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Case Report

Jaffe-Campanacci syndrome presenting as young stroke

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ABSTRACT

Jaffe-Campanacci syndrome is a rare clinical entity characterized by multiple non-ossifying fibromas, café-au-lait macules, and various extraskeletal manifestations, which have a debated association with neurofibromatosis type 1. Fewer than 30 cases have been reported in the literature, making the diagnosis and characterization of this disease challenging. We report a rare case of a 16-year-old girl who presented with an acute onset of headache, left-sided weakness, slurring of speech, and sudden loss of consciousness. Neuroimaging revealed a right gangliocapsular haemorrhage with a midline shift, consistent with a haemorrhagic stroke. MRI confirmed a large subacute intracerebral haemorrhage, and genetic analysis revealed positivity for the NF1 gene mutation. This case is noteworthy because haemorrhagic stroke is an extremely uncommon presentation of Jaffe-Campanacci syndrome and highlights the possible association between vascular abnormalities and the syndrome. Early identification of the characteristic skeletal and cutaneous manifestations is essential for the correct diagnosis and correct medical management. Reporting such unusual presentations of Jaffe Campanacci syndrome contributes to a better understanding of the disease spectrum and its relationship with neurofibromatosis type 1.

Keywords: Jaffe-Campanacci syndrome, Neurofibromatosis 1, Intracranial hemorrhages, Non-ossifying fibroma, Café-au-Lait spots

INTRODUCTION

Jaffe-Campanacci syndrome is characterized by multiple non-ossifying fibromas of the long bones, mandibular giant cell lesions, and café-au-lait macules.[1] This syndrome is most commonly identified in children and adolescents with recurrent fractures. Radiological findings typically show multiple lytic lesions in the metaphyseal region of the long bones.[2] It also includes other extraskeletal congenital abnormalities, such as intellectual disability, ocular deformities, hypogonadism, and cryptorchidism.[3]

Many Jaffe-Campanacci syndrome cases lack the presentation of slit lamp (Lisch nodules), pigmented patches known as café au lait spots, and family evaluation of the disease.[4] Currently, there are no accepted diagnostic criteria for Jaffe-Campanacci syndrome, and one of the previously published case reports included NF1 genetic testing.[3] Although a relationship with neurofibromatosis type 1 has been suggested, there is still no proof that Jaffe–Campanacci syndrome is a subtype or variant of neurofibromatosis type 1.[5] Fewer than 30 cases have been reported in the literature, highlighting their rarity.[1] Due to its rare and variable clinical presentation, Jaffe-Campanacci syndrome mimics other skeletal dysplasias or metabolic bone disorders. Here, we report a case of Jaffe-

Campanacci syndrome presenting with haemorrhagic stroke.

CASE REPORT

A 16-year-old girl with no known comorbidities was brought in with complaints of headache for the past 2 days and weakness of the left upper and lower limbs for 6 h. The patient also had a history of slurred speech and sudden loss of consciousness. The patient had a history of not attaining menarche. The patient had a limping gait since the age of 10. The patient had no history of previous trauma. On examination, the patient was drowsy but oriented to place and time. She had multiple café-au-lait spots (Figure 1B) in various areas of the body and alopecia (Figure 1C) was present. The patient also had microphthalmia (Figure 1A) and solitary orbital neurofibroma (Figure 2).

Figure 1: (A) Microphthalmos in the patient; (B) depicts the presence of café au lait spots in the neck region; (C) shows the alopecia in the patient.

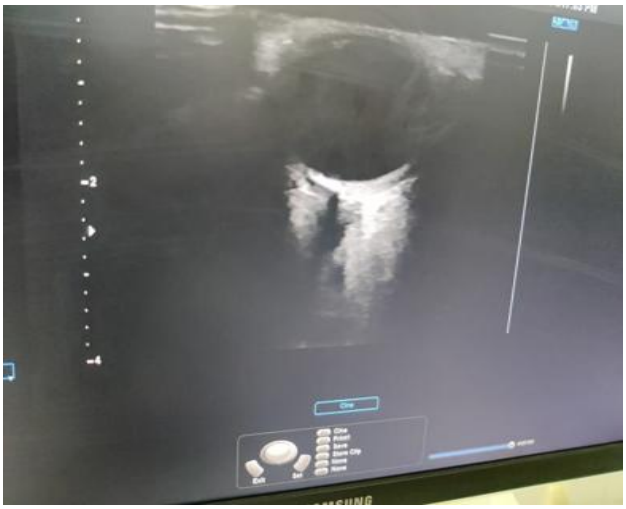

Figure 2: USG depicts a solitary orbital neurofibroma.

