 mg intravenous labetalol every 10–15 minutes to reduce blood pressure from 255/130 mmHg to 180/113 mmHg while achieving a target heart rate of approximately 61 beats per minute. Additional interventions may include 1 g of levetiracetam for seizure prophylaxis and 40 μg of desmopressin intravenously to reduce the risk of hemorrhage [10–12].

Patients with spontaneous acute SDH, symptoms of intracranial hypertension, and cranial nerve dysfunction, such as abducens nerve palsy, often require emergency hemicraniectomy. Pathological samples may also be analyzed to detect atypical cellular changes, such as

hematopoietic neoplasms, although no such pathological findings are typically observed in Marfan syndrome. The underlying cause in such cases is likely attributable to arterial wall weakness associated with the condition [13–16].

However, in a study of 56 patients with acute subdural hematomas, the incidence of low density in the hematoma according to the initial CT scan and the spontaneous resolution of up to 32% were reported. The majority of the patients did not even undergo surgery because of their poor prognosis, and some studies found significant differences in the use of antiplatelets [7,17].

CONCLUSION

This case underscores the rare occurrence of spontaneous acute SDH in the setting of Marfan syndrome, emphasizing the need for prompt recognition and surgical intervention. The patient outcome was unfavorable, highlighting the challenges associated with managing neurological complications.

Marfan syndrome is considered a volatile syndrome. The loss of elasticity and other cardiovascular and arterial changes brought on by its fibrillin gene abnormality are so severe that they are linked to the aneurysms that result in these spontaneous SDHs. Therefore, any other type of emergency was expected in the patient's life record, but not this type of SDH. In other words, he was treated as professionally as possible using all available equipment and medical services. From a medical perspective, there are questions, but the management of Marfan syndrome should be done in conjunction with the neurological, emergency, and neurosurgery services without neglecting psychiatric consultation.

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Ethics Statement

(1) All the authors mentioned in the manuscript have agreed to authorship, read and approved the manuscript, and given consent for submission and subsequent publication of the manuscript.
(2) The authors declare that they have read and abided by the JEVTM statement of ethical standards including rules of informed consent and ethical committee approval as stated in the article.

Conflicts of Interest

The authors declare that they have no conflicts of interest.

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Author Contributions

Conceptualization, MP, DRC, ES, and DES; methodology, DES and GS; software, DES and GC; validation, IB, GS, and BC; formal analysis, GS and GF; investigation, DES; resources, MB and ES; data curation, GS; writing—original draft preparation, DES; writing—review and editing, GS and DES; visualization, AR and NB; supervision, GC, EC, IB, GS, and BC.

Availability of Data and Materials

The manuscript has been read and approved by all authors, and availability of data and materials will be facilitated for reasonable requirements.

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