The patient also had a non-ossifying fibroma at the lower end of the tibia (Figure 4). The results of Other general examinations were normal. The patient did not exhibit axillary freckling or shagreen patches. The patient's vital signs were normal; however, the blood pressure was slightly elevated. On central nervous system examination (Table 2), higher mental functions and cranial nerve examination were found to be normal. The Spino-motor bulk was equal on both sides, and the sensory system was normal. However, power was decreased on the left side, and the plantar extensor was decreased on the left side. The Cerebellar signs were normal, and the spine and cranium appeared normal. Signs of meningeal irritation were absent. 2D ECHO and chest radiography findings were normal. Non-contrast computed tomography (Figure 3) revealed a gangliocapsular bleed with a midline shift to the left.

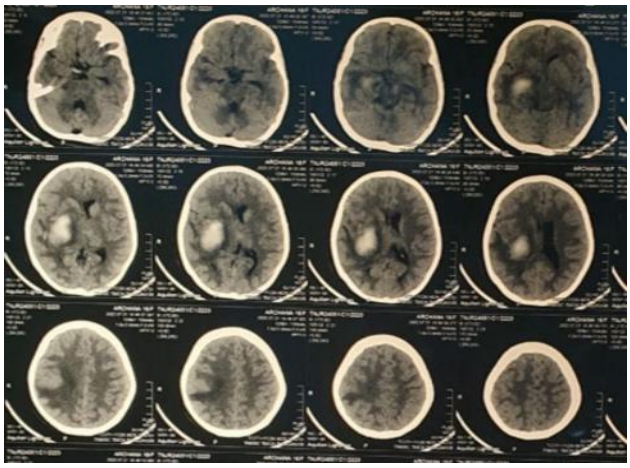

Figure 3: CT brain shows a gangliocapsular bleed which led to a 6 mm midline shift.

Table 1: Lab parameters.

Parameters	Values
Complete blood count	
RBC	3.41 million/µl
Hemoglobin	11.0 g/dl
HCT	30.4 %
MCV	84.6 fl
PLT	3 Lakh cells/mm^3
Random blood sugar	81 mg/dl
Renal function	
Urea	25 mg/dl
Creatinine	0.7 mg/dl
Liver function test	
SGOT	37 U/l
SGPT	32 U/l
Total bilirubin	0.9 mg/dl
Direct bilirubin	0.7 mg/dl
Indirect bilirubin	0.2 mg/dl
ALP	67 U/l
Total protein	6.1 g/dl
Albumin	3.6 g/dl
HBsAg	Negative
Anti-HCV	Negative
VCTC	Non-reactive

Table 2: Clinical examinations.

		Right	Left
Motor power	Tone	Normal	Normal
	Power UL	5/5	3/5
	Power LL	5/5	3/5
	Plantor	Flexor	Extensor
Sensory system	Spinothalamic sensation		
	Lateral spinothalamic tract -pain and temperature	Intact	Intact
	Anterior spinothalamic tract	Intact	Intact
	Crude touch	Intact	Intact
	Dorsalcolumn sensation		
	Position sense	Normal	Normal
	Joint sense	Normal	Normal
	Vibration sense	Normal	Normal
	Pressure sense	Normal	Normal
	Romberg's Sign	Negative	Negative
Cortical sensation	Two-point discrimination	Intact	Intact
	Tactile localization	Intact	Intact
	Graphesthesia	Intact	Intact
	Stereognosis	Intact	Intact
Cerebellar functions	Nystagmus	No nystagmus	No nystagmus
	Tremor	No tremor	No tremor
	Finger nose test	No past pointing	No past pointing
	Finger-finger nose test	No past pointing	No past pointing

A complete blood count and coagulation profile were performed, and no significant abnormalities were observed in either test. Peripheral smears showed normal cells with no other abnormalities. Liver and renal function tests were normal (Table 1).

Urinary Bence Jones Protein was absent in the patient. The results for ANA and APLA were negative. An MRI BRAIN was performed, which showed a subacute large haemorrhage in the right GC region with a midline shift of 6 mm. Finally, on suspicion, gene sequencing was done, which revealed a positive result for the NF1 gene.

On conservative management the patient symptoms improved however minimal weakness was present and was subsequently discharged advising to continue limb physiotherapy. On subsequent visits the patient recovered completely with no neurological deficits.

DISCUSSION

The present case of Jaffe-Campanacci Syndrome (JCS) highlights several critical aspects of this rare disorder, especially its overlap with neurofibromatosis type 1 (NF1) and its implications for clinical management. Clinical Features and Diagnosis: Jaffe-Campanacci Syndrome is characterized by multiple non-ossifying fibromas (NOFs), café-au-lait spots, and variable extraskeletal abnormalities, such as ocular and cardiovascular malformations.[1] In this case, the patient presented with typical features, such as café-au-lait spots, solitary neurofibroma, short stature, and microphthalmia. The absence of axillary freckling and Lisch nodules, typically associated with NF1, highlights the diagnostic complexity and overlap between Jaffe-Campanacci and NF1.

A similar observation was made by Ahmed et al who described multiple café-au-lait spots and a distal femur fracture in a 13-year-old boy. He also had hypogonadism and short stature; however, no Lisch nodules or cutaneous neurofibromas were found, thus emphasizing that JCS is a separate entity from NF1; however, there is no clear demarcation between them.[2] Observations made by him provided a similar presentation of Jaffe-Campanacci syndrome as in our case, but our case presented with a hemorrhagic stroke, which is a rare presentation.

Hemorrhagic stroke in JCS

The most unusual and significant feature of this case is the presentation of hemorrhagic stroke in a young individual with JCS. Although stroke is a major cause of morbidity and mortality worldwide, it is rarely associated with Jaffe-Campanacci Syndrome. In this case, the patient suffered from a gangliocapsular bleed, as revealed by neuromaging. This finding underscores the potential for serious vascular complications in patients with JCS, which may be an underreported aspect of this syndrome. The possibility of vascular anomalies contributing to severe events in JCS should be further explored, as the presentation of a stroke is quite unusual. Previous reports have focused on its skeletal and dermatological manifestations, and little is known about its vascular complications. This case emphasizes the importance of vascular complications, which might be presenting in this syndrome, and provides insight of stroke in patients, especially presenting with a headache

Management and prognosis

The management of Jaffe-Campanacci syndrome is conservative, which focuses on preventing pathological fractures due to non-ossifying fibromas.[6] In the case of hemorrhagic stroke, immediate medical intervention is required. Recognition of this rare syndrome is crucial to avoid misdiagnosis and facilitate appropriate multidisciplinary management involving orthopedic surgeons, radiologists, pathologists, geneticists, and dermatologists. Furthermore, reporting additional cases will contribute to the existing literature and may help clarify the uncertain relationship between JCS and NF1. Given the rarity of JCS and its overlap with NF1, genetic counselling plays a crucial role in managing patient expectations and informing them of potential complications.

CONCLUSION

In conclusion, Jaffe-Campanacci syndrome is an uncommon disorder characterized by multiple non-ossifying fibromas associated with café-au-lait macules and systemic manifestations. Owing to its overlap with neurofibromatosis type 1, careful clinical, radiological, and histopathological evaluation is essential for an accurate diagnosis. Early identification of the syndrome helps prevent complications, such as pathological fractures, and enables timely, multidisciplinary management. Reporting rare cases contributes to a better understanding of the disease spectrum, improves diagnostic awareness, and aids in establishing optimal management strategies for affected patients.

